First-Trimester Detection of Down Syndrome Using Fetal Ultrasound Markers Combined with Maternal Serum Assessment

DESCRIPTION

Definitive diagnosis of Down syndrome and other chromosomal abnormalities requires amniocentesis or chorionic villus sampling (CVS), both of which are invasive procedures that carry a risk of miscarriage estimated at 0.5% to 1%. Less invasive screening programs have been developed with the use of biochemical markers and ultrasound that show an association with Down syndrome.

First trimester screening for Down syndrome may include measurement of nuchal translucency (ultrasound detection of subcutaneous edema in the fetal neck) combined with maternal serum assessment including free beta subunit of human chorionic gonadotropin (β-hCG) or total human chorionic gonadotropin (hCG), along with pregnancy-associated plasma protein-A [PAPP-A]. Cell-free DNA testing may also be performed. A specific risk estimate is calculated using these results as well as maternal factors such as maternal age, prior history, weight, race, and number of fetuses. Measurement of nuchal translucency alone is less effective for first-trimester screening than the combined testing.

Another potential ultrasound marker is fetal nasal bone examination. The technique for assessing the nasal bone using ultrasound involves viewing the fetal face longitudinally and exactly in the midline. The nasal bones are considered to be present if the line within the bridge of the nose is more echogenic than the overlying skin and absent if the echogenicity is the same or less than the skin, or if it is not visible. The absence of fetal nasal bone is considered a positive test result, indicating an increased risk of Down syndrome.

POLICY

- First-trimester screening for detection of Down syndrome incorporating maternal serum assessment and measurement of fetal nuchal translucency may be considered ***medically necessary*** if the medical appropriateness criteria are met. **(See Medical Appropriateness below.)**

- First-trimester screening for detection of Down syndrome using measurement of nuchal translucency alone is considered ***investigational.***

- First-trimester screening for detection of Down syndrome using fetal nasal bone assessment translucency is considered ***investigational.***

MEDICAL APPROPRIATENESS

- First-trimester screening for detection of Down syndrome is considered **medically appropriate** if **ALL** of the following are met:

    - Screening includes measurement of fetal nuchal translucency with **ANY ONE** of the following:
        - Maternal serum markers (free beta subunit of human chorionic gonadotropin [β-hCG] or total human chorionic gonadotropin (hCG), and pregnancy-associated plasma protein-A [PAPP-A])
        - Maternal plasma using cell-free fetal DNA (cffDNA) with singleton pregnancy

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 is insufficient evidence on the performance of fetal nasal bone assessment to determine its impact on health outcomes. Additional studies are needed before conclusions can be drawn about its utility.

SOURCES

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