PRESBYTERIAN

Medical Policy

Subject: Genetic Testing for Carrier Testing and Prenatal Diagnosis

Medical Policy #: 7.13 **Original Effective Date: 05/22/2024**

Status: Reviewed **Last Review Date: 05-28-2025**

Disclaimer

Refer to the member's specific benefit plan and Schedule of Benefits to determine coverage. This may not be a benefit on all plans, or the plan may have broader or more limited benefits than those listed in this Medical Policy.

Description

Genetic testing may be performed on prospective parents to identify potential diseases that may be passed on to their offspring. This is known as carrier screening. Carriers are usually themselves unaffected by the disease, showing no symptoms, however, they may be at risk for passing the disease onto their children. Preferably, carrier screening takes place before pregnancy (preconception) but can occur during the early stages of pregnancy.

A consensus from the professional organization guidelines and recommendations is used when considering which genetic conditions may be appropriate for carrier screening and includes the following:

- Ability to be diagnosed prenatally and allow opportunities for antenatal intervention to improve perinatal outcomes, changes in delivery management to optimize newborn and infant outcomes and education of the parents about special care needs after birth.
- Carrier frequency of 1 in 100 or greater with a well-defined phenotype that would have a detrimental effect on quality of life (eg, cause cognitive or physical impairment, require surgical or medical intervention).
- Should not include conditions primarily associated with adult-onset disease.
- Should not replace newborn screening or risk-based genetic testing (eg, known family history).
- Targeted testing based on an individual's race, ethnicity or family history for single-gene disorders that have an autosomal or X-linked recessive inheritance pattern. Examples of these diseases include, but may not be limited to alpha thalassemia, cystic fibrosis (CF) and fragile X syndrome.

PHP recognizes that the field of genetic testing is rapidly changing and that other tests may become available.

See also, Genetic Testing for Non-Invasive Prenatal Testing (NIPT), MPM 20.15 for Prenatal genetic screening and testing of a fetus (Aneuploidy),

Coverage Determination

Prior Authorization may or may not be required. (CF, SMA, Fragile X do not require prior auth) Logon to Pres Online to submit a request: https://ds.phs.org/preslogin/index.jsp

Any state mandates for genetic testing for carrier screening take precedence over this medical coverage policy. For New Mexico, Human Services Letter of Direction (LOD) Prenatal Genetic Screening for Cystic Fibrosis, Spinal Muscular Atrophy (SMA) and Fetal Chromosomal Aneuploidy Billing and Guidance, LOD #32 and Biomarker Coverage, LOD #42- outline guidance for covered state (Medicaid) mandates. These LOD have been considered and incorporated into this policy for services that are mandated as covered, or shown to meet all coverage criteria, including services that are not considered investigational or experimental or proven not medically necessary. Please see the LOD for details on criteria for coverage. Please also see Presbyterian MPM 7.1 for breakdown of our Biomarker Hierarchy for Prior Auth Determination when a service is not specifically listed as covered or non-covered in this policy

GERMLINE CARRIER TESTING FOR FAMILIAL DISEASE

Preconception or prenatal carrier testing for an individual who has the capacity and intention to reproduce is considered medically necessary when **ANY** of the following criteria is met:

- There is an identified pathogenic genetic variant in a blood relative specific to the conditions listed below.
- An individual's reproductive partner is a known carrier of a disease-causing pathogenic or likely pathogenic variant in a recessively inherited condition.
- A genetic diagnosis is clinically suspected by a genetic specialist in an affected relative, AND the affected relative has not had genetic testing and is unavailable for testing.

When **ANY** of the above criteria is met, preconception or prenatal carrier testing is considered medically necessary for the following indications (list may not be all inclusive) with the additional restrictions listed below:

Nuclear mitochondrial genes	Sickle cell disease
Muscular dystrophies (DMD, BMD, EDMD, DM1, DM2, SM)	Alpha and beta thalassemia
Fragile X syndrome	Gaucher disease
Rett syndrome	Niemann-Pick disease
PTEN-related disorders	Canavan disease
Von Hippel-Lindau disease	Tay-Sachs disease
Long QT syndrome	DFNB1/ GJB6 for nonsyndromic hearing loss and deafness
SMN1	GJB2 for nonsyndromic hearing loss and deafness
Retinoblastoma	Huntington disease
21-hydroxylase deficiency	Cystic fibrosis

Fragile X:
No Prior Auth required for (81243, 81244, 81171, 81172)
Preconception or prenatal genetic testing of a prospective biologic female parent for **Fragile X** (i.e., FMR1) gene mutations for the purpose of reproductive screening as described by the American College of Obstetrics and Gynecology (ACOG) and American College of Medical Genetics (ACMG is considered medically when the individual has the capacity and intention to reproduce and testing has not been previously performed.

