CLINICAL POLICY AND PROCEDURE MANUAL

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PA.239.CC – Noninvasive Genetic Testing During Pregnancy

Summary:
ACOG estimates that 6-11% of stillbirths and neonatal deaths result from aneuploidies (fetus with missing or extra chromosomes). Most aneuploidies involve the presence of an extra chromosome, also referred to as trisomy.

Down syndrome, which is most commonly caused by trisomy 21 (T21), is routinely evaluated as the standard of care for the majority of the 4 million women who give birth each year in the United States. Conventional screening tests typically involve measurement of blood serum markers in conjunction with ultrasound followed by recommendation for diagnostic invasive procedures for abnormal results from screening.

First trimester combined screening (FTS) and integrated screening (INT) have the best screening performance, yet still only have T21 detection rates of 82-87% and 88-95%, respectively, at false positive rates of 5%. Invasive testing with amniocentesis or CVS is highly accurate but has up to a 3% risk of procedure related miscarriage. The reported complication rates have come down in the last 10 years, but there is still some procedure-related risk.

A prenatal test that evaluates cell-free DNA (cfDNA) in maternal blood has been shown to be highly accurate, with T21 detection rates >99%, 98% detection rate for fetal trisomy 18 and 99% detection rate for fetal trisomy 13 with a combined false positive rate of 0.13%.

cfDNA testing, can detect more T21 cases and at the same time reduce unnecessary invasive procedures and in turn fewer procedure related fetal losses. cfDNA testing, when used as a follow-up test for an abnormal result from the FTS or INT screening test can spare the vast majority of the 5% of women with false positive results from undergoing invasive diagnostic testing. Any woman with an abnormal result from

cfDNA test should undergo diagnostic testing by amniocentesis or chorionic villus sampling.

Clinical Criteria:

CountyCare considers cell-free fetal DNA-based prenatal screening for fetal aneuploidy (trisomy 13, 18, and 21) medically necessary for women with a current single or twin gestation pregnancy.

OR

Cell-free fetal DNA-based prenatal testing for fetal sex determination is considered medically necessary for singleton pregnancies at increased risk of a sex (X)-linked condition or congenital adrenal hyperplasia.

Limitations:

Cell-free fetal DNA-based prenatal screening for fetal aneuploidy (trisomy 13, 18, 21) is considered not medically necessary for individuals not meeting the criteria above, including pregnancies involving 3 or more fetuses.

OR

Cell-free fetal DNA-based prenatal screening for fetal aneuploidy (trisomy 13, 18, and 21) in twin pregnancies is considered not medically necessary when the current pregnancy is affected by fetal demise, vanishing twin, or one or more anomaly detected in one or both of the twins.

OR

Cell-free fetal DNA-based prenatal testing for fetal sex determination is considered not medically necessary for pregnancies without an increased risk of a sex (X)-linked condition or congenital adrenal hyperplasia.

OR

Cell-free fetal DNA-based prenatal testing is considered not medically necessary for all other indications, including testing for microdeletion syndromes.

Cell-free fetal DNA-based screening for indications other than those listed in above clinical coverage criteria will not be covered.

Only one cell-free DNA testing will be covered per pregnancy. Only one screening modality will be covered per pregnancy.

In patients with a positive screen, providers should consider evaluation with a maternal fetal medicine specialist, genetic counseling, comprehensive ultrasound and diagnostic testing.

Applicable Codes:

CPT codes covered if selection criteria are met:	
Code	Description
81420	Fetal chromosomal aneuploidy (eg, trisomy 21, monosomy X) genomic sequence analysis panel, circulating cell-free fetal DNA in maternal blood, must include analysis of chromosomes 13, 18, and 21
81479	Unlisted molecular pathology procedure
81507	Fetal aneuploidy (trisomy 21, 18, and 13) DNA sequence analysis of selected regions using maternal plasma, algorithm reported as a risk score for each trisomy

References

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Revision History:

Revision	Date
Policy Created	April 2021
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