Prenatal Screening
Clinical Coverage Criteria

Overview

Approximately 3% to 5% of pregnancies are complicated by birth defects or genetic disorders. Chromosomal abnormalities are present in approximately 1 in 150 live births, and congenital malformations remain the leading cause of infant death and a leading cause of childhood death. These chromosomal abnormalities include aneuploidy (defined as having one or more extra or missing chromosomes), translocations, duplications, and deletions. The most common chromosomal disorder is trisomy 21 (Down syndrome), with an incidence of 1 per 800 live births.4 Trisomy 13 and 18 can also result in live births, though with a significantly lower incidence. Sex chromosome aneuploidies are less common than autosomal aneuploidies. The only known viable monosomy is monosomy X (Turner syndrome). Risk of aneuploidy increases with maternal age. Other factors also influence patients' risk in any given pregnancy, including having a previous pregnancy with a chromosomal abnormality. A past family history of aneuploidy increases current pregnancy risk of aneuploidy, especially if a parent is a balanced Robertsonian translocation carrier, though most cases are sporadic and secondary to chromosomal nondisjunction (Carlson and Bora, 2017).

All patients should be offered aneuploidy screening or diagnostic testing during pregnancy. Just as importantly, available options should be explained to patients and families in depth, most notably including the risks and benefits of each option, and how results might be reported. Patients who choose cell-free DNA screening should be counseled that the test remains a screening test for aneuploidy at this time and that microdeletion testing continues to have poor positive predictive values due to the low prevalence of these disorders. It is not recommended that patients undergo more than one screening modality but rather that women who have positive screens and wish to pursue further testing be counseled on diagnostic testing with amniocentesis and chorionic villus sampling (CVS) so as not to delay diagnosis. Amniocentesis and CVS are increasingly safe with low rates of pregnancy loss and should continue to be available to all women who desire diagnostic testing regardless of risk factors or presence or absence of anomalies (Carlson and Bora, 2017).

Policy

This Policy applies to the following Fallon Health products:

☒ Medicare Advantage (Fallon Medicare Plus, Fallon Medicare Plus Central)

☒ MassHealth ACO

☒ NaviCare HMO SNP, NaviCare SCO

☒ PACE (Summit Eldercare PACE, Fallon Health Weinberg PACE)

☒ Community Care

Effective for dates of service on or after November 21, 2024, screening for fetal chromosomal aneuploidy (CPT 81420) does not require prior authorization when the claim is submitted by a contracted provider.

Fallon Health Clinical Coverage Criteria

Screening for fetal chromosomal abnormalities should be an informed patient choice based on provision of adequate and accurate information, and the patient's clinical context, accessible

health care resources, values, interests, and goals. Prenatal screening (serum screening with or without nuchal translucency ultrasound or cell-free DNA screening) and diagnostic testing (chorionic villus sampling or amniocentesis) options should be discussed and offered to all pregnant patients regardless of age or risk for chromosomal abnormality. After review and discussion, every patient has the right to pursue or decline prenatal screening and diagnostic testing. Pretest and posttest counseling is essential (ACOG Practice Bulletin, Number 226).

Single time point prenatal screening approaches include cell-free DNA screening, first-trimester screening and second-trimester screening. Combined screening approaches in which samples are obtained in the first and second trimesters include integrated, serum integrated, sequential, and contingent screening (ACOG Practice Bulletin, Number 226).

If prenatal screening is accepted, patients should choose one screening approach and should not undergo multiple screening approaches (ACOG Practice Bulletin, Number 226).

Regardless of screening approach, all patients should be offered a second-trimester ultrasound for fetal structural defects, since these may occur with or without fetal aneuploidy; ideally this procedure is performed between 18 and 22 weeks of gestation (with or without second-trimester maternal serum alpha-fetoprotein) (ACOG Practice Bulletin, Number 226).

Cell-Free DNA Screening

Effective for dates of service on or after November 21, 2024, cell-free DNA screening for the detection of aneuploidies in chromosomes 13, 18, 21, X and Y, is considered medically necessary when:

1. The member has a singleton or twin pregnancy, and
2. The member has not previously had cell-free DNA testing in the current pregnancy.

