Strategies for Medical Care for Persons with Intellectual Disabilities (“ID”)

Handbook for Providers
Caring for Persons in the VSHP SelectCommunity Program
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About This Guide

This resource guide is designed for providers serving persons with ID in the VSHP SelectCommunity Program. Most clinicians have had little training or exposure to individuals with ID. They are supported by federally funded state services through the Division of Developmental Disabilities (DIDS), a system which is often confusing for PCPs. This handbook is not intended to prescribe practice. Every provider must determine best interventions based on the unique needs of individual patients.

Information is not meant to be exhaustive or even complete but is designed to provide specific information about care of people with ID to help the provider integrate information into common generic medical practice. It is also meant to help the provider understand care issues for people in non-independent (i.e. residential placement) settings of Tennessee’s care system for adults with ID. These systems include federally funded systems of Intermediate Care Facilities for the Mentally Retarded (ICF/MR), and HCBS Waiver programs supported by nurse case managers in the VSHP SelectCommunity Program.

This handbook is a resource to help providers quickly access resources and information which may facilitate efficient treatment and integration into this new program that links medical care (through VSHP) with the overall programs (federally funded) provided by Tennessee. It is strongly recognized that PCPs may not have extensive information about the ID population, who often are medically complex, have communication deficits, may have behavior variances, and often have low incidence or rare diagnoses (e.g. Lennox-Gastaut seizures, Fragile X syndrome, or DS). The PCP is encouraged to consult and interact with clinicians in these or other programs (R.N., nurse case manager for VSHP, therapists and clinicians working with DIDS, and through HCBS Waiver programs). In addition, this handbook provides resources to assist practitioners in remaining up to date with current, best practices.
Introduction

Most doctors have a few patients with ID in their practice. These patients may present difficulties with care based on issues beyond the control of the PCP. This includes lack of information about the patient (past medical history or current pertinent information), lack of exposure or experience with people with ID, lack of modification of procedures for persons with physical, behavioral and communication issues, or confusion about programs (residential, therapy, work, etc.) provided by “the State” (generally referred to as “Waiver programs”).

Recently, TennCare designated a single Managed Care Organization (MCO) to develop a person-centered system of medical care for persons with ID. The goal of this approach is to integrate TennCare/MCO health services with the HCBS, a federally funded program with TennCare as the “lead agency.” The program is administered through the DIDS with a delivery system of daily supports in a holistic way. It is a partnering with multiple agencies responsible for health care. This is not a stand-alone service, but a generic health care system modified to address the specific needs of people with ID. Although there are around 60,000 people with ID in Tennessee, only about 8-10,000 are served through state programming.

The plan is to provide individual-specific health care for this often misunderstood population by making relatively small adjustments to current health care systems, while pairing responsibilities with Waiver requirements for health supports. This is a new approach that integrates health care (necessary for optimum quality of life), with a service system (the Waiver), that provides supports for improving the lives of people with ID. These services include clinical services, (though not medical care) paid through federal programs and not private insurance.

"Good health" may be visualized in the broad sense, but is not well defined for each individual. This is even more of a problem for someone with ID where "health" is often much less understood or defined. In many instances, for some people, ID is synonymous with poor health and a short lifespan. In fact, many death certificates state the cause of death is “mental retardation,” “DS” or “CP.” These are never causes of death, although secondary conditions or associated problems may be. If someone dies at age 43 from "mental retardation," why not at age 3, or 6, or 28?

Recent national data collected by National Association of State Developmental Disabilities Services (NASDDS) demonstrates the complexity of caring for people with ID. Of people served, 27 percent are in the severe to profound range of ID. It is well understood that the majority of people in this functional range have an identifiable "organic" cause for their ID (often a genetic syndrome such as DS) which may be associated with treatable medical conditions. Unfortunately, many people are not diagnosed, their diagnosis may be "rare" or poorly understood, and most clinicians do not learn about these issues in their training.

The same study noted 24 percent of patients are completely nonverbal, and, since all ID is, by definition, a cognitive deficit, this includes communication. A person with ID is likely to lack the ability to relay past medical history to a clinician. Information about past medical history (illnesses, hospitalizations, results of tests, response to medication) is
the cornerstone of good medical treatment. Significant medical history is also often lost when people go to a new doctor without transferring records, or were transferred to and from residential agencies.

The leading medical issues in this study were dual diagnosis (someone with ID who also has mental health issues) at 27 percent and 29 percent with a seizure disorder. (The prevalence of seizures in the general population is around 1 percent). Other studies note the high rate of problems with the Gastro Intestinal (G.I) system (mainly GERD, and especially constipation), osteoporosis (many risk factors in this population), sensory issues (vision and hearing), and skin issues (although this has not been problematic with people who are supported by DIDS). In addition, “diagnosis specific” issues, especially with genetic syndromes, often determine medical needs.

A recent series of focus groups, conducted to understand health promotion practices and what helps or hinders people with disabilities in maintaining healthy lives, found that participants described health and wellness as:

- Being able to function and having the chance to do what you want to do
- Being independent, having self determination regarding choices and opportunities
- Having physical and emotional state of well-being
- Not being held back by pain

However, persons in the group emphasized that having a disability did not preclude health and wellness, but understood that health and wellness exists within the experience of disability, including the social and physical barriers, as well as the attitude of others. They also noted that disabilities may pose additional challenges to health and wellness, including secondary conditions, as well as medical challenges unrelated to the disability, just as experienced by the general population. This emphasizes the often overlooked principle that health is not a fragmented part of life, but needs to be integrated within a person's entire life, encompassing the emotional, physical, social, intellectual, and spiritual areas of life. Too often for people with ID and/or health issues are reviewed as some separate component “over there” and viewed as something “not really part of life.” This is often reflected by significant lack of integration of health into daily life.

The 8 Principles of Health Care in Adults with ID are:
(from Tyler, C. and Baker, S. Intellectual Disabilities at Your Fingertips)

1. Adults with ID have unmet health needs
2. Adults with certain syndromes have additional health care needs
3. Understanding the cause of developmental disability may help with health care planning and with medical diagnosis
4. Physical and mental health problems often coexist and contribute to each other
5. Behavior problems may be due to underlying medical conditions
6. Health care providers tend to underestimate decision-making capacity
7. Some conditions occur more frequently in adults with ID
8. All adults with ID need a PCP

Clinical services for persons in the Waiver programs are different from those provided by traditional insurance but are often critical to the well-being of this unique population. These services often enhance and support medical care and may be helpful to PCPs.
Throughout this handbook, these services will be delineated with ideas on how the PCP may use them to improve health, and solve clinical problems. In general, these services are not for acute care issues (which fall under traditional MCO modes of care), but are used for maintenance and prevention to preserve health and quality of life.

Because of the uniqueness, complexity and low incidence of many health issues, each person will have a specially-trained nurse case manager who will be the glue to put these plans into action. The nurse case manager will, first and foremost, support the health of the person by both helping with specific issues and being a medical advocate. The nurse case manager will also be of help to the PCP to understand the person, special topics related to the person's ID, and to help with understanding and navigating the care system (HBCS Waiver) in which the person is enrolled.

All this will be accomplished by: 1) Integrating health care into the person's life using the principles of the Patient-Centered Medical Home model; 2) Developing partnerships between DIDS programs and professionals with clinical providers (TennCare, MCOs, doctors) to support the health of persons served by this system; 3) Evaluating or monitoring the effectiveness of the program and outcomes achieved.

**Definition**

Intellectual Disability ("ID") is a diagnostic term denoting deficits in cognitive functioning. Over the past few decades the term "mental retardation" has acquired pejorative and shameful connotations, and currently most professionals use the term ID.

Three criteria must be met for the diagnosis: 1) an IQ (on formal testing) below 70; 2) significant limitations in two or more areas of adaptive function (measured by an adaptive rating scale, i.e., communication, self-help skills, interpersonal deficits); and 3) occurring before the age of 18.

An identifiable etiology may be important in considering treatment and may be prenatal, perinatal, or postnatal. As a general rule, the lower the IQ, the more likely it is to identify an etiology.

Unique disorders and problems beyond those found in typical populations fall into three main areas: cognitive, motor, and neurobehavioral. Individuals may have difficulties in only one area, or in all three. Understanding these areas may be easier when considering the principal diagnoses of ID, CP and autism.
Additionally, most people with an ID have other disorders and problems not visually obvious. This may be understood by considering the overall diagnosis as “the tip of the iceberg” where underlying and associated problems or complications may not be appreciated.

Taking care of someone with ID is similar to taking care of the typical population. However, some modifications may be necessary: Care may require extra time, modifications for communication, adaptations for physical limitations, understanding and compensating for the environment in which they live, understanding social support systems, and seeking specific information about little seen (mainly genetic) diagnoses. It is important to make reasonable adjustments to ensure each person has the same opportunity for health, whether they have an ID or not. Everyone has a right to expect and receive appropriate health care.

Issues of consent often arise and capacity is specific to a particular decision and time. You must assume the person has capacity if there is no legal evidence otherwise. In addition, people may have partial capacity (e.g., to choose whether to be treated with a shot or pill) but decisions for more complicated issues (e.g., surgery) may be made by someone legally appointed. Medical decisions may only be made by the patient or his/her legal representative.
Overview of Program
Although people with ID have the same acute medical problems as the rest of the population, the majority of their problems relate to chronic issues which began early in life, often at birth. People with ID, by definition, have an abnormal brain which may contribute to difficulties in behavior (originating in the brain), and problems with physical status (since muscles are innervated by nerves which come from the brain), and may affect both use and appearance.

Childhood Onset Brain Disorders

Complications
  Cognitive Dysfunction
  Neuromotor Dysfunction
  Seizures
  Abnormal Behavior

Consequences

Individuals with ID, over the span of their lifetime, learn to adapt to these disabilities and need help to maximize their abilities through appropriate clinical treatment. These problems are often viewed by the general population as chronic "non-curables" problems. Because of their lack of communicative abilities (either because they don't speak or don't express themselves well), many people with ID also pose problems in dealing with the clinical world in defining and treating their medical problems. In addition, there are many misconceptions about people with ID relating to their ability to enjoy life, their ability to have emotions, their lifespan, and in particular, concern about their behavior or what their behaviors mean.

Clinical services for persons in the Waiver program are different from those provided by traditional insurance but are often critical to the well-being of this unique population. These services often enhance and support medical care and may be helpful to PCPs. Throughout this handbook, these services will be delineated with ideas of how the PCP may use them to improve health and solve clinical problems. In general, these services are not for acute care issues (which fall under traditional MCO modes of care), but are used for maintenance and prevention to preserve health and quality of life.

People with ID generally have disabilities beyond those seen in the general population which fall into three distinct areas, often not considered in traditional clinical settings. These include: 1) cognitive abilities, 2) physical abilities, and 3) neurobehavioral functions. An example of the impact of cognitive deficits ID on health and well-being would be someone who is unable to talk, and would have difficulty explaining symptoms to a physician. In addition, they would have associated problems related to following through with health care recommendations.
In the physical realm, many have difficulties with normal positioning or movement which may impact other activities such as eating, or gross motor activities such as sitting and walking, and fine motor activities such as hand use (i.e., writing, buttoning, zipping). Most people are familiar with traditional therapies when a person has an injury. So, for instance, if someone breaks a leg, they may need short-term physical therapy to learn to walk again after healing. If someone has a stroke, they may receive speech therapy to help them speak, and occupational therapy to help them rehabilitate back to their prior abilities for normal functions of daily living. For the most part, these therapies are short-term, and when people complete their goals or stop making progress, these services are discontinued.

In people with ID and developmental disabilities, the goals of treatment are often different and relate to maintenance, promoting function, or showing gains in new abilities. Therapies may need to be continuous rather than time-limited. For example, someone who is developing or has scoliosis, or contractures of their joints might benefit from ongoing therapy and positioning to prevent progression of these problems which could result in significant medical issues such as respiratory problems, fractures or pain.

In addition, some therapies allow continuation of routine activities of daily living which may contribute to overall health. An example of this would be someone who has difficulty self-feeding or chewing, and problems maintaining an upright position for eating. Maintaining as much independence for eating, in a person such as this, would contribute not only to the well-being and self-image, but also to decreased health problems. Many people who have eating problems such as swallowing and chewing may benefit from development of a dining plan with specialized equipment and specialized procedures which allow them to continue feeding without the risk of choking and subsequent aspiration pneumonia.

Some individuals who do not have swallowing problems may benefit from therapies allowing them to use their hands for feeding themselves to maintain independence. In addition, sometimes positioning alone will reduce the possibility for choking, and contribute to decreased chronic constipation, which may lead to obstruction, surgical emergencies or death. These sorts of therapies require specialized training and specialized evaluations which often do not fit into routine short-term therapy people without these challenges may require.

Individuals with ID may need specialized nursing to integrate their medical treatments. As an example, some people in the Waiver program are fed by enteral feeding tubes. These tubes are not placed because people are in a terminal state, as most of the general population is familiar with. Enteral feeding tubes in this population are usually used for a reason, such as a swallowing problem or chronic pneumonia or because the individual is unable to eat safely by mouth. Most of these individuals with feeding tubes go on to live productive, happy lives, functioning at their level for many years after placement of the tube. However, having a tube requires nursing oversight for feeding regimens as well as awareness of potential medical problems and emergencies.

There are also many people with ID who have multiple and sometimes unusual combinations of medical problems. It is not uncommon to have patients who have seizures, GI problems (such as GE reflux disease and constipation), osteoporosis, respiratory problems, spasticity requiring medication, and many behavior problems requiring psychotropic medications. Oversight of these complicated, but medically
complex people, requires nursing oversight by individuals who understand these problems.

People with ID often have problems with behavior based on their lack of coping ability and the ability to control situations. These are often not related to an underlying psychiatric problem but are often a means of communication or expression of emotions. Within the DIDS system, there are professionals who utilize special formats for addressing these behaviors. This may help avoid inappropriate medication use or unnecessary hospitalizations. This service is often not present in other treatment sectors such as the mental health system and is often not required or utilized by the general population.

In addition, there may be other significant medical challenges which require integration of clinical support and care provided by clinicians and agencies serving people with ID. Some of these issues might be as simple as modifications to obtain routine preventive care which is available to the rest of us. Other challenges are a lack of consistently available medical history and information. Many people in this system have been to a variety of doctors, in a variety of settings, and/or with a lack of transfer of information. Often people with ID have unusual presentations of common problems because of their inability to either speak or express themselves. For instance, GE reflux (heartburn) is a very common finding in the general population who either treat themselves by going to the local drugstore, or who present themselves to a physician describing their problems after which they generally receive adequate treatment.

Some challenges in medical care for people with ID are related to difficulties in monitoring the effect and side effects of medications. For example, the side effects of many seizure medications are symptoms such as pain, dizziness, or nausea, which are difficult to observe externally in a nonverbal population. This requires other indirect methods for assessment. In addition, there are many people with very low incidence genetic diagnoses which may have as components unrecognized medical problems. This requires understanding of the entire person and integration of the genetic diagnoses which might provide a roadmap for medical care.

People with DS have very well-defined medical problems which are easily diagnosed and treated if they are anticipated. An example is hypothyroidism which occurs in a high percentage of people with DS and increases with age. This leads to obesity (not an expected finding for DS), decreased activity, and, in severe cases, bradycardia, which has been misinterpreted as a heart problem. Having the knowledge to look for this contributes to early diagnoses and treatment and prevents significant discomfort and complications.

There are many studies highlighting poorer health for many people with ID arising from:

- Undetected ill-health
- Occurrence of other coexisting health problems
- Barriers to access to health care services
- Their need for support in seeking health care
- Poor contact with primary health care services
- Few health care professionals having specific training in caring for people with ID
Medical problems for people with ID are the same as with the general population, but may have an unusual presentation: e.g., anemia as a presentation for reflux, constipation as a potentially life-threatening condition.

Physical, cognitive and/or behavioral problems may interfere with care:
- physical, e.g., difficult to examine a person with severe contractures
- cognitive, e.g., not able to communicate or understand treatment
- behavior interferes with compliance, e.g., uncooperative, disruptive

Medical records are often unavailable or inaccurate. Doctors are often working “blind” without adequate information, e.g., historical medical information lost or purged from record, medical history not communicated to doctor, person transferring from one doctor to another without a transfer of records, etc.

Confusion between Division of Mental Retardation Services (DMRS) and the medical world created by the fact that what each system regards as a medical record is not the same. What DMRS requires as medical documentation is not useful to a clinician for medical care.

There is a high percentage of rare and unusual medical conditions such as genetic syndromes, including behavioral phenotypes, with unique care needs, e.g., Angelman syndrome, William’s syndrome, Fragile X syndrome, Prader-Willi, Tuberous sclerosis, DS, etc.

There is often a high complexity of medical needs, such as persons with multiple, complex diagnoses, e.g., seizures, hypothyroidism, on psychotropic medications, constipation, reflux, etc. It is difficult to integrate care with different specialists who don’t communicate with each other. There is often no central responsible person, and care becomes fragmented, counter productive, and more expensive because it is not integrated. People are subjected to repeated testing, previously used medications that have adverse side effects, and proper medical care may even be delayed.

Existence of multiple diagnoses may be complicated by the care of each one, e.g., medication for reflux that interferes with seizure medications.

There is no specialty back up for people with ID, e.g., most doctors can take care of 80 percent to 90 percent of all medical issues for their patients and refer out the balance of the issues to specialists. There are few specialty doctors specifically trained to care for people with ID, and most are pediatricians.

Lack of adequate compensation for care:
- More complex conditions take longer to treat
- Special adaptations are often needed, e.g., examinations such as mammogram or CT-scan, physical exams, getting weights for people in wheelchairs, etc.
- Additional requirements of Waiver programs are a disincentive, e.g., yearly physical, paperwork, etc.

The health of many people with ID is quite complex. These individuals can have medical problems that cross over a number of medical specialties. The PCP and the medical home concept is one that provides integration and coordination of all specialists. The PCP does not need to have the same knowledge as the specialists, but needs to coordinate care to prevent specialties from working at odds with each other, e.g., many medications for behavior may cause increased seizure activity, and many seizure medications have behavioral side effects; psychotropics may cause constipation and other GI problems which may lead to behavior problems and increased psychotropics.
The Waiver system, through supports, acts as the “patient” by assuming or facilitating all the tasks a patient might do for himself/herself in creating a partnership with the PCP. This includes carrying out the patient’s responsibility in health care as partnered with the PCP. For the PCP, this means access to accurate information about concerns, response to treatment, side effects, etc. This also means that due to Waiver program regulations, the PCP has some assurance that recommendations will be followed. Appointments are more likely to be planned in advance with the proper information needed for treatment provided to the PCP. Since the systems will be working together to support optimum health, tasks not usually routine for the typical patient will be important for efficiency and good outcomes.

Completion of a yearly evaluation which involves a partnership between the patient and his/her “health supporters” and the doctor is important. This is not a typical component of a person’s health care in today’s medical environment. This is not limited to an “Annual Physical” but is a comprehensive medical evaluation with development of a treatment plan. The goal of the yearly evaluation is to ensure accurate diagnoses, ensure the person is getting needed specialty care, ensure the person is getting the appropriate medications, and ensure that everyone in the “system” has an accurate picture of the person’s needs in order to support health. This is a task the nurse case manager will be able to facilitate. The best quality of care is provided not in episodic, illness-oriented, complaint-based care, but through patient-centered, physician-guided, cost-effective, longitudinal care that encompasses and values both the art and science of medicine. (A manual for this procedure has been developed by DIDS.)

In addition to the medical home model, case management services have been effective in managing chronic conditions. Nurse case managers, employed by VSHP, will manage the health plan and medical needs of the person, and interface with DIDS systems to facilitate good health. The RN Case Manager will work with both systems in integrating health care.

Consults are available through regional DIDS clinical staff and can help with filling in “specialty gaps.” Such consults by an MD or a RN (especially experienced with the specific needs of this population) may be particularly useful when the PCP has questions, needs help or when there is a new diagnosis or change in condition. DIDS consultations may be of use to the PCP by providing suggestions on how to better serve people with ID in a community office setting, including how to assist the patient in the exam setting, simplifying documentation, and providing general support to the patient during the visit.

The Individual Support Plan (ISP) document is used as a plan or road map to life for persons supported by the Waiver program. This document is created by people supporting the patient (direct care staff, Waiver therapists, Independent Support Coordinator (ISC), advocate, family and/or conservator, or natural supports called the Circle of Support or COS), and has a section to focus on integration of health into the person’s life, and the development of a Health Action Plan. The Health Action Plan is not a stand-alone product, but needs to be integrated into every aspect of a person’s life. To achieve this, the person’s health and health issues need to be completely and accurately included in the ISP for purposes of life management. This occurs through the partnership facilitated by the VSHP nurse case manager in linking the PCP and medical treatment with the Waiver program and the rest of the person’s life. In this way, working together through a partnership achieves positive health outcomes for persons with ID.
Medical Home Model Proposal

Continue regular activities with adjustments to support health and partner with MCO

Program

Medical Care

New service to support link to health care

Continue to do regular activities but adjust services to meet unique needs of ID and partner with DMRS system to support health

Case Management
**HBCS Waiver**

Persons served under DIDS auspices include those in both residential and non-residential settings. However, it is important to note that less than half of persons with ID in Tennessee are enrolled in services provided by DIDS. In recent years, there has been a major shift in thinking in the field of developmental disabilities. Emphasis is now on people living in their own homes, controlling their own lives and being an integral part of their home community. The state of Tennessee is no longer admitting people to state-run ICF/MR facilities.

The Waiver program supports people in many living arrangements. They may continue to live with family or live semi-independently and receive Personal Assistance Services. Individuals may live with families or companions who receive payment for services. Individuals may live in other settings such as homes with up to four people. There are a few settings with up to eight people. These settings may be specialized, for instance for people with medical problems, but the settings are homes where people are living regular lives.

For those individuals supported by DIDS who are not living independently or with family, residential alternatives include:

- **Family Model Residential Services**: A family, other than family of origin, is paid to provide care and supports for an adult or child in their home.
- **Group home**: A setting with four or more individuals living in a facility with staff present. DIDS contracts with agencies that provide this service.
- **Supported Living Alternatives**: Residential services in a home with three or fewer people. The control and selection of service providers is maintained by the person receiving services with the assistance of a support coordinator and the family or guardian.
- **ICF/MR residential program including both private and state-run facilities**. The ICF/MR benefit is an optional Medicaid benefit. The Social Security Act created this benefit to fund "institutions" (four or more beds) for people with ID and specifies that these institutions must provide "active treatment." Most individuals residing there have other disabilities in addition to ID. Many of the people are non-ambulatory, have seizure disorders, behavior problems, mental illness, visual or hearing impairments or a combination of these problems. All must qualify for Medicaid assistance financially. These programs are part of the long-term care (such as nursing homes) programs paid for by Medicaid.

Medicaid funds are used to serve or support individuals in the ICF/MRs and HCBS Waiver program. To qualify for either, a person must meet financial eligibility for Medicaid, and medical criteria through the State’s Pre-Admission Evaluation (PAE) process. (PCPs are often called upon to participate in this process). HCBS is called a Waiver program because certain requirements of the ICF/MR Medicaid program are waived or set aside in order for individuals with ID to live in community settings rather than in ICF/MR centers.

The Waiver program also provides other services to support regular lives, such as programs for employment, environmental accessibility adaptation and AT. Other supports such as transportation, housing assistance, home modification, guardianship/conservatorship and special equipment are provided to those who qualify.
The Waiver program also provides a variety of services to help people with ID live a fulfilling and productive life to the best of their abilities. Special clinical services are available, but all aspects of medical care are provided outside the system. Although many people in the Waiver system are medically fragile and may receive specialized clinical services, this system is for living and supporting day-to-day life. Settings within this program are not nursing homes and usually are not staffed by medical personnel such as nurses. Since good health care is necessary to assure maximum independence and function, the system is required to support healthy lives and has procedures and requirements to meet this responsibility. Persons in this system obtain their health care services in generic medical systems, the same way the general population does. Waiver program responsibility for medical support needs to mirror (albeit informally) what anyone would do for themselves to support their own health and well-being. This would include considering health in everyday life (e.g., exercise, sleep, avoidance of toxic materials, etc.), maintaining a relationship with the health care system (i.e., personal physician, specialty clinics), partnering with the health care system (providing necessary information for treatment, asking questions, following through with recommendations) and maintaining appropriate documentation. The primary responsibility for health care maintenance lies with the person or agency supporting someone with ID with the help of DIDS in the areas of technical assistance, monitoring, education and consultation.

The Role of Waiver Therapeutic Services

Therapeutic services provided by the Waiver program include physical therapy, occupational therapy, speech language pathology, audiology, nutrition, and O&M services. Therapeutic service clinicians assist individuals to overcome barriers to accomplishing personal goals, and to attain and maintain optimal health and safety related to day-to-day tasks and activities.

Some of the many things therapeutic services providers may assist an individual with include improving balance when walking; communicating wants, needs, and feelings; learning to eat a healthy diet; eating safely; adapting tasks and environments to meet physical and/or sensory needs; improving work-related skills and developing independence in getting around a home when there are visual impairments.

The need for a therapeutic service assessment may be identified in a variety of ways. People moving from a developmental center may have received therapeutic services and supports that may need to be reassessed as they move into the community and adjust to their new environments and establish new daily routines. For others who are currently living in the community, those working with the person (the ISC, case manager, family, guardian, residential or day service providers, advocates and/or the primary care provider) may identify therapeutic service needs.
Following are common areas of concern for individuals with ID and developmental disabilities and may be assessed by the corresponding clinician:

Occupational therapist
- Eating and drinking
- Sensory defensiveness, self-injurious behaviors
- Oral hygiene
- Bathing and other activities of daily living

Physical therapist
- Wheelchair positioning
- Alternate positioning
- Transfers
- Mobility

Speech language pathologist
- Eating and drinking
- Communication

Audiologist
- Unrecognized hearing loss
- Adapting environments to help with hearing issues
- Adapting to hearing aids

Nutritionist
- Eating healthy, following special diets and menu planning
- Impact of genetic disorders, etc. on dietary needs
- Drug-nutrient interactions
- Impact of nutrition on chronic diseases

Orientation and Mobility (O&M)
- Impact of severe visual impairment/blindness on daily function
- Making use of other sensory systems (i.e., vision, hearing, touch, etc.) in order to make sense of the day
- Accessing the environment

When there are issues of concern in the above areas, an assessment may be indicated. An assessment should clearly identify how individual limitations are impacting function in order to justify the need for any recommended services. A plan of care is then drafted to include recommended goals, interventions and timeframes for completion. Assessment recommendations should identify interventions needed to support the person across environments (i.e., home, work, community) as appropriate. Therapeutic services are provided when they are necessary to provide strategies or techniques for promoting an individual’s health and safety, or to address a barrier to the individual achieving an important outcome or goal. When the individual is at risk due to health and safety issues, direct support staff and clinical providers need to collaborate on the development of staff instructions, which outline necessary techniques for carrying out certain daily activities.
Another therapeutic service available in the Waiver program is behavioral support. Not all negative behavior in persons with ID is a result of a psychiatric problem, but it may be problematic and may be an important communication strategy (as, for instance, pain), or impact participation in activities important to the person. There are two types of providers in the Waiver system: Behavior Analysts and Behavior Specialists. Behavior Analysts are trained at the graduate level and write functional assessments and behavior support plans. Behavior Specialists are trained at the undergraduate level and assist the Behavioral Analyst in training and monitoring behavior plans.