SMN1:
No Prior Auth required for (81329, 81336, 81337)
Preconception or prenatal carrier testing for spinal muscular atrophy by **SMN1** gene variant analysis for the purpose of reproductive screening as described by the American College of Obstetrics and Gynecology (ACOG) American College of Medical Genetics (ACMG is considered medically necessary when the individual has the capacity and intention to reproduce, and testing has not been previously performed.

Cystic fibrosis (CF):
No Prior Auth required for (81220, 81221,81222, 81223, 81224)
Preconception or prenatal carrier testing for **cystic fibrosis** (CF) with targeted variant analysis of CFTR gene variants as described by the American College of Medical Genetics (ACMG) is considered medically necessary for a prospective biologic parent with the capacity and intention to reproduce and testing has not previously been performed. American College of Medical Genetics (ACMG)

Hemoglobinopathies:
No Prior Auth required for (CPT Codes 81257, 81259, 81361)
Preconception or prenatal carrier testing for hemoglobinopathies (i.e., **thalassemias**, sickle cell disease) is considered medically necessary when the individual has the capacity and intention to reproduce, and testing has not been previously performed.

Tay-Sachs:
Prior Auth is required.
Preconception or prenatal carrier testing for the HEXA gene* for carrier screening for Tay-Sachs disease for reproductive decision making when **ANY** of the following criteria are met:

- Pre- and post-test genetic counseling; **AND**
- And **one** of the following:
    - Individual to be tested has an abnormal or inconclusive beta-hexosaminidase A enzyme activity, **OR**
    - Individual to be tested has an affected or carrier family member in whom a variant has been identified, **OR**

- Individual to be tested is of Ashkenazi Jewish ancestry* or the reproductive partner of an individual of Ashkenazi Jewish ancestry*, **OR**
- Individual to be tested is the reproductive partner of an individual affected with or carrier of Tay-Sachs disease.

*Testing for this condition begins with a targeted gene panel. If negative, gene sequence analysis may be considered.

Ashkenazi Jewish (AJ):
Prior Auth is required.
Preconception or prenatal carrier testing for a prospective biologic parent of Ashkenazi Jewish (AJ) descent is considered medically necessary for the conditions specified by the American College of Medical Genetics, including but not limited to the following:

- Targeted panel testing for variants found in an individual of Ashkenazi Jewish descent.
- Familial dysautonomia
- Tay-Sachs disease
- Canavan disease
- Fanconi anemia group C
- Niemann-Pick disease, type A
- Bloom syndrome
- Mucolipidosis IV
- Gaucher disease, type 1

Panel testing must assess at minimum, mutations associated with **ALL** the following diseases (additional genes also may be appropriate):

- Canavan disease (ASPA gene)
- Cystic fibrosis (CFTR gene)
- Familial dysautonomia (Riley-Day syndrome; ELP1 gene)
- Tay-Sachs disease (HEXA gene)

If only one individual of the couple is of Ashkenazi Jewish ancestry, then testing begins with the individual of Ashkenazi Jewish ancestry. If positive for a disease listed above, proceed to test the non-Ashkenazi Jewish partner for that disease using the most appropriate technology for his/her ethnicity. Expanded genetic panel testing for additional genetic conditions not listed above will require MDR review.

Other Inherited Conditions:
Presbyterian members may be eligible under the Plan for genetic testing for carrier screening of other inherited conditions including, but not limited to: Canavan disease, Fabry disease, Gaucher disease, and Mucolipidosis IV for reproductive decision making when **ANY** of the following criteria are met:

- Pre- and post-test genetic counseling; **AND**
- Individual to be tested has an affected or carrier family member in whom a variant(s) has been identified; **OR**
- Individual to be tested is of reproductive age with a family history of a genetic condition that puts that individual at higher risk than the general population to be a carrier, **OR**
- Individual to be tested is the reproductive partner of an individual affected with or carrier of an inherited condition.

Note: The criteria for genetic testing for carrier screening are not consistent with the Medicare National Coverage Policy and therefore may not be applicable to Medicare members. Refer to the CMS website for additional information.

