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Prenatal Testing for Fetal Aneuploidy

I. Policy Description

Aneuploidy is defined as an abnormal number of chromosomes present in the cell. Fetal aneuploidy is a condition where the fetus has one or more extra or missing chromosomes leading to either a nonviable pregnancy, offspring that may not survive after birth, or surviving newborn with congenital birth defects and functional abnormalities. The most common fetal aneuploidies associated with an additional chromosome are Down syndrome (trisomy 21), Edwards syndrome (trisomy 18), and Patau syndrome (trisomy 13). Prenatal screening for fetal aneuploidy is an assessment of the pregnant individual's risk of carrying a fetus with fetal aneuploidy using markers found in maternal serum.[1] Noninvasive-invasive prenatal screening is a method for screening for chromosomal abnormalities using a maternal blood sample where cell-free fetal DNA (cffDNA) is extracted and screened for aneuploidies.[2]

For guidance on the use of chromosomal microarray in the confirmation of fetal aneuploidy, please see AHS-M2033-Chromosomal Microarray and Low-pass Whole Genome Sequencing.

II. Indications and/or Limitations of Coverage

Application of coverage criteria is dependent upon an individual's benefit coverage at the time of the request. Specifications pertaining to Medicare and Medicaid can be found in the "Applicable State and Federal Regulations" section of this policy document.

1) For pregnant individuals who desire information on the risk of having a child with fetal aneuploidy, the following screening tests to detect fetal aneuploidy of chromosomes 13, 18, and 21 **MEET COVERAGE CRITERIA**:
   a) First-trimester (defined as 11-14 weeks) screening incorporating maternal serum markers (hCG, PAPP-A with nuchal translucency (NT)).
   b) Second-trimester (15-22 weeks) screening incorporating triple maternal serum markers (hCG, AFP, uE3 with NT) and quad maternal serum markers (hCG, AFP, uE3, DIA with NT).
   c) First (11-14 weeks) and second (15-22 weeks) trimester integrated screening incorporating maternal serum markers (PAPP-A with NT) and quad maternal serum markers (hCG, AFP, uE3, DIA with NT).
   d) First (11-14 weeks) and second (15-22 weeks) trimester sequential screening incorporating maternal serum markers (PAPP-A, hCG with NT) and quad maternal serum markers (hCG, AFP, uE3, DIA with NT).

e) First (11-14 weeks) and second (15-22 weeks) trimester contingent screening incorporating maternal serum markers (PAPP-A, hCG with NT); if positive, quad maternal serum markers (hCG, AFP, uE3, DIA with NT).

2) For pregnant individuals who desire information on the risk of having a child with fetal aneuploidy, non-invasive prenatal screening (NIPS) to detect fetal aneuploidy of chromosomes 13, 18, 21, X, and Y (singleton or twin pregnancies of at least 10 weeks gestation) **MEETS COVERAGE CRITERIA**.

3) For pregnant individuals who desire information on the risk of having a child with fetal aneuploidy or to pursue additional confirmatory testing of equivocal or positive results from the above testing, karyotyping to confirm fetal aneuploidy **MEETS COVERAGE CRITERIA**.

4) To detect fetal aneuploidy, the use of the "penta" screen (hCG, AFP, uE3, DIA with NT, and hyperglycosylated hCG) **DOES NOT MEET COVERAGE CRITERIA**.

5) Screening for the detection of fetal aneuploidies **DOES NOT MEET COVERAGE CRITERIA** in any of the following situations:

a) Parallel or simultaneous testing with multiple screening methodologies for fetal aneuploidy.

b) For the screening of pregnant individuals with higher order multiple gestation pregnancies.

c) Repeat screening for pregnant individuals with negative screening results.

d) For the detection of other chromosomal abnormalities (e.g., microdeletion syndromes, unbalanced translocations, deletions, duplications) not addressed above.

e) For the determination of fetal sex.

The following does not meet coverage criteria due to a lack of available published scientific literature confirming that the test(s) is/are required and beneficial for the diagnosis and treatment of an individual's illness.

6) For the diagnosis of fetal aneuploidy, the use of single cell genotyping in trophoblasts isolated from maternal serum (e.g., Luna Prenatal Test) **DOES NOT MEET COVERAGE CRITERIA**.

III. Applicable State and Federal Regulations

DISCLAIMER: If there is a conflict between this policy and any relevant, applicable government policy for a particular member [e.g., Local Coverage Determinations (LCDs) or National Coverage Determinations (NCDs) for Medicare and/or state coverage for Medicaid], then the government policy will be used to make the determination. For the most up-to-date Medicare policies and coverage, please visit the Medicare search website: https://www.cms.gov/medicare-coverage-database/search.aspx. For the most up-to-date Medicaid policies and coverage, visit the applicable state Medicaid website.