Cell-free DNA screening is not medically necessary in the following clinical scenarios:

- Higher order gestations (≥ 3 fetuses)
- Fetal demise
- Co-twin demise (vanishing twin)
- Multiple fetal anomalies
- Concurrent screening with other prenatal screening approaches
- Prior to 9 weeks gestation
- To determine fetal sex only
- Single genes (e.g., CFTR, HBB, SMN1, RhD)
- Microdeletions (e.g., DiGeorge syndrome, Cri-du-chat syndrome)
- Twin zygosity (monozygotic versus dizygotic)
- Genome-wide copy number variants
- Aneuploidies of other autosomal chromosomes, e.g., trisomy 7, trisomy 15, trisomy 16, trisomy 22, etc.
- Polygenic risk assessment

Testing in some of the scenarios listed above may have a role under certain circumstances, but not in routine prenatal screening.

First-Trimester Screening, Second-Trimester Screening, and Combined First- and Second-Trimester Screening

Although cell-free DNA screening is frequently used to screen for the common fetal aneuploidies, the following screening approaches remain available are considered medically necessary as alternatives to cell-free DNA screening. Patients should choose one screening approach and should not undergo multiple screening approaches.

1. First-trimester screening – Measurement of maternal serum analytes [pregnancy-associated plasma protein-A (PAPP-A), free beta human gonadotropin (hCG), with or without alpha-fetoprotein (AFP)] and ultrasound measurement of nuchal translucency (NT) performed between 10 and 14 weeks.

2. Second-trimester screening - Also known as quad screen, includes measurement of maternal four serum analytes (hCG, AFP, inhibin A, and unconjugated estriol) performed between 15 and 22 weeks.
3. Combined First-Trimester and Second-Trimester Screening:
    a. Integrated Screening - First-trimester NT ultrasound measurement and maternal serum analyte screening (PAPP-A) followed by a second-trimester quad screen (hCG, AFP, inhibin A, and unconjugated estriol) with single test result in the second trimester.
    b. Serum Integrated Screening – First-trimester maternal serum analyte screening (PAPP-A) followed by a second-trimester quad screen (hCG, AFP, inhibin A, and unconjugated estriol) with single test result in the second trimester.
    c. Stepwise Sequential – First-trimester screening with results provided: if first trimester screening is positive, patient is offered additional testing (diagnostic testing or cell-free DNA), if first trimester screening is negative, patient is informed that they have received a negative first-trimester screening result and quad screen is planned for the second trimester with a final combined risk assessment that incorporates first- and second-trimester results.
    d. Contingent Screening – First-trimester screening, after which women are stratified into high-, medium-, and low-risk groups. The high-risk group is offered additional testing (diagnostic testing or cell-free DNA). The low-risk group has no further testing. The intermediate-risk group is offered second-trimester quad screening. First-trimester and second-trimester results are combined to calculate a final risk of aneuploidy in patients at intermediate risk.

With all combined first- and second-trimester screening approaches, physicians must ensure that nondisclosure is acceptable to patients if they choose to accept one of these approaches.

Medicare Variation

Items and services which are not reasonable and necessary for the diagnosis or treatment of illness or injury are not covered under Medicare (Medicare Benefit Policy Manual, Chapter 16, Section 20 – Services Not Reasonable and Necessary). Medicare Part B pays for the specific preventive (screening) services listed in section 1861(ww)(2) of the Social Security Act. Additional preventive services not described in the definition of “preventive services” under § 410.2, are covered when the Secretary determines through the national coverage determination process (as defined in section 1869(f)(1)(B) of the Act) that these services are all of the following:
(1) Reasonable and necessary for the prevention or early detection of illness or disability.
(2) Recommended with a grade of A or B by the United States Preventive Services Task Force.
(3) Appropriate for individuals entitled to benefits under part A or enrolled under Part B.

Prenatal screening for fetal chromosomal aneuploidy is not listed as a covered preventive (screening) service in section 1861(ww)(2) of the Social Security Act. Medicare does not have an NCD for prenatal screening for fetal chromosomal aneuploidy. National Government Services, Inc., the Part A and B Medicare Administrative Contractor (MAC) with jurisdiction in the Plan’s service area has an LCD for Molecular Pathology Procedures (L35000). Per NCD L35000, many applications of the molecular pathology procedures are not covered services under Medicare given the lack of benefit category (e.g., preventive service or screening for a genetic abnormality in the absence of a suspicion of disease) and/or failure to meet the reasonable and necessary threshold for coverage (e.g., based on quality of clinical evidence and strength of recommendation or when the results would not reasonably be used in the management of a beneficiary). CPT 81420, 81422 and 81507 are Tier 1 Non-Covered Codes, unlikely to impact therapeutic decision-making in the clinical management of the beneficiary and are denied automatically as not medically necessary. (Medicare Coverage Database search 01/27/2025).