LIMITATION/EXCLUSION

PHP members may **NOT** be eligible under the Plan for the following:

- Reproductive carrier screening based on the general population risk, other than conditions noted above, is considered not medically necessary.
- Reproductive carrier screening for nonmedical traits (e.g., eye color, hair color) is considered not medically necessary.
- If a provider is requesting a multigene reproductive carrier screening panel, the requesting provider must provide supportive documentation for each genetic condition along with CPT codes upon request.

- Expanded carrier screening refers to the practice of screening for many conditions in a panethnic approach (without regard to race or ethnicity) and can include testing for many genetic disorders depending on specific laboratory offerings. Expanded carrier screening panels for multiple heritable conditions including, but may not be limited to: (the following proprietary tests may change, this is not an all-inclusive list)
    - Foresight Universal Carrier Screens
    - Invitae (Broad and Comprehensive) Carrier Screens
    - Natera Horizon Carrier Screen
    - Genesys Carrier Panel
- Detection of genetic susceptibility to adult-onset/late-onset disorders including, but not limited to, genetic testing for breast cancer (eg, BRCA gene testing) is non-covered.
- BillionToOne Unity Carrier Screen Examples of single-gene disorders include various skeletal dysplasias (0449U)

Coding

The coding listed in this medical policy is for reference only. Covered and non-covered codes are within this list.

CPT	DESCRIPTION
81171	AFF2 (ALF transcription elongation factor 2 [FMR2]) (eg, fragile X intellectual disability 2 [FRAXE]) gene analysis; evaluation to detect abnormal (eg, expanded) alleles
81172	AFF2 (ALF transcription elongation factor 2 [FMR2]) (eg, fragile X intellectual disability 2 [FRAXE]) gene analysis; characterization of alleles (eg, expanded size and methylation status)
81220	CFTR (cystic fibrosis transmembrane conductance regulator) (eg, cystic fibrosis) gene analysis; common variants (eg, ACMG/ACOG guidelines)
81221	CFTR (cystic fibrosis transmembrane conductance regulator) (eg, cystic fibrosis) gene analysis; known familial variants
81222	CFTR (cystic fibrosis transmembrane conductance regulator) (eg, cystic fibrosis) gene analysis; duplication/deletion variants
81223	CFTR (cystic fibrosis transmembrane conductance regulator) (eg, cystic fibrosis) gene analysis; full gene sequence
81224	CFTR (cystic fibrosis transmembrane conductance regulator) (eg, cystic fibrosis) gene analysis; intron 8 poly-T analysis (eg, male infertility)
81234	DMPK (DM1 protein kinase) (eg, myotonic dystrophy type 1) gene analysis; evaluation to detect abnormal (expanded) alleles
81239	DMPK (DM1 protein kinase) (eg, myotonic dystrophy type 1) gene analysis; characterization of alleles (eg, expanded size)
81274	HTT (huntingtin) (eg, Huntington disease) gene analysis; characterization of alleles (eg, expanded size)
81312	PABPN1 (poly[A] binding protein nuclear 1) (eg, oculopharyngeal muscular dystrophy) gene analysis, evaluation to detect abnormal (eg, expanded) alleles
81329	SMN1 (survival of motor neuron 1, telomeric) (eg, spinal muscular atrophy) gene analysis; dosage/deletion analysis (eg, carrier testing), includes SMN2 (survival of motor neuron 2, centromeric) analysis, if performed
81336	SMN1 (survival of motor neuron 1, telomeric) (eg, spinal muscular atrophy) gene analysis; full gene sequence
81337	SMN1 (survival of motor neuron 1, telomeric) (eg, spinal muscular atrophy) gene analysis; known familial sequence variant(s)
83080	b-Hexosaminidase, each assay

