Whole Exome and Genome Sequencing

DESCRIPTION

 Whole exome sequencing (WES) analyses the portion of the genome that contains protein-coding DNA, known as exons; whole genome sequencing (WGS) analyses both coding and noncoding regions of the genome. WES and WGS have been proposed for use in children presenting with disorders and anomalies that have not been explained by standard clinical workup. While exons represent only about 2% of the genome, they account for approximately 85% of disease-causing genetic variants.

Genetic counseling is primarily aimed at individuals who are at risk for inherited disorders. Experts recommend formal genetic counseling when genetic testing for an inherited condition is considered.

POLICY

- Whole exome sequencing (WES) may be considered medically necessary if the medical appropriateness criteria are met (See Medical Appropriateness below).

- Whole exome sequencing (WES) when used as a screening tool for asymptomatic individuals is considered investigational.

- Whole genome sequencing (WGS) is considered investigational.

- Pre- and post- genetic counseling as an adjunct to genetic testing is considered medically necessary.

MEDICAL APPROPRIATENESS

- Whole exome sequencing (WES) may be considered medically appropriate if ALL of the following are met:
  - Individual is 18 years of age or younger
  - There is potential for change in treatment or management of the individual being tested
  - Congenital anomalies and/or neurodevelopmental disorder(s) cannot be explained by standard non-invasive tests
  - Whole exome sequencing (WES) would be ANY ONE of the following:
    - Genetic etiology is the most likely explanation, even though no genetic testing has been done
    - A better alternative than standard invasive testing (e.g. muscle biopsy)
    - Previous genetic testing (e.g. targeted single gene testing, panel testing) has failed to yield a diagnosis

IMPORTANT REMINDERS

- Any specific products referenced in this policy are just examples and are intended for illustrative purposes only. It is not intended to be a recommendation of one product over another, and is not intended to represent a complete listing of all products available. These examples are contained in the parenthetical e.g. statement.

- We develop Medical Policies to provide guidance to Members and Providers. This Medical Policy relates only to the services or supplies described in it. The existence of a Medical Policy is not an authorization, certification, explanation of benefits or a contract for the service (or supply) that is referenced in the Medical Policy. For a determination of the benefits that a Member is entitled to receive under his or her health plan, the Member's health plan must be reviewed. If there is a conflict between the Medical Policy and a health plan, the express terms of the health plan will govern.
ADDITIONAL INFORMATION

There are no regulations required by the U.S. Food and Drug Administration (FDA) for genetic testing. Laboratories performing clinical tests must be certified for high complexity testing under the Clinical Laboratory Improvement Amendments (CLIA) of 1988.

Published evidence of well-designed studies in peer review journals is insufficient to determine whether the use of whole genome sequencing tests improve health outcomes.

SOURCES


This document has been classified as public information.


**EFFECTIVE DATE**

11/1/2017

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