Prenatal Maternal Serum Screening

Procedures addressed

The inclusion of any procedure code in this table does not imply that the code is under management or requires prior authorization. Refer to the specific Health Plan's procedure code list for management requirements.

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What is prenatal maternal serum screening

Definition

Some factors predict an increased risk for Down syndrome and NTDs, such as maternal age, family history, and maternal diabetes or seizure disorder. However, there are no recognizable risk factors to explain the vast majority of babies born with these birth defects.\(^4,5\) As a result, prenatal screening to identify affected pregnancies is routinely offered to all pregnant women.\(^5,6\)

- About 3% of pregnancies have some type of birth defect.\(^1\) Down syndrome and neural tube defects (NTDs) are among the most common serious birth defects. Down syndrome affects about 1 in 700 live births.\(^2\) NTDs, such as spina bifida and anencephaly, affect about 3000 pregnancies per year in the United States.\(^3,4\)
- While not the focus of maternal serum screening programs, other birth defects (such as abdominal wall and heart defects) and general risks for poor pregnancy outcome may also be identified.
Test information

- Prenatal screening relies on maternal serum markers, and sometimes nuchal translucency ultrasound data (ACOG recommended technique when available)\(^6\) to predict a pregnancy's risk for Down syndrome, open neural tube defects, and other rarer birth defects such as trisomy 18.

Typical marker patterns for these birth defects are seen in the first and second trimesters. Measurements are provided as multiples of the median (MoM), which compare results to normal population medians. Therefore, values are higher or lower relative to 1.0. Risk assessment algorithms evaluate several factors, so pregnancies may be at-risk without each marker being abnormal.

- Screening results are generally reported as “screen positive” for Down syndrome or trisomy 18 if the predicted risk exceeds a laboratory-determined risk cut-off (often about 1 in 270 for Down syndrome and 1 in 100 for trisomy 18). A pregnancy is screen-positive for neural tube defect if the maternal serum AFP exceeds a cut-off, which is usually 2.5 MoM.\(^4\) However, different MoM calculations or cut-offs may be used for those with recognized risk factors or multiple gestations.\(^7\)

Guidelines and evidence

- Practice guidelines from the American College of Obstetricians and Gynecologists (ACOG, 2016) address prenatal screening for chromosome abnormalities. ACOG recommends the following:
  - “Aneuploidy screening or diagnostic testing should be discussed and offered to all women early in pregnancy, ideally at the first prenatal visit.” [evidence level C: “consensus and expert opinion”]\(^6\)
  - “All women should be offered the option of aneuploidy screening or diagnostic testing for fetal genetic disorders, regardless of maternal age.” [evidence level C]\(^6\)
  - Several other level A and B recommendations are made about test effectiveness, choice, patient counseling, and follow-up.

- The American College of Medical Genetics (ACMG)\(^7\) and the American Academy of Family Physicians (AAFP)\(^5\) have also published prenatal screening statements similar to ACOG’s recommendations.

- While the ACOG guidelines focus primarily on Down syndrome screening, they do include this recommendation about ONTD screening: “Women who undergo first-trimester screening should be offered second-trimester assessment for open fetal defects (by ultrasonography, MSAFP screening, or both) and ultrasound screening for other fetal structural defects.” [evidence level A]\(^6\) A 2003 ACOG practice guideline more directly addressed NTD screening: “Maternal serum alpha-fetoprotein evaluation is an effective screening test for NTDs and should be offered to all pregnant women.” [evidence level A]\(^4\)
Criteria
Screening for aneuploidy by ONE of the following methods is covered one time per pregnancy:

- First trimester screening – Total or free beta-HCG and PAPP-A levels performed on a maternal serum sample performed in conjunction with an ultrasound measurement of fetal nuchal translucency (NT)**
- Second trimester screening – human chorionic gonadotropin (hCG), alpha-fetoprotein (AFP), unconjugated estriol (uE3), and dimeric inhibin-A (DIA) performed on a maternal serum sample
- Integrated, step-wise sequential, or contingent sequential screening – combines results of first and second trimester screening in various testing algorithms.

**Limits on prenatal ultrasonography will depend on the insurer’s ultrasound coverage policy and are outside the scope of this program.

References