Applicable To:
- ☑ Medicare
- ☑ Medicaid (excluding AZ and KY)
- ☑ Florida CMS Health Plan

Claims and Payment Policy: Fetal Aneuploidy Frequency Over Time

Policy Number: CPP-122

Original Effective Date: 5/2/2019
Revised Effective Date(s): 12/18/2019

BACKGROUND

Humans have 23 pairs of chromosomes. Aneuploidy is an abnormal number of chromosomes. Trisomy is a type of aneuploidy in which there are three copies of a chromosome instead of two. Trisomy 21, also called Down syndrome, is a chromosomal condition that is associated with intellectual disability, a characteristic facial appearance and poor muscle tone (hypotonia) in infancy. The degree of intellectual disability varies, but it is usually mild to moderate. Individuals with Down syndrome may be born with a variety of birth defects including heart defects and digestive abnormalities. The risk of having a child with trisomy 21 increases with a mother's age.

Down syndrome can also be caused by translocation, which occurs when a part of chromosome 21 breaks away and becomes attached to another chromosome. In a balanced translocation, pieces of chromosomes are rearranged but no genetic material is gained or lost in the cell. In these cases, the parent's health is not affected.

Trisomy 18, also called Edwards syndrome, is a chromosomal condition associated with slow growth before birth and a low birth weight. Affected individuals may have heart defects and abnormalities of other organs that develop before birth. Other features of trisomy 18 include a small, abnormally shaped head; a small jaw and mouth; and clenched fists with overlapping fingers. The risk of having a child with trisomy 18 increases with a mother's age.

Trisomy 13, also called Patau syndrome, is a chromosomal condition associated with severe intellectual disability and physical abnormalities in many parts of the body. Individuals with trisomy 13 often have heart defects, brain or spinal cord abnormalities, very small or poorly developed eyes (microphthalmia), extra fingers and/or toes, an opening in the lip (a cleft lip) with or without an opening in the roof of the mouth (a cleft palate) and weak muscle tone (hypotonia). The risk of having a child with trisomy 13 increases with a mother's age. Patau syndrome can also be caused by translocation.

Original Effective Date: 05/02/2019 - Revised: 12/18/2019
PRO_48137E Internal/State Approved 04022020 NA9PROWEB48137E_0000

Routine screening tests for trisomies 21, 18 and 13 include first-trimester screening (which involves an ultrasound and a blood test), maternal serum screening (a blood test) and a high-resolution ultrasound evaluation in the second trimester. These tests may identify women with an increased risk of having a child with trisomy 21, 18 or 13, but they cannot diagnose, confirm or exclude the possibility of a chromosomal disorder. Conventional prenatal diagnosis (i.e., chorionic villus sampling (CVS) or amniocentesis) can definitively diagnose fetal trisomies, although these invasive procedures are associated with a risk of miscarriage.

Tests that detect fetal trisomies, analyze cell-free DNA (cfDNA) fragments in maternal blood. During pregnancy, there are cfDNA fragments from both the mother and fetus in maternal circulation. The tests detect an increased amount of chromosomal material in maternal blood and can be offered as early as 10 weeks of pregnancy. Available tests use different methodologies and algorithms for data analysis. Depending on the test, the methodology may involve massively parallel sequencing (MPS), targeted sequencing of specific chromosomal segments or directed sequence analysis of single nucleotide polymorphisms (SNPs). All tests were validated in high-risk couples. It is unknown whether they are as accurate for low- to average-risk couples

The Centers for Disease Control and Prevention (CDC) created the ACCE model process for evaluating genetic or genomic-based tests. The 4 main components of the ACCE process include analytic validity, clinical validity, clinical utility and ELSI. Analytic validity refers to how accurately and reliably the test measures the genotype of interest. Clinical validity refers to how consistently and accurately the test detects or predicts the intermediate or final outcomes of interest. Is what's measured associated with the outcome of interest? Clinical utility refers to how likely the test is to significantly improve patient outcomes. What is the clinical value? ELSI refers to the ethical, legal and social implications that may arise in the context of using the test.

Fetal aneuploidy testing is not for diagnostic testing and should not be the basis on making a decision for pregnancy termination. It is recommended follow-up testing for positive results such as amniocentesis or chorionic venous sampling.