Prenatal screening for fetal chromosomal aneuploidy (CPT 81420, 81422 and 81507) is not covered/not reasonable and necessary for Medicare Advantage plan members.

MassHealth Variation

Effective for dates of service on or after November 21, 2024, noninvasive prenatal screening (cell-free DNA prenatal screening) to ascertain if a pregnancy has a risk of fetal chromosomal aneuploidy is covered for MassHealth members. The Plan will not limit availability and coverage for such screening based on the age of the pregnant patient or any other risk factor, unless the limitation is part of the generally accepted standards of professional practice as recommended by the American College of Obstetricians and Gynecologists (Massachusetts General Laws, Chapter 118E, Section 10R - Coverage for noninvasive prenatal screening).

Exclusions

- Noninvasive prenatal screening for microdeletions (CPT 81422, 0060U) is considered experimental and investigational.
- Noninvasive prenatal screening to predict twin zygosity (CPT 0060U) is considered experimental and investigational.
- Noninvasive prenatal screening for single-gene disorders (CPT 81302, 81404, 81405, 81406, 81407, 81408, 81442) is considered experimental and investigational
- Noninvasive prenatal screening for other aneuploidies such as trisomy 16 and trisomy 22 is considered experimental and investigational.
- Genome-wide cell-free DNA screening for large deletions or duplications is considered experimental and investigational.

Summary of Evidence

Cell-Free DNA

Cell-free DNA screening, commonly referred to as noninvasive prenatal screening (NIPS) or noninvasive prenatal testing (NIPT), screens for fetal aneuploidies by analyzing the fetal fraction, i.e., the percentage of fetal to total cell-free DNA in the maternal circulation. Fetal fraction increases with gestational age and is affected by many factors, including maternal influences and fetal influences (notably multiple gestation and fetal aneuploidy).Depending on the laboratory, cell-free DNA screening can be performed as early as 9 weeks gestation, although results are more reliable at 10 weeks and beyond. Cell-free DNA screening has the highest available detection rate of all available screening tests for trisomy 21 with a detection rate of 99% according to a recent meta-analysis (Gill et al. 2015). Detection rates for trisomy 18, 13, and sex chromosome abnormalities are significantly lower than for trisomy 21. Cell-free DNA is the most sensitive and specific screening test for the common fetal aneuploidies, nevertheless, it has the potential for false-positive and false-negative results, and furthermore, cell-free DNA testing is not equivalent to diagnostic testing. Ultrasound is recommended before testing as some ultrasound findings detectable early in pregnancy may affect the timing of cell-free DNA screening, the appropriateness of performing cell-free DNA screening, or the ability to interpret cell-free DNA test results. These findings include an earlier than expected gestational age, confirmation of viability, number of fetuses, presence of a vanishing twin or empty gestational sac, or presence of a fetal anomaly (ACOG Practice Bulletin, Number 226).

First-trimester Screening

First-trimester screening includes a combination of maternal serum analytes measurement and nuchal translucency (NT) ultrasound measurement performed between 10 and 14 weeks gestation. Serum analytes include free-beta human gonadotropin (hCG) and pregnancy-associated plasma protein-A (PAPP-A), with or without alpha-fetoprotein (AFP), A risk estimate for common trisomies (generally trisomies 13, 18, and 21) is then developed that incorporates maternal age, past pregnancy history, number of fetuses in the current gestation, weight, race, serum markers, and NT measurement (ACOG Practice Bulletin, Number 226).

NT refers to the fluid-filled space on the dorsal aspect of the fetal neck. An enlarged NT (often defined as 3.0 mm or more or above the 99th percentile for the crown–rump length) is significantly associated with both fetal aneuploidy and structural malformations such as cardiac anomalies. The risk of adverse fetal outcome is proportional to the degree of NT enlargement (ACOG Practice Bulletin, Number 226).