Behavior Analysts are trained to analyze behavior through the use of a functional assessment. Based upon observation and data collection in settings where the behavior takes place, a functional assessment provides information on the purpose of the behavior, and may often indicate an appropriate treatment such as a medical evaluation, referral for a psychiatric assessment, or development of a behavior support plan. Behavior Analysts have worked with a variety of clinicians to help solve problems creating negative behaviors that interfere with quality of life.

Assistive Technology (AT) is the generic term that includes assistive, adaptive and rehabilitative devices for people with disabilities, and includes the process used in selecting, locating, and using them. AT promotes greater independence as well as contributing to health. Someone who is positioned properly is less likely to fall and has better alignment of his/her internal organs. A person who feeds himself/herself has a lower incidence of choking than if fed by someone else. AT may include mobility devices such as walkers and wheelchairs, as well as hardware, software, and peripherals that assist people with disabilities in accessing computers or other information technologies. For example, people with limited hand function may use a keyboard with large keys or a special mouse to operate a computer. People who are blind may use software that reads text on the screen in a computer-generated voice. People with low vision may use software that enlarges screen content. People who are deaf may use a TTY (text telephone). People with speech impairments may use a device that speaks out loud as they enter text via a keyboard. A formal, legal definition of AT was first published in the Technology-Related Assistance for Individuals with Disabilities Act of 1988 (The Tech Act). The Tech Act was amended in 1994. In 1998, it was repealed and replaced with the Assistive Technology Act of 1998 ("AT Act").

AT devices are usually covered by Medicaid and Medicare, but there are some needs for customized equipment. Clinicians specially trained to evaluate people with ID are few. DIDS, through the Waiver program, has AT programs with trained therapists in each region. Workshops are also available to fabricate customized equipment which may cost less and be more effective than generic products. There are also several unique treatment approaches in the Waiver program unavailable outside the Waiver program. This includes position devices for very physically involved patients who, as a result, have had marked improvements in their health. People with severe contractures and severe scoliosis have been treated over time (sometimes up to a year) and these conditions have been greatly corrected. These were individuals who were not deemed eligible for surgery but had significant secondary health problems. Outcome examples include improved body alignment to reduce constipation and ileus, and significant improvement of severe scoliosis to improve swallowing and respiratory function to reduce aspiration pneumonia and hospitalization.
Resources
Family Guide to Assistive Technology is available at www.pluk.org/AT1.html (Parents, Let's Unite for Kids)

Independent Support Coordination
An essential and required feature of the DIDS way of serving people is Independent Support Coordination. A support coordinator acts on behalf of the person with ID to arrange and monitor services and supports. Support coordinators (and the agencies they work for) are distinct from DIDS, TennCare or VSHP.

Support coordination is the process of:
- assisting individuals and families in identifying preferences (what the person wants), capacities (what the person needs) and resources
- finding and gaining access to necessary supports and services
- developing an Individual Support Plan (ISP) for the person
- coordinating the delivery of supports and services
- monitoring the delivery of supports and services to determine the extent to which they meet needs and expectations

The ISP is used by the support coordinator to outline and obtain needed supports and services. The ISP document:
- Reflects the person’s vision of the future
- Reflects the person and his/her current lifestyle and needed supports
- States the outcomes necessary to achieve the person’s vision of their desired life
- Describes the actions, supports and services required
- Identifies the persons and providers responsible for the desired outcomes
- Is the central document for planning and reviewing the supports and services to be provided

The ISP also includes a medical section with a listing of all diagnoses and conditions, brief information about these, and how these may affect the person’s life. Also included in the ISP is essential information on clinical services provided through the Waiver program. These are clinical services (Occupational Therapy, Physical Therapy, Speech Language Pathology, nursing, nutrition, mobility, behavior, dental, etc.) which are paid for by the federally funded Waiver program (not the MCO). These must be very specific and are reviewed before approval for funding is given.

All services require a doctor’s order, and this may be confusing for the PCP who is used to initiating orders. In general, the process begins with asking for an evaluation by a Waiver provider. The request usually follows a specific concern by the COS, and the evaluation will be done by a qualified Waiver provider (OT, dietician, behavior analyst, etc.). Waiver providers are clinicians who have had specialized training and experience in treating persons with ID. After the evaluation, the clinician makes recommendations and develops a plan of care. This is given to the PCP for review and to obtain orders for this service to be provided through the Waiver program. At any time, Waiver clinicians are available for consultation with the PCP.
The Medical Home Concept

The concept of the Medical Home has evolved since its introduction by the American Academy of Pediatrics in the late 1960s. It was first used for children with disabilities. Positive outcomes and cost containment were such that some states and some insurance plans have adopted the model for chronic care conditions in the general adult population. This concept shifts the paradigm from episodic acute care to a continuous comprehensive model. The basic premise of the Medical Home Concept is that care managed and coordinated by a personal physician, with the right tools and supports, will lead to better outcomes. This provides a higher degree of personal care coordination, access beyond the acute-care episodes, and identification of key medical and community resources to meet patients' needs.

The Medical Home concept has been used since 1967, when it was first used in caring for children with special needs, so it is already adapted for the ID population. The components of this model focus on a single physician to coordinate and integrate care. This physician is also the "go to" person for general medical care and integrating other care, including specialty care and routine well care such as immunizations. This medical professional provides first contact for all medical issues and has continuous involvement with specialists – this approach integrates all aspects of comprehensive care. Central to the Medical Home approach is the premise that patient-centered care requires a fundamental shift from a fragmented relationship with many specialists, to a partnership between patients and their PCPs. This involves a shift from the specialty care model to an integrated, PCP-directed model.

Many people with ID are quite complex – they have medical problems that cross over a number of medical specialties. The Medical Home PCP provides integration and coordination of all of these aspects. The PCP does not need to have the same knowledge as the specialists, but needs to coordinate care so specialties do not work at odds with each other, e.g., many medications for behavior may cause increased seizure activities, and many seizure medications have behavioral side effects; psychotropics may cause constipation and other GI problems which may lead to behavior problems and thus increased psychotropics. Integration is also important as some issues and problems may “fall through the cracks” (e.g., preventive care).

In addition to medical needs, a PCP who knows the person well may manage or address other issues that may impact a person’s total health and well-being (e.g., school, work, SSI and other benefits, familiarity of PCPs with DIDS "rules," etc.). The PCP in this situation understands the "workings" of the Waiver program and DIDS systems in which the person is supported.

Studies show coordination may improve overall outcomes and quality of life. In addition, care is more efficient with a reduction in specialty visits, unnecessary emergency department visits and hospital admissions. The basic principles of the medical home that promotes better outcomes for patients, efficiency for PCPs, and better use of health systems are:

- **Personal physician:** Each patient has an ongoing relationship with a personal physician trained to provide first contact, and continuous and comprehensive care.
• **Physician directed medical practice:** The personal physician leads a team of individuals (all involved clinicians) at the practice level who collectively take responsibility for appropriately arranging care with other professionals.

• **Whole person orientation:** The personal physician is responsible for providing for all of the patient’s health care needs, or taking responsibility for appropriately arranging care with other professionals.

• **Care:** Coordinated and/or integrated across specialists, hospitals, home health agencies, the Waiver program, and/or nursing homes.

• **Quality and safety:** Assured by a care planning process; evidence-based medicine; clinical decision support tools; performance measurement; active participants of patients (or their representatives) in decision making; information technology; quality improvement activities, and other measures.

• **Enhanced access to care** is available (e.g., via open scheduling, expanded hours, and new options (e.g., a home visit)) for communication.

• **Payment must appropriately recognize the added value to patients** who have a patient-centered medical home.

**Resources**


**Roles and Responsibilities of the Nurse Care Managers (NCM) (VSHP)**

• Leader of the care management team

• Primary point of contact for physical and behavioral health needs for both member and providers

• All assessments and reassessments of a member’s physical and behavioral health needs, including face-to-face contacts

• Development of individualized Integrated Plan of Health Care for each member

• Coordination of the full array of covered physical and behavioral health services members need

• Provision of timely access to and monitoring of covered physical and behavioral health services, including authorization of TennCare covered benefits, and verification the service has been provided

• Collaboration with ISCs or Waiver Case Managers, as applicable, and MR Waiver, ICF/MR, and/or NF providers in implementing the Integrated Plan of Health Care which operates in conjunction with the member’s ISP

• Provision of disease management activities (if indicated) for members

• Identification and immediate addressing of service gaps

• Assistance in resolving concerns about service delivery or providers, including the quality of care rendered by providers, workers or NCM staff

• Provide information to contract providers, including, but not limited to, physicians and behavioral health providers and caregivers regarding the role of the NCM

• Request providers and caregivers to notify the member’s NCM as expeditiously as warranted by the member’s circumstances of any significant changes in the
• Receive all relevant information from other internal departments regarding his/her members (e.g., member services, disease management, utilization management and claims processing). The NCM shall follow-up on this information as appropriate (e.g., documentation in the member’s plan of care, monitoring of outcomes, and, as appropriate, needs reassessment and updating the plan of care)
• Monitor and evaluate a member’s Emergency Department and behavioral health crisis service utilization to determine the reason for these visits
• Active involvement in discharge/transition planning when a member is hospitalized or transitioned to alternative levels of care
• Ensure coordination with Medicare payers, Medicare Advantage plans, and Medicare providers, as appropriate, to coordinate the care and benefits of members who are also eligible for Medicare

**Supported Medical Home**

VSHP recognizes that access to effective and patient-centered primary care is a significant challenge for ID adults.

With the support of TennCareSelect, the primary care medical home may provide much of the care and coordinate specialized care for ID adults.

VSHP will strongly support a PCP medical home for Arlington members by:
• Assisting the PCP in obtaining medical records
• Linking the PCP with specialists
• Providing access to Mental Retardation and Developmental Disabilities (MR/DD) expert advice and consultation
• Information support
• Identification of resources
• Providing Clinical Practice Guidelines

**General Considerations**

Taking care of an adult with ID may often be a daunting task for the PCP. Problems usually encompass two areas: health itself and the system of support (federally supported system) in which they live, where the main emphasis is housing, working, hiring, recreation, etc., and not health care. For the busy PCP, these patients are time-consuming and may present atypical issues. Health is really the same for persons with ID as that of the general population, and most medical problems fall into well-defined areas. What is different is difficulty with information; for example, the patient's inability to give information, a lack of records and often presenting signs and symptoms later in disease progression, usually related to the nonverbal status of the patient which poses difficulty in early diagnoses and treatment. A typical patient may present complaining of common symptoms such as heartburn, and they probably have tried over-the-counter medications. A patient with ID, who cannot complain because of a lack of communication skills, may present with weight loss or anemia. These symptoms in a typical patient usually prompt the PCP to look for serious problems and embark on
extensive testing. As another example, persons with DS have a very high incidence of hypothyroidism. Many people think people with DS are "naturally overweight". They are not. They gain weight in the same ways as the typical population: having hypothyroidism, not getting enough exercise or eating too much.

The “system” which often asks PCPs for a lot of paperwork may also be confusing because of all its requirements. However, if the PCP understands "the system," it will be evident that it may help with medical care. Much information is kept (e.g., weights, sleep data, elimination charts, food intake) which may be available to the PCP if needed. Also, Waiver program requirements mandate many aspects (e.g., following doctor's orders, health care oversight and maintenance) which may be very helpful in diagnosing and treating patients.

In addition, there are clinical services provided by the Waiver program, not paid for through MCOs, to cover many critical areas often not available to typical patients. Examples of this include behavior support and assessment, which may be of immense value when assessing sleep problems or diagnosing medical problems when nonverbal patients use behavior as communication; or nutrition (help with persons with enteral tubes, significant obesity or special diets).

In this new program, the R.N. Case Manager (VSHP) may provide a link between the two systems. This person has the knowledge and training in health care for persons with ID, how health is supported in programs, and understands what clinical services are available through the Waiver program and the MCO.

**Treatment principles to consider:**

- See the person – not the disability. Find time to address the person and the best way to communicate. Avoid using jargon.
- People with ID feel pain and have emotions. Learn to appreciate how they express their feelings.
- Diseases are the same; presentation may be different (e.g., GERD may present with anemia rather than a complaint of heartburn since the individual may be non-verbal).
- The genetic diagnosis may provide a “medical roadmap” to care (e.g., hypothyroidism in DS, cancer in TS, Attention Deficit Hyperactivity Disorder (ADHD) in Fragile X syndrome).
- Medical issues which are normally minor in the general population may be life-threatening in this population (e.g., constipation, aspiration).
- Behavior problems may often be secondary to unrecognized medical problems or related to environmental issues (e.g., lack of coping skills, control, etc.).
- Significant medical issues may be overlooked when “diagnostic shadowing” (attributing everything to the disability) occurs.
- People with ID and genetic diagnoses vary widely and often do not “fit” the short paragraph in a medical textbook.
- Much is known about children with most of these diagnoses but less about adults, including life span. For example, the median life span for DS is now 55 years.
- Because of advances in technology and information, adults may look different and have different issues than younger individuals with the same diagnosis. For instance, it is rare to find someone under 30 with PKU who has significant
• Many patients with ID experience the “White Coat” syndrome. It might be worthwhile reconsidering the need for a white coat.
• Many patients with ID also associate masks and gloves with negative experiences and become anxious. If possible, do not use them.

**Suggested Office Strategies for the PCP**

Since DIDS and the RN case manager will be working with the PCP for optimum health care, the PCP may want to consider procedures to promote working with this model with patients served by DIDS Waiver system. All persons in the Waiver program are supported by people in the DIDS system (ISC, residential personnel, etc.) that may help with information, modifications and support. This would include obtaining records, helping with follow-through of recommendations, support with treatment and procedures, planning for emergencies, communicating with COS, and PCP use, among others.

The PCP may make this arrangement go smoothly by considering some simple procedures for the office. These could include:

1. Establishing a longer first visit with a checklist for planning strategies and outlining the PCP’s and patient’s (or representative’s) requirements and responsibilities.
2. Discussing medical records so DIDS personnel may facilitate acquisition of past medical history to include hospitalization and ER data, procedures (e.g., MRIs, genetic testing, etc.), prior medication use, allergies, family history, and consent status (conservator or guardian).
3. Reviewing communication issues such as when and how to call, agency records and system requirements (e.g., DIDS requires a written prescription for all medications, including over-the-counter, sunscreen, special shampoos, etc., and use of prescriptions (PRNs) must be very specific – e.g., not one or two tabs every 4-6 hours, but 2 tablets every 6 hours), who in DIDS is responsible for managing direct health care, (e.g., agency nurse, house manager), who the conservator is or if family is involved, and current clinical services provided by the Waiver program.
4. Specialist involvement should be reviewed and responsibility (as a consultant or primary prescriber) should be clear.
5. Review decision-making strategy: patient competence, involvement of conservator/guardian or family involvement.
6. Clinical issues such as accurate diagnoses, current medications (including OTC and PRN use), use of Waiver clinicians, and need for crisis plans should be reviewed.
7. It is imperative to discuss plans for emergency and acute-care, as well as issues of hospitalization, including post-hospital review guidelines (follow-up, medication reconciliation, care issues, diagnosis changes, etc.). This should include issues of sick/acute care and issues of hospital discharge planning.
8. Following are suggestions to help things go more efficiently and be productive:
   • Offer the first or last appointment of the day to someone who has ID.
   • Offer double consultation time, especially with the first visit.
   • Speak to the person with ID first. Only check with the caregiver if something is not clear. Be sensitive to the person’s feelings. Ask open-ended questions or changing the question around to see if you get the same response.
• Explain the process of the visit and procedures, including physical exam, before you start. This reduces anxiety and improves cooperation, even in nonverbal patients.
• Use language the patient understands at a simple level, or use a communication aid like pictures or symbols.
• PCPs should require accurate and concise information from agencies and families.
• Sometimes it may be useful to get information from supporters as well. Direct care staff may help with preparation and positioning for examinations.
• When you are talking about time, use events the person might understand.
• Do not assume the person will understand the connection between the illness and something they have done, or something that has happened to them.
• Have front office staff ensure the patient’s chart is available, all supporting documents are present, and the person comes with pertinent health information and someone who knows them and details about the issue.
• Review the chart before the patient is seen.
• Don’t allow a person with behavioral problems to remain in the waiting area or an examining room for long periods of time. Provide the person with an examining room or a larger area (indoors and outdoors) where the caregiver may take the person if needed.

People with ID are people first. Their disability is a part of who they are and how they function, but it does not define them. Each person has his/her own individual personality, ability and experience. They want to be treated like anyone else, but with some understanding and modification. Using negative language, not including them in the medical process, or ignoring them is offensive to them, their families and their caregivers. Appreciate the rights of people with ID to live as independently as possible and participate in their communities. The role of the PCP is to support them to do so.

Specific Components for the Care of People with ID

In addition to the usual types of care such as preventive, acute and chronic, a patient’s etiological diagnosis for ID and complications often guide treatment issues. For example, people with lifelong seizures are often on medications predisposing to osteoporosis. Non-ambulatory, non-self-feeding individuals are often predisposed to GERD but may be unable to express early symptoms. A genetic diagnosis often provides a “roadmap” for care and often a “medical checklist” may be available for the identified syndrome.

1) **Acute care** – Since many individuals with ID do not communicate well, early symptoms of illness may not be noted. Discussion of non-verbal cues may be warranted, as well as development of crisis plans. Discussion, in advance of when and where to go for acute issues (PCP’s office or the ER) is also helpful. Emphasis should also be on medical information beyond just presenting problems to include all medical issues. Below is a generic information outline which addresses the most often asked questions associated with medical illnesses which, though not always used, should always be available. This information should include a discussion with caregivers who will not accompany the patient. A simple format for history (may discuss process with supporting agency) is:
   a) Information about acute issue
   b) All current diagnoses
   c) All current medications, reasons for use, prescriber.
   d) Available data: weights, behavior, elimination, etc.
   e) Information on general issues with verification if there are any changes regarding sleep, eating/diet, elimination, recent emotional state/behavior, seizures, or life changes (e.g., staff, family, program, residence, roommates).
   f) Recent medical issues including dates: illness, ER visits, last hospitalization, recent medication changes and/or recent labs/tests.

2) **Chronic care** – Chronic care may be complicated in some individuals with ID who have several diagnoses and are on multiple medications. Also, if they are seen by multiple specialists, care may be fragmented and not integrated. To avoid complications, a yearly evaluation with a review of all diagnoses and a short plan for each is helpful in both saving time later and avoiding problems. A format for a short but comprehensive yearly evaluation could be:
   a. Review of past year’s medical encounters
   b. Review of significant events, health changes
   c. Review of collected health data (e.g., weights, elimination, etc.)
   d. Complete physical exam (P.E.) noting changes from last P.E., including weight
   e. Accurate verification of all current diagnoses, including etiology for ID
   f. Brief plan for each active medical problem

3) **Preventive care, including immunizations** – This care is particularly lacking for people with ID. This may also be difficult in some situations secondary to problems with body position in people with deformities. There are adaptations (e.g., use of ultrasound in lieu of mammograms, heel scans for hip densitometry, etc.). In many cases, there are indications for preventive care based on diagnosis (e.g., DS for hypothyroidism).
4) Diagnosis specific care – There are many issues unique to the care of people with ID. This includes specific problems related to the diagnosis or modifications necessary due to disabilities. For instance, there are many health guidelines (e.g., DS) for identified genetic syndromes. There are many modifications and adaptations for interactions (e.g., communication and positioning) with individuals that may be provided by staff working with the individual. There is often data (e.g., weight, elimination, sleep, seizures, etc.) routinely kept by staff which may help solve problems. There are also modifications or accommodations that may be made for appointments (e.g., first appointment of the day to avoid waiting, desensitization, etc.) and procedures. Over time, secondary complications may cause more problems than the initial diagnosis.
Predicting Health Related Risks

Certain medical issues may predict other health risks. These issues should be discussed with the COS by the PCP after the general examination. This may only occur if knowledge of important events of the past year, with respect to a person’s health, is available. This will enable the PCP and the ISP teams to more effectively plan for the future needs of the individual. This provides a documentation mechanism for the ISP team to review the current interventions, whether they are sufficient to meet the individual’s needs or not, and any anticipated future supports. The following are typical for persons with ID:

- Multiple medical or psychiatric hospitalizations in a year
- Multiple visits to the emergency room (whether admitted or not)
- A person living alone or with little supports who takes multiple medications
- Taking three or more medications for a chronic medical condition, including a psychiatric diagnosis, with reduced supports
- Medical benefit loss
- Poor follow through on post hospitalization discharge orders
- Significant changes in health or mental status
- Significant changes in sleeping or eating patterns
- Unmet medical needs (e.g. appointments not scheduled, follow-up appointments missed)
- Information shared with medical personnel by support staff is inadequate (e.g., reason for referral)
- Poor compliance or non-compliance with medical regime
- Refusal of services
- Inability to tolerate a medical examination/procedure
- Multiple falls/fractures, a history or diagnosis of osteoporosis
- Mobility impairment and/or skin breakdown
- Significant weight gain or loss
- Swallowing disorders
- History of choking and/or aspiration
- Genetic history
- Obesity and/or diabetes
- History or diagnosis of thyroid issues
- Stroke or cardiovascular issues
- Compromised communication skills (especially in relation to being able to indicate physical pain)
- Pica
- Lifestyle choices that negatively affect health, (e.g., smoking, or drinking when contraindicated by medications)
- Multiple contacts with mobile crisis

Medical risk may be anticipated and discussed based on medical diagnoses and genetic disease. Many medical conditions have predictable progressions, complications and associated conditions. Many of these may be avoided or the severity lessened if anticipated before they are a significant problem. In addition, genetic diagnosis often provides a “roadmap” to medical care, including diagnosis-specific growth parameters, biochemical abnormalities, and medication sensitivities.
Medical risk also is an issue with lack of medical records (past medical history, test results, current care, hospitalizations and ER visits, etc.). This puts people at risk when accurate diagnoses are not known, or people are subjected to repeat testing, prior medication side effects, or medication failure.

Examples of areas of risks include:

1. Medical risk in people with ID and associated common diagnoses:
   a) Constipation puts people at risk for megacolon, obstruction, and eventually, Ogilvie’s Syndrome (gut shutdown). It also creates other problems such as increased seizures, hydronephrosis due to compression, abdominal pain, decreased appetite and weight loss, and increased behavior problems. Many medications contribute to constipation (iron) as well as many medical diagnoses (hypothyroidism).
   b) Non-ambulatory status places people at risk for osteoporosis (no weight bearing activity), constipation (variety of mechanisms) and weight loss (inadequate free access to food). Also, lack of free access to fluids may contribute to chronic or acute dehydration and constipation.
   c) Hypothyroidism puts people at risk for osteoporosis (treatment side effects) and constipation.
   d) Long-term administration of phenobarbital and Dilantin (standard seizure treatment for many years) puts people at risk for osteoporosis.
   e) GERD puts people at risk for anemia (from bleeding, which is more common in nonverbal persons), weight loss (from food refusal), Barrett’s esophagus, and adenocarcinoma.
   f) Seizures place people at risk for SUDEP (Sudden Death from Epilepsy). This occurs most often in persons between 20 and 40, and the incidence is 1/350 to 1/800 person years. It is not related to severity of seizures.
   g) Aspiration puts people at risk for aspiration pneumonia (acute) or chronic lung disease (when continuous over time). It may or may not accompany GERD.
   h) Use of many psychotropic medications and Reglan may put people at risk of Tardive Dyskinesia, which may be non-reversible.
   i) Use of the Selective Serotonin Reuptake Inhibitors (SSRI) class of psychotropics puts people at risk for obesity and diabetes mellitus.
   j) Chronic lung disease puts people at risk for pulmonary hypertension and significant cardiac problems.
   k) Poor oral hygiene puts people at risk for dental problems and heart attacks.
   m) Duodenal ulcers (usually caused by H. pylori) put people at risk of gastric cancer.
   n) Spasticity puts people at risk of pain, decreased energy, contracture and scoliosis.
   o) Scoliosis places people at risk for pain and discomfort, as well as pulmonary compromise in severe cases.
   p) Prior abdominal surgery (e.g., enteral tube placement) puts people at risk for abdominal obstruction.
   q) Intracranial shunts place people at risk for infection and behavior problems.
   r) Multiple medications increase the likelihood of negative drug interactions and side effects.
s) Incontinence increases the risk of perineal skin problems. Chronic use of catheters predisposes people to bladder cancer.

2. Genetic risk: Many genetic syndromes have medical checklists and medical guidelines for care. New information about these problems is continually evolving. Many genetic conditions have support groups where this information may be accessed. The majority of these medical problems are easily diagnosed and treated when anticipated. Some examples of medical risk based on a genetic diagnosis are:
   a) Down syndrome: Hypothyroidism (up to 40 percent to 50 percent over lifetime), vision problems (>50 percent), hearing problems (>70 percent over lifetime), and Alzheimer’s disease (25 percent). Also increased incidence of GERD, depression, and atlanto-axial instability at any age.
   b) Lesch-Nyhan disease: “Hard-wired” self-mutilation and kidney stones, causing kidney failure
   c) Fragile X syndrome: High incidence (80 percent) of ADHD
   d) Prader-Willi: Hyperphagia with morbid obesity and secondary problems
   e) TS, Neurofibromatosis: High incidence of cancers
   f) Velo-cardio-facial syndrome: High incidence of schizophrenia and depression
   g) Ehlers-Danlos syndrome: Easy bruisability
   h) Smith-Magenis-syndrome: “Hard-wired” behavior consisting of insertion of foreign bodies into body orifices, Self-Injurious Behavior
   i) Osteogenesis imperfecta: Pathological fractures
   j) Rett syndrome: Apnea and breathing problems
   k) Septo-optic-dysplasia: Endocrine problems
   l) Usher’s syndrome: Retinitis pigmentosa
   m) William’s syndrome: Cardiac problems, hypertension, and hypercalcemia in infancy
   n) Behavioral phenotypes: Many “hard-wired” behaviors are mistaken for psychiatric symptoms

3. Lack of Medical records/information:
   a) EVERY medical encounter begins with gathering a history. Having no, incomplete or inaccurate information hampers medical care.
   b) Without prior information, people are at risk of repeating diagnostic tests (e.g., CT scan) which could subject them to discomfort or side effects, and are not cost-effective.
   c) Without prior medication information (particularly true for anticonvulsants and psychotropics), people are at risk of having repeated adverse events.
   d) Without proper immunization information, people are at risk of repeat injections which may be painful or cause side effects, and is not cost-effective.
   e) Without proper information on follow-up medical visits, problems are at risk of not being treated correctly or resolved.
   f) Without proper information on “routine” follow-up for chronic problems, people are unlikely to receive care that is maximally effective and cost effective.
   g) Lack of information of all medications a person is receiving to all practitioners the individual sees contributes to side effects, as well as lack of effectiveness, and may increase cost of care.
   h) Inefficient care caused by lack of information has the potential to significantly interfere with a person’s daily life, and increase the burden on those who support the individual.
**Special Issues**

**PHENYLKETONURIA (PKU)**

PKU is an inborn error (enzyme deficiency) that results in an increase in the phenylalanine which, in high doses, causes damage to the developing brain. This results in ID. Brain damage may be prevented by managing the patient’s diet to keep phenylalanine levels low, but this treatment needs to begin very early in life. For over 30 years, babies have been diagnosed at birth by mandatory testing and treated with special formulas and diets to prevent brain damage. Therefore, there are actually two populations of people with PKU: young people who are not cognitively impaired (and not in the Waiver system), and older individuals who did not begin treatment at an early age and have brain damage associated with typical features of cognitive impairment.