CPT	DESCRIPTION
0218U	Neurology (muscular dystrophy), DMD gene sequence analysis, including small sequence changes, deletions, duplications, and variants in non-uniquely mappable regions, blood or saliva, identification and characterization of genetic variants
0236U	SMN1 (survival of motor neuron 1, telomeric) and SMN2 (survival of motor neuron 2, centromeric) (eg, spinal muscular atrophy) full gene analysis, including small sequence changes in exonic and intronic regions, duplications and deletions, and mobile element insertions
0449U	Carrier screening for severe inherited conditions (eg, cystic fibrosis, spinal muscular atrophy, beta hemoglobinopathies [including sickle cell disease], alpha thalassemia), regardless of race or self-identified ancestry, genomic sequence analysis panel, must include analysis of 5 genes (CFTR, SMN1, HBB, HBA1, HBA2)
81243	FMR1 (fragile X mental retardation 1) (eg, fragile X mental retardation) gene analysis; evaluation to detect abnormal (eg, expanded) alleles
81244	FMR1 (fragile X mental retardation 1) (eg, fragile X mental retardation) gene analysis: characterization of alleles (eg, expanded size and promoter methylation status)
81161	DMD (dystrophin) (eg, Duchenne/Becker muscular dystrophy) deletion analysis, and duplication analysis, if performed
81187	CNBP (CCHC-type zinc finger nucleic acid binding protein) (eg, myotonic dystrophy type 2) gene analysis, evaluation to detect abnormal (eg, expanded) alleles
81200	ASPA (aspartoacylase) (eg, Canavan disease) gene analysis, common variants (eg, E285A, Y231X)
81205	BCKDHB (branched-chain keto acid dehydrogenase E1, beta polypeptide) (eg, maple syrup urine disease) gene analysis, common variants (eg, R183P, G278S, E422X)
81209	BLM (Bloom syndrome, RecQ helicase-like) (eg, Bloom syndrome) gene analysis, 2281del6ins7 variant
81242	FANCC (Fanconi anemia, complementation group C) (eg, Fanconi anemia, type C) gene analysis, common variant (eg, IVS4+4A>T)
81251	GBA (glucosidase, beta, acid) (eg, Gaucher disease) gene analysis, common variants (eg, N370S, 84GG, L444P, IVS2+1G>A)
81252	GJB2 (gap junction protein, beta 2, 26kDa, connexin 26) (eg, nonsyndromic hearing loss) gene analysis; full gene sequence
81253	GJB2 (gap junction protein, beta 2, 26kDa, connexin 26) (eg, nonsyndromic hearing loss) gene analysis; known familial variants
81254	GJB6 (gap junction protein, beta 6, 30kDa, connexin 30) (eg, nonsyndromic hearing loss) gene analysis, common variants (eg, 309kb [del (GJB6-D13S1830)] and 232kb [del (GJB6-D13S1854)])
81255	HEXA (hexosaminidase A [alpha polypeptide]) (eg, Tay-Sachs disease) gene analysis, common variants (eg, 1278insTATC, 1421+1G>C, G269S)
81256	HFE (hemochromatosis) (eg, hereditary hemochromatosis) gene analysis, common variants (eg, C282Y, H63D)
81257	HBA1/HBA2 (alpha globin 1 and alpha globin 2) (eg, alpha thalassemia, Hb Bart hydrops fetalis syndrome, HbH disease) gene analysis; common deletions or variant (eg, Southeast Asian, Thai, Filipino, Mediterranean, alpha3.7, alpha4.2, alpha20.5, and Constant Spring)
81258	HBA1/HBA2 (alpha globin 1 and alpha globin 2) (eg, alpha thalassemia, Hb Bart hydrops fetalis syndrome, HbH disease), gene analysis; known familial variant
81259	HBA1/HBA2 (alpha globin 1 and alpha globin 2) (eg, alpha thalassemia, Hb Bart hydrops fetalis syndrome, HbH disease), gene analysis; full gene sequence