POSITION STATEMENT

Coverage

DNA-based noninvasive prenatal tests of fetal aneuploidy **are considered proven and/or medically necessary as screening tools for trisomy 21 (Down syndrome), trisomy 18 (Edwards syndrome) or trisomy 13 (Patau syndrome)** in ANY ONE of the following circumstances:

- Fetal ultrasound findings indicating an increased risk of aneuploidy; **OR**
- History of a prior pregnancy with a trisomy; **OR**
- Positive first- or second-trimester screening test results for aneuploidy; **OR**
- Parental balanced Robertsonian translocation with an increased risk of fetal trisomy 13 or trisomy 21; **OR**
- Allows only one test per pregnancy

Frequency: Routine screening tests for trisomies 21, 18 and 13 include first-trimester screening (which involves an ultrasound and a blood test).

Alternatives to Procedure:

- Conventional screening with serum markers and ultrasound; **OR**
- Invasive testing (e.g., amniocentesis) for confirmation of diagnosis of aneuploidy when conventional screening test are positive for aneuploidy.

CODING & BILLING

Covered ICD 10 Codes

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G91.2	(Idiopathic) normal pressure hydrocephalus
O09.291	Supervision of pregnancy with other poor reproductive or obstetric history, first trimester
O09.292	Supervision of pregnancy with other poor reproductive or obstetric history, second trimester
O09.293	Supervision of pregnancy with other poor reproductive or obstetric history, third trimester
O09.299	Supervision of pregnancy with other poor reproductive or obstetric history, unspecified trimester
O09.511	Supervision of elderly primigravida, first trimester
O09.512	Supervision of elderly primigravida, second trimester
O09.513	Supervision of elderly primigravida, third trimester
O09.519	Supervision of elderly primigravida, unspecified trimester
O09.521	Supervision of elderly multigravida, first trimester
O09.522	Supervision of elderly multigravida, second trimester
O09.523	Supervision of elderly multigravida, third trimester
O09.529	Supervision of elderly multigravida, unspecified trimester
O28.3	Abnormal ultrasonic finding on antenatal screening of mother
O28.5	Abnormal chromosomal and genetic finding on antenatal screening of mother
O28.9	Unspecified abnormal findings on antenatal screening of mother
O35.0XX0	Maternal care for (suspected) central nervous system malformation in fetus, not applicable or unspecified
O35.1XX0	Maternal care for (suspected) chromosomal abnormality in fetus, not applicable or unspecified
O35.1XX1	Maternal care for (suspected) chromosomal abnormality in fetus, fetus 1
O35.1XX2	Maternal care for (suspected) chromosomal abnormality in fetus, fetus 2
O35.1XX3	Maternal care for (suspected) chromosomal abnormality in fetus, fetus 3
O35.1XX4	Maternal care for (suspected) chromosomal abnormality in fetus, fetus 4
O35.1XX5	Maternal care for (suspected) chromosomal abnormality in fetus, fetus 5
O35.1XX9	Maternal care for (suspected) chromosomal abnormality in fetus, other fetus
O35.2XX0	Maternal care for (suspected) hereditary disease in fetus, not applicable or unspecified
Q90.0	Trisomy 21, nonmosaicism (meiotic nondisjunction)
Q90.1	Trisomy 21, mosaicism (mitotic nondisjunction)
Q90.2	Trisomy 21, translocation
Q90.9	Down syndrome, unspecified
Q91.0	Trisomy 18, nonmosaicism (meiotic nondisjunction)
Q91.1	Trisomy 18, mosaicism (mitotic nondisjunction)
Q91.2	Trisomy 18, translocation
Q91.3	Trisomy 18, unspecified
Q91.4	Trisomy 13, nonmosaicism (meiotic nondisjunction)
Q91.5	Trisomy 13, mosaicism (mitotic nondisjunction)
Q91.6	Trisomy 13, translocation
Q91.7	Trisomy 13, unspecified

Covered CPT Codes

0009M	Fetal aneuploidy (trisomy 21, and 18) DNA sequence analysis of selected regions using maternal plasma, algorithm reported as a risk score for each trisomy
81420	Fetal chromosomal aneuploidy (e.g., 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 (e.g., DiGeorge syndrome, Cri-du-chat syndrome), circulating cell-free fetal DNA in maternal blood

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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

Note: For NE Medicaid, CPT codes **81420**, **81422**, and **81507** are non-covered under Fee For Service (FFS). CPT code **81479** is covered under FFS for Severe Combined Immunodeficiency (SCID) new born screenings only.