First-trimester screening gives the potential for earlier diagnosis as well as the ability to screen for other structural, genetic, or placental disorders. The detection rate for trisomy 21 varies from 82% to 87% depending on the laboratory, using a 5% screen positive rate (ACOG Practice Bulletin, Number 226). All patients should be offered second-trimester assessment for open fetal defects (by ultrasonography, with or without second trimester serum AFP) and ultrasound screening for other fetal structural defects (ACOG Practice Bulletin, Number 226).

Second-Trimester Screening

Second-trimester screening is a quadruple marker screen also known as the quad screen. The quad screen can be performed between 15 and 22 weeks gestation and is a single point in time test that involves measurement of proteins secreted by the pregnancy, including hCG, AFP, inhibin A, and unconjugated estriol. These protein measurements are combined with the patients' age, race, weight, number of fetuses in the current gestation, diabetes status, and gestational age to provide a risk estimate. The detection rate for trisomy 21 is slightly lower than that of the first-trimester screen, with a reported detection rate of 81% using a 5% screen positive rate (ACOG Practice Bulletin, Number 226). Advantages of the quad screen include its ability to screen for open neural tube defects in addition to aneuploidy. Serum AFP is secreted by the fetus and is present in the amniotic fluid and, therefore, also maternal serum (ACOG Practice Bulletin, Number 226).

Combined First-Trimester and Second-Trimester Screening

Combined first-trimester and second-trimester screening with either integrated, serum integrated, stepwise sequential, or contingent screening involving maternal serum analytes, NT, or both measurements provides a higher detection rate for trisomy 21, 18, and 13 than first- or second-trimester single point in time screening. Depending on the test selected, there is variable timing of results available to the patient (ACOG Practice Bulletin, Number 226).

Integrated Screening

With integrated screening, the patient undergoes a first-trimester NT ultrasound measurement and serum analyte screening (PAPP-A) followed by a second-trimester quad screen (hCG, AFP, inhibin A, and unconjugated estriol) and receives a single test result in the second trimester. All of these values are then incorporated into a single risk estimate to provide patient a second trimester risk of aneuploidy. The detection rate for trisomy 21 is 96%, the highest of any available serum screens other than cell-free DNA, with a 5% screen positive rate. Downsides to this approach include the relatively late availability of results, limiting the time in which patients and their provider may have to make important decisions about future care (ACOG Practice Bulletin, Number 226).

Serum Integrated Screening

In locations where an NT measurement by a certified ultrasonographer is unavailable, or if fetal position, maternal body habitus, or imaging properties preclude an accurate NT measurement, serum integrated screening, which includes only the first-trimester and second-trimester serum analytes, also is an option. Serum integrated screening has a lower detection rate than integrated screening that includes an NT measurement, but a similar detection rate to first-trimester screening (ACOG Practice Bulletin, Number 226).

Stepwise Sequential

The stepwise sequential screen involves performing the first-trimester screen (serum analytes and NT measurement). If the first-trimester screening result indicates that the risk of aneuploidy is greater than the laboratory's positive screening cutoff, the patient is notified and offered additional testing (diagnostic testing or cell-free DNA). If the first-trimester screening result indicates a lower risk than the cutoff level, the patient is informed that they have received a negative screening test result and quad screening is planned in the second trimester to receive a final combined numerical risk. The detection rate for trisomy 21 is 95%, with a 5% screen positive rate (ACOG Practice Bulletin, Number 226).

Contingent Screening

The contingent screen involves performing a first-trimester screen, after which women are stratified into high-, medium-, and low-risk groups. The high-risk group is offered additional testing

(diagnostic testing or cell-free DNA). The low-risk group has no further testing. The intermediate-risk group is offered second-trimester quad screening. First-trimester and second-trimester results are used together to calculate a final risk of aneuploidy in patients at intermediate risk. The detection rate for trisomy 21 varies between 80% and 94% for this screening method, with a 5% screen positive rate (ACOG Practice Bulletin, Number 226).

Analysis of Evidence (Rationale for Determination)

N/A

Coding

The following codes are included below for informational purposes only; inclusion of a code does not constitute or imply coverage or reimbursement.

