

**Medical Coverage Policy** | First-Trimester Detection of Down Syndrome Using Fetal Ultrasound Markers Combined with Maternal Serum Assessment



**EFFECTIVE DATE:** 11 | 17 | 2005  
**POLICY LAST UPDATED:** 06 | 07 | 2016

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

## MEDICAL CRITERIA

Not applicable

## PRIOR AUTHORIZATION

Not applicable

## POLICY STATEMENT

### BlueCHiP for Medicare 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 medically necessary as there is insufficient peer-reviewed literature that demonstrates that the procedure is effective.

First-trimester screening for detection of Down syndrome incorporating fetal nasal bone assessment translucency is not medically necessary as there is insufficient peer-reviewed literature that demonstrates that the procedure is effective.

## 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 of alpha-fetoprotein, human chorionic gonadotropin, and unconjugated estriol (i.e., triple screen). More recently, there has been the option of a fourth marker, 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 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 human chorionic gonadotropin (B-hCG) and pregnancy-associated plasma protein-A (PAPP-A). These are different serum markers than 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 using US involves viewing 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.

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.

There is sufficient evidence from 2 large prospective multicenter studies (SURUSS, FASTER) and several smaller studies that first-trimester screening for Down syndrome with measurement of fetal NT and maternal serum markers is at least as accurate as alternative tests and may allow earlier confirmation or exclusion of Down syndrome. Therefore, use of this test in the first trimester is a reasonable approach and may be considered medically necessary. The SURUSS and FASTER studies also found that overall first-trimester screening with NT alone is inferior to either 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 in these cases.

Studies have found a high rate of successful imaging of the fetal nasal bone and an association between absent nasal bone and the presence of Down syndrome in high-risk populations. However, there is insufficient evidence on the performance of fetal nasal bone assessment in average-risk populations. Of particular concern is the low performance of fetal nasal bone assessment in a subsample of the FASTER study conducted in a general population sample. Two studies conducted outside of the United States have found that, when added to a first-trimester screening program evaluating maternal serum markers and NT, fetal nasal bone assessment can result in a modest decrease in the false-positive rate. Several experts in the field are proposing that fetal nasal bone assessment be used as a second stage of screening, to screen women found to be of borderline risk using maternal serum markers and NT. Additional studies using this contingent approach are needed before conclusions can be drawn about its utility. In summary, given the uncertainty of

test performance in average-risk populations and the lack of standardization in the approach to incorporating this test into a first-trimester screening program, detection of fetal nasal bone is considered not medically necessary as there is insufficient peer-reviewed literature that demonstrates that the procedure is effective.

## **CODING**

### **BlueCHiP for Medicare and Commercial Products**

The following CPT codes are covered if the conditions cited above are met:

**76813      76814**

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

## **RELATED POLICIES**

Preauthorization via Web-Based Tool for Genetic Testing

## **PUBLISHED**

Provider Update, August 2016

Provider Update, December 2015

Provider Update, February 2009

Policy Update, January 2008

## **REFERENCES**

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