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Many labs have developed specific tests that they must validate and perform in house. These laboratory-developed tests (LDTs) are regulated by the Centers for Medicare and Medicaid (CMS) as high-complexity tests under the Clinical Laboratory Improvement Amendments of 1988 (CLIA '88). LDTs are not approved or cleared by the U. S. Food and Drug Administration; however, FDA clearance or approval is not currently required for clinical use.

In April 2021, the FDA released a statement titled, "Genetic Non-Invasive Prenatal Screening Tests May Have False Results: FDA Safety Communication," that detailed safety concerns regarding the use of NIPS tests outside of standard screening purposes. Specifically, they reinforced that NIPS tests are laboratory-developed tests not regulated by the FDA and that the use of NIPS should be confined to screenings, rather than diagnoses. "The accuracy and performance of NIPS tests have not been evaluated by the FDA and these tests can give false results, such as reporting a genetic abnormality when the fetus does not actually have one. NIPS tests are screening tests, which means the NIPS test may only tell you the risk of the fetus having certain genetic abnormalities. They are not diagnostic tests, which are generally used to more definitively confirm or rule out a suspected genetic abnormality." Additionally, "The FDA recommends that patients discuss the benefits and risks of NIPS tests with a genetic counselor or other health care provider before deciding to get these tests. Patients should also discuss the results of NIPS tests with a genetic counselor or other health care provider before making any decisions about their pregnancy. Health care providers should be aware of the risks and limitations of using these screening tests and should not use the results from these tests alone to diagnose chromosomal (genetic) abnormalities or disorders."[72]

IV. Applicable CPT/HCPCS Procedure Codes

CPT	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
81422	Fetal chromosomal microdeletion(s) genomic sequence analysis (eg, DiGeorge syndrome, Cri-du-chat syndrome), circulating cell-free fetal DNA in maternal blood
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 Proprietary test: Harmony™ Prenatal Test Lab/Manufacturer: Ariosa Diagnostics
81508	Fetal congenital abnormalities, biochemical assays of two proteins (PAPP-A, hCG [any form]), utilizing maternal serum, algorithm reported as a risk score
81509	Fetal congenital abnormalities, biochemical assays of three proteins (PAPP-A, hCG [any form], DIA), utilizing maternal serum, algorithm reported as a risk score
81510	Fetal congenital abnormalities, biochemical assays of three analytes (AFP, uE3, hCG [any form]), utilizing maternal serum, algorithm reported as a risk score

81511	Fetal congenital abnormalities, biochemical assays of four analytes (AFP, uE3, hCG [any form], DIA) utilizing maternal serum, algorithm reported as a risk score (may include additional results from previous biochemical testing)
81512	Fetal congenital abnormalities, biochemical assays of five analytes (AFP, uE3, total hCG, hyperglycosylated hCG, DIA) utilizing maternal serum, algorithm reported as a risk score
81599	Unlisted multianalyte assay with algorithmic analysis
82105	Alpha-fetoprotein (AFP); serum
82106	Alpha-fetoprotein (AFP); amniotic fluid
82677	Estriol
84163	Pregnancy-associated plasma protein-A (PAPP-A)
84702	Gonadotropin, chorionic (hCG); quantitative
84703	Gonadotropin, chorionic (hCG); qualitative
84704	Gonadotropin, chorionic (hCG); free beta chain
86336	Inhibin A
88235	Tissue culture for non-neoplastic disorders; amniotic fluid or chorionic villus cells
88267	Chromosome analysis, amniotic fluid or chorionic villus, count 15 cells, 1 karyotype, with banding
88269	Chromosome analysis, in situ for amniotic fluid cells, count cells from 6-12 colonies, 1 karyotype, with banding
88271	Molecular cytogenetics; DNA probe, each (eg, FISH)
88280	Chromosome analysis; additional karyotypes, each study
88285	Chromosome analysis; additional cells counted, each study
0327U	Fetal aneuploidy (trisomy 13, 18, and 21), DNA sequence analysis of selected regions using maternal plasma, algorithm reported as a risk score for each trisomy, includes sex reporting, if performed Proprietary test: Vasistera™ Lab/Manufacturer: Natera, Inc
0341U	Fetal aneuploidy DNA sequencing comparative analysis, fetal DNA from products of conception, reported as normal (euploidy), monosomy, trisomy, or partial deletion/duplication, mosaicism, and segmental aneuploid Proprietary test: Single Cell Prenatal Diagnosis (SCPD) Test Lab/Manufacturer: Luna Genetics, Inc

Procedure codes appearing in Medical Policy documents are included only as a general reference tool for each policy. They may not be all-inclusive.

V. Evidence-based Scientific References

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VI. Revision History

Revision Date	Summary of Changes
12/03/2025	Initial RTM Policy Presentation