Code	Description
59000	Amniocentesis; diagnostic
59001	Amniocentesis; therapeutic amniotic fluid reduction (includes ultrasound guidance)
59015	Chorionic villus sampling, any method
76805	Ultrasound, pregnant uterus, real time with image documentation, fetal and maternal evaluation, after first trimester (> or = 14 weeks 0 days), transabdominal approach; single or first gestation
76810	Ultrasound, pregnant uterus, real time with image documentation, fetal and maternal evaluation, after first trimester (> or = 14 weeks 0 days), transabdominal approach; each additional gestation
76813	Ultrasound, pregnant uterus, real time with image documentation, first trimester fetal nuchal translucency measurement, transabdominal or transvaginal approach; single or first gestation
76814	Ultrasound, pregnant uterus, real time with image documentation, first trimester fetal nuchal translucency measurement, transabdominal or transvaginal approach; each additional gestation (List separately in addition to code for primary procedure)
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
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
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
82105	Alpha-fetoprotein (AFP); serum

82106	Alpha-fetoprotein (AFP); amniotic fluid
82107	Alpha-fetoprotein (AFP); AFP-L3 fraction isoform and total AFP (including ratio)
84163	Pregnancy-associated plasma protein-A (PAPP-A)
84702	Gonadotropin, chorionic (hCG); quantitative
84704	Gonadotropin, chorionic (hCG); free beta chain

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Policy history

Origination date:	03/28/2006
Review/Approval(s):	Technology Assessment Committee: 06/21/2006, 05/22/2012, 2/26/2014 ICD 10 CM codes mapped; 4/23/2014 correction due to ICD 10 CM implementation, 03/25/2015 (updated coding, references) 03/23/2016 (removed ICD-9 codes, added new code 0009M requiring prior authorization, updated references), 04/26/2017 (updated references), 02/28/2018 (added exclusion to NIPT criteria for multiple gestations, updated references), 02/27/2019 (removed ICD-10 codes, updated references), 03/27/2019 (added direction related to Spinal Muscular Atrophy testing to Genetic Testing Policy),, 07/10/2021 (Added clarifying language related to Medicare Advantage, NaviCare and PACE under policy section), 01/28/2025 (annual review, added new paragraph for

MassHealth ACO in Policy section; updated coverage criteria for cell-free DNA effective for dates of service on or after November 21, 2024; updated Exclusions, Coding, References).
Utilization Management Committee: 02/18/2025 (review and approval).

Instructions for Use

Fallon Health complies with CMS's national coverage determinations (NCDs), local coverage determinations (LCDs) of Medicare Contractors with jurisdiction for claims in the Plan's service area, and applicable Medicare statutes and regulations when making medical necessity determinations for Medicare Advantage members. When coverage criteria are not fully established in applicable Medicare statutes, regulations, NCDs or LCDs, Fallon Health may create internal coverage criteria under specific circumstances described at § 422.101(b)(6)(i) and (ii).

Fallon Health follows Medical Necessity Guidelines published by MassHealth when making medical necessity determinations for MassHealth members. In the absence of Medical Necessity Guidelines published by MassHealth, Fallon Health may create clinical coverage criteria in accordance with the definition of Medical Necessity in 130 CMR 450.204.

For plan members enrolled in NaviCare, Fallon Health first follow's CMS's national coverage determinations (NCDs), local coverage determinations (LCDs) of Medicare Contractors with jurisdiction for claims in the Plan's service area, and applicable Medicare statutes and regulations when making medical necessity determinations. When coverage criteria are not fully established in applicable Medicare statutes, regulations, NCDs or LCDs, or if the NaviCare member does not meet coverage criteria in applicable Medicare statutes, regulations, NCDs or LCDs, Fallon Health then follows Medical Necessity Guidelines published by MassHealth when making necessity determinations for NaviCare members.

Each PACE plan member is assigned to an Interdisciplinary Team. PACE provides participants with all the care and services covered by Medicare and Medicaid, as authorized by the interdisciplinary team, as well as additional medically necessary care and services not covered by Medicare and Medicaid. With the exception of emergency care and out-of-area urgently needed care, all care and services provided to PACE plan members must be authorized by the interdisciplinary team.

Not all services mentioned in this policy are covered for all products or employer groups. Coverage is based upon the terms of a member's particular benefit plan which may contain its own specific provisions for coverage and exclusions regardless of medical necessity. Please consult the product's Evidence of Coverage for exclusions or other benefit limitations applicable to this service or supply. If there is any discrepancy between this policy and a member's benefit plan, the provisions of the benefit plan will govern. However, applicable state mandates take precedence with respect to fully-insured plans and self-funded non-ERISA (e.g., government, school boards, church) plans. Unless otherwise specifically excluded, federal mandates will apply to all plans.