Coding information is provided for informational purposes only. The inclusion or omission of a CPT, HCPCS, or ICD-10 code does not imply member coverage or provider reimbursement. Consult the member's benefits that are in place at time of service to determine coverage (or non-coverage) as well as applicable federal / state laws.

DEFINITIONS

Aneuploidy	The presence of an abnormal number of chromosomes in a cell, for example a human cell having 45 or 47 chromosomes instead of the usual 46. It does not include a difference of one or more complete sets of chromosomes.
Trisomy 21 (Downs Syndrome)	The most common form of Down syndrome, caused by an extra copy of chromosome number 21.
Trisomy 18 (Edwards Syndrome)	A rare, serious genetic disorder caused by an error in cell division at the embryo stage, resulting in three copies of chromosome 18 instead of a normal pair.
Trisomy 13 (Patau Syndrome)	A rare, serious genetic disorder caused by having an additional copy of chromosome 13 in some or all of the body's cells
DNA	Deoxyribonucleic acid, a self-replicating material which is present in nearly all living organisms as the main constituent of chromosomes. It is the carrier of genetic information.
Amniocentesis	The sampling of amniotic fluid using a hollow needle inserted into the uterus, to screen for developmental abnormalities in a fetus.
Chorionic Venous Sampling	A test made in early pregnancy to detect congenital abnormalities in the fetus. A tiny tissue sample is taken from the villi of the chorion, which forms the fetal part of the placenta.

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7. Koster MP, Wortelboer EJ, Cuckle HS, et al. Placental protein 13 as a first trimester screening marker for aneuploidy. Prenat Diagn. 2009; 29(13):1237-1241.
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IMPORTANT INFORMATION ABOUT THIS DOCUMENT

Claims and Payment Policies (CPPs) are policies regarding claims or claim line processing and/or reimbursement related to the administration of health plan benefits. They are not recommendations for treatment, nor should they be used as treatment guidelines. Providers are responsible for diagnosing, treating, and making clinical recommendations to the member. CPPs are subject to, but not limited to, the following:

- State and federal laws and regulations;
- Policies and procedures promulgated by the Centers for Medicare and Medicaid Services, including National Coverage Determinations and Local Coverage Determinations;
- The health plan's contract with Medicare and/or a state's Medicaid agency, as applicable;
- Other CPPs and clinical policies as applicable including, but not limited to, *Pre-Payment and Post-Payment Review*.
- The provisions of the contract between the provider and the health plan; and
- The terms of a member's particular benefit plan, including those terms outlined in the member's Evidence of Coverage, Certificate of Coverage, and other policy documents.

In the event of a conflict between a CPP and a member's policy documents, the terms of a member's benefit plan will always supersede the CPP.

The use of this policy is neither a guarantee of payment, nor a prediction of how a specific claim will be adjudicated. Any coding information is for informational purposes only. No inference should be made regarding coverage or provider reimbursement as a result of the inclusion, or omission, in a CPP of a CPT, HCPCS, or ICD-10 code. Always consult the member's benefits that are in place at time of service to determine coverage or non-coverage. Claims processing is subject to a number of factors, including the member's eligibility and benefit coverage on the date of service, coordination of benefits, referral/authorization requirements, utilization management protocols, and the health plan's policies. Services must be medically necessary in order to be covered.

References to other sources and links provided are for general informational purposes only, and were accurate at the time of publication. CPPs are reviewed annually but may change at any time and without notice, including the lines of business for which they apply. CPPs are available at www.wellcare.com. Select the "Provider" tab, then "Tools" and then "Payment Guidelines".

RULES, PRICING & PAYMENT COMMITTEE HISTORY AND REVISIONS

Date	Action
10/30/2019	• Approved by RGC