Many patients with PKU stop the therapeutic diet when brain growth is complete. Published studies show that some young adults, particularly those with behavior changes, should continue on the PKU diet, which is both expensive and unpalatable for many. This diet, however, will not correct existing damage and requires very close oversight to prevent additional metabolic problems. Use of the diet might be considered in an older individual with PKU if significant behavior issues exist. If the diet is used, a clear reason for use and goals should be in place (e.g., specific behaviors). Careful monitoring of both behaviors and metabolic parameters is mandatory. General diets modified to lower the overall phenylalanine level should be considered, and Waiver program dieticians may recommend and monitor these.

**Spina Bifida (SB)**

SB is a general term that encompasses a wide range of malformations in the brain and spinal cord. While children may have life-threatening issues and complications, adults are generally stable with chronic problems, but secondary problems or complications may become life-threatening.

Until recently, people with SB continued to be cared for by pediatricians, but now that they are living longer, care is being transferred to generic adult services. Multidisciplinary clinics exist, but are mainly for children, and are usually located with a medical school. While most individuals with SB have normal intelligence, there are many especially complex cases, in the Waiver program. PCPs will be faced with caring mainly for co-morbid conditions and complications.

Major issues are:

1. Intracerebral Shunts – A great number of children with SB have hydrocephalus and are treated with shunts. These are not removed and remain in place in adulthood. (They are often overlooked but there are clues noticeable during a physical exam such as an obvious tube subcutaneously located in the neck and/or palpation of a “bulb” in the scalp under the hair). Shunts are considered to be “nonfunctional” in adults, but there is no way to be sure. The possibility of shunt malfunction with accompanying symptoms, sometimes quite severe, should be considered. It should also be remembered that the shunt is a foreign body, and some people recommend prophylaxis as used for valve issues in the heart. Symptoms of shunt failure include development of seizures or increase in
2. MRI evidence of Chiari malformation is present in up to 75 percent of patients with SB, and one third of these may have compression with development of apnea, dysphagia, headache, quadraparesis, scoliosis and gait abnormalities. This may develop at any time; it is usually progressive but may be acute and constitute a medical emergency.

3. Sequelae of secondary neuromuscular deficits include deformities of limb and spine and may have similar physical progression of neuromuscular status as CP.

4. Bladder abnormalities are significant and progressive. Because of the underlying neurological deficit, urinary elimination is compromised leading to retention, infection, hydronephrosis and renal damage. Treatment should be aggressive to avoid progression. Eighty percent have a neurogenic bladder. Some adults have urinary diversion procedures such as ureterostomy, which may present secondary problems. Renal damage over time may occur, especially if urinary excretion is blocked over a period of time. Because of decreased muscle mass, the use of routine blood creatinine to diagnose renal involvement may not be helpful and at least baseline. More definitive investigations such as a 24-hour creatinine clearance study should be performed.

5. Bowel abnormalities (secondary to nervous system dysfunction) may result in constipation, impaction and co-morbidities. An aggressive bowel management program should be in place.

6. Individuals with SB have no sensory feeling below the neurological lesion’s nerve innervation level, and as a result, are unaware of bruises, lacerations, etc. This puts them at great risk of secondary problems including significant decubitus ulcers and infections.

7. A high percentage of people with SB also have problems with obesity, short stature, lower extremity atrophy and visual acuity.

8. A particular problem in SB is latex allergy.

9. A high percentage of people with SB also demonstrate "cocktail party chatter," a condition where speech "sounds normal" but "says little." This may result in a situation where the person appears to understand information when they don't. This may compromise medical treatment when the patient is unable to follow through with misunderstood recommendations.

10. Physical limitation (movement) and treatment issues (incontinence) may limit participation in many activities and put individuals at risk of other problems (e.g., depression, weight gain, and/or poor self-image).

**Down Syndrome (DS)**

DS is one of the most common etiologies for ID. This diagnosis represents 15 to 18 percent of those receiving Waiver program services. One of the most-studied
There are several medical checklists available. In the past, persons with DS were treated by pediatricians since it was felt they would not live long. Now, since the median lifespan is 55, they are largely taken care of by PCPs who treat adults. For the most part, problems seen in people with DS have been well-defined; though people with DS may have any medical problem seen in the typical population. There are, however, a few exceptions which should be kept in mind. There is a decrease in the incidence of solid tumors such as lung and breast cancer. This is believed to be due to increased tumor suppressant activity secondary to an extra chromosome 21. They are also not at risk of myocardial infarction, also due to the extra chromosome increasing the mechanisms to address free radicals, a factor in atherosclerosis. Alzheimer’s disease occurs in a large portion, 25 to 50 percent, and occurs about 20 years earlier than in the typical population. However, Alzheimer’s disease is not inevitable in persons with DS. There are also individuals with DS who have consequences of adult congenital heart disease, usually secondary to lack of treatment as children. Following is a medical checklist based on published information and mortality information of people with DS in Tennessee’s Waiver program.

An interesting point to note is that 5 percent of this cohort was being treated for gout. Although not reported in the literature, other programs treating adults with DS have noted this finding.

<table>
<thead>
<tr>
<th>ISSUE/PROCEDURE</th>
<th>FREQUENCY</th>
</tr>
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<tbody>
<tr>
<td>Preventive Care - should follow that for typical population; immunizations should address risk of hepatitis</td>
<td>Yearly</td>
</tr>
<tr>
<td>Height and Weight - DS has higher pounds per inch; may refer to DS growth curves</td>
<td>Yearly</td>
</tr>
<tr>
<td>Thyroid - hypothyroidism occurs in 10-40%; slightly higher incidence of hyperthyroidism</td>
<td>Yearly</td>
</tr>
<tr>
<td>Vision - 25-43% have refractive error; cataracts in 12%, keratoconus in 15%</td>
<td>Every 2 years</td>
</tr>
<tr>
<td>Hearing- A. cerumen- small ear canals B. audiology - up to 70% with conductive and sensorineural loss</td>
<td>A - yearly; B – Every 2-3 years</td>
</tr>
<tr>
<td>Dental - gingivitis, periodontal disease and bruxism most common</td>
<td>Consider re Physical Exam (PE)</td>
</tr>
<tr>
<td>Cervical Spine - atlantoaxial instability in up to 14%; cervical stenosis and arthritis also common. Hx and PE of greater value than x-rays</td>
<td>Consider re PE</td>
</tr>
<tr>
<td>Obstructive Sleep Apnea - occurs in up to 50%; not necessarily associated with obesity</td>
<td>Consider re History</td>
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<tr>
<td>9</td>
<td><strong>GERD</strong>- common and may present with extra-system symptoms (e.g.,&quot;asthma&quot;)</td>
</tr>
<tr>
<td>10</td>
<td><strong>Constipation</strong> - very common and may present risk of obstruction, hemorrhoids</td>
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<tr>
<td>11</td>
<td><strong>Nutrition</strong> - must also consider celiac disease; risk of obesity is high</td>
</tr>
<tr>
<td>12</td>
<td><strong>Cardiac</strong> - congenital malformations either treated or untreated (risk of secondary lung problems); valve dysfunction common in adults without known disease (requiring Subacute Bacterial Endocarditis prophylaxis)</td>
</tr>
<tr>
<td>13</td>
<td><strong>Gait</strong> - about 5% with lower extremity subluxations. Cervical compression must also be ruled out</td>
</tr>
<tr>
<td>14</td>
<td><strong>Feet</strong> - may have significant issues with skin, hallux, fungus</td>
</tr>
<tr>
<td>15</td>
<td><strong>Skin</strong> - folliculitis, dyshidrosis, sensitivities and significant seborrhea including eye lashes, xerodermatitis, onychomycosis</td>
</tr>
<tr>
<td>16</td>
<td><strong>Seizures</strong> - peaks in childhood and in older adults; may indicate diagnosis of Alzheimer's Disease</td>
</tr>
<tr>
<td>17</td>
<td><strong>Behavior</strong> - look for medical issues, psychiatric diagnosis (most common OCD, depression, conduct disorder) or Alzheimer's dementia. &quot;Self-talk&quot; is common and not psychosis.</td>
</tr>
<tr>
<td>18</td>
<td><strong>Loss of Skills</strong> - look for medical issues; consider Alzheimer's disease. All individuals should have baseline evaluation.</td>
</tr>
<tr>
<td>19</td>
<td><strong>Diabetes</strong> - more common than typical population</td>
</tr>
<tr>
<td>20</td>
<td><strong>Abuse</strong> - consider with significant sudden change in behavior or functional skills</td>
</tr>
<tr>
<td>21</td>
<td><strong>Cancer</strong> - solid tumors are less common; leukemia occurs in young children</td>
</tr>
<tr>
<td>22</td>
<td><strong>Laboratory</strong> - leukopenia and macrocytosis are quite common (without treatable pathology); hyperuricemia may indicate gout or renal disease</td>
</tr>
<tr>
<td>23</td>
<td><strong>Immunizations</strong> - some people recommend pneumovax at 50; at greater risk for hepatitis</td>
</tr>
<tr>
<td>24</td>
<td><strong>Gout</strong> - noted in 5% of deaths in TN</td>
</tr>
</tbody>
</table>
| 25 | **Individuals with DS:**  
  - Have decreased tendency to complain of pain |   |
- Often manifest medical problems with behavior change
- Have increased risk for Alzheimer's Disease: 0-10% age 30-39; 10-25% age 40-49; 28-55% age 50-59; 30-75% age 60-69

### End-of-Life Issues

End-of-life planning is important for everyone, including those with ID, and should start early when patients are relatively healthy and discussion creates less anxiety. This may begin by discussing prognosis of major diagnoses (including etiology for ID), possible complications, and expected course. Life span may be discussed but only in general terms (no one can predict death accurately) and using similar populations for reference. As an example, the median age of survival for persons with DS is about 55 years, although generally shorter for those with congenital heart disease. Individuals with DS have been reported to live into their 80s. Obviously, this is a wide range, but may be used to begin discussion. So, in discussing medical issues of a healthy person with DS, it is likely the person will outlive their parents, and understanding this may prompt planning for the future. Because of the uncertainty of life in general, and also to facilitate planning for the future, discussion of end-of-life issues should start early for everyone, including those with ID. No one can predict when someone will die, but for people with ID, many people think their lives will be shorter. This is shown by death certificates that indicate the cause of death as MR, ID, SB, or DS. These are not terminal illnesses and not a cause of death. People with these medical disorders usually die from secondary (complications) medical problems such as kidney problems in SB, respiratory problems secondary to chronic dysphagia in CP, and Alzheimer’s disease associated with DS, all of which may be predicted in the course of routine medical care, and should be used to predict prognosis, complications and secondary problems.

There are some cases of ID with genetic or metabolic etiologies who may have a shorter life span, but some of these expectations should be known in advance. There are some people with severe and profound ID who develop significant secondary medical problems or complications which may lead to a shorter life span. Examples of this are individuals with severe CP with dysphagia (swallowing problems) that may develop GE reflux disease which progresses to Barrett’s Esophagus, ending in adenocarcinoma. Also, there are some significant problems which are often uncommon in the general medical community that may shorten life, such as respiratory problems secondary to chronic aspiration from dysphagia, and significant bowel problems, including obstruction with long-term constipation, that are a very common finding in individuals with disabilities and ID.

Another issue to be discussed is who makes decisions. Persons with ID in the Waiver system may have families (many have no family involvement or contact), paid guardians, or have not been judged incompetent (meaning, theoretically they can make informed decisions themselves). Also as a general rule, persons in the system may not have a living will which requires competence to execute, and cannot be done by another person, even a legal conservator.
End-of-life issues are not just deciding treatment at the time of a crisis or serious illness, but planning for all aspects of the future. Most people with ID will outlive their parents, even though most parents usually have a deep desire that their child with ID may precede them in death. Often, this is because they are concerned about who might look after the person with ID. The life span for people with ID is very close to the life span of people without ID, except for people with particular genetic diseases or perhaps, severe ID with associated medical complications. The public, and sometimes the medical community, often receive misinformation and assume that people with ID will have shorter lives.

As with the general population, persons with ID wish to end their lives surrounded by people they know who care about them. This includes spending their last years in a place where they are comfortable (aging in place). As a rule, Waiver services may be provided to support these wishes.

Another critical issue to consider is does the individual have a terminal diagnosis. This is an area that may become a “slippery slope” for people with ID. There are certain misconceptions that people with ID have terminal diagnoses which are not treatable and, occasionally, treatment is denied (e.g., they die of “terminal DS,” “terminal CP; but these diagnoses are NOT terminal diagnoses). The etiologies of ID are usually not terminal themselves, or even the cause of death. Causes of death and terminal illnesses match similar categories as that of the typical population. This requires very careful discussion. Examples of this are the diagnoses of various forms of cancer, end stage renal disease, cardiac problems, pulmonary problems and Alzheimer’s disease, among other diseases and medical problems. Unfortunately, sometimes, people with ID are denied treatment of cancer, kidney dialysis, cardiac treatment and other common forms of treatment just because they are ID. There is also the misconception that people with ID do not respond to treatment or will not cooperate with treatment. This is not true. They actually may do better because of the supports they have in the Waiver system which may ensure following treatment regimens, recognizing complications earlier, and other advantages. As an example, studies have shown that of the less than 50 people with ID who have received kidney transplants, as a group they have done better, cooperated more and lived longer than the rest of the population who received kidney transplants who did not have ID. Therefore, when someone with ID receives a terminal diagnosis with the declaration that no treatment is available, a second opinion should be sought. This should be discussed with people knowledgeable about both the medical issues and the response of people with ID.

The hospice movement has been very positive in promoting death and dying, as well as the use of palliative care, and is appropriate in many situations. It is also helpful for people with ID, but there are a few issues to consider. First, is qualification and prediction of closeness to death. One of the critical elements of hospice evaluation both for admission and to estimate closeness to death is based on a functional assessment (e.g., Karnofsky scale). With the typical population, healthy people function at 100 percent and those near death at 20 to 30 percent. It is important to understand that with people with ID, 20 to 30 percent may be their baseline functioning and not represent a terminally ill state. It is important to ensure that the person really has a terminal disease and is not just written off because they are “mentally retarded.” In addition, diagnoses such as “failure to thrive” and “debility,” which are poorly understood in people without ID, are even less understood in people with ID. These diagnoses should be viewed with great caution in people with ID whose baseline function before illness may qualify them
as appearing terminal compared to someone without ID. In these people, their baseline function may not have changed.

Treatment issues should also be reviewed. In some settings, all or most medications are discontinued. However, discontinuing seizure medications or those that treat GERD may cause considerable distress and be counterproductive. Treatment of pain should follow general guidelines and have a defined plan. Persons with ID do have issues with pain and may be assessed and followed, although procedures may require adaptation. Pain medications for comfort should only be given if discomfort (e.g., pain) is really present. Remember that constipation is already common in people with disabilities, and constipation as a side effect of most pain medications.

Discussion of a possible “do not resuscitate” or “do not attempt to resuscitate” (DNR) order needs to take place when someone is in a terminal state. Generally, “do not resuscitate” orders are not given unless someone has a terminal illness, or in cases in which resuscitation may cause more harm than good. ID alone is not sufficient reason for a DNR order. Consideration of this and other treatment issues may be facilitated by using the Physician Order for Scope of Treatment (POST) developed by the Tennessee Department of Health which is equivalent to the old “do not resuscitate” orders, and stays with the patient whether he or she is in the hospital, at home, or in another setting. People with severe deformities may pose significant procedural issues when considering CPR since it may be difficult to perform the procedure itself or if damage such as rib fracture is likely to occur. There are no guidelines for this situation, so each case must be carefully discussed. In addition, unless other decisions have been made, it must be clear that for persons with ID, DNR does not mean “Do Not Treat.”

End-of-life planning is extremely important for individuals with ID, although often it is difficult to discuss. It is prudent to start discussing end-of-life issues when people are generally in good health. A good way to do this might be beginning to discuss end-of-life issues each year when the ISP is written. Another way to make this discussion more fruitful and less stressful might be for parents and family members to discuss their own end-of-life issues.

**Resources**


Resources: TN dept of Health POST for Guidelines

**Treatment of Pain**

In general, pain is under-treated in this country, and this is also an issue for people with ID. In studies surveying people with developmental disabilities who can communicate, it was found this population experiences pain more often than the typical population, including both acute and chronic pain. Many people think this population does not experience pain; this is a myth. This misconception is probably related to habituation where, over the course of years, pain is not treated, and the person’s pain response has been extinguished. In addition, it may be difficult to distinguish non-pain behavior from pain behavior, particularly in a non-verbal patient; behaviors typically associated with pain may be caused by non-painful stimuli. Most people in the Waiver system have
someone who knows them well who can usually identify stress associated with pain and
discomfort. Specific behaviors usually related to pain include: vocalizations like crying,
screaming and moaning; grimacing or other facial distortions; changes in eating and
sleeping; changes in posture and tone, such as arching and stiffening; becoming
withdrawn; being inconsolable; not wanting to be touched; and changes in physiological
responses such as blood pressure, heart rate and diaphoresis. There are several pain
checklists* developed for children or non-verbal patients which, together with discussion
of caretakers’ perceptions, may be very helpful in evaluating pain.

Types of pain may be nociceptive, neuropathic, visceral hyperalgesia, or central.
Nociceptive pain includes such acute diagnoses as fractures, UTI, lacerations, and
chronic causes such as GERD, constipation, joint pain, or dental pain. Nociceptive pain
may be treated by the usual pain medications. Neuropathic pain is associated with
alterations of Central Nervous System CNS, and can be identified by presence of
spasm, dystonia and tremors or autonomic disturbances such as diaphoresis, erythema
or mottling. Neuropathic pain may be treated by other drugs such as gabapentin, elavil,
or benzodiazepines. Visceral hyperalgesia is an altered response to stimuli most often
in the GI system. This type of pain most often responds to treating the cause (e.g.,
constipation), or symptoms (e.g., gas). Common issues include enteral tube feeding
intolerance, intestinal gas, prolonged response to GERD, and pain associated with
elimination. Central pain is associated with insults to the CNS (e.g., Cerebrovascular
accident (CVA) and is not well defined in patients with childhood neurological
impairment.

Treatment strategies begin with an assessment for treatable causes of pain such as UTI
or dental issues. It would also include evaluation of pain potential for chronic problems
such as constipation (very common in people with disabilities), spasticity and fixed
contractures. Common unrecognized causes of pain in the ID population, especially
those with neurological impairment, include Ear, Nose, and Throat (ENT) infections,
corneal abrasion, ventriculoperitoneal shunt malfunction, esophagitis, intussusception,
rectal fissure, cholecystitis, pancreatitis, neurogenic bladder, obstructive uropathy,
inguinal hernia, menstrual cramps, gonad torsion, hip subluxation, and spinal stenosis.

Treatment obviously starts with treating the underlying cause if it can be identified. The
use of pain medications must involve understanding the interactions of other
medications the patient is taking as adverse effects are common. In addition, side
effects of the pain medication must be kept in mind. As an example, opioids may cause
constipation, which may result in pain that clouds evaluation of effectiveness of
treatment. Drug interactions such as through the P450 system may cause problems.
Plan of treatment, as with any patient, should be built on a thorough evaluation and a
comprehensive plan, and should be monitored on a regular basis. In general, treatment
should begin with the milder medications, but stronger medications should not be held
on the basis of ID. Nonpharmacologic treatment such as massaging, repositioning,
supportive equipment, use of warm baths, etc., should be considered. Waiver therapists
may be of help with identification of symptoms and response, and may help with
nonpharmacologic treatments.

*Pain scales: Face, Legs, Activity, Cry, Consolability (FLACC), Non-communication
Children’s Pain Checklist-Revised (NCCPC-R), Pediatric Pain Profile (PPP), or FACES
Pain Scale – all available online via the Internet.
Oral Health Care

Persons with ID may be at more risk for oral pathology (decay, periodontitis, pain, etc.) for several reasons:

- Mental capabilities vary in people with developmental disabilities and influence how well they can follow directions at the dentist and at home.
- Behavior problems may complicate oral health care. Anxiety and fear about dental treatment may cause some patients to be uncooperative. Behaviors may range from fidgeting or temper tantrums, to violent, self-injurious behaviors, such as head banging. This is challenging for everyone and may curtail care.
- Mobility problems are a concern for many people with disabilities; some rely on a wheelchair or a walker to move around. Just sitting in a dental chair may be difficult.
- Neuromuscular problems may affect the mouth. Some people with disabilities have persistently rigid or loose masticatory muscles. Others have drooling, gagging and swallowing problems that complicate oral care.
- Uncontrolled body movements may jeopardize safety and the ability to deliver dental care.
- Cardiac disorders may affect the delivery of oral health care. Many people with DS, for example, have congenital heart disorders that place them at risk for bacterial endocarditis. Prescribe antibiotic prophylaxis when indicated.
- Gastroesophageal (GE) reflux sometimes affects people with central nervous system disorders such as CP. Teeth may be sensitive or display signs of erosion.
- Seizures accompany many developmental disabilities. The mouth is always at risk during a seizure. Patients may chip teeth or bite the tongue or cheeks. Persons with controlled seizure disorders may easily be treated in the general dental office.
- Latex allergies may be a serious problem. People who have Spina Bifida or who have had frequent surgeries are especially prone to developing an allergic reaction or sensitivity to latex which may be life-threatening.

People with developmental disabilities typically have more oral health problems than the general population. This is complicated by a lack of access to dental care created by payment issues, dental caries, dentists’ unwillingness to treat them, and issues of individual patients with physical or behavioral limitations.

Dental caries are common and are associated with diet and oral hygiene, and sometimes medication (e.g., medications that reduce saliva or contain sugar). Periodontal disease also occurs more often in people with ID. Gingival hyperplasia caused by medications such as some anticonvulsants, antihypertensives and immunosuppressants increase the risk. Malocclusion may be associated with intraoral and perioral muscular abnormalities, underdevelopment of the maxilla, and oral habits such as bruxism and tongue thrusting. Malocclusion can make chewing difficult, and increase the risk of periodontal disease, dental caries and oral trauma. Damaging oral habits such as bruxism, food pouching, mouth breathing, and tongue thrusting, as well as self-injurious behaviors involving mouth structures, rumination and pica may cause significant problems. Oral malformations such as enamel defects; variations in size, shape, and number of teeth; cranial facial abnormalities; and hyperplasia of the mid-facial region are also contributory factors. Trauma and injury secondary to falls, accidents, and sometimes, seizures, also affect oral health. Physical abuse often...
presents as oral trauma and should be addressed. Abuse is reported more often in people with ID than in the general population.

Oral hygiene may be a real challenge, but there are many adaptations and approaches for success. These include:

- evaluation for the use of adaptive equipment (toothbrushes, mouth guards, aids for staff to brush teeth)
- proper position for success
- desensitization both for tooth brushing (oral sensitivity) and dental appointments
- staff training in special techniques for oral hygiene

Waiver clinicians can help in all these areas with training, evaluation and treatment by therapists experienced with treating people with ID.

In addition, Waiver funds may be used for dental treatment for adults in the Waiver program. This may include sedation anesthesia for those who need it. A desensitization program may avoid the need and expense of sedation; and Waiver clinicians may help with this.

Dental issues are known to be important for general health. As an example, it is now known that mouth pathology may increase the likelihood of myocardial infarction. This means that people who are fed through enteral tubes continue to need daily oral hygiene, even if they are not eating by mouth.

An oral exam should be part of the PCP's annual evaluation. As well as diagnosing oral and dental needs, a clue to other diseases (e.g., enamel erosion with reflux) may be noted.

**Resources**

Adults with Cerebral Palsy (CP)

Although CP is a non-progressive disorder of motor function with onset in childhood, adults with this diagnosis differ from children secondary to complications and physical changes, due to abnormal muscle tone. Surprisingly, about 80 percent of persons with CP in the United States are adults aged 21 and older. These patients were often followed by pediatric specialists long into their adult years, but this is rarely done now. These patients also had access to specialized clinics for CP and received many supportive services through school programs. The majority of these patients now are cared for by a PCP with occasional, fragmented care by specialists with little experience with adults with CP.

In many instances, adult patients in the Waiver system do not have a formal diagnosis of CP but are just noted to be “spastic” or have “spasticity”. As children, they may not have had access to diagnostic evaluations or the diagnosis has been dropped from their diagnostic problem list. In some adults, individuals may have spasticity that is secondary to events and pathological processes that is not CP because they did have problems as a child. CP is a developmental disability and therefore begins in childhood. An example of this is someone with lower extremity contractions of hips, knees and feet (usually of 90 degrees) as a result of sitting in a wheelchair for years. This condition does not resolve with treating the supposed “spasticity.”

Most adults have not had diagnostic studies which would corroborate a diagnosis or provide an etiology which might dictate treatment. If one exists it is usually metabolic abnormalities or a progressive encephalopathy. Most adults have not had imaging studies such as a CT scan of the brain or MRI to correlate pathology with physical deficits and other pathological processes.

The central feature of CP is neuromuscular problems of movement, function and balance. This may appear as gait problems, balance problems, asymmetries, secondary contractures, scoliosis, spasticity and clonus. It is fairly standard that over time, movement and positioning problems progress. It is not unusual for an adult to have progression that interferes with function or activities of daily living. This usually manifests itself as contractures or fatigue, secondary to the extreme effort required for activity, mainly walking. Many people with CP, who were independent ambulators when young, elect to use wheelchairs in their 30s because it is too hard to walk. These patients are beginning to report symptoms in their 30s and 40s that are often seen in people much older. Their energy levels are declining, muscles are getting stiffer, joints sorer and problems with balance and coordination are increasing. Degenerative hip disease and dislocation are common which result in increased pain when sitting and/or standing. Foot deformities may progress and interfere with standing and walking. Scoliosis occurs in 25 to 64 percent and progresses with growth and asymmetry but is usually stable by adulthood. Other secondary deformities such as scissoring or windswept hips cause discomfort. Obviously, the risk of pain is high with advancing age, and nonverbal patients cannot complain.

The secondary physical problems may contribute to problems with daily living or even the ability to obtain health care. In addition to the problems with sitting, standing and walking, people with CP may have functional issues with hand use (buttoning clothes, cutting meat, shaving, brushing teeth) and hygiene (toileting, menstruation, bathing).
Some necessary help with these tasks may affect privacy and self-image. Inability to cooperate with procedures and medical examinations may contribute to missed diagnoses or lack of screening procedures. If a special examination table is unavailable, staff may help with positioning and preparation. The staff supporting people in the Waiver system who have physical challenges have all been trained in physical management with the person they support, and they can help (e.g., they can undress them and help them onto the examination table). Alternate positioning for some exams and procedures may be necessary, and it would be a good idea to have office staff consider issues of access to the office.

For some adults with CP, aging intensifies problems that did not cause difficulty at a younger age. Physical decline often begins in the 30s with premature deterioration of their musculoskeletal functioning. These patients begin to become less able to exert the extra effort required for movement. Pain and increasing tightness around hips and knees may result in increased crouching (inability to stay upright), and they may stop independent ambulation. Asymmetries like hemiplegia or hand weakness may become more prominent and affect function with loss of abilities. An arthritis type picture may develop, and discomfort from contractures and limitation of movement may cause pain. Back pain is more common and more difficult to treat than in the general population. Carpal tunnel syndrome is common, as well as an increase in problems with spinal stenosis. Progressive hip dislocation and early hip degeneration are possibilities. Scoliosis may become more pronounced due to asymmetries of tone creating progressive orthopedic disability. Most of these problems are not amenable to surgery, and pain is often not addressed. Most adults with significant contractures and deformities, when referred for orthopedic surgery, return with a recommendation of “no surgery as risks outweigh benefits.” Most orthopedists have had little training or exposure to adults with CP. These individuals should be referred for evaluation to the regional Assistive Technology Resource Centers where they can be seen by experienced Waiver therapists.

Persons with CP also have increased problems in other body systems which progress over time. 25 to 39 percent have vision problems, and 8 to 18 percent have hearing problems. Extremities may be cold or swollen due to abnormal vascular reflexes. Aspiration, due to poorly coordinated muscles in the pharynx, may cause choking and drooling, and result in chronic aspiration. There is a high incidence of GERD, esophagitis and constipation. Osteoporosis is common even at younger ages. Seizures are common (reported in 30 to 60 percent of people with CP) and occur more often in patients with greater brain damage and those with hemiplegia. Dental issues are significant, secondary to both oral deformations and difficulty brushing teeth. Life expectancy may be shorter than the general population and can be related to the type of CP. Of the common CP descriptions, those with quadriplegia are likely to have the shortest lifespan, followed by those with diplegia, then hemiplegia. Those with the choreoathetoid type have the longest lifespan on average. There is also an increased rate of death from cancer and heart disease at a younger age. This is felt to be due to delay in diagnosis, secondary to communication issues, and difficulty with physical examination. Most people with CP rarely exercise. Under the auspices of the United Cerebral Palsy Association, a group of adults with CP, along with research scientists and physical fitness experts developed a set of exercise guidelines available on the UCP website, www.ucp.org. The benefits of exercise for people with CP are the same as that for the typical population.
Treatment of Spasticity

Treatment of spasticity in people with ID may be very helpful in reducing discomfort and increasing function. Treatment is likely to be more effective and long-lasting if a careful plan is developed. This should be addressed in an interdisciplinary fashion and involve a therapist knowledgeable of disabilities. (Waiver therapists have vast experience, and DIDS therapists can offer consultation to persons in the Waiver system.) The underlying diagnosis and treatment goals need to be specific, and effects and side effects must be followed on a regular basis.

To consider spasticity treatment:

1. Spasticity must be documented to be primary spasticity. For example, individuals with tight muscles secondary to prolonged sitting do not have primary spasticity. In some situations, however, short-term treatment combined with physical therapy may be beneficial.

2. Goals must be specific and measurable as well as reasonable. For instance, increased range of motion is not a reasonable goal with someone who already has fixed contractions.

3. Development of a program should involve:
   a. Consideration by the PCP after discussion with the Intellectual Disability Team (IDT)
   b. A thorough and accurate physical therapy evaluation
   c. Development of goals of treatment, including treatment effects, means to follow the effects, and a way to follow side effects.

4. If an oral medication trial is elected, these steps should be followed:
   a. A plan with clear responsibility should include a basic diagnostic evaluation, goals of treatment, and plans for treatment, following effects of treatment and following side effects.
   b. Medication is gradually introduced with thorough evaluation in each stage.
   c. Treatment needs to be followed regularly. Ideally, a physical therapist or other therapists should follow progression towards goals, and the PCP should monitor the side effects.
   d. It is imperative that the PCP follow the effects on seizures, GE reflux, and psychiatric/behavioral programs, as well as other side effects encountered with medications.
   e. If it is decided to taper off long-term medication use, the taper should be slow. The evaluation should continue with withdrawal and to determine if there is a change. The effect of medication may be better appreciated after decreasing or discontinuation if the effect was subtle. At this time, if there is felt to be a positive effect of the medication, the medication may...
5. If oral Baclofen side effects are a concern, the patient may be referred for an intrathecal Baclofen trial. Stopping Baclofen without tapering may result in withdrawal seizures.

6. Use of Botox needs to be discussed with a health care professional, usually by a physiatrist or neurologist, who is qualified to administer the treatment. It is also important to remember that while treatment is ongoing, patients usually need injections every three to four months. This treatment is often valuable in short-term use with therapies such as inhibitive casting. This treatment should also have a plan with functional or medical goals and monitoring. It’s important to remember that correction of spasticity with Botox has little effect on fixed contractures.

**Cancer**

Carcinomas and other proliferative diseases occur in persons with ID as in other populations, but there are some specific situations which require unique approaches

Individuals with DS have a lower incidence of solid tumors (e.g., lung and breast) than the general population. Recent research has shown some protection on a genetic basis (such as an extra copy of cancer-suppressing genes with 3 copies of chromosome 21). Infants and young children with DS (with an incidence of about 1 in 500) are at risk of leukemia. There is NOT an increased incidence of leukemia in adults with DS. Males with DS have an increased incidence of testicular cancer.

Some syndromes are associated with high cancer risk. An example of this is Tuberous sclerosis (TS), where tumors occur in greater numbers in several organs. A patient with TS should have a surveillance plan for detection. Other syndromes such as Neurofibromatosis also have a risk of certain kinds of cancers and should have regular surveillance.

Some medical issues are associated with a higher incidence of cancer. People with ID have frequent reflux, and appear to have a higher risk of Barrett’s esophagus and subsequent adenocarcinoma. Long-term use of indwelling bladder catheters is also associated with an increased incidence of bladder cancer. Chronic constipation may also be associated with a higher incidence of colon and rectal cancers.

Patients with ID have a right to treatment and should not be denied treatment "because they are mentally retarded." Because these individuals have support staff, they may actually be more compliant with treatment measures. There is no evidence that people with ID respond less effectively to treatment than the general population. Issues of determining quality of life, response to treatment and personal wishes are difficult to decide for another person. This may pose problems in making decisions for people with ID whose caregiver is a family member or guardian. Individuals without family or a
For a variety of reasons, patients with disabilities often do not receive regular screenings, like Pap smears, colonoscopies or prostate exams, which can detect early, treatable forms of cancer. They should have regular screenings as recommended for the general population. It may be necessary to make modifications for testing procedures. Patients may need desensitization or sedation. It may be necessary to modify positions (e.g., lateral sideling for a vaginal exam in a woman with CP). Breast exams may be done by ultrasound rather than mammogram. Sedation may be needed as well as extra time in some situations. Exams may be done under general anesthesia, but that should not be done on a routine basis. If general anesthesia is used, an attempt should be made to combine multiple examinations if more than one is needed.

Obstacles to procedures should be discussed with the Circle of Support (COS) and even perhaps with VSHP. For instance, a CT scanner for morbidly obese people may not be locally available, but the patient can get the procedure elsewhere.

**Hydration Issues**

Many individuals with ID do not have an adequate fluid intake. This is usually a consequence of the inability to ask for fluids or physical limitations that preclude free access to fluids. In some cases, individuals will avoid liquids if they have problems with choking. Conversely, they may be on thickened liquids as part of a dining plan. Some individuals find this unpalatable and do not consume adequate amounts.

Obviously, inadequate fluid intake predisposes the patient to dehydration and associated symptoms if not outcomes of morbidity and mortality. In addition, chronic dehydration affects concentration levels of antiepileptic drugs which may result in frequent dosage changes secondary to erratic blood levels. Dehydration also may contribute to problems with constipation.

**Fall Prevention**

Falls are the leading cause of injury death, and the number one cause of non-fatal injury among older adults. They are associated with considerable medical costs, loss of mobility and a reduction in independence. Falls are also a predominant cause of serious injury for persons with ID. Many individuals with ID tend to experience the aging process at a faster rate, and often experience challenging risk factors such as impaired cognition, impaired mobility, aggressive or impulsive behaviors, and inadequate protective reflexes or reactions, all of which increase the likelihood of a serious injury occurring during a fall.

According to incident data compiled through the Waiver program, falls are the third most frequent type of incident reported to DIDS. Furthermore, falls accounted for approximately 46 percent of the total number of incidents resulting in serious injuries. Falls and serious injuries resulting from falls tend to occur more during business hours on weekdays than at other times. They also tend to occur more in the service recipient’s home than in other locations. Fractures of extremities (e.g., ankle, wrist, arm, fingers) and lacerations, especially in the head and face area, are the most frequent serious injuries associated with falls.
In addition, falls that do not result in injury and “near falls” are of significant concern. Statistics show that once a person falls, there is an increased risk of the person falling again. In addition, the person may develop a fear of falling which may significantly impact his or her activity level. Finally, direct support professionals and family that support a person who is at risk for falling may also sustain injuries trying to help “break” the person’s fall, or when helping him or her to get up after a fall.

Over the past several years, there has been increased focus on risk management within the Division of Intellectual Disabilities Services (DIDS). Risk assessment is part of annual pre-planning, as well as an ongoing process, for every person receiving DIDS supports and services. When falls are identified as a risk factor for a person, there are a number of resources available through the Waiver program to help the person and his/her planning determine the reason for the risk, and ways to reduce the risk of falling or prevent future falls.

Prevention is a key factor in managing risk for falls. Developing a Fall Prevention Plan may help reduce injury by focusing on preventing falls, reducing the number of falls, or decreasing the number of serious injuries due to falls. An extensive amount of information is available regarding causes of falls, risk factors that increase the likelihood of someone falling, and ways to prevent falls from occurring. Waiver therapists may help with developing a Fall Prevention Plan:

The PCP should be aware of falls or near falls, and the VSHP nurse case manager can help facilitate this. Because falls may be associated with medication side effects, a new onset diagnosis, or evidence of health decline, the PCP should consider a medical workup to search for a treatable cause. Sometimes falls are not considered by support staff because “this is how he/she is.” The PCP may have to ask for this information directly if there is a concern, or the RNCM may have concerns.

**Resources**

*DMRS Fall Prevention Resources*, available from DIDS.

**PRN Medication Use**

PRN medication use is a standard medical practice in developing an approach to treatment, mainly to address symptoms. In the general population, a person makes his own decisions about this approach and might not even discuss it with his PCP. When people are in the care of others as they are in the Waiver system, things are a little different because of liability issues and the risk of “practicing medicine without a license.” Waiver regulations exist to address these issues. Use of PRN medications must be supported with a written prescription from the PCP (even for over-the-counter medications), and must be written very specifically so support staff do not have to make medical judgments. An example would be: instead of writing a prescription for the patient to take 1 or 2 tabs every 4-6 hours, the order should read one tab every 4 hours or 2 tabs every 6 hours. This is a little more work for the PCP but, in the end, protects the patient – and may help promote monitoring of the use of PRN medications.
Another issue which may require understanding is the use of PRN psychotropics. Because this population has, in the past, been subject to chemical restraints without clear indication, the Waiver system monitors the use of PRN psychotropics closely. Although PRN psychotropics may be used, orders must be very specific so nurses do not have to diagnose, and the patient must be evaluated by a nurse before the medication is given.

Another area of frustration is that, in the Waiver system, service recipients require an order for anything used which may have a physical or medical component. This, unfortunately, includes the use of sunscreen, lip balm, special shampoos, etc. This requirement may often be less frustrating if these needs are discussed at the annual evaluation, and done all at once. Be cautious, however, about “routine” PRNs. It might seem efficient to have lots of available PRNs (cough, temperature, fever, etc.) for convenience for both the agency and the PCP. However, this process may create problems by missing critical early symptoms if given multiple times for an illness needing medical attention, or used frequently without the PCPs knowledge to integrate into overall health issues. Examples of this would be several days’ use of PRN medications for diarrhea while the patient develops serious dehydration or use of cough medicines two or three times a week which may be a symptom of reflux. If PRNs are used, orders should be very specific as to when to contact the PCP.

**Lifestyle Issues**

People with disabilities are more susceptible to compromised health status and preventable secondary conditions. Health and wellness are the foundations that allow people to participate in many of the most important aspects of life. Current approaches to wellness recommend a variety of activities and lifestyle changes to achieve optimum health. Unfortunately, the information, practices and resources needed to realize a healthy lifestyle are not available for most people with disabilities.

Many health care and service providers do not address fitness and lifestyle issues for persons with ID. Community resources may not be easily accessible. Health promotion campaigns have largely neglected people with disabilities, resulting in less access to health promotion and maintenance than the general population. There are three main areas of importance for the health of people with ID: physical fitness, combating obesity and smoking cessation.

Special Olympics are available to people with ID at all functioning levels. These programs provide year-round activities. As an added bonus, Special Olympics have health screenings under the Healthy Athlete Program to address sensory issues, dental problems, and assess foot and other problems. These screenings are done by volunteer professionals and may be valuable for integration into a person’s overall plan of health care. In addition, Waiver therapists are available for consultation to address barriers to physical activity programs. Local YMCAs usually make allowances for people with ID which may include allowing their support staff to accompany them. Many private practitioners (e.g., yoga) will also make adaptations to work with people with disabilities often at low or no cost. A little push in the right direction with fitness recommendations by the PCP can go a long way.
Obesity is also a significant issue in this population as it is in the general population. While some issues may be medical (e.g., hypothyroidism in DS, metabolic syndrome in persons treated with psychotropics) and possibly amenable to treatment, a plan developed by the COS with the PCP is beneficial. It may address appropriate activities or change the patient’s present sedentary activities. United Cerebral Palsy (UCP) has developed exercise programs for persons with CP and physical disabilities. Because many people have neuromuscular issues, discussion of the possibility of pain and limitations with exercise should be initiated by the PCP. The PCP may also ask the patient’s support staff (with help from Waiver therapists) to develop a reasonable diet to promote weight loss. If behavior issues involve food, a behavior analyst may be of help.

Many people with ID who have behavior problems also have problems with smoking and pica involving cigarette butts. As with the general population, the PCP can help with a smoking cessation plan which might include appropriate medication support. It may be that this represents some sort of “self-treatment” like nicotine, and an evaluation and other treatment might be considered.

**Weight Issues**

Weight issues may have a significant impact on persons with disabilities. These issues may be due to obesity or situations in which a patient may be significantly underweight and nutritionally compromised.

For the most part, people with ID become overweight through the same mechanisms as the general population (e.g., increased calories, lack of exercise). However, in addition to weight gain associated with taking psychotropic medication, there are two other scenarios of excessive weight often seen in this population: hypothyroidism and Prader-Willi, or other syndromes with both obesity and ID.

Hypothyroidism is very common, especially in persons with DS (approximately 25 percent) and should be considered in any overweight patient.

Prader-Willi syndrome and obesity may become life-threatening with significant cardio respiratory compromise (Pickwickian syndrome). One of the cardinal features of this syndrome is hyperphagia because of defects in leptin protein hormone that plays a key role in regulating energy intake and energy expenditure, including appetite and metabolism. These individuals never feel full, and cannot stop eating. They may develop behavior issues around food, and it is not unusual for them to forage in the garbage. Some may need to have kitchen restrictions with locks on cabinets and the refrigerator. These restrictions may require a presentation at the Waiver Human Rights Committee to approve their use and will require PCP support. The Waiver system can provide services of the behavior analyst to address food-seeking behavior. Diet plans and an exercise program will be important, and development of these supports can be assisted by Waiver therapists.

There are also many individuals in the Waiver program who have issues with weight loss, being underweight and being fed by enteral tubes. This group of patients is most often complex medically and physically compromised. These individuals require a thorough medical workup, although the most common causes for weight loss are
dysphagia, GERD and hyperactivity. Waiver professionals may assist and support the PCP in diagnosis and treatment.

**People Who Are Deaf/Blind**

People who have dual sensory loss of vision and hearing are a special population among those with development disabilities, and often have associated ID. Impairments of vision and hearing obviously present difficulties in communication, which could impact health care. These disabilities also affect support persons helping the individual function in and enjoy life. In many individuals, some sensory abilities are still present but diminished; these people may benefit from vision support and hearing aids. Etiologies are often concomitant with ID, i.e., Congenital Rubella Syndrome. There are many adaptations and treatment approaches which may help the deaf-blind person. These include touch cues and gestures which may be helpful for medical examinations, procedures and other adaptations. DIDS, through the Waiver program, has specially qualified therapists to help with training, education, evaluation and treatment, and may be of value to the PCP in caring for someone with this special disability.

**Transitions**

Transitions are hard for anyone, and this is especially true for persons with ID. Transitions may involve moving to a different agency, changing doctors, going to the hospital or other events. All of these transitions involve medical considerations. Although it is the responsibility of the support agency to transfer medical records, this is often overlooked. The PCP can help by asking for previous records. This is an area where the Nurse Care Manager (NCM) may be of help.

Transition to and from the hospital should also follow a formal procedure. Most agencies provide basic, including health, information if a patient goes to the ER, or is admitted to the hospital. The PCP should be aware of this and could possibly share important information and data that would help with care in the hospital setting.

The PCP may not be the treating physician while the patient is hospitalized. If this is the case, the PCP should be asked to be notified when the patient is discharged. The PCP should encourage agencies to use a medication reconciliation process for hospital discharge and all transitions.

**Sleep Problems**

Patients with ID are often brought to the PCP with a complaint of a sleep problem. Before writing a prescription, it is important to evaluate the situation. Waiver behavior analysts can help with data collection which should start with a 24-hour chart done in hourly increments. This allows one to see a bedtime. People going to bed at 8 p.m. may awaken at 4 a.m. after 8 hours of sleep, which is not a sleep problem. The chart may also track daytime sleep often seen with sleep apnea, breaks in sleep and overall consistency in sleep. Ideally, people should go to sleep and awaken at the same time every day. A sleep diary may also include other events such as the time of medications, food and drink consumed at bedtime, moods and behavior at bedtime, etc.

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Ask if the person is awakened at night, e.g., to go to the bathroom. An environmental survey should be done to ask where the person sleeps, if there is anyone else up at night, etc. A review of systems should be done to look for any medical cause which may affect sleep such as GERD, urinary frequency, medication changes, positional discomfort due to contractures, pulmonary symptoms, stool incontinence (requiring awakening to address by staff), signs of pain, etc. It may also be revealing to see if there are any daytime behavior changes associated with the sleep problems. One should also ask about snoring, leg movements during sleep, and signs of narcolepsy.

The hallmark symptoms of insomnia are:
- Difficulty falling asleep—tossing and turning for an hour or more
- Waking up at night and being unable to go back to sleep
- Waking up too early in the morning
- Feeling un-refreshed upon awakening
- Daytime sleepiness, irritability or anxiety

After a diagnosis is made, treatment can address the medical cause, environmental issues, or develop a sleep hygiene program (consistency, rituals, environmental changes, etc.). If medications are used, patients should be reviewed regularly as most sleep medications are not recommended for long-term use and have side effects. Solving problems with a pill looks appealing, but, in general, medication does not cure insomnia and can often make things worse in the long run. Concerns with using sleep medications are:
- Drug tolerance
- Drug dependence
- Withdrawal symptoms
- Side effects
- Drug interactions
- Rebound insomnia
- Masking an underlying problem
- Interfering with other therapeutic treatments

Most OTC preparations for sleep problems have an antihistamine as their main ingredient. Antihistamines can cause:
- Drowsiness the next day
- Dizziness and forgetfulness
- Clumsiness, feeling off-balance
- Constipation and urinary retention
- Blurred vision
- Dry mouth and throat

Prescription sedatives or hypnotics such as benzodiazepines pose problems for people with ID. Chronic use of benzodiazepines can be troublesome for several reasons:
- Physical and psychological dependence
- Loss of effectiveness as brain receptors become less sensitive to their effects
- Overall reduced quality of sleep (less restorative deep and dream sleep)
- Rebound insomnia after discontinuance

Some newer medications are not benzodiazepines and are thought to have fewer side effects and a lower risk of dependency. Trazodone, approved by the FDA for treatment
of depression (not insomnia), is the most commonly prescribed medication for the treatment of insomnia. It might raise the possibility of a diagnosis of depression, and it has a small, but significant, risk of suicidal thoughts or worsening of depression. Some studies have shown trazodone to be effective for short-term use.

Melatonin is a medication which affects the circadian rhythm. It has been reported as being effective in treating jet lag and has been shown to be effective in children with autism. There are no studies on the effect of long-term melatonin use.

Restless leg syndrome also occurs in people with ID and may have many possible causes including kidney failure, nerve disorders, vitamin and iron deficiencies, or be a side effect of some medications such as antidepressants. About 50 percent of people who have restless leg syndrome have relatives with the condition if a family history is available.

**Adults with Congenital Heart Problems**

Congenital Heart Disease is the most common birth defect in the U.S., and is a component of many genetic syndromes including those with ID such as DS, Williams syndrome, Velo-cardio-facial syndrome and others. The past 60 years have brought dramatic surgical and medical advances, and many children with congenital heart disease survive into adulthood. There are many patients in the Waiver system with a uniquely wide spectrum of cardiac defects, with varying degrees of severity and prognosis, and who are at risk for complications. The most common congenital heart disorders affecting adults are congenital valve defects, atrial and ventricular septal defects, and patent foramen ovale.

Congenital heart disease complications may not develop until years after initial treatment. Some common problems or complications that may develop in adulthood include:

- Arrhythmias
- Heart infections (endocarditis)
- Stroke
- Heart Failure
- Pulmonary Hypertension
- Heart valve problems

Symptoms or signs of congenital heart disease may not show up until later in life. They may recur years after treatment for a heart defect. Some typical congenital heart disease symptoms that may occur in adults include:

- Abnormal heart rhythms (arrhythmias)
- A bluish tint to the skin (cyanosis)
- Shortness of breath
- Tiring quickly upon exertion
- Dizziness or fainting
- Swelling of body tissue or organs (edema)
- Poor exercise tolerance
Treatment of Adult Congenital Heart Disease is based on the severity of the congenital heart disease. Some mild heart defects do not require any treatment. Others can be treated with medications, invasive procedures or surgery. Most adults with congenital heart disease should be monitored by a heart specialist and take precautions to prevent endocarditis (an infection of the heart) throughout their life. Those with congenital heart disease are at risk for getting endocarditis, even if the heart was repaired or replaced through surgery. Antibiotic prophylaxis is important for dental work and other invasive procedures. For complicated cases, special clinics for adults with congenital heart disease are available, usually situated at medical schools.

Resources

**Adults with Intracranial Shunts**

Many developmental disabilities (especially Spina Bifida) have associated hydrocephalus and are treated with intracerebral shunts in early childhood. As a general rule, these shunts are felt to be nonfunctional by the time the child grows up, but there is no way to be sure. If the shunt is still functioning, the adult is at risk for shunt malfunction which can be a medical emergency. Another compounding issue is that often the shunt is “forgotten” and is not even listed as a diagnosis. Shunts are easy to miss on the physical exam. Hints of a shunt are the presence of a subcutaneous tube in the neck and a soft small “bulb” somewhere on the scalp, usually laterally. Shunt systems come in a variety of models but have similar functional components. Catheters (tubing) and a flow-control mechanism (one-way valve) are components common to all shunts.

The parts of a shunt are named according to where they are placed in the body. The portion of the tube which is inserted into the ventricles is called the ventricular catheter. The peritoneal catheter is the portion of the tube which passes the cerebrospinal fluid (CSF) into the abdomen (peritoneal cavity). If the tube is placed into the right atrium of the heart, it is called the atrial catheter. The valve regulates the pressure of the CSF flow, and prevents backward flow of spinal fluid toward the ventricles. Many shunt systems also have a flexible flushing chamber (reservoir) which may be housed within the same unit as the valve, or may be a separate unit along the shunt, depending on the design of the shunt system. In children, this reservoir may be depressed to assess shunt function, but this is rarely helpful years later.

Shunt malfunctions can occur gradually or suddenly which may create an emergency situation. A missed diagnosis can result in permanent neurological sequelae or even death. Symptoms of shunt malfunction are headaches, lethargy, swelling at the shunt site, vomiting, irritability, loss of function, vision changes, or loss of coordination or balance. A shunt infection can also cause the shunt not to work properly, and cause CSF to back up, leading to enlarged ventricles. Signs and symptoms of shunt infection also relate to signs of shunt malfunction. Significant constipation in patients with shunt-dependent hydrocephalus may often be enough to bring a sub-clinical shunt malfunction to clinical attention, or be the cause of temporary distal peritoneal shunt malfunction.
The treatment of the constipation may address the symptomatic shunt dysfunction so as to avoid operative intervention.

There are no specific guidelines for treating a shunt placed during childhood in the adult, but if there are concerns, the patient should be referred to a neurosurgeon. A shunt is never removed as tissue has grown around it and removal would cause damage. Since the shunt is a foreign body, some people may consider prophylactic antibiotics for dental work or other procedures.

**Resources**


**Skin Problems**

Among those supported in the Tennessee Waiver programs, the incidence of decubitus ulcers is actually quite low -- a credit to the care they receive. This condition is also monitored carefully by DIDS. When it does occur, it usually involves very physically challenged or medically complex people who have little subcutaneous tissue, or persons with Spina Bifida in their lower extremities, who have no sensory innervation. Decubitus ulcers may develop while individuals are hospitalized for acute illness. Wound care for this population is generic, and they may benefit from a referral to wound care clinics. Some people may also benefit from a referral to Waiver clinicians when skin problems are related to body position. Risk for ulcers can be determined by use of either the Braden Scale or the Modified Norton Scale.

PCPs may also note unusual skin patterns or pigmentation which may relate to genetic syndromes and provide clues to other medical problems. Commonly seen are:

- Portwine stains on face in Sturge-Weber syndrome where glaucoma and seizures may be present
- Café au lait spots seen in Neurofibromatosis where progression of tubers may present serious medical problems
- Streaky pigmentations found in Incontinentia Pigmenti where a variety of CNS and other problems may be present
- Butterfly-like nodular paranasal permanent rash seen in TS where seizures may be severe and progressive, and there is a high risk of malignancy.

These are only a few possibilities. Any peculiar permanent skin changes in a person with ID should prompt consideration of a genetic etiology which might outline risks for other medical problems.
Mottling, as well as cold extremities (different from Raynaud’s disease) due to autonomic causes are often seen in patients with significant Central Nervous System (CNS) problems. This rarely causes any problem or responds to any treatment.