CPT	DESCRIPTION
81260	IKBKAP (inhibitor of kappa light polypeptide gene enhancer in B-cells, kinase complex-associated protein) (eg, familial dysautonomia) gene analysis, common variants (eg, 2507+6T>C, R696P)
81269	HBA1/HBA2 (alpha globin 1 and alpha globin 2) (eg, alpha thalassemia, Hb Bart hydrops fetalis syndrome, HbH disease), gene analysis; duplication/deletion variants
81271	HTT (huntingtin) (eg, Huntington disease) gene analysis; evaluation to detect abnormal (eg, expanded) alleles
81290	MCOLN1 (mucolipin 1) (eg, Mucolipidosis, type IV) gene analysis, common variants (eg, IVS3-2A>G, del6.4kb)
81302	MECP2 (methyl CpG binding protein 2) (eg, Rett Syndrome) gene analysis; full sequence analysis
81303	MECP2 (methyl CpG binding protein 2) (eg, Rett Syndrome) gene analysis; known familial variant
81304	MECP2 (methyl CpG binding protein 2) (eg, Rett Syndrome) gene analysis; duplication/deletion variants
81321	PTEN (phosphatase and tensin homolog) (eg, Cowden syndrome, PTEN hamartoma tumor syndrome) gene analysis; full sequence analysis
81322	PTEN (phosphatase and tensin homolog) (eg, Cowden syndrome, PTEN hamartoma tumor syndrome) gene analysis; known familial variant
81323	PTEN (phosphatase and tensin homolog) (eg, Cowden syndrome, PTEN hamartoma tumor syndrome) gene analysis; duplication/deletion variant
81324	PMP22 (peripheral myelin protein 22) (eg, Charcot-Marie-Tooth, hereditary neuropathy with liability to pressure palsies) gene analysis; duplication/deletion analysis
81325	PMP22 (peripheral myelin protein 22) (eg, Charcot-Marie-Tooth, hereditary neuropathy with liability to pressure palsies) gene analysis; full sequence analysis
81330	SMPD1 (sphingomyelin phosphodiesterase 1, acid lysosomal) (eg, Niemann-Pick disease, Type A) gene analysis, common variants (eg, R496L, L302P, fsP330)
81331	SNRPN/UBE3A (small nuclear ribonucleoprotein polypeptide N and ubiquitin protein ligase E3A) (eg, Prader-Willi syndrome and/or Angelman syndrome), methylation analysis
81361	HBB (hemoglobin, subunit beta) (eg, sickle cell anemia, beta thalassemia, hemoglobinopathy); common variant(s) (eg, HbS, HbC, HbE)
81362	HBB (hemoglobin, subunit beta) (eg, sickle cell anemia, beta thalassemia, hemoglobinopathy); known familial variant(s)
81363	HBB (hemoglobin, subunit beta) (eg, sickle cell anemia, beta thalassemia, hemoglobinopathy); duplication/deletion variant(s)
81364	HBB (hemoglobin, subunit beta) (eg, sickle cell anemia, beta thalassemia, hemoglobinopathy); full gene sequence
81400	Molecular pathology procedure, Level 1 (eg, identification of single germline variant [eg, SNP] by techniques such as restriction enzyme digestion or melt curve analysis)
81401	Molecular pathology procedure, Level 2 (eg, 2-10 SNPs, 1 methylated variant, or 1 somatic variant [typically using nonsequencing target variant analysis], or detection of a dynamic mutation disorder/triplet repeat)
81403	Molecular pathology procedure, Level 4 (eg, analysis of single exon by DNA sequence analysis, analysis of > 10 amplicons using multiplex PCR in 2 or more independent reactions, mutation scanning or duplication/deletion variants of 2-5 exons)

CPT	DESCRIPTION
81404	Molecular pathology procedure, Level 5 (eg, analysis of 2-5 exons by DNA sequence analysis, mutation scanning or duplication/deletion variants of 6-10 exons, or characterization of a dynamic mutation disorder/triplet repeat by Southern blot analysis)
81405	Molecular pathology procedure, Level 6 (eg, analysis of 6-10 exons by DNA sequence analysis, mutation scanning or duplication/deletion variants of 11-25 exons, regionally targeted cytogenomic array analysis)
81406	Molecular pathology procedure, Level 7 (eg, analysis of 11-25 exons by DNA sequence analysis, mutation scanning or duplication/deletion variants of 26-50 exons
81408	Molecular pathology procedure, Level 9 (eg, analysis of > 50 exons in a single gene by DNA sequence analysis)
81412	Ashkenazi Jewish associated disorders (eg, Bloom syndrome, Canavan disease, cystic fibrosis, familial dysautonomia, Fanconi anemia group C, Gaucher disease, Tay-Sachs disease), genomic sequence analysis panel, must include sequencing of at least 9 genes, including ASPA, BLM, CFTR, FANCC, GBA, HEXA, IKBKAP, MCOLN1, and SMPD1
81430	Hearing loss (eg, nonsyndromic hearing loss, Usher syndrome, Pendred syndrome); genomic sequence analysis panel, must include sequencing of at least 60 genes, including CDH23, CLRN1, GJB2, GPR98, MTRNR1, MYO7A, MYO15A, PCDH15, OTOF, SLC26A4, TMC1, TMPRSS3, USH1C, USH1G, USH2A, and WFS1
81431	Hearing loss (eg, nonsyndromic hearing loss, Usher syndrome, Pendredsyndrome); duplication/deletion analysis panel, must include copy number analyses for STRC and DFNB1 deletions in GJB2 and GJB6 genes
81437	Hereditary neuroendocrine tumor disorders (eg, medullary thyroid carcinoma, parathyroid carcinoma, malignant pheochromocytoma or paraganglioma); genomic sequence analysis panel, must include sequencing of at least 6 genes, including MAX, SDHB, SDHC, SDHD, TMEM127, and VHL
81438	Hereditary neuroendocrine tumor disorders (eg, medullary thyroid carcinoