

Medical Coverage Policy | First-Trimester Detection of Down Syndrome Using Fetal Ultrasound Markers



EFFECTIVE DATE: 11 | 17 | 2005

POLICY LAST REVIEWED: 07 | 16 | 2025

OVERVIEW

Ultrasound (US) markers can potentially increase the sensitivity of biochemical measures for first-trimester detection of Down syndrome. Nuchal translucency (NT) refers to the US detection of subcutaneous edema in the fetal neck between weeks 10 and 13 of gestation. Fetal nasal bone examination involves US assessment at 11 to 14 weeks of gestation to identify the presence or absence of the nasal bone.

Note: This policy only addresses the US markers nuchal translucency and fetal nasal bone assessment.

MEDICAL CRITERIA

Not applicable

PRIOR AUTHORIZATION

Not applicable

POLICY STATEMENT

Medicare Advantage Plans and Commercial Products

First-trimester screening for detection of Down syndrome incorporating maternal serum markers and measurement of fetal nuchal translucency may be considered medically necessary for women who are adequately counseled and desire information on the risk of having a child with Down syndrome.

First-trimester screening for detection of Down syndrome using measurement of nuchal translucency alone is not covered for Medicare Advantage Plans and not medically necessary for Commercial products as the evidence is insufficient to determine the effects of the technology on health outcomes.

First-trimester screening for detection of Down syndrome incorporating fetal nasal bone assessment translucency is not covered for Medicare Advantage Plans and not medically necessary for Commercial products as the evidence is insufficient to determine the effects of the technology on health outcomes.

COVERAGE

Benefits may vary between groups/contracts. Please refer to the appropriate member Benefit Booklet, Evidence of Coverage, or Subscriber Agreement for applicable radiology benefits/coverage.

BACKGROUND

Definitive diagnosis of Down syndrome and other chromosomal abnormalities requires amniocentesis or chorionic villus sampling, both of which are invasive procedures that carry a risk of miscarriage estimated at 0.5% to 1%. Because of this risk, before biochemical screening existed, diagnosis was generally only offered to women aged 35 years or older, for whom the risk of the procedure approximated the risk of Down syndrome. However, most babies with Down syndrome are born from mothers younger than 35 years, even though the mothers are at lower individual risk. This situation created interest in developing less invasive screening programs based on assessment of serum markers that have shown associations with Down syndrome. In the late 1980s, biochemical screening at 16 weeks of gestation was developed and began to be offered to all pregnant women. Biochemical screening consists of maternal serum measurements α -fetoprotein, human chorionic gonadotropin (hCG), and unconjugated estriol (ie, triple screen). More recently, a fourth marker has been used, inhibin-A (quadruple screen). The triple screen identifies approximately 69% of Down syndrome pregnancies and the quadruple screen 81%, both at a 5%

false-positive rate. This false-positive rate refers to the proportion of all tests administered that are falsely positive at the cutoff point that produces that particular value of sensitivity. Among women who test positive, only about 2% actually have a fetus with Down syndrome.

There has been interest in Ultrasound (US) markers to improve the accuracy of biochemical screening. One potential marker is fetal NT. This refers to the US detection of subcutaneous edema in the fetal neck and is measured as the maximal thickness of the sonolucent zone between the inner aspect of the fetal skin and the outer aspect of the soft tissue overlying the cervical spine or the occipital bone. In the early 1990s, screening studies of pregnant women reported an association between increased NT in the first trimester of pregnancy (10-13 weeks of gestation) and chromosomal defects, most commonly Down syndrome (trisomy 21), but also trisomy 18 and 13. NT could be done alone as a first-trimester screen or in combination with maternal serum markers, free beta subunit of hCG and pregnancy-associated plasma protein-A. These serum markers differ from those used in the second-trimester triple or quadruple screen.

Another potential US marker is fetal nasal bone examination. The technique for assessing the nasal bone is to view the fetal face longitudinally and exactly in the midline. The nasal bone synostosis resembles a thin echogenic line within the bridge of the nose. The nasal bones are considered to be present if this line 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 to be a positive test result, indicating an increased risk of Down syndrome. In some cases, the sonographer will not be able to visualize the nasal area of the fetus's face and thus cannot make a determination of the presence or absence of nasal bone. The inability to visualize the nasal bone is regarded as an unsuccessful examination, rather than a positive test result. Fetal nasal bone examination can be done from 11 weeks to just before 14 weeks of gestation. It is sometimes recommended that, if the nasal bone is absent on US done between 11 and 12 weeks of gestation, a second examination be done 2 weeks later. Fetal nasal bone assessment can be done along with NT, or in the second step of a 2-stage screen for cases that are borderline using other first-trimester markers.

For individuals who are pregnant and in the first trimester who receive first-trimester Down syndrome screening of maternal serum markers and nuchal translucency, the evidence includes observational screening studies. Relevant outcomes are test accuracy and validity and resource utilization. There is sufficient evidence from 2 large multicenter prospective studies the Serum, Urine, and Ultrasound Screening Study (SURUSS) and the First and Second Trimester Evaluation of Risk (FASTER) trials well as several smaller studies, that first-trimester screening for Down syndrome with measurement of fetal nuchal translucency (NT) and maternal serum markers is at least as accurate as alternative tests and may allow earlier confirmation or exclusion of Down syndrome. The evidence is sufficient to determine qualitatively that the technology results in a meaningful improvement in the net health outcome.

For individuals who are pregnant and in the first trimester who receive first-trimester Down syndrome screening of nuchal translucency alone, the evidence includes observational screening studies. Relevant outcomes are test accuracy and validity and resource utilization. The large multicenter prospective studies SURUSS and FASTER found, overall, that first-trimester screening with NT alone is inferior to first or second-trimester combined screening. Additional testing may not be necessary in those few cases when NT is at least 4.0 mm due to the high likelihood of Down syndrome, but this would affect only a very small number of cases (0.09%-0.3%). The evidence is insufficient to determine the effects of the technology on health outcomes.

For individuals who are pregnant and in the first trimester who receive first-trimester Down syndrome screening of fetal nasal bone, the evidence includes several observational studies. Relevant outcomes are test accuracy and validity and resource utilization. The accuracy of testing in the published literature is variable, with some studies reporting relatively low sensitivity rates. The variability in accuracy reported may reflect the difficulty in performing and interpreting this test, and test results are likely prone to differences in operator characteristics. Limited evidence has suggested that there may be modest incremental benefit when the test is used in combination with NT measurement and serum markers, but the degree of benefit is unclear. The evidence is insufficient to determine the effects of the technology on health outcomes.

CODING

Medicare Advantage Plans and Commercial Products

The following CPT code(s) are covered if the conditions cited above are met:

- 76813** Ultrasound, pregnant uterus, real time with image documentation, first trimester fetal nuchal translucency measurement, transabdominal or transvaginal approach; single or first gestation
- 76814** Ultrasound, pregnant uterus, real time with image documentation, first trimester fetal nuchal translucency measurement, transabdominal or transvaginal approach; each additional gestation (List separately in addition to code for primary procedure)

There is no specific CPT code(s) for ultrasound assessment of fetal bone translucency. It should be reported with an unlisted code.

RELATED POLICIES

Unlisted Procedures

PUBLISHED

Provider Update, September 2025

Provider Update, September 2024

Provider Update, April 2023

Provider Update, June 2022

Provider Update, May 2021

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