Skin issues are often associated with enteral tubes and ostomies. These are most commonly hypergranulation, skin irritation, candidiasis and cellulitis. Hypergranulation is very common and may be treated with topical steroids, silver nitrate (which may be painful), or, occasionally, surgical excision. Skin irritations often have drainage and are unsightly (and usually not infected) but may respond to barrier treatment or tube size change. Cellulitis is fairly uncommon, but candidiasis may be a source of infection elsewhere.

**Modified Norton-Scale**

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<th>Readiness for co-operation / motivation</th>
<th>Age</th>
<th>Condition of skin</th>
<th>Additional diseases</th>
<th>Physical condition</th>
<th>Mental condition</th>
<th>Activity</th>
<th>Mobility</th>
<th>Incontinent</th>
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<td>none</td>
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</tr>
<tr>
<td>less</td>
<td>&lt; 30</td>
<td>scaly, dry</td>
<td>undermine of resistance, fever, diabetes</td>
<td>fair</td>
<td>apathetic</td>
<td>walk-help</td>
<td>slightly limited</td>
<td>occasional</td>
<td>3</td>
</tr>
<tr>
<td>partly</td>
<td>&lt; 60</td>
<td>moist</td>
<td>multiple ulcers, ulcers</td>
<td>poor</td>
<td>confused</td>
<td>chair-bound</td>
<td>very limited</td>
<td>usually</td>
<td>2</td>
</tr>
<tr>
<td>none</td>
<td>&gt; 60</td>
<td>wounds, allergic lacerations</td>
<td>artery occlusion</td>
<td>very bad</td>
<td>stupor</td>
<td>stupor</td>
<td>immobile</td>
<td>doubly</td>
<td>1</td>
</tr>
</tbody>
</table>

Please assign the condition of the patient to one of the 4 possible descriptions stated in the scale. The single points shall be added up. At a sum of 25 or less, a risk for pressure ulcers is existing. The less points reached, the higher the risk. In order to avoid the development of pressure ulcers, the necessary caring measurements shall be planned, initiated and executed.

**Risk for pressure ulcers acc. to modified Norton-Scale:**
- low (25 - 24 points)
- medium (23 - 19 points)
- high (18 - 14 points)
- very high (13 - 9 points)
Common Medical Problems in Patients with ID

The PCP who works with large numbers of people with ID, such as those who work with developmental centers or ICF/MRs, quickly recognizes that most medical problems fall into about ten different categories. Within the categories, many medical problems can be easily predicted once the PCP gains a little familiarity with caring for people with ID. One area of importance is that people with developmental disabilities may not present with early symptoms of common diagnoses, but with later symptoms, thereby creating confusion about the differential diagnosis (e.g., GERD presenting as anemia, constipation as ileus, and reflux as wheezing, etc.).

The ten most common categories of medical diagnoses are:

1. Nutrition and feeding problems
2. GI diseases
3. Constipation and bowel obstruction
4. Seizure disorders
5. Aspiration and pneumonia
6. Neuromuscular disorders
7. Musculoskeletal problems
8. Sensory impairment
9. Genetic syndromes
10. Behavior and psychiatric problems

Nutrition

People with ID have similar nutritional issues as the typical population – most notably a high prevalence of obesity. It may be that this is under-addressed secondary to the multiple persons supporting the individual, and the myth that obesity is “part of mental retardation.” There are also nutritional issues not often seen in the typical population that are unique to this population, especially those with genetic syndromes. In addition, there is a higher incidence of hypothyroidism, contributing to obesity, which is often overlooked. Because of the higher prevalence of nutritional issues, the Waiver system can be accessed to use dieticians familiar with both the special issues and how the system works. Division of Intellectual Disabilities Services (DIDS) has also developed a resource guide for the Waiver system.

Most people in the Waiver system have been regularly weighed, and support agencies can provide this information to the PCP. This is also helpful for the PCP who may not have the equipment to weigh persons in wheelchairs or someone who has difficulty standing on equipment in the office. Addressing obesity should follow the procedures for the typical population: beginning with a medical evaluation, creating a plan and monitoring progress. As a general rule, it is more effective to have a plan to lose weight gradually, paired with some sort of exercise plan; dropping overall calories precipitously may result in behavior issues. For, instance, a person freely eating food who might be consuming 3,000+ calories will likely fuss if placed immediately on a 1,000-calorie diet.

Because of underlying diagnoses and/or treatment, there are a number of people in the Waiver system that are either overweight due to the Metabolic syndrome secondary to psychotropic medications, or underweight secondary to complications of neuromuscular disease. The latter group is at risk of receiving an enteral tube. Care of this usually falls
to the PCP, but Waiver dieticians are an invaluable resource. Information about enteral tubes is available elsewhere in this handbook.

There are also several special nutrition issues in the Waiver program. There are a few people on specialized diets due to a metabolic problem (e.g., maple sugar urine disease-MSUD) who are mostly followed by specialized clinics. People in the Waiver system with PKU are rarely on the PKU diet since the diet only prevents brain damage early in life, and that period is long past for these people. People with DS are at a higher risk for Celiac Disease than the general population. Individuals with Prader-Willi syndrome are often overweight (some significantly and with complications) due to hyperphagia. Some individuals with hyperactivity, a common symptom in ID, have high calorie needs and may be underweight.

Some people who are underweight are prescribed supplements. There are a few issues to consider with this practice. First, if additional calories are the goal, this can often be accomplished by increasing regular food. A cheeseburger and fries or an ice cream sundae may be more palatable than the same supplement day after day. Giving supplements at meals may also not accomplish increasing calories if the person cannot eat everything. Orders such as “if 50 percent of meal not consumed, give supplement,” may be non-productive, particularly if the supplements begin to replace the regular meal and mask diagnostic symptoms. Most supplements are soothing for GERD, like antacids, and persons with GERD may prefer them over a regular meal. Treatment of the GERD is a better alternative.

**Resources**


**Dysphagia**

Dysphagia consists of difficulty swallowing and may occur in the mouth (oral), in the throat (pharyngeal), or in the esophagus (esophageal). This is very common in persons with ID, especially those with motor problems or who are the most medically involved. Since these are largely internal problems, they are often difficult to diagnose in someone who is non-verbal or has communication issues. Some factors that make dysphagia common in this population are:

- Neurologic deficits
- GERD
- Prolonged use of psychotropic and other medications
- Environment
- Previously learned behaviors
- Physical disabilities

Prolonged dysphagia, which is unrecognized, can result in serious problems such as weight loss, dehydration, aspiration, chronic pneumonia, COPD, choking, increased secretions, upper respiratory infections and otitis media.
In addition, drug-induced dysphagia may also be a problem. This includes drugs that have primary side effects like dry mouth with antihistamines, tri-cyclic antidepressants, or secondary side effects like drowsiness with benzodiazepines).

Patients may present with the following complaints:
- Frequent episodes of gagging, coughing or choking
- Coughing or choking during or after eating and/or drinking
- Increased time required for eating and/or drinking
- Spillage, drooling, nasal regurgitation
- Intolerance of certain textures
- “Gurgly” (wet) voice during or after eating and/or drinking
- Watery eyes during or after drinking
- Regurgitation, vomiting after eating
- Problems chewing and swallowing
- Storing food (“pouching”) in mouth
- Frequent “allergies,” upper respiratory infections or pneumonia
- Wheezing or “asthma”

The most effective way to diagnose dysphagia is by getting an oral motor evaluation usually done by an Occupational Therapist (OT) or Speech-Language Pathologist (SLP). This can be done by an experienced Waiver therapist. Definitive testing also involves using a variety of positions and food textures. Waiver therapists can help this procedure go smoothly.

The most common treatment modality is a dining plan which may be written, taught and monitored by an experienced Waiver therapist. This may include modifications of food texture (including use of thickening agents), positioning, feeding techniques, environmental modifications, adaptive utensils, or behavior modifications (e.g., fast eater). This approach treats the majority of patients. It is also important to remember that dysphagia may have an effect on medication administration and modifications may be necessary.

Occasionally, the patient needs to be Nil Per Os (NPO) but this decision is not to be made lightly. It is important to remember that all of us occasionally aspirate, and people are more likely to have problems when they are sick. It is not recommended to do a definitive evaluation while someone is hospitalized if it can be avoided. Individuals who are NPO can be fed by tube, but it is important to remember that tubes create a different set of problems and do not treat dysphagia “from above” (i.e., aspiration of oral secretions).

Long-term complications from dysphagia can be serious and even lead to death. Aspiration pneumonia is a common cause of death in all debilitated groups. Chronic microaspiration (sometimes asymptomatic) can lead to chronic lung disease (COPD), cardiopulmonary problems, and occasionally ARDS (acute respiratory distress syndrome).

**GERD**

Gastroesophageal Reflux Disease (GERD) is very common in individuals with ID but often is not recognized due to absence of information commonly used for diagnosis such
as lack of records and communication issues. Presenting symptoms may be those far
down on the differential diagnosis scale – such as anemia rather than heartburn.
Because this problem is so common, it is often productive to first rule out GERD before
embarking on more extensive investigations.

GERD may present with many of the symptoms seen in dysphagia, but most often the
presentation is weight loss, anemia or behavior problems, particularly in non-verbal
patients. Respiratory issues such as chronic upper respiratory infections, (review
frequent use of PRNs for “cough”), “bronchitis,” “asthma,” and even otitis media are often
secondary extra-organ (i.e., not the GI system) presentations. Sleep issues, such as
nightly discomfort from reflux (bedtime Benzodiazepines make this worse) are common,
as well as other behavioral presentations in attempts to communicate discomfort. Even
chronic constipation can exacerbate symptoms of discomfort with GERD.

There is an increased occurrence in the following situations:
1. Immobility and positioning - Lying in the supine position for long periods of time,
and lack of proper upright positioning, particularly during eating
2. Abnormal postures - Scoliosis, hip contractures
3. Neurological dysfunction - Esophageal motility and sphincter function is
controlled by the autonomic nervous system
4. Medication use - Medications may affect esophageal function, decrease saliva,
delay gastric emptying, promote constipation, and directly irritate the esophagus.
This includes, but is not limited to, theophylline, benzodiazepines, calcium
channel blockers, iron and opioid narcotics.
5. Excessive drooling - Saliva neutralizes stomach acids so this protective
mechanism is lost.

Significant symptoms suggesting reflux to watch for:
- Drooling
- Meal refusals, beyond the occasional occurrence
- Unexplained weight loss; individuals who remain significantly underweight even
with high caloric intake
- Coughing/agitation during the night
- Crying/irritability within 30 minutes of mealtime or during the night
- Nighttime awakening and seeking water/milk to soothe burning
- Self-injurious behavior
- Change in complete blood cell count: lab results show drops in hemoglobin levels
if there is bleeding from the lining of the esophagus or stomach. This may be
slow, and if unrecognized, may lead to a large bleed with significant blood loss
and drop in Hemoglobin/Hematocrit (H/H) prompting emergency treatment
- Hands in mouth (“hand-mouthing”)  
- PICA behaviors – craving to ingest any material not fit for food, e.g., starch, dirt,
clay, wood, paper, plaster, or cigarette butts)
- Rumination (bringing up a previously swallowed food and re-chewing it), or
periods of vomiting
- Chewing and swallowing movements outside of mealtime
- Body posturing – stiffening or changing position to relieve pain
- Noisy or wet respirations/“asthma” attacks
- Holding breath
• For individuals with feeding tubes, formula in the back of the throat, or formula on the breath
• History of antipsychotic medication use; current use of medications which have the side effect of relaxing the smooth muscle or delaying stomach emptying

Diagnosis and treatment of GERD for persons with ID is the same as any other person. The cornerstones are an eating and positioning plan, medication, and surgery, and, in some cases, enteral feeding. Many patients are treated empirically, but a significant number of persons show structural problems such as esophagitis, gastritis, esophageal narrowing, ulcers, and Barrett’s esophagus on esophagogastroduodenoscopy (EGD) probably due to a delay of symptom recognition.

Therapists in the Waiver program are quite experienced with GERD in persons with ID and may help the physician with both plans and monitoring. In addition, nutritionists are available to help with specific issues.

Non-pharmacological interventions include:
• Positioning – elevating head of bed, upright position after eating; therapeutic posturing while eating, including side lying
• Dietary changes – frequent small meals, not eating before bedtime; avoidance of certain foods such as high-fat meals and carbonated beverages
• Lifestyle changes – weight loss, smoking cessation
• Dining plan

Medical interventions include:
• Eliminating exacerbating medications – Benzodiazepines are some of the worst offenders
• Medications
  a. Acid suppressors – antacids contribute to constipation, cimetidine interacts with many medications, especially carbamazepine, causing toxicity; and many people on 24-hour preparations need a nighttime dose for breakthrough symptoms.
  b. Prokinetic agents – these also help with constipation which may increase GERD: reglan (consider the risk of tardive dyskinesia, especially in patients on psychotropics), erythromycin and colchicine.
• Surgery – Nissen fundoplication is very effective and may be done laparoscopically, except in individuals with significant body deformities. Paraesophageal hernias are rare but appear to be more common in people with ID in Tennessee. Corrective surgery has been very effective.
• Enteral feeding tubes. This decision should be carefully made, and Waiver personnel can help with data and decision making.

While tubes may solve some problems, they introduce a whole new set of problems like intestinal obstruction. It is important to understand that abdominal surgery has a significant long-term risk factor for intestinal obstruction and perforation. In addition, gastric tubes do not treat reflux, and in fact, there is a high incidence of development of reflux when tubes are placed for dysphagia. Complications such as skin infections, leakage, plugging and dislodgment occur. Tubes also need to be replaced periodically. If a tube is placed, it is imperative that a plan of care include problem solving for tube complications. Plans for
replacement, both acutely and routinely, should be available. Tubes put in by specialists are usually referred to the PCP for management. Most people in the Waiver system who get tubes do not have a terminal disease and live many years after placement.

Complications of GERD pose serious problems for people with ID since symptoms are generally not noticed in early stages. Esophagitis may cause bleeding and anemia, and may get worse over time in a non-verbal patient. It is also important to keep in mind that use of iron to treat anemia may also pose problems. Iron is very caustic, and many of these patients remain mainly in a supine position. Dysphagia may predispose iron pills to lodge in the esophagus, and liquid iron may be refluxed continuously. Strictures may be a consequence of esophagitis and may be severe enough to almost totally restrict flow of food and liquids. Barrett’s esophagus and subsequent adenocarcinoma is also a risk. Secondary respiratory problems with aspiration pneumonia and COPD are common. Non-GI symptoms of GERD such as coughing, wheezing or otitis media are often overlooked and can be misdiagnosed.

**Enteral Tubes**

Enteral tubes are usually placed in adults for terminal conditions, and in children for growth and development. Tubes placed for specific conditions are often followed by a specialty feeding team, usually hospital-based. Tubes are placed in people with ID primarily for GI complications of dysphagia and reflux, and are for maintenance. The person is not terminal and goes on with his or her life. These individuals are usually referred back to their PCPs for care, usually without a plan of care for the enteral tubes. Unfortunately, the average community doctor is not familiar with tube care complications and feeding regimens, nor does the average dietician have extensive experience in these areas beyond nutritional advice. Care of enteral tubes requires both a plan of care and an ad hoc team. This is an area where Waiver clinical personnel may be of significant help.

Nutritional risk factors which might be appropriate for treatment with an enteral tube include:

1. Underweight: <90 percent IBW
2. Significant weight loss: 5 percent in 6 months, 10 percent in 1 year; >5 lb./month, >2 lb./month for three successive months
3. Increased metabolic needs (e.g., sepsis, surgery)
4. Nutrient loss secondary to persistent nausea, vomiting, diarrhea or malabsorption
5. Frequent NPO or clear diet
6. Chronic disease (e.g., COPD, cancer, renal failure)
7. Delayed maturation or non-menopausal amenorrhea
8. Known GERD or severe chronic constipation
9. Increased caloric needs secondary to activity, behavior or neurological movement disorder
10. Severe dysphagia
11. Decreasing laboratory nutritional parameters – cholesterol, iron, total protein/albumin
12. Abnormal (decreased) laboratory parameters
13. Decubiti
14. Dehydration

Enteral tube use in persons with dementia should follow general guidelines, although support may be necessary to understand the effectiveness of the procedure and quality of life issues.
**Tube Placement Options**

**Nasoenteric Feeding Tubes**

Tubes are passed through the nose to various points in the GI tract and are named with reference to the location of the terminal end of the feeding tube. Examples include nasogastric, nasoduodenal, and nasojejunal tubes.

**Advantages**
- Avoids general anesthesia or surgical procedure
- Low incidence of complications

**Disadvantages**
- Risk of aspiration (may be less with nasoduodenal and nasojejunal tube)
- X-ray confirmation of correct tube placement required
- Suited only to short-term (less than two weeks) use; long-term use in people with disabilities usually leads to complications

**Tube Enterostomy**

Tubes are placed either laparoscopically, operatively or percutaneously. Examples include esophagostomy, gastrostomy (PEG), percutaneous endoscopic jejunostomy (PEJ), needle catheter jejunostomy (NCJ), operative laparoscopic gastrostomy, and operative laparoscopic jejunostomy.

**Advantages**
- May be used immediately or within hours of placement
- May be used for long-term support
- May be used in presence of significant disease of upper GI tract (esophagus, stomach and duodenum)
- Percutaneously placed tubes avoid risks of surgery and general anesthesia
- Laparoscopic feeding tubes allow patients to return home the same day of procedure

**Disadvantages**
- May require endoscopy, abdominal ultrasound, or radiologic procedure with contrast media
- Endoscopy may be difficult or impossible in presence of altered anatomy
- Laparoscopically or operatively placed tubes require general anesthesia
- Potential for chronic wound complications

**Formula Selection**

Formula choice should be made by the physician with input from the dietitian. In general, isotonic feedings are best tolerated. Feedings should be started full-strength at a rate of 25 cc to 30 cc per hour. There is no need to dilute formulas that are isotonic. This was a procedure done in the past when formulas were hypertonic. Advance the
formula by 20 cc every six hours, being sure that gastric residuals are not above 100cc. Once the patient is stable at the desired rate, gastric residuals generally do not have to be monitored. Keep in mind that most tube feeding protocols in print were developed in hospitals for acutely ill patients. In long-term care, tube feedings are being given due to lack of ability to swallow not because there is GI dysfunction.

Tube feedings may be given via pump or gravity. The main difference is usually the ease of administration. However, in some cases of gastric paresis, dumping syndrome, and diabetes, the slow and controlled delivery of formula is necessary to prevent aspiration, diarrhea and blood sugar swings, respectively. With either type of administration, the set should be changed daily to prevent bacterial contamination of formula. Pumps must be kept clean to ensure proper operation.

Waiver dieticians are available to assist with formula selection and monitoring. The dietician should not write orders for times, amounts, or method (bolus v. continuous) as these decisions are based on medical issues.

Patients should initially be given caloric amounts based on a general formula according to weight and activity (may be calculated by the dietician) but adjusted according to individual response. Someone who exclusively receives nutrition by tube will gain or lose weight only with changes in total caloric intake. Fluids should be separately calculated, including fluids used for the flushing of the tube.

**Enteral Feeding Guidelines**

Nutritional issues should be discussed by the PCP/RN/RD team, and an integrated plan of care developed to include specific goals, intervention, and reevaluation until the individual has reached the goal or been placed on stable status.

Calculated nutritional needs and laboratory values should be reviewed regularly (biweekly, monthly, quarterly, annually) or as the individual’s status changes. A stable status may change with illness, new medications, change in feeding skills, behavior, environment, etc.

As a rule, most individuals do not need specialized formulas. If specialized formulas are ordered, it should be discussed by the PCP/RN/RD team, and the rationale should be well-documented.
Standard enteral formulas are in the following table:

### Standard Enteral Formulas

<table>
<thead>
<tr>
<th>Product</th>
<th>Calories /cc</th>
<th>Osmolality</th>
<th>Protein gm/L</th>
<th>Fat gm/L</th>
<th>CHO gm/L</th>
<th>Na+/K+ mg/L</th>
<th>Water cc</th>
<th>Calories to meet RDI for Vitamins/Minerals</th>
</tr>
</thead>
<tbody>
<tr>
<td>Ensure</td>
<td>1.06</td>
<td>555</td>
<td>37</td>
<td>26</td>
<td>169</td>
<td>840/1554</td>
<td>845</td>
<td>1000</td>
</tr>
<tr>
<td>Ensure w/ Fiber (12g)</td>
<td>1.06</td>
<td>500</td>
<td>37</td>
<td>26</td>
<td>176</td>
<td>840/1554</td>
<td>819</td>
<td>1000</td>
</tr>
<tr>
<td>Jevity Plus w/Fiber (12g)</td>
<td>1.2</td>
<td>450</td>
<td>56</td>
<td>393</td>
<td>1350/1850</td>
<td>810</td>
<td>1000</td>
<td></td>
</tr>
<tr>
<td>Ensure Plus</td>
<td>1.5</td>
<td>690</td>
<td>55</td>
<td>53</td>
<td>200</td>
<td>1050/1940</td>
<td>769</td>
<td>1420</td>
</tr>
<tr>
<td>Perative</td>
<td>1.3</td>
<td>385</td>
<td>67</td>
<td>37.4</td>
<td>177</td>
<td>1040/1730</td>
<td>789</td>
<td>1500</td>
</tr>
<tr>
<td>TwoCal HN</td>
<td>2.0</td>
<td>690</td>
<td>84</td>
<td>89</td>
<td>216</td>
<td>1460/2450</td>
<td>713</td>
<td>1900</td>
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</table>

### Oral Supplements

<table>
<thead>
<tr>
<th>Product</th>
<th>Calories</th>
<th>OSM</th>
<th>Protein</th>
<th>Fat</th>
<th>CHO</th>
<th>Na+/K+ mg/6 Oz.</th>
<th>Water H2O</th>
<th>Flavors</th>
</tr>
</thead>
<tbody>
<tr>
<td>Ensure</td>
<td>1.06</td>
<td>555</td>
<td>8.8</td>
<td>6.1</td>
<td>40</td>
<td>840/1554</td>
<td>845</td>
<td>Vanilla Chocolate</td>
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<tr>
<td>Ensure w/ Fiber (12g)</td>
<td>1.06</td>
<td>555</td>
<td>8.8</td>
<td>6.1</td>
<td>40</td>
<td>840/1554</td>
<td>33.1</td>
<td>Strawberry Butter Pecan</td>
</tr>
<tr>
<td>Enlive</td>
<td>1.25</td>
<td>840</td>
<td>10</td>
<td>0</td>
<td>65</td>
<td>65/40</td>
<td>191</td>
<td>Peach Apple</td>
</tr>
</tbody>
</table>

### Medication Pass Supplement

| TwoCal HN | 475 | 690 | 20 | 22 | 511 | 345/580 | 159 | Vanilla Butter Pecan |

If the individual is exclusively on enteral feeding, an evaluation must ensure the individual is receiving adequate vitamins and minerals, especially iron, calcium and Vitamin D. Refer to “calories to meet RDI for vitamins and minerals” column at the right. **Cal/cc x ccs per hour x hrs fed/day** should be equal to or slightly above the caloric nutrient base, otherwise consider vitamin supplementation or another formula.

As a general rule, if diet is adequate, supplemental protein does not usually result in an increase in blood protein and albumin levels. If protein or albumin levels are low and intake is adequate another medical cause for low total protein/albumin should be considered, such as liver disease.
Oral Supplementation
Nutritional issues should be discussed by the PCP/RN/RD, and an integrated plan of care developed to include specific goals, intervention, and reevaluation until the individual has reached the goal or is placed on stable status. In addition to caloric and protein needs, an individual’s risk factors and laboratory parameters should be assessed.

Oral supplements should be started only after careful evaluation by the PCP/RN/RD/OT/SLP team. Oral supplements should not be a substitute for scheduled meals, but offered to the individual in addition to their meals as ordered. Individuals with undiagnosed GERD often respond to supplemental formulas because they may mimic antacids.

Individuals needing nutritional supplementation may first be started on “60 cc of TwoCal HN QID” with medication. A goal (weight gain, improved protein status, etc.) should be established. Once the goal or the stable status has been reached, the supplementation program should be evaluated to reduce the volume of supplement necessary to maintain the individual status or discontinue the program.

Individuals who required supplements in addition to the above program should be evaluated by the PCP/RN/RD team and monitored regularly.

Laboratory Parameters
In addition to periodic weights and physical examination, periodic laboratory investigations related to nutrition should be followed. If placed on a flow sheet, multiple parameters are easily followed.

Important laboratory findings, including those suggesting need for evaluation are:

- **Cholesterol**
  - levels above 200
  - low levels should be evaluated: levels below 130 may be serious, and levels below 100 are evidence for homeostatic compromise

- **Iron**
  - a drop in serum iron precedes anemia
  - anemia is also associated with a decrease in ferritin, and increases in TIBC and absorption

- **Total Protein/Albumin**
  - albumin usually drops over time (half-life 21 days); prealbumin shows current protein status (half life-1.2 days); levels less than 17 are considered evidence of protein compromise

- **Alkaline Phosphatase**
  - elevated levels may be seen in bone disease (fractures, osteoporosis), as well as gall bladder and liver disease
  - levels should be elevated during periods of growth and may be correlated with bone age

- **Calcium and Magnesium**
  - bound to albumin and will show a concomitant drop in values

- **Dehydration**
  - suspicion should be followed with specific gravity; BUN may be falsely low in individuals with decreased muscle mass
Avoiding the Refeeding Syndrome

Refeeding syndrome is a series of fluid and electrolyte disturbances occurring within the first few days of increased caloric intake in a malnourished individual. Electrolyte disturbances, particularly hypophosphatemia, may occur with neurologic, cardiac, pulmonary, and hematologic complications. Some tips to avoid refeeding syndrome include:

1. Understand what refeeding syndrome is and the population it affects.
2. Monitor electrolytes prior to and after initiating oral, enteral or parenteral nutrition support
3. Vigorously replete any electrolyte deficiencies with special emphasis on potassium, phosphorous and magnesium levels
4. Maintain glucose control
5. Monitor pulse rate, intake and output carefully, while judiciously restoring circulatory volume
6. Assess caloric needs, ideally with a metabolic cart. Aim for meeting the measured needs first, and then slowly increase calories to assist in weight gain.
7. Administer vitamin and mineral supplements routinely

Administration Guidelines

1. Initiation and Progression
   a. To promote tolerance, enteral tube feedings should be initiated at rates of 50 cc/hr (or sometimes less) in adults. Most currently available formulas are isotonic (300 mOsm/L) and are well-tolerated at full strength when delivered into the stomach or small intestine. The rate of administration of isotonic formulas may usually be advanced in 20-25 cc/hr increments every eight hours until the goal rate is achieved. It is often more realistic to calculate goal rates based on 20-22 hours/day, allowing for interruptions in delivery.

2. Calculate Additional Free Water Requirements
   a. Most patients on enteral nutrition therapy will require additional fluids to meet minimum fluid requirements. To calculate additional fluid requirements, begin by determining the patient’s total fluid needs.

3. Transition to Cyclic or Bolus Feedings
   a. Hospitalized patients may initially benefit from a continuous infusion to establish tolerance to enteral nutrition therapy, and later transition to an intermittent infusion schedule. They may not be in the hospital long enough for this to occur, so it may need to be done after being discharged by the PCP. Intermittent infusion (bolus feedings) may be administered by gravity drip or syringe bolus for those patients with gastric feeding tubes. This is a more normal schedule for nutrition if it can be tolerated. It may also be adjusted intermittently, depending on activities – just as most people adjust meals to their daily lives. Cyclic feeding using a pump may also be used in patients with intestinal feeding tube sites (duodenum or jejunum). Cyclic feedings infuse formula for a set number of hours (e.g., 8-12 hours overnight). A cyclic feeding schedule should be considered for patients for whom free time off the pump is desired. Orders for bolus feedings should be adapted according to the person’s schedule. Variance for special situations, such as when the patient is away from home, should be discussed.
Methods of Delivering Enteral Feedings

Continuous Drip

<table>
<thead>
<tr>
<th>Advantages</th>
<th>Disadvantages</th>
</tr>
</thead>
<tbody>
<tr>
<td>Ability to increase volume of formula more</td>
<td>More expensive feeding method because a pump is required delivery</td>
</tr>
<tr>
<td>Rapidly</td>
<td></td>
</tr>
<tr>
<td>Improved absorption of major nutrients in</td>
<td>Restricts patient ambulation</td>
</tr>
<tr>
<td>infants with intestinal diseases</td>
<td></td>
</tr>
<tr>
<td>Reduced stool output in hypermetabolic patients</td>
<td>Less physiologic</td>
</tr>
<tr>
<td>Associated with a reduced incidence of vomiting</td>
<td></td>
</tr>
<tr>
<td>in infants with GE reflux</td>
<td></td>
</tr>
<tr>
<td>Greater caloric intake when volume tolerance</td>
<td></td>
</tr>
<tr>
<td>may be a problem</td>
<td></td>
</tr>
</tbody>
</table>

Intermittent

<table>
<thead>
<tr>
<th>Advantages</th>
<th>Disadvantages</th>
</tr>
</thead>
<tbody>
<tr>
<td>More physiologic because a normal feeding</td>
<td>Associated with a longer time to reach nutritional goals</td>
</tr>
<tr>
<td>schedule is mimicked</td>
<td></td>
</tr>
<tr>
<td>Less expensive because an enteral pump is not</td>
<td>Reduced weight gain and nutrient absorption in infants with malabsorption</td>
</tr>
<tr>
<td>Required</td>
<td></td>
</tr>
<tr>
<td>Greater flexibility in feeding schedule</td>
<td>Larger bore tube may be required for gravity administration</td>
</tr>
<tr>
<td>Freedom from infusion equipment</td>
<td>More nursing time required for administration than for pump-delivered feedings</td>
</tr>
</tbody>
</table>

Medications and Enteral Nutrition

The following are general guidelines for ordering and administering medications to patients receiving enteral nutrition:

- If the patient is able to take medications by mouth, the oral route is preferred over administration via the feeding tube.
- For certain medications that pose a particular problem for enteral administration (e.g., repeatedly clogs tube, unavailable in suitable liquid or crushable form, unpredictable absorption, etc.), alternative routes of administration that bypass the tube or even therapeutic alternatives should be considered. Alternative routes may include IV, IM, PR, SL or transdermal.
- For individual doses of most medications, the tube should be flushed with at least 30 mL of sterile water before and after administration. This serves to clear the tube for drug delivery, facilitates drug transport to the intestine, and indicates whether the tube is cleared. In general, all medications should be given separately.
- Most drugs in suspension, elixir or other liquid form are hypertonic. Highly concentrated drug solutions and suspensions should be diluted before administration to decrease gastric mucosal irritation and prevent osmotic diarrhea.
• For patients receiving many medications via the feeding tube, the volume required for diluting, flushing, and administering medications may be significant. Alternative routes of administration may need to be considered.

• Medications should never be added directly to the feeding formulation. The potency, stability, and availability of the medication, as well as the stability of the enteral formulation cannot be ensured.

• For most medications, the enteral feeding should be stopped for at least 15 minutes before and after drug administration. Certain drugs have increased bioavailability, produce more predictable blood levels, and/or are better tolerated on an empty stomach. Some drugs may require feedings to be held for longer intervals. For example, Dilantin administration requires the feeding be stopped for one hour prior to and after dosages. Multiple interruptions in formula delivery may compromise nutrition support and should be avoided. Other routes of drug administration may need to be considered.

• Stop gastric feedings ½ hour prior to and after treatment or procedures requiring the Trendelenburg position, (e.g., chest physiotherapy, central line insertion).

Complications of Enteral Nutrition Therapy

Patients should be monitored frequently for evidence of complications from enteral nutrition support. The following table lists potential complications of tube feedings, and offers suggestions for intervention.
### Complications of Enteral Nutrition Therapy: Possible Causes & Management

<table>
<thead>
<tr>
<th>Complications</th>
<th>Possible Cause</th>
<th>Suggested Management</th>
</tr>
</thead>
<tbody>
<tr>
<td>Diarrhea (&gt;4 BM per day or large loose stool)</td>
<td>Medications</td>
<td>• Eliminate antibiotics or antacids if possible</td>
</tr>
<tr>
<td></td>
<td></td>
<td>• Eliminate liquid formulations containing sorbitol</td>
</tr>
<tr>
<td>Fat intolerance</td>
<td></td>
<td>• Change to low-fat formula</td>
</tr>
<tr>
<td>Fat intolerance</td>
<td></td>
<td>• Stool culture for pathogens RX L. acidophilus/L. bulgarious (Lactinex if patients receiving antibiotics)</td>
</tr>
<tr>
<td>Bacterial overgrowth</td>
<td></td>
<td>• Discontinue current formula</td>
</tr>
<tr>
<td></td>
<td></td>
<td>• Replace bag and tubing using aseptic techniques</td>
</tr>
<tr>
<td>Contaminated formula</td>
<td></td>
<td>• Adhere to clean standard when changing or manipulating feeds</td>
</tr>
<tr>
<td>Osmotic overload</td>
<td></td>
<td>• Decrease concentration of formula</td>
</tr>
<tr>
<td></td>
<td></td>
<td>• Change to isotonic formula</td>
</tr>
<tr>
<td></td>
<td></td>
<td>• Further dilute hypertonic medications, administer medications by alternate route</td>
</tr>
<tr>
<td>Decreased bulk</td>
<td></td>
<td>• Change to high-fiber formula</td>
</tr>
<tr>
<td></td>
<td></td>
<td>• Administer bulking agents (e.g., psyllium) but not through small bore (10 French) feeding tubes</td>
</tr>
<tr>
<td>Nausea or vomiting</td>
<td>Patient position</td>
<td>• Position patient on right side to facilitate passage of gastric contents through pylorus; decrease volume; check residuals</td>
</tr>
<tr>
<td>Volume overload</td>
<td></td>
<td>• Decrease total volume</td>
</tr>
<tr>
<td></td>
<td></td>
<td>• Decrease delivery rate to one tolerated previously; advance delivery rate slowly over 12-24 hours</td>
</tr>
<tr>
<td>Delayed gastric emptying</td>
<td></td>
<td>• Stop feeding for two hours and check residuals</td>
</tr>
<tr>
<td></td>
<td></td>
<td>• Change to low-fat formula</td>
</tr>
<tr>
<td></td>
<td></td>
<td>• Administer prokinetic agent to stimulate GI motility</td>
</tr>
<tr>
<td>Specific nutrient intolerance</td>
<td></td>
<td>• Change to lactose-free or low-fat formula</td>
</tr>
<tr>
<td>GI tract obstruction</td>
<td></td>
<td>• Stop feeding</td>
</tr>
<tr>
<td>Constipation (no stool x 3 days)</td>
<td>Dehydration &amp; impaction</td>
<td>• Provide free water</td>
</tr>
<tr>
<td></td>
<td></td>
<td>• Remove impaction</td>
</tr>
<tr>
<td>Constipation (no stool x 3 days)</td>
<td>Decreased fiber</td>
<td>• Change to fiber formula</td>
</tr>
<tr>
<td></td>
<td></td>
<td>• Administer bulking agents (e.g., psyllium) but not through small bore feeding tubes (&lt;10 French)</td>
</tr>
<tr>
<td>Constipation (no stool x 3 days)</td>
<td>GI tract obstruction</td>
<td>• Stop feeding</td>
</tr>
</tbody>
</table>
Care of Tubes

Tube Complications

- Aspiration
- Buried bumper syndrome
- Colocutaneous fistula
- Gastric outlet obstruction
- Ileus
- Intraperitoneal Placement
- Necrotizing fasciitis
- Tube deterioration
- Bleeding
- Candida cellulitis
- Dehydration
- Hematoma
- MRSA skin infection
- Peritonitis
- Pneumonia
- Suture/wire breakage
- Tube displacement
- Pneumoperitoneum
- Wound infection
- Bowel obstruction
- Clogged tube
- Incisional pain
- Multiple punctures of stomach
- Small bowel perforation
- Subcutaneous emphysema
- Volvulus
- Stomal leakage

To ensure optimal usage, safety and patient comfort, enteral feeding tubes must be cleaned and maintained properly on a daily basis. There are many nurses in agencies who are experienced in tube care, as well as Waiver clinical personnel who can provide advice.

External Site Care

Proper care of the tube exit site is critical. Be aware of any labels or markings indicating an externally removable PEG tube. Direct care staff should understand procedures for avoiding positioning, manipulation, and protection issues to avoid tube dislodgement. Dislodgement requires immediate attention, and plans to address this should be in place.

Flushing the Tube

This is a daily procedure which helps prevent clogging and should be done on a regular basis. In general, only water should be used. While mildly acidic foods such as carbonated beverages or cranberry juice are commonly used to maintain feeding tube patency, recent research has shown that these acids can cause whole protein formulas to coagulate and clog the tube. As a result, some experts discourage this practice.

Tube Maintenance

The balloon-in-balloon gastrostomies require periodic maintenance. Saline, which may clog the access port, should not be used. Do not use air as it tends to seep out, causing the balloon to collapse, which can lead to migration and dislodgement. Under-inflation of balloons will not secure the tube properly, while over-inflation could cause the gastric lumen to be obstructed, or necrosis to occur.

Low-profile devices should have the auto-reflux valve periodically cleaned.
Nursing orders for care should be specific and include any measurements to verify positioning.

Dislodgement of tubes presents a problem; tubes should only be replaced by a clinician or nurse who is properly trained to do so. Remember that if a gastrostomy tube is pulled out, the tube tract begins to close in several hours unless a new tube is inserted into the gastrostomy. A plan to address removal should be in place. If the person is taken to the emergency room, it is best to provide a similar “back up” tube as most ERs do not stock all available tubes. If an “interim” tube is placed in the ER, the patient should be seen in the near future for definitive placement.

Clear orders and plans should be made for routine replacement including when, where, by whom, and what (which tube). A flow sheet to monitor tube replacement is often helpful.

For a person with ID who has reached stability with an enteral tube, the consideration of a gastrostomy button (commonly used in pediatrics when patients with tubes go on about their lives) should occur. This will allow the person greater freedom and reduce the possibility of the tube accidentally becoming dislodged with activity.

**Resources**


**Constipation**

Constipation is a very common for people with ID, especially those who are medically fragile and those on long term psychotropic medications. In a significant percentage, constipation may contribute to behavior problems (secondary to unrecognized discomfort) and GI complications (e.g., obstruction) which can lead to death. For many of these patients, constipation has been present for many years, can lead to serious morbidity, and can be a contributing factor in mortality.

The pathophysiology of constipation in adults with ID often starts in childhood. By the time the person is an adult, he/she has been on multiple and increasingly stronger laxatives long term. It is not unusual to see secondary megacolon, which poses additional risks for complications such as volvulus, obstruction and perforation. Some patients may develop secondary problems of prolapse, fissures and hemorrhoids. Occasionally, after many years, some will develop chronic pseudo-obstruction or even complete gut shutdown (Ogilvie syndrome).

Obviously, the best approach is prevention, but by the time this population presents to the PCP, constipation has been longstanding. In addition, other factors often contribute to the problem. Patients who are non-ambulatory may not have movement and gravity to help with good bowel habits. Many individuals do not have free access to water, and diets may be lacking in fiber. Some do not have the physical or cognitive abilities for good bowel habits or regular natural elimination. Constipation may result from a poor diet, poor bowel habits, or abnormalities in elimination of stool, whether physical, functional, or voluntary.
Risk factors in people with ID include:

- Poor diet, including low fiber and decreased fluids
- Poor bowel habits – ignoring the desire or opportunity may initiate a cycle of constipation
- Medications – especially antacids and calcium, antispasmodic drugs, antidepressants, iron, anticonvulsant drugs, pain killers containing narcotics, and/or other medications with constipation side effects
- Irritable bowel syndrome (an overlooked diagnosis in a non-verbal patient or in someone with primary bowel incontinence)
- Laxative abuse, generally secondary to long-term treatment
- Structural problems such as anal fissure, hemorrhoids, and stenosis, Hirschsprung’s disease, or strictures following earlier abdominal surgery
- Intestinal obstruction – volvulus is a risk with megacolon; people with abdominal surgery (gastrostomy placement, prior obstruction) are at increased risk
- Lack of movement and exercise, and persons who are primarily bedridden
- Lack of a consistent bowel movement plan
- Fecal impaction, anal fissures which may cause pain or discomfort
- Other medical diagnosis such as hypothyroidism, diabetes, renal failure; increase in GERD symptoms may be seen as well as UTIs (secondary to ureter compression and backflow of urine).

The cause is usually multifunctional. Constipation and impaction may present in unusual ways in people with ID. These are:

- Behavioral problems – discomfort can cause significant changes in behavior including sleep, SIB, tantrums.
- Chronic diarrhea may actually be secondary to constipation. This occurs when newly digested food flows around (overflow diarrhea) the impacted stool in the rectum.
- Anorexia – essentially patient “feels full,” and there is little room for more food
- Abdominal distention and ileus
- Vomiting, aspiration and hiccups
- Clinicians working with this population also see an increase in seizures associated with constipation
- Fecal smelling breath
- Increase in GERD symptoms may be seen as well as UTIs (secondary to ureter compression and backflow of urine)

Assessment may be difficult, and the presenting complaint may not be constipation. Therefore, in patients at risk, especially those on chronic laxatives, routine assessment may be prudent. The most valuable piece of information is bowel quality and frequency. Most individuals supported by residential agencies have regular recordings of bowel function, and these records should be requested for every visit.

Physical examination of the abdomen and rectum may reveal distention or even palpable stool especially in the right lower quadrant (RLQ). Rectal examination should be performed to assess rectal tone, feel for the presence of hard or impacted stool, and to test for fecal occult blood. Anal fissures and/or external hemorrhoids should be noted if present. In cases of severe constipation, a supine flat plate view may be helpful. This may be difficult in individuals with deformities and positioning problems, and should not
be used routinely to avoid excessive radiation. An evaluation should address the following:

- Predisposing medical problems
- Medications contributing to constipation
- Diet
- Fluids: intake, specific gravity, BUN (not valid with significant muscle atrophy)
- Positioning, activity
- Natural pattern: urination, bowel movements
- Presence of gastro-colic reflex

Treatment should always include a comprehensive plan with regular follow up and address:

- Diet modifications
- Fluid management
- Positioning/activity programming
- Toileting program
- Consideration of medication
- Medications

**Epilepsy**

Epilepsy is a common co-morbidity in persons with ID reported in 30 to 60 percent of persons. A three-year review of epilepsy noted in mortality reviews for DIDS showed the incidence in the Tennessee Waiver population at close to 40 percent. It is well recognized that people with ID who have brain pathology and function at lower levels are more likely to have seizures. Some types of seizures, such as Lennox-Gastaut Syndrome and hypsarrhythmia, are seen primarily in individuals with ID. Further complicating treatment is the fact that people with ID may have several types of seizures and be refractory to good control.

For treatment to be optimum, the patient should have an accurate seizure diagnosis and be on antiepileptic medications used for the specific type of seizures present. This is sometimes difficult when past medical history is not available, and the patient presents on many seizure medications with the diagnosis of “seizure disorder.”

Here again, partnership of the two systems, linked by the nurse case manager may help the PCP. Old records can be obtained, and nearly everyone in the system in residential care has been monitored with data available about their seizures (frequency, timing and what the seizure looks like).

There may be challenges in managing patients with ID and seizures using standard approaches to treatment. This is an area where the RN/CM and Waiver system professionals can fill in the gaps. Testing (MRI, CT, video EEG’s, etc.) may require preparation, positioning, or behavioral support which may be facilitated by Waiver clinicians. Accurate recording may be clouded by misinterpretation of other movements seen in the ID population such as tics, startle reflex, clonus, etc. A brief video from a cell phone may often be helpful. And, finally, some common side effects may be difficult to interpret in a non-verbal person without a caretaker’s input. Side effects such as nausea, drowsiness, sleep problems, double vision, and mood changes are subjective in nature, but if discussed beforehand, staff who know the patient may be able to identify these symptoms.
Treating Seizures is a Partnership

<table>
<thead>
<tr>
<th>Physician Responsibility</th>
<th>Support Responsibility</th>
</tr>
</thead>
<tbody>
<tr>
<td>Diagnosis – includes Hx, PE, testing</td>
<td>Provide accurate information</td>
</tr>
<tr>
<td>Treatment – decide on specialist</td>
<td>Follow medical recommendations</td>
</tr>
<tr>
<td>Advise COS</td>
<td>Integrate problems and treatment into life</td>
</tr>
<tr>
<td>Consider Emergency Plan</td>
<td>Understand problems and effect on life</td>
</tr>
<tr>
<td>Consistent follow up</td>
<td></td>
</tr>
</tbody>
</table>

A common seizure pattern seen in people with ID but not regularly seen by PCPs is Lennox-Gastaut Syndrome (LGS). LGS is a difficult-to-treat form of childhood-onset epilepsy that most often appears between the second and sixth years of life, and is characterized by frequent seizures, and different seizure types. It is often accompanied by ID, and psychological and behavioral problems. The syndrome shows clear parallels to West syndrome (infantile spasms or hypsarrhythmia) – enough to suggest a connection. Daily multiple seizures are typical in LGS. Also typical are the broad range of seizures that may occur, larger than that of any other epileptic syndrome. The most frequently occurring seizure types are tonic, which are often nocturnal (90 percent). The second most frequent are myoclonic seizures, which often occur when the patient is over-tired and atonic. Atypical absence, complex partial, focalized, and tonic-clonic seizures are also common.

Additionally, about half of patients will suffer from status epilepticus, usually the non-convulsive type, which is characterized by dizziness, apathy and unresponsiveness. The seizures may cause sudden falling (or spasms in tonic, atonic, and myoclonic episodes) and/or loss of balance, which is why patients often wear a helmet to prevent head injury. In addition to daily multiple seizures of various types, children with LGS frequently have arrested/slowed psycho-motor development, and behavior disorders resulting in adults with LGS having longstanding problems. The syndrome is also characterized by an interictal (between-seizures) EEG, featuring slow spike-wave complexes.

Another issue is when patients with ID are treated with phenobarbital and phenytoin (Dilantin) which are usually considered to be “old” medications for seizures. Phenobarbital is no longer covered under Medicaid or Medicare. These medications also have significant side effects. Phenobarbital has behavioral side effects in about 40 percent of patients with ID (which may not be evident until a person who has been taking phenobarbital for 20 or more years is off the drug). These side effects include hyperactivity and irritability, which may contribute to problem behavior; inattentiveness; depression; personality change, argumentativeness, stubbornness; and slowing of verbal and motor responses. If a person is taken off phenobarbital, it should be done very slowly, about 10 percent reduction/month, and take about a year, or withdrawal seizures may occur.

Phenytoin also poses problems. Many people have physical side effects, which although not life-threatening, affect self-image and may need treatment. These include hirsuteness (a real problem in females), coarsening of facial features, and gingival hypertrophy (which can create problems with eating, drooling and dental hygiene). Phenytoin also interacts with many common medications such as theophylline, including other anti-epileptic drugs (AEDs) which may affect blood levels.
The absorption of phenytoin is also affected by food. If someone on phenytoin is being fed by enteral tube, the feeding needs to be stopped an hour before and after the administration of phenytoin. Phenytoin's non-linear saturation kinetics may cause toxicity with medication increase, even at small amounts, which may produce sharp rises in serum level at higher doses. Toxicity may be difficult to diagnose clinically in a non-verbal, non-ambulatory patient. A little known cause of toxicity, especially in these patients, is free phenytoin. Phenytoin binds with albumin in the blood. When the albumin is low (common in nutritionally compromised patients or those with liver disease) there is no place for phenytoin to bind, and it is “free.” The total phenytoin level (as routinely measured) may be normal even though the “free” phenytoin is elevated and causes toxicity. The free phenytoin normally should be about 10 percent of total phenytoin, so therapeutic levels are between one and two. A free phenytoin level should be considered if a patient has a low albumin level.

Current recommendations for treatment of seizures in people with ID are for monotherapy. Multiple medications are sometimes used as “add-on” therapy, or when a patient has more than one type of seizure. In general “polypharmacy” is non-productive. Studies actually show decrease in seizures with reduction of numbers of AEDs. Side effects and quality of life are also impacted when a patient is on multiple medications.

There are currently at least 20 AEDs to choose from for treatment. Seizure classification is important in choosing an AED and prognosis. Principles of AED treatment are the same for patients with ID who have epilepsy as they are for those with epilepsy alone, and IQ should not be a limiting variable. No single seizure type is characteristic in people with ID, although the most common seizure type is partial, with secondarily generalized tonic-clonic seizures. Also common and often refractory is Lennox-Gastaut seizures which include tonic-atonic and myoclonic (mixed) seizures. “Weird seizures” (e.g., frontal lobe) may also occur more often than in the typical population.

Goals of therapy should be “reasonable,” and derived by discussion with the patient and Circle of Support (COS) with the PCP (in consultation with the neurologist). Although a goal for optimal therapy would be complete seizure control and no adverse events, this is often difficult and unrealistic with some patients with ID – although this goal should be considered first. This is because of quality of life issues. Uncontrolled seizures and long postictal states interfere with quality of life. But no seizures accompanied by serious side effects such as drowsiness, tremors, nausea, vomiting, and double vision, etc. also interfere with quality of life.

Therefore, reasonable goals of therapy should be:
- Accurate diagnoses
- Seizure control
- Reduction of polypharmacy
- Reduction of side effects
- Improved safety
- Improved quality of life

Individuals with ID and epilepsy also need regular follow up care. Much of this may be accomplished by the PCP with intermittent consultation from the neurologist and include:
- Regular review
- Accurate recording of seizures
• Periodic blood tests (there is no mandatory frequency and should be at PCP discretion)
• Periodic medical evaluation with adequate and accurate information
• Attention to quality of life
• Integration of treatment with other medical problems

Intervals for review are usually based on severity, stability, types of medications, doctors’ practices, and patient issues. Significant change should prompt an early review.

When seizures increase, a review should promote a review for any treatable cause. This includes blood levels and compliance review. Some “triggers” which increase seizures in people with ID are: illness, infections, stress, emotional upset, temperature changes (fever, hypothermia), photosensitivity, hunger, and constipation.

Status epilepticus occurs when someone has prolonged seizures, or clusters of two or more seizures without complete recovery in between. Status may result in permanent neuronal damage and is responsible for significant morbidity and mortality. Definition by the World Health Organization defines status as a continuous seizure lasting 20 minutes. There is evidence that injury may occur earlier, and that earlier treatment may be more effective. Because of this, most recommendations are to consider emergency treatment after 5 or 10 minutes of continuous seizures. This usually involves I.V. medication, but in many patients with a crisis plan developed in advance, treatment may begin outside of the hospital, and be done by supporting staff. The standard approach is to give rectal diazepam (Valium), and this procedure has been used for over 30 years, especially in children. Diazepam, is usually given in gel form under the trade name of Diastat in a pre-filled administration device. However, since benzodiazepines are no longer covered by Medicaid or Medicare, this can be quite expensive. To reduce cost, use of liquid diazepam may be given via syringe (without the needle) as a rectal bolus at 0.5 mg/kg or 10-30 mg for an adult. There is a very small risk of respiratory depression with either form. Even though respiratory depression is very rare, use of rectal diazepam warrants transfer to the emergency room.

There are long term risks and effects of taking AEDs. One of the most serious is the development of osteoporosis in patients who have taken phenytoin (Dilantin) and/or phenobarbital for many years, which is another reason to discontinue these medications. Long-term behavior issues are well-described in all patients with epilepsy who have been on long-term treatment with AEDs. These include aggression, anxiety, impulse control disorder, pica, somatization, memory problems, hyperactivity, stereotypies, SIB, and especially depression.

Safety issues are of paramount importance, and the risk of injuries from seizures (falls, fractures, bruises, broken teeth, lacerations, etc.) is common. Medically fragile and physically compromised individuals are at high risk of aspiration during seizures with subsequent pneumonia which can be fatal. Activity restrictions should be carefully discussed and consider principles of quality of life and dignity of risk. Restrictions should be reasonable and prudent. Certain activities like climbing or swimming may sometimes be done under supervision. Use of helmets to protect the head may affect self-image.

Another serious risk for patients with epilepsy is Sudden Death in Epilepsy (SUDEP). Approximately 50 percent of persons with epilepsy die from causes either directly or
indirectly related to seizures. About a third of these deaths are sudden and unexpected, and no cause is identified at autopsy. Most experts believe this is an associated autonomic response affecting heart rhythm though visible seizures are rare with this event. About half these deaths occur during sleep, and the person is found the next morning. The overall reported incidence is 1/370-1100 individuals with epilepsy. Although some risk factors are known, the causative mechanism has yet to be pinpointed. The maximum incidence occurs in persons between 20 and 40 years of age, and severity of the condition is not a factor. Most have longstanding seizures and about 50 percent have structural abnormalities in the brain. In the majority, subtherapeutic levels of AEDs have been found but no link for this has been established.

The use of AEDs, especially in the case of polypharmacy, may lead to problems with medication interactions and side effects. This is particularly a problem when each physician prescribing medications is not aware of all the drugs a person is taking. There is significant interaction with blood levels of the AEDs themselves, and several AEDs are affected by other medications commonly taken. As an example, cimetidine (Tagamet) has a significant effect on lowering blood levels of the AED carbamazepine (Tegretol) through the P450 mechanism, a common pathway for medication metabolism. Psychotropics may lower seizure threshold, and many have behavioral side effects. An example is a newer AED, levetiracetam (Keppra), where it is reported that 10 percent of persons with ID have shown irritability as a side effect. This points out a non-productive outcome when multiple specialists treat this population without communication, and highlights the value of the PCP integrating overall care. A not unusual scenario is to see some patients develop negative behaviors from a new medical problem which is undiagnosed, and referred to psychiatry. The psychiatrist (usually having inadequate information) will prescribe medications which may upset seizure control, prompting a change in AEDs with a behavioral side effect, and the patient is referred back to the psychiatrist and the medical problem is still undiagnosed and untreated.

A significant medication issue among persons with ID is the use of benzodiazepines which are not covered under Medicaid and Medicare. While benzodiazepines are the first line in treating status epileptics, their use on a regular basis is sometimes not clear or not monitored. For instance, use of clonazepam (Klonopin) only at bedtime is suspect (unless a clear diagnosis and rationale for treating nocturnal seizures is in place) as it does not have a 24-hour therapeutic effect, and it’s usefulness for seizures is not reviewed. Many patients also are placed on clonazepam as an “add on” after an increase in seizures without thorough review of the overall picture (diagnosis, medical, problems, behavior issues, etc.). Benzodiazepines, most often Ativan (lorazepam), are also used for behavior issues in persons with ID, but it is sometimes unclear what the drug is being prescribed for, or if prescribed for behavior, how it affects seizure control. Probably the biggest worry is lorazepam’s side effect profile and the development of tolerance. When used for both seizures and behavior, patients develop a tolerance and need higher doses of effect diminishers. This increases dependency and withdrawal, especially if done suddenly, and may result in withdrawal seizures. This may even occur in someone without previously diagnosed seizures, and in someone on long-term high-dose treatment, may be delayed up to weeks secondary to the drug’s long half-life. Benzodiazepines are stored in lipid tissue. If someone is being treated with benzodiazepines, it should be clear what the plan and rationale is, and understood by all. In addition, benzodiazepines have side effects which may affect health, behavior, and day-to-day function. Sedation is a major problem. Hypersalivation may occur causing excessive drooling which predisposes the person to both aspiration and social
shunning. Ataxia and confusion may interfere with daily life. And, in patients with psychiatric diagnoses and behavior problems, disinhibition caused by benzodiazepines may increase negative behaviors, and possibly result in unnecessary prescription of psychotropics.

Other treatment modalities should be considered in patients who are refractory to medication treatment: usually defined as an inability to completely control seizures after using three appropriate AEDs for the identified seizure type. This occurs more often in persons with ID than in the general population. Surgery has been helpful for persons who qualify on standard criteria. Level of function does not preclude a patient for this modality, and many patients with ID who have had surgery have fewer seizures and a better quality of life.

Another helpful treatment alternative is the use of the vagal nerve stimulator. This mode of treatment is often not offered to people with ID but has been shown to be both effective and cost-efficient. A positive side effect has been improvement of symptoms of depression.

Pseudoseizures also occur in people with ID, and may be hard to diagnoses and treat. These are defined as “an epileptic-like event, without EEG changes” consistent with epilepsy, which results from emotional conflict. These are often distinguished by the following observations:

- Asynchronous (out of phase) movements.
- Pelvic thrusting
- Side-to-side head movements
- Individual will exercise enough control to prevent injury and incontinence
- More prone to vocalization during tonic-clonic events
- Events more likely to occur in the presence of others.

Pseudoseizures often occur in patients who have bona fide seizures compounding treatment. Etiologies often involve post-traumatic stress disorder (particularly childhood physical or sexual abuse), somatization of repressed anger/stress/anxiety, attention-getting, and secondary gain (work avoidance). The diagnosis is difficult to make, especially in patients with real seizures, and patients often need psychiatric counseling.

**Resources**


**Aspiration and Pneumonia**

Aspiration, and subsequent pneumonia, is a common cause of death in individuals with chronic disorders, impairments and disabilities. In published reports, respiratory complications were the cause of death in 50 percent of persons with severe ID – more than seven times that of the general population. Individuals with ID have risk factors for aspiration secondary to neuromuscular disorders, altered level of consciousness (seizures), GI dysfunction, and perhaps side effects of benzodiazepines.

There are four aspiration syndromes that occur:
1. Acute airway obstruction - food or an object can totally block the airway
2. Chemical pneumonitis - gastric contents mixed with food enter the lungs
3. Bacterial pneumonia - oral secretions enter the lungs (greater risk with poor oral hygiene)
4. Repeated trace amounts of aspiration over long periods of time can cause serious complications and may result in a COPD-like condition.

There can be a combination of factors. Aspiration can result in pneumonia, upper respiratory infections, chronic lung disease (similar to COPD), choking (obstruction of airway), and possible death. In people with ID, risk of aspiration in people who aspirate may be predicted by:
- Dependence on others for feeding and assistance with meals
- Multiple health problems
- Smoking
- Enteral nutrition
- Dependent on others for oral care
- Number of decayed teeth
- Number of medications
- Use of muscle relaxants, particularly benzodiazepines

The following conditions often seen in persons with ID are also risk factors:
- Difficulty with muscle control
- Weak muscles, tongue, lips, cheeks, and in general
- Poor skeletal muscle development
- Birth defects
- Lack of mobility
- Obesity
- GI disorders
- Behavioral or situational problems
- Craniofacial abnormalities
- Weak cough
- Dependency on caregiver
- Seizures
- Some medications
- Scoliosis or restrictive lung disease

Dysphagia is usually present in chronic aspiration, but acute events may also cause aspiration in a person without dysphagia (e.g., seizures). Many individuals have silent aspiration and show no symptoms. They may, however, develop serious complications.

Diagnosis of dysphagia is necessary to treat and prevent chronic problems and progression. Professionals in the Waiver system may help with prevention plans such as positioning and dining plans. Risks of complications, and issues of diagnosis based on symptoms, can be initiated by the PCP during annual evaluations.

**Neuromuscular Disorders**

Neuromuscular disease is a very broad term that encompasses many diseases and ailments that either directly, via intrinsic muscle pathology, or indirectly, via nerve pathology, impairs the functioning of the muscles. Neuromuscular diseases affect the
muscles and/or their nervous control. In general, problems with nervous control can cause either spasticity or some degree of paralysis depending on the location and the nature of the problem. The most common neuromuscular problem in the Waiver system is CP, but this does not cover all neuromuscular problems. Immobility may produce increased tone and contractures, but is not CP.

In this population, the majority of neuromuscular problems have a central nervous system origin. This may result from brain damage due to prenatal insult, prematurity complications, traumatic brain injury, meningitis, etc.; malformation or brain defects like porencephalic cysts, hydrocephalus, schizencephaly, neural tube defects (Spina Bifida), metabolic or, in some cases, genetic causes.

Since the origin of the problem is in the brain but affects muscles via their motor control, deficits are usually manifested in problems with balance and movement, position and gait, and activities of daily living. The external defects usually correlate with brain damage and may be predicted by CT or MRI scans. As an example, the left side of the brain controls the right side of the body, so a person with brain damage in the left motor cortex will demonstrate a right hemiplegia. Hemiplegics have a greater incidence of seizures, and since the language center is in the same area on the left side, in 90 percent of people, the person may also be at risk for language and communication difficulties. Imaging studies may be valuable in predicting complications.

Although the basic brain damage mechanism is static, the functional picture may change dramatically with growth and development, so these problems look very different in adults as compared to children. Adults may have significant contractures or neuromuscular scoliosis. Even patients who had hypotonia as children will develop contractures and may appear spastic when in fixed positions. (e.g., an adult “fixed” in a “frog-legged” position present in childhood caused by severe hypotonia).

These long-term physical problems have significant complications which may create significant secondary medical problems. They may also affect positioning and movement, and have negative health effects - constipation, eating, movement, balance, perception, pain, sleep, as well as social responses, and the ability to respond to health care events.

These patients have little to receive for treatment options. Pain is common, and should be considered and treated. Treatment may also be required for caretaking issues such as perineal care. Use of muscle relaxants may be helpful, but will have little positive outcomes if contractures are fixed. Surgery is rarely done to correct contractures. Positioning programs done through the Waiver system may be the most effective way to address these problems.

Another serious problem of increasing frequency in persons with neuromuscular disorders is spinal stenosis; this condition is also increasing worldwide in people over the age of 60. Spinal stenosis occurs when the spinal canal narrows and compresses the spinal cord and nerves. It usually occurs with spinal degeneration due to aging but may be intensified in those with pre-existing neuromuscular problems. It can affect the cervical, thoracic or lumbar spine. It may be difficult to diagnose in this situation or may be overlooked. Symptoms are similar to those found with claudication, but are termed pseudoclaudication, and include atrophy, pain, weakness, and tingling in the legs. The first symptoms, however, are often bouts of lower back or neck pain. Physical
examination may reveal signs of a myelopathy, which may be difficult to interpret in a patient with pre-existing hypertonia if the patient has not been seen previously. Treatment is usually surgery, although salmon calcitonin has shown to be promising in treatment of non-operative cases.

Atlantoaxial instability is a well-reported problem in people with DS. There are recommendations for screening in children with DS, and screening is required for participation in Special Olympics. Little is known about incidence in adults, and people with DS are also at risk for spinal stenosis. While in childhood, neck X-rays are recommended for diagnosis. In adults with DS, physical changes and findings will point the way to a diagnosis. The patient will have signs of a myelopathy which is progressive (over weeks or months) with increased tone (people with DS are basically hypotonic) and increased deep tendon reflexes (DTRs). Patients will also have a change in gait, most often dragging their feet, usually asymmetrically, and may develop torticollis. Direct care staff will usually notice and report these physical changes.

**Musculoskeletal Issues**

The most common musculoskeletal problem in the people with ID receiving Waiver-based services is osteoporosis, which may result in pathological fractures. In the general population, osteoporosis is thought of as a medical problem associated with aging. In persons with ID, it can occur much earlier, even in adolescence, with medically complex, non-ambulatory patients. The presence of a dowager’s hump in patients of middle age, including males, may be a tip-off.

Osteomalacia and osteopenia occur frequently and may have a variety of causes. Non-ambulatory people do not bear weight. Low-functioning patients may not get enough calcium in their diets, and may not be outside in the sunshine enough to produce enough vitamin D. Of most concern, however, are the large numbers of patients on phenytoin (Dilantin) and phenobarbital. Both predispose the person to developing osteoporosis. Genetic heritage may also play a role, as well as aging. Of course, there are other causes of osteoporosis, such as hormone deficiency, so a full workup should be done. Although the Dexascan is the “gold standard” for diagnosis, positioning may create a problem. Use of calcaneus measures is acceptable in this situation. Often reports of osteopenia in a chest X-ray may be a clue. The cardinal key, however, is suspicion.

Treatment is the same as the general population. The risk of GERD should be kept in mind with this population already at high risk. Pathological fractures occur in thin or weak bones with little force or movement and are not unusual occurrences in the Waiver system. The most common fractures are in the small bones (fingers, toes) and may be related to bumps associated with daily activities. Fractures of major bones such as the femur have occurred when the patient turned over in bed. While all fractures in the Waiver system are investigated, abuse and mistreatment are rarely found to be the cause and should not be assumed to have taken place. Obviously, fall prevention programs are very important in people with osteoporosis.

Another very common group of medical problems involves feet. Non-ambulatory persons still need foot care including clipping of nails. Neuromuscular problems may cause increasing problems over time with weakness (e.g., in ankle with excessive pronation) and flat feet. Problems with asymmetries and walking may predispose these
patients to foot deformities. Spasticity causes contractures. Special Olympics, as part of its Healthy Athlete Program, examine feet and have developed a manual to address these issues.

**Sensory Deficits**

Sensory deficits are common in people with ID. Vision and hearing problems are a component of many syndromes as evidenced by those with DS who have upwards of 50 percent prevalence in adults (myopia, keratoconus, cataracts, secondary problems of diabetes, sensorineural hearing loss, etc). These deficits are magnified by aging, and even patients without genetically associated problems are affected by sensory loss with decline in function as they age.

Vision and hearing are difficult to test in some people with physical or communicative problems, but some evidence of problems can be found in everyone with ID where there is a concern. As an example, a baby may be discharged with a diagnosis of “cortical blindness” based on evidence (MRI, eye exam) which is an evaluation not based on function. Later on, parents or therapy staff may question this when the baby is noted to reach for toys or feeding utensils. Similarly direct care staff may be important in assessing sensory abilities due to their intimate knowledge of the individual’s day-to-day function. However, definitive testing should be done as direct care staff may over- or underestimate visual acuity.

There are existing myths or confusion as to whether sensory deficits should be treated in patients with ID. A person who is not ambulatory may be felt not to need glasses because they don’t walk, but better vision may help with independent skills such as eating or decrease anxiety caused by not being able to assess the surrounding environment. Some people feel that in people who have ID, correcting vision or hearing is unnecessary when the opposite is true. People with typical cognitive function adapt to a vision or hearing problems much better than someone who already has a disability.

Hearing deficits are reported as occurring in approximately 40 percent of individuals with ID. This also affects communication with health care providers. Hearing impairment can be classified into four types:

1. Conductive hearing loss which results from abnormalities in the external or middle ear. This type of hearing loss may respond to corrective surgery or medical treatment.
2. Sensorineural hearing impairment which results from a dysfunction of the cochlea of the inner ear or auditory nerve, and may be treated with a cochlear implant or hearing aids.
3. Central hearing impairment which is caused by pathology in the nervous system.
4. Mixed hearing impairment which is a combination of any of the above.

There are many hearing tests both direct (e.g., audiometer) and indirect (e.g., Impedance testing) that can provide information about hearing. The etiology of those with ID may provide clues for hearing problems such as congenital rubella syndrome, post-meningitis, PKU, DS, etc.
Treatment of a hearing deficit in an adult with ID will require a team approach. In addition to the possible use of a hearing aid, a program for use and training to address the overall issues needs to be in place. Waiver therapists may help with this.

Sensory problems are often overlooked in people with ID but should be part of the yearly discussion. In addition, sensory deficits may mask, look like, or interfere with evaluation of other medical problems, particularly dementia.

The Role of Genetics in Clinical Issues in ID
All of us are affected by our basic genes: including the color of our eyes, our height, metabolism, response to drugs, and most specifically, health risks, and certain diseases. All people carry a few significantly abnormal hidden genes which are not expressed, usually because the disease is recessive, and the person has only one gene. An example most people are familiar with is PKU. This is a recessive genetic disease which, without treatment, results in severe ID. For approximately 30 years, most states have screened for this disease in newborns to identify babies for treatment in order to avoid the consequences of ID. There are also other genetic mechanisms which are either related to reproduction, or what might be described as “accidents” consisting of mutations and other “mistakes.”

A genetic etiology for ID is fairly common, particularly in individuals who have low IQs or low functioning levels. It is estimated that in this population, 30 to 50 percent of individuals probably have a genetic diagnosis, which may contribute to and predict clinical issues. An accurate diagnosis is important for several reasons. First, most people want an answer as to why someone is the way they are – looking different or having an ID. This might include families when the individual is young; the individual when he/she is older; or the people who are taking care of the individual. In addition, and probably most important from a clinical standpoint, is that having an accurate diagnosis often provides a “roadmap” to medical care and predicts prognosis and other life issues such as risks for certain cancers and/or dementia.

A good example of this is DS. Thirty to forty years ago, people felt that individuals with DS did not live very long, or do very much. We now know the average life span of someone with DS is normally 55 years, and based on what is known about the syndrome, physicians can predict most of their medical problems. The majority of these problems are easy to diagnose and treat. In addition, in other genetic diseases, surveillance may help prevent serious outcomes such as a number of cancers associated with a genetic disease such as TS.

The last reason for having an accurate diagnosis is that it helps contribute to the body of knowledge that helps other people with similar problems. Knowing the diagnosis may help researchers with finding a test for the syndrome, understanding the course of that genetic syndrome, and even coming up with preventive and/or curative treatments.

There are several very common genetic syndromes among individuals with ID supported by the Waiver program. These include DS, which is usually identifiable to most people (and accounts for about 15 percent of those receiving Waiver services), and Fragile X syndrome, which is the most common and under-diagnosed genetically inherited condition and may present with behavior issues. Other common syndromes include
Prader-Willi syndrome, Smith-Magenis syndrome, Cornelia de Lange syndrome, Rett syndrome, Myotonic Dystrophy, Tuberous sclerosis, and other syndromes such as Neurofibromatosis and Williams’ syndrome, which also occur in individuals who do not always have ID as a component. The average IQ is below 100.

There are several features in the clinical profile of individuals which might hint at a genetic disease. This would include family history of similar issues, or individuals with ID; dysmorphic features, as in DS; specific functioning as occurs in behavioral phenotypes; and specific clinical issues such as a maternal history of an increased number of miscarriages, neonatal deaths, and early significant medical problems. In addition, a large number of genetic syndromes associated with ID are associated with short stature, and that is often a key finding suggesting a genetic evaluation.

There are also several genetic syndromes which cause behavioral differences, which is somewhat unique to the field of developmental disabilities. We refer to this group of syndromes as “behavioral phenotypes,” or stereotypical behaviors that are characteristic of a specific genetic syndrome. These include unique behaviors often mistaken for psychiatric disease. Such problems might include “gaze avoidance” as seen in Fragile X syndrome, often mistaken for autism; “hand wringing” often seen in Rett syndrome and often mistaken for Obsessive Compulsive Disorder; and hyperphagia, the “inability to stop eating” as seen in Prader-Willi syndrome.

The question is how an individual should be diagnosed, and what is done with the compiled information. Diagnosis can either be made through a genetic evaluation, or in many cases, a simple laboratory test after suspicion is raised, often by staff supporting the individual in the Waiver system. A genetic evaluation is most often done in the prenatal period or around birth when there is a suspicion based on either family history, or parents with a genetic problem. The process for this evaluation involves a careful family and medical history, and a thorough physical examination including meticulous evaluation for dysmorphic or unusual features, particularly in the face. These features would include such issues as widely set eyes, low set ears, a slant to the eyes, and other abnormalities of facial features. Other areas of dysmorphic features include lines on the hand, most specifically the Simian Crease (single horizontal crease), and other appearances which include skin lesions, abnormal limbs or evidence of abnormalities of internal organs such as abdominal distention. Height, weight and head circumference are also recorded.

After this is done, certain laboratory tests may be performed to rule out certain diagnoses. Examples of this could be an enzyme test to rule out PKU, a uric acid level to rule out Lesch-Nyhan syndromes, or a cholesterol level to point in the direction of Smith-Lemli-Opitz syndrome. Often, however, some type of chromosome test is done. The most common test performed for genetic evaluation is a karyotype which may yield unexpected information to allow diagnosis. There are also specific tests such as DNA probes which focus on specific genes, and many other tests which focus on functions of genetic metabolism.

With adults, genetic diagnosis is somewhat more difficult, and this is sometimes an issue when we have individuals who were placed into the Waiver system for care many years ago when these tests and syndrome information were not available. With increasing information provided to the lay public about genetic issues, questions are often raised. In some cases, syndromes are well known. Perhaps support staff has heard about a
specific syndrome, and the question is raised by the individual’s support staff. This might be the case, for instance, with Fragile X syndrome or other common syndromes. Often, when the issue is raised, it may be presented to the PCP who can review the concerns and order simple genetic tests to complete the diagnosis.

The biggest issue with genetic disease is keeping up with new developments. This usually is part of an individual’s personal responsibility for their health; in the Waiver system it is the responsibility of support staff and caregivers. With the explosion of information in genetics on a daily basis, new information is discovered about syndromes including diagnostic and treatment issues. The bottom line with genetic issues is that the more one knows about one’s self, particularly when it involves clinical issues which may impact health; the more likely the person is to lead a longer more productive life.

In summary, genetic disease is common to all of us but may hold more prominence in an individual with ID, especially when it contributes to decisions about their health and life. The important areas to consider are the value of a diagnosis and associated information, and to keep up with changes in the field. One resource which may be important for people who support these individuals is a website called Family Village – www.familyvillage.com. This website provides both technical and general information, other people with whom to chat, and most specifically, organizations related to a specific genetic syndrome.
<table>
<thead>
<tr>
<th>Condition</th>
<th>Cerebral Palsy</th>
<th>Down Syndrome</th>
<th>Fragile X</th>
<th>Prader-Willi</th>
<th>Phenyketonuria</th>
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<tbody>
<tr>
<td>Nerve Damage</td>
<td>1:1000</td>
<td>1:1000</td>
<td>1:100</td>
<td>1:10 000-25 000</td>
<td>1:10 000-1:20000</td>
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<td>Hearing Impairment</td>
<td>Audiovisual</td>
<td>Eye pathology and visual impairment (multifactorial), cataracts hearing impairment (multifactorial) Both common annual assessments recommended</td>
<td>Visual impairment (multifactorial) Hearing impairment Recurrent ear infections</td>
<td>Strabismus Myopia</td>
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<tr>
<td>Endocrine</td>
<td>Osteoporosis</td>
<td>Hypothyroidism (annual TFT recommended) Osteoporosis</td>
<td>NIDDM (secondary to obesity) Hypogonadism Delayed puberty</td>
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<tr>
<td>Psychiatric/Psychological</td>
<td>Depression</td>
<td>Depression Alzheimer's type dementia (20 years earlier than general population) Variable intellectual capacity</td>
<td>Attention deficit/ hyperactivity Variable intellectual capacity Disabled in social functioning Poor eye contact Hand flapping</td>
<td>Hyperphagia needs active monitoring to prevent morbid obesity, associated morbidities and death Impulse control difficulties Self injury</td>
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</tr>
<tr>
<td>Central Nervous System</td>
<td>Epilepsy</td>
<td>Epilepsy Usually dystonic Epilepsy Usually dystonic, complex partial</td>
<td>Hypotonia at neonate Small hands and feet Short stature Almond shaped eyes</td>
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<tr>
<td>Cardiovascular</td>
<td>Congenital heart defects (&lt; 6%) Aortic dilation Mitral valve prolapse</td>
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<tr>
<td>Musculoskeletal and Skin</td>
<td>Orthopaedic problems Neurovascular problems Pressure areas skin atlantoaxial instability &lt;1% Skin disorders, alopecia, eczema Connective tissue dysplasia, scoliosis Congenital hip dislocation</td>
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<tr>
<td>Other</td>
<td>Genito-urinary problems Incontinence Constipation Dental problems Recurrent aspiration Oesophagus, gastrooesophageal reflux V/B bleeding, anemia Swallowing, eating difficulties Blood dyscrasias Childhood leukemia Sleep apnoea Obesity Susceptibility to infections Coeliac disease Hernia Anomalies of speech and language Characteristics more distinctive in males may not appear until late childhood Microtia, cleft, hare lip, prominent ears, long face Infinitesimal failure to thrive, thrashing, hyperphagia and severe obesity High tolerance to pain Decreased ability to vomit Poor body temperature control Sleep apnoea Osteopenia Undescended testes Dental abnormalities</td>
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<tr>
<td>Inheritance</td>
<td>50% cause by antenatal problem 10% anoxia at birth</td>
<td>95% recdision at multiple loci leading to trisomy chromosome 21; 4% translocation of C21 or rared parental mosaicism C21</td>
<td>X-linked Chromosome 15 autosomal recessive Econa Improved significance of previously untested autosomal recessive individuals after introduction of a phenylalanine restricted diet</td>
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<tr>
<td>Disorder/Condition</td>
<td>Angelman syndrome</td>
<td>Williams syndrome</td>
<td>Rett syndrome</td>
<td>Noonan syndrome</td>
<td>Tuberous sclerosis</td>
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<tr>
<td></td>
<td>&lt;1:10 000 &lt;80 000</td>
<td>&lt;1:10 000 females</td>
<td>&lt;1:10 000</td>
<td>&lt;1:6 000-17 000</td>
<td>1:2000</td>
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<td>Audiovisual</td>
<td>Glaucoma</td>
<td>Hypersusitue Strabismus</td>
<td>Refractory errors</td>
<td>Strabismus refractive errors Vision/hearing impairments</td>
<td>Retinal tumours Eye rhabdomyomas</td>
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<tr>
<td>Endocrine</td>
<td></td>
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<tr>
<td>Psychiatric/ psychological</td>
<td>Easily excitable Hyperactive</td>
<td>Variable intellectual capacity Attention deficit problems in childhood Seizures Epilepsy</td>
<td>Severe intellectual disability</td>
<td>Mild intellectual disability</td>
<td>Variable intellectual capacity Behavioral difficulties Sleep problems</td>
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<td>Central nervous system</td>
<td>Severe development delay-apparent by 1 year Microcephaly Epilepsy</td>
<td>Perceptual and motor function reduced Epilepsy</td>
<td>Epilepsy Vasomotor instability Epilepsy</td>
<td>Epilepsy</td>
<td>Cerebral astrocytomas Epilepsy</td>
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<td>Cardiovascular</td>
<td></td>
<td>Cardiac abnormalities Hypertension CVAs Chronic hemiparesis</td>
<td>Prolonged QT interval Pulmonary valvular stenosis ASD, VSD, PDA</td>
<td>Hypertension Rheobteryomus</td>
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<td>Musculoskeletal</td>
<td>Joint contractures and scoliosis in adults Truncal hypotonia Limb hypotonia</td>
<td>Joint contractures Scoliosis Hypotonia</td>
<td>Osteopenia Fractures Scoliosis</td>
<td>Scoliosis Talipes exostosis Pesito cruris/male exostomas</td>
<td>Bone rheobteryomus</td>
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<tr>
<td>Other</td>
<td>Speech impairment Movement and balance disorder Characteristic EEG changes</td>
<td>Renal abnormalities Hyperventilation Apnoea Pott’s disease Facial difficulties Growth failure</td>
<td>Abnormal clotting factors, platelet dysfunction Undescended testes, deficient spermatogenesis Lymphoedemaoma Hepatolelophromegaly Cubitus valgus, hand abnormalities</td>
<td>Kidney and lung hamartoma Polycystic kidneys Liver rhabdomyomas Dental abnormalities Skin lesions</td>
<td>Variable clinical phenomena depending on location of the neurofibroma Tumours are susceptible to malignant change Other varieties of tumours may be associated</td>
</tr>
<tr>
<td>Inheritance</td>
<td>Variety of genetic mechanisms on chromosome 15</td>
<td>Microdeletion on chromosome 7</td>
<td>Mainly sporadic</td>
<td>Autosomal dominant may be sporadic</td>
<td>Autosomal dominant</td>
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</table>
Behavior/Psychiatric Issues

People with ID have a high prevalence of challenging behaviors which may limit life opportunities and result in comorbidities such as injury, medication side effects, etc. In the past, many people ascribed these behaviors to the underlying ID; this called diagnostic overshadowing. These behaviors are not always representative of a psychiatric problem; although persons with ID have a higher incidence of psychiatric problems. They may have the same mental health diagnoses, as per DSM-IV, as the typical population. Diagnosis is sometimes difficult because of the cognitive deficit, but there is now available an adaptation of diagnostic criteria for use with people who have ID.

Many behavioral issues in ID are due to the inability to communicate (“If I can’t tell you I don’t want ice cream, I might throw the bowl.”) or difficulties with coping and control (“If you make me go to the park and I don’t want to, I might become aggressive.”).

A significant cause of behavioral problems, especially if the behavior issue is new, may be from an underlying medical problem. This is a particular problem in nonverbal people.

Challenging behaviors often have a variety of contributing factors, and evaluation may involve several clinicians. The PCP should start with a medical evaluation for organic factors leading to behavioral disturbances as outlined below. The Waiver system offers the services of behavioral analysts. These professionals can assess behaviors to evaluate whether they are organic (medical or psychiatric) or behavioral (with a variety of underlying driving factors: attention seeking, avoidance, etc.), or a combination of both. This evaluation usually takes place in the natural setting of the patient. Behavior analysts may also write a behavioral program to be carried out by support staff to ameliorate the behavior.

After the prior issues are addressed, if no diagnosis is made, the person should be seen by a psychiatrist for a comprehensive assessment.

Therefore, assessing an individual with ID with challenging behaviors is truly a biopsychosocial exercise. It has multiple and complex etiological and contributing factors. It is important to assess possible contributions from the following:

- Medical disorders – Constipation, toothache, earache, GERD, fracture, UTI, other sources of pain, or bodily discomfort, etc.
- Problems in expectations and supports – Individuals with ID are dependent on external structures. Emotional and behavior problems often arise when expectations and supports change or are inappropriate (e.g., recent move, change in staff, change in daily life schedule, change in program activities, inappropriate expectations to complete tasks or travel independently, illness or decline in function).
- Emotional upsets – Loss of family (usually death which may be poorly understood by the person), response to co-resident or staff leaving home (grief may be delayed), illness in patient or other close persons, seasonal pattern/anniversary reaction, trauma, and abuse (including sexual, verbal), or triggers to past abuses.
The most frequent new onset disorders are an ongoing (chronic) psychiatric condition; adjustment, mood, anxiety, or post traumatic stress. Autism is the most frequent comorbid psychiatric disorder across the range of functioning, where stereotypes, self-injurious, and compulsive behaviors are often seen as chronic comorbid conditions, especially in lower functioning individuals.

In evaluating persons with ID and behavior problems, one should be aware of the following:

- Persons with developmental disability show great heterogeneity in terms of functioning level, understanding, an ability to communicate needs, discomforts and concerns.
- If the individual has disruptive behavior, meet briefly with caregiver first. With the caregiver, establish the individual’s level of functioning, and seek advice from caregivers on the best way to meet and interact with the patient. Identify any circumstances that might be specifically upsetting for that individual such as asking too many questions; a noisy/busy environment; moving too close; reflecting surfaces such as the eyeglasses you may be wearing.
- Many individuals may be unable to communicate verbally, but are very aware of nonverbal behaviors in others. They may be very sensitized to negative attitudes in others toward them. Such individuals often depend on others to help modulate their emotions and will quickly pick up fear and anxiety in you. A warm, accepting, calm, reassuring attitude will help.
- Evaluation settings like the ER and physician’s office are strange and unfamiliar environments. A person with ID has even greater difficulty understanding what is going on and is likely to be very apprehensive and scared. The process of getting to the setting may also have been traumatic for both for the individual and their caregivers/family. Waiting itself can generate anxiety and contribute to behavioral difficulties. Take a moment to explain the reason for the wait to the patient and to the caregivers accompanying him/her. Assessment of the individual with ID takes time. Research indicates that assessing an individual with ID may take four times that required for the person without such disabilities.
- Appearances may be deceptive: individuals with ID may appear to be hearing impaired or unable to speak, but that does not mean they cannot hear or speak. They may have frequently overheard comments about them that have exacerbated the presenting problems.
- Practical tips in conducting the interview:
  - Try to make the individual as comfortable as possible
  - Familiarity helps – if possible, have the caregiver present
  - Use suggestions previously identified by caregiver
  - Encourage use of “comforter” items and behaviors – does the individual have a favorite item he/she likes to carry, or like to engage in self-soothing, like rocking or needing to stand?
  - Try to find a quiet spot, without interruptions
  - Try to establish a positive relationship with the individual:
    - Be interested in the precious object being clutched
    - Show warmth, positive regard
    - Be sensitive to cues, tone of voice
    - Be aware of your own nonverbal behavior as the patient may generalize to a previous experience
    - Indirect questions are often more productive than direct ones
• If the patient seems fearful, give him/her time to size you up
• Respect personal space
• Ask permission, explain, and pre-warn for examination procedures
• Find ways to communicate effectively
• Use simple words
• Speak slowly
• Do not shout
• Pause, do not overload individual with words
• Be sensitive to individual’s nonverbal clues and adjust your behavior accordingly
• Use visuals like drawings
• Use gestures
  o Remember that persons with ID have a variable and limited ability to interpret their own internal cues, and may not be able to give you an accurate picture of their internal state. Involvement of caregivers, who know the individual well, may be helpful in your attempts to better understand his/her subjective experiences. Reporting internal symptoms like hallucinations may be difficult, but there may be visible cues possibly representing these symptoms. Individuals with ID, especially DS, often use “self-talk,” which is not hallucinations.

At times, patients are taken to the ER and may need hospitalization. Although crises will be handled by the local (generic) mobile crisis teams, Division of Intellectual Disabilities Services (DIDS) has an Intensive Consultation Team in each region that may help with crisis plans, prevention and acute behavior issues.

Occasionally, a patient requires inpatient psychiatric hospitalization. This is often a problem as there is no specialized unit specifically designed and staffed for people with ID; psychiatrists, as other physicians, lack training and exposure to ID, and usual daily supports (behavior programs, feeding and dining programs, therapies) are not available.

Meeting patients’ needs and achieving treatment goals may be facilitated by:

• **Attention to inpatient routines** as they impact the individual. The patient’s caregivers may assist in adapting the hospital routine and procedures to be consistent with those that occurred in the patient’s home environment. For example, attention to how and when the client normally sleeps, how he/she engages in hygiene routines and other activities of daily living, or the best way to ensure medication is taken.

• **Attention to the physical environment.** For example, locating the patient in an end room/bed to reduce distress for the patient, possible disruption to other patients, and ensuring there is space for caregivers. If the patient requires restraint, it is usually helpful to ensure this is offered in a manner similar to that experienced in his/her home as in the community. Detailed procedures outlining the use of restraints are required, and caregivers should be able to provide this information to hospital staff.

• **Attention to staffing resources.** It is wise to find ways to provide consistency of nursing staff to the patient, and to identify nursing staff to the patient. Identify nursing staff with the responsibility to work with community caregivers so as to optimize the inpatient stay. Good communication with the community team and community physicians (e.g., family physician, community psychiatrist) while the patient is in the hospital usually contributes to a more comprehensive
DIDS Waiver personnel, especially the Intensive Consultation Teams, may help with providing in-hospital supports, discharge planning and transition back into the community.

The high incidence of behavior issues is a result of complex behaviors, and may often stem from undiagnosed medical conditions, and medication side effects that may interfere with psychiatric diagnosis and effective treatment. The first consideration in a person with ID, who displays behavior problems, especially if they are new, is a medical review. This should include investigation for a physical problem as well as a review of possible medication side effects.

The PCP may receive a request to rule out a medical problem. This will require a comprehensive review, but perhaps the most important element is the patient history provided by Waiver support staff. This should include Past Medical History (PMH), current diagnoses and problems, all medications patient is receiving, review of data kept in home (sleep, weight, diet, elimination, behavior, etc.), changes in health or functioning, and staff concerns (beyond “behavior”). If a behavior analyst has been involved, there may be data to support organicity, and clues to point in the direction of the problem.

Common undiagnosed medical problems are:

- Epilepsy, especially of new onset. This may occur in patients with dementia, especially persons with DS. This may not have been previously diagnosed.
- Hypo/hyperthyroidism
- Sensory deficits associated with behavior such as vision changes or hearing problems are easily treated. This is of particular importance in older patients, especially those with DS, where sensory deficits are often misdiagnosed as dementia.
- Functional decline. Individuals who have physical decline (motor functioning in persons with CP, cardiac in others, etc.) may become frustrated when expected to perform tasks. Individuals with early dementia (loss of short-term memory and loss of spatial abilities) may have trouble doing tasks such as dressing, taking the bus independently and when they refuse, are felt to be non-compliant. In general, the diagnosis of dementia has not been considered, again. This is seen in persons with DS.
- Reflux disease may cause pain and food refusal. The responses are often seen by staff as intentional and are reported as “behavior problems.” GERD may also interfere with sleep; this also is seen by staff as volitional behavior.
- Constipation is a major contribution to behavior problems secondary to physical discomfort. This may not be recognized by staff who chart “smears” (an attempt to expel hard stools) as a regular bowel movement.
- Unrecognized joint and bone pain. It is important to understand that negative behavior is often a form of communication about discomfort, and the problem is often due to treatable medical causes.
- Pain of any sort - toothache, fracture, etc.
Medications, particularly many used in people with ID, often have behavioral side effects or interactions leading to behavioral side effects. Multiple medications prescribed by several practitioners without clear communication creates risk for adverse effects. This problem may be further complicated when patients cannot express their discomfort. Many side effects are internally subjective. A doctor or caregiver cannot see double vision, nausea, dizziness, memory problems, limb tingling, headaches, etc. in someone else. In some situations, however, discussion of potential side effects with staff may produce development of an external sign that might indicate internal symptoms.

The incidence of psychiatric diagnoses in people with ID is equal to that found in the typical population (since the genetics are the same), but probably occur more often due to defects and variations in the central nervous system and experience.

An especially hard aspect of diagnosis in people with ID is the presence of a neurobehavioral phenotype where the behavior is “hard-wired,” not volitional, and does not respond to medication. In these patients, dysmorphic features may be a tip off to diagnosis. Symptoms noted include biting oneself (Lesch-Nyhan syndrome), pulling out fingernails or sticking foreign bodies in nose or ears (Smith-Magenis syndrome), gaze avoidance (Fragile X syndrome), hand wringing (Rett syndrome), uncontrollable laughter (Angelman syndrome), hyperphagia (Prader-Willi syndrome), and others.
The following is advice written by a psychiatrist to address psychiatric issues (written as a special contribution for SelectCommunity providers)

This is a brief guide to evaluating common situations that arise in the psychiatric care of people with ID.

The evaluation of people with ID is often more complex than for people without ID, but the general approach is the same. Though the patient may come with family, caregivers, nurses, behavior analysts, or an entire entourage, it is important to make proper introductions, and to establish rapport with the patient directly. One should describe the basic purpose of the appointment. Though there will be a range of abilities to answer questions about his or her condition, one should initially give the patient an opportunity to answer questions him or herself. It is important to pay attention to affect and nonverbal communication, as well as verbal communication. Most of the time, the patient has not made the appointment for him or herself, and he or she may not know why the referral was made. At the same time, by the time a referral to a psychiatrist has been made, most have some understanding that there is a problem with their “behavior.”

At some point, it will probably be necessary to obtain further history from the other caregivers. As with most patients, the history begins with development, and where development deviated from normal, expected development. While it is important to know previous diagnoses, it is important to try to obtain the signs and symptoms that led to the diagnoses, in case there have been changes in the presentation that may lead to a different diagnosis.

In general, there are four areas to consider when evaluating people with ID: medical conditions, environment, disorders usually diagnosed in childhood, and acquired psychiatric disorders.

**Medical conditions** that have an impact on behavior are myriad, but include seizure disorders, endocrine disorders, medications that have central nervous system effects, and pain from any cause. This is a wide and expansive list, but it truly exemplifies the fact that the psychiatrist evaluating a person with ID must also think like a PCP, as well as psychiatrist. One must keep in mind that although the patients often have around-the-clock care, caregivers are often unsophisticated in terms of medical knowledge.

**Environment** refers to conditions external to the patient. This includes the physical environment, but also the people who work with the patient. There is high turnover in this field, and it is always good to find out how long a person has been working with the patient. Environment also includes people that the patient lives with, whether peers, or family. Environment includes the conditions in which a behavior occurs. Is it when someone makes a demand? Does it seem to be for attention?
Disorders that are usually diagnosed in childhood would include ID, autistic spectrum disorders, ADHD, etc.

Acquired psychiatric disorders include mood disorders, psychotic disorders, anxiety disorders, etc.

Of course, things are rarely so simply deconstructed. There may be the presence of two, three, or even four active issues at the same time, and it may be difficult to tease out the most important factors at a given time.

There are seven rules of thumb that are helpful in providing psychiatric care for people with ID:

1. Gather as much information as possible. Listen to the patient, the parents, and the staff caregivers. Obtain as many records of previous evaluations as possible.
2. Make the best diagnosis possible with the information available. Unfortunately, people with ID who live with an agency may have lived with several different agencies over their lifetime, and getting all of the records may be difficult. Keep in mind that a diagnosis is a hypothesis, not a determination.
3. Use as few medications as possible. Many psychiatric medications have cognitive side effects, especially those with anticholinergic activity. As in general, treatment should be chosen based on the diagnosis, not the symptoms. Consider whether some of the patient's medications prescribed by other providers are causing the symptoms.
4. Be willing to review the diagnosis when more information becomes available. The diagnosis is a hypothesis. If the treatments aren't working, it could be that the diagnosis guiding the treatment is incorrect. There are sometimes new symptoms that arise which may be overlooked or discounted, and when seen together with the previous symptoms, change the differential diagnosis completely.
5. Don't mess with success. There are times when the patient seems to be doing well, and there are medications that seem to be superfluous to the treatment, and one wants to "clean things up." It is important to go slowly with changes in medications if there is no reason to make aggressive changes. There are plenty of times when there is a medication prescribed by another physician that should not be having any effect on the behavior, but changes in that medication cause definite deterioration in behavior.
6. Mess with success. Sometimes everyone is satisfied with the status of a patient, but it seems that they are 80 percent back to baseline, not 100 percent. Be willing to consider what it would take to go that last 20 percent to get a better result. Sometimes, there are good reasons to maintain treatment; other times, more aggressive treatment is well worth it.
7. Don't mess up people's holidays. People with ID who live with a provider agency may have limited contact with their families. These are often very important, and may be centered around holidays. Be mindful of the patient's social schedule, and realize that good visits with families are immensely important.
Other things to consider in the treatment of persons with ID include:

1  Threatening Suicide - In assessing a patient with ID, who has made threats of suicide, utilize many of the same methods for assessing risks as for people without ID.

First of all, assess the risk for suicide in the patient. Is there a history of suicide attempts? Was there lethal intent or capacity? Is there a mood or psychotic disorder present? Is there a substance abuse disorder present? Is there a family history of suicide? What are the precipitating factors? Are there psychological factors at work, such as suicide threats enabling access to tangibles like increased attention or reduced responsibilities? Recognize that there may be a combination of psychological factors as well as psychiatric conditions.

Secondly, assess the safety of the environment. Can the patient be managed safely at home during a time of increased risk? Are sharp objects locked up? Is there adequate staffing to monitor and respond the risk level? Understand that staffing available on a psychiatric inpatient unit is often less intense than the patient has on a routine daily basis, so there should be a clear reason to consider inpatient treatment.

Thirdly, communicate the treatment plan to the team, along with expectations of how the changes will reduce the risk. Communicate what conditions to watch for which would indicate that the situation should be reassessed.

2  Threatening to harm others - As in threats of suicide, in assessing a patient with ID who has made threats of harm to others, utilize many of the same methods for assessing risks as for people without ID.

First of all, assess the risk for danger to others in the patient. Is there a history of violence towards others? Was there lethal intent or capacity? Have others been injured? How severely? Is there a mood or psychotic disorder present? Is there a substance abuse disorder present? What are the precipitating factors? Are there psychological factors at work such that the threats are enabling access to tangibles like increased attention or reduced responsibilities? Recognize that there may be a combination of psychological factors, as well as psychiatric conditions.

Secondly, assess the safety of the environment. Can the patient be managed safely at home during a time of increased risk? Are sharp objects locked up? Is there adequate staffing to monitor and respond the risk level? Understand that staffing available on a psychiatric inpatient unit is often less intense than the
3 Poor Impulse Control/Running away - elopement - Poor impulse control, running away or elopement is often a reason for psychiatric referral. The first task is to evaluate the severity of the elopement. Generally in psychiatry, we refer to elopement when a patient has left the grounds, and no one knows where the patient is. For the population with ID, elopement often means that the patient is headed towards the door, or walked through the door in plain sight of everyone, but is not out of sight and missing. It is important to make that distinction. Think through the checklist of medical, environmental, child onset, and acquired psychiatric disorders. Medical explanations are less likely in this case, but there could certainly be environmental factors that lead to or maintain the poor impulse control or elopement behavior. Child onset diagnoses could include ID, autistic spectrum disorders, or ADHD. Acquired disorders could include mania or psychotic disorders. Of course, there are not specific medications for this symptom, but based on the diagnosis, or hypothesis, many different medications may improve the symptom (a stimulant for ADHD, atypical antipsychotic for autistic spectrum disorder, or mood stabilizer for mania). It is important to communicate the expectations of how the effectiveness of the medication will be evaluated to the patient and caregivers.

4 Refusal - noncompliance. Refusal or noncompliance is a common reason for referral to a psychiatrist. The first question that comes to mind is whether this is new behavior, and if it is not, why does it warrant evaluation at this time? If the behavior is new, there may be many reasons for the change in behavior. If the behavior is not new, the reason for referral is more likely due to the environment than the patient.

It is important to have a full description of the refusal or noncompliance. Are there medical conditions causing discomfort and behavioral changes? Are there changes in medications? What environmental changes are present? Is the noncompliance something that occurs every day? At particular times during the day? With particular people or with everyone? Changes in staffing ratios? New staff? What level of dangerousness occurs with the refusal?

Finally, refusal or noncompliance may or may not be part of an acquired psychiatric syndrome. While the caregivers may be focusing on the patient's noncompliance, there may be other symptoms which would suggest depression, mania, psychosis, obsessive compulsive disorder, or other disorders. If one of
these diagnoses can be made, then treatment of the disorder may reduce the symptoms of refusal or noncompliance.

5 Self-Injurious Behavior - Self-injurious behavior is complex to diagnose and treat. As with the other behaviors discussed, it is important to establish whether the behavior is new or has been going on for years. It is important to know how it has previously been assessed and treated. The typography of the self-injurious behavior can vary widely from flicking one’s finger against another to pulling out one’s finger and toenails, to banging one’s head against the corner of the door. What is the nature of the injuries? How frequently does the behavior occur?

Are there medical issues? The most common medical issue associated with this behavior is probably allergic rhinitis. It is important that the primary care doctor has evaluated the patient for allergies as well as other medical issues. It is important that the PCP know his/her evaluation is part of a multidisciplinary evaluation of problematic behavior, not just a search for a signature on a page.

Environmental issues are often important for self-injurious behavior. All of the same questions about the conditions for other behaviors apply in this case as well. A particularly important question about the environment in self-injurious behaviors is how the person handles transitions from one activity to another. For some people with severe or profound ID or with autistic spectrum disorders, they have difficulties in transitioning from one activity to another. This may be transitioning from home to a day program, or from leaving a store, or preparing for bed. The transition may even be external to them, for instance, when the patient’s roommates return from the day program and the house becomes more crowded with peers and staff, or the change in shift.

As for the other behaviors discussed, one would evaluate for chronic and new psychiatric disorders. There is a diagnosis, rarely used outside people with ID, of Stereotypic disorder with self-injurious behavior, which may be appropriate.

6 Aggression/ Property Destruction - The assessment of aggression and property destruction is similar to that for threats to others.

First of all, there needs to be a description of property destruction. Sometimes, behavior is called property destruction when there is only a risk of property destruction. For example, the person hit an object but nothing broke, and there is no sign of damage. Sometimes property destruction means there is damage, but it is cosmetic. The term property destruction implies there is intent to destroy. A frequent presentation in someone with autistic spectrum disorder would be pulling at loose threads until the article unravels. This does not involve willfully destroying property, but if one T-shirt is destroyed every day or every week, then the amount of money involved may be significant.
Suspected Hallucinations - The evaluation of hallucinations in people with ID may be challenging. There may often be a complaint that a patient talks to themselves. When the person’s talking to themselves consists of talking about their planned activities or past conversations, it is usually not pathologic. Usually the goal in terms of psychiatric diagnosis is to determine whether a psychotic disorder is present, such as schizophrenia or bipolar disorder. Certainly one can directly ask if the patient hears voices that no one else can hear, but, as with people without ID, the reliability of the answer is often suspect. Input from caregivers may help determine whether the person appears to respond to internal stimuli. At times, this may be observed during the appointment. Additionally in evaluating for a diagnosis of psychosis in schizophrenia or mania, it is important to remember that hallucinations are only one of the criteria. If “talking to herself” is the only criterion, then it is less likely that the person has a diagnosis consistent with schizophrenia or mania.

If there is a new presentation of hallucinations, the possibility of delirium should be entertained. Is there a current illness or new medications that have been started? Is there a change in mental status or sleep-wake cycle that would suggest delirium?

Anxiety - Anxiety can be difficult to assess. Anxiety disorders such as panic disorder, obsessive compulsive disorder, and post-traumatic stress disorder can be seen in persons with ID, but anxiety as a symptom may be seen in medical conditions, side effects of medications, or as a symptom in the context of other psychiatric disorders. The history should provide some help in the differential diagnosis. Are the symptoms new onset or longstanding? Have there been any medication changes? Anxiety may be a sign of withdrawal for medications such as benzodiazepines or barbiturates. Anxiety may be a symptom in the context of depression, psychosis, mania, or even autistic spectrum disorders. The presence of other signs and symptoms of those disorders should help in
Mood Symptoms - Mood disorders are some of the most common disorders in persons without ID, as well as those with ID. The diagnosis of depression includes depressed mood, but it also includes neurovegetative signs as well. These are observable, and sometimes even quantifiable. Decreased interest in pleasurable activities, reduced appetite or weight, insomnia or hypersomnia, psychomotor retardation, or lack of energy are all observable behaviors. Feelings of worthlessness may be expressed by self-deprecating statements. Recurrent thoughts of death may be seen in people with ID by talking about death, or talking about family members who have previously died.

Similarly, the diagnosis of mania has many signs which are observable by others. Increased interest in pleasurable activities, change in appetite, reduced need for sleep, and psychomotor agitation are all observable. Grandiosity may be manifested in statements that the individual owns all of the cars in the parking lot, or other statements that are clearly elevated beyond attainment.

Overuse of 911 - Some people repetitively call 911 for emergencies. Some of those people have ID. They may call 911 because of the excitement that comes with the emergency call. The police or ambulance staff may have given them a ride around the block previously, and the person is hoping for another ride. Some like the increased attention that comes with the visit to the ER. There is probably nothing that can be done about the fact that when a person calls 911, the emergency responders respond, unless legal action has been taken. Mostly there needs to be a behavioral plan in place that provides attention to the person that can replace the need to call 911. As part of the behavioral plan, there may be included a visit to the fire station or emergency room if the other contingencies are met.

However, sometimes the use of 911 may signal another symptom. On one occasion, inquiry into the use of 911 revealed the patient was reporting that she was pregnant each time she called 911. The patient had a diagnosis of schizophrenia, and a delusion that she was pregnant was a recurrent delusion for her. The patient was not pregnant, and was not calling just to get attention, but did have a symptom which responded to an increase in antipsychotic medication.

Accusations of abuse - Whenever a person with ID makes an accusation of abuse or neglect, it has to be investigated. The psychiatrist needs to be sure that the accusation was reported. Some people make frequent accusations, and make accusations that are essentially in retaliation for not getting what they want. Their accusations are still going to be investigated. This is a population that has an increased level of abuse and neglect perpetrated against them.
There are several important and pertinent publications. Tennessee Department of Mental Health and Disabilities has best practice guidelines for adolescents and children. These guidelines are on diagnosis and comorbid disorders in the population with ID, and although they were written for children, much is applicable to adults with ID. Recently, the DSM-IV has been adapted for the population with ID by a joint project of National Association for the Dually Diagnosed (NADD) in association with the American Psychological Association (APA), and is published as “DM-ID” by the NADD press. This is a diagnostic manual designed to facilitate an accurate DSM diagnosis in persons with ID, and to provide a discussion of the issues involved in reaching an accurate diagnosis using the evidence-based model, supported by the expert-consensus model.

**Preventive Services**

Preventive services are as important for people with ID as the general population. Unfortunately, a family history is not often available, but many parents are still living, and this information should be sought. Preventive care should follow that for anyone else, and guidelines exist as written by a number of medical organizations, state public health departments, and insurance companies. In addition, depending on medical care (e.g. medications such as Dilantin or psychotropics), diagnosis (e.g., DS, TS), and other factors, additional tests and procedures should be considered. For instance, because of the high incidence of Alzheimer’s disease in people with DS, screening for Alzheimer’s disease should begin at age 40. This would be an evaluation for loss of function so the surveillance should begin with a simple baseline for this population. Several simple tests exist. At least two states, Massachusetts and South Carolina, have published preventive care guidelines for people with ID.

Lack of preventive care is one of the biggest issues in the care of individuals with ID. Many people die of diseases like breast cancer, colon cancer, atherosclerosis, and etc., that could have been diagnosed and treated early. There are several reasons for this. One reason is that it is felt that people with ID don’t live very long. This should not be a barrier since most people with ID, except for a few with specific genetic disease, have the potential to live to the same life spans as the typical population, with good medical care. Another reason is obstacles created by either physical disabilities making procedures difficult to perform, or behavior problems that interfere with care. The majority of these problems may be overcome by adaptations (some just as simple as more time), or behavior approaches, both of which may be addressed with the PCP by Waiver therapists.

Another problem is failure to keep up with immunizations. This often stems from not transferring medical records. This either results in unneeded, and perhaps painful and anxiety-producing, immunizations, or lack of important immunizations like hepatitis A, pneumococcal disease, etc.

Preventive care and possible need for adaptations should be reviewed yearly by the PCP, preferably with adequate information being provided, and consideration of use of Waiver therapists to help with difficult procedures.
Resources
Health Screening Recommendations, Massachusetts Department of Mental Retardation.

Abbreviations, Mnemonics and Terms
AT - Assistive Technology
COS - Circle of Support
DIDS - Division of Intellectual Disability Services
HCBS - Home and Community Based Services
ID - Intellectual Disabilities
ISC - Independent Support Coordination
ISP - Individual Support Plan
MCO - Managed Care Organization
Medical Home Model - Concept of care managed/coordinated by a personal physician
MR - Mental Retardation
PAE - Pre-Admission Evaluation
PCP - Primary Care Provider
Waiver Programs - Residential program, therapy, work, etc. provided by the “State”
WHO - World Health Organization
Resources
A short clinical guide specific to the developmentally disabled population will be available for SelectCommunity providers, in addition to this handbook, which is merely a simple bridge to point efforts in the right direction:

Another resource is recommended:

Individuals at both VSHP and DIDS have many resources available on request. Other resources:
1) Go to the Medscape Today website and type in “mental retardation” or “emedicine,” and 573 up-to-date scientific articles will be available for developmentally disabled diagnoses, including most of the common genetic diagnoses.
2) The Family Village site, www.familyvillage.wisc.edu/, provides a “library” of both scientific articles and resources for family and caregivers, including diagnostic support groups.
3) The California DIDS has an extensive site www.ddhealthinfo.org for health care providers; which includes free category CME credits.
4) Special Olympics, www.specialolympicstn.org, has a “healthy athletes” program which includes health screenings and information on difficult to address/overlooked issues such as dental care, foot care, vision and hearing screening, etc.
5) Textbooks such as Rubin and Crocker’s Medical Care for Children and Adults with Developmental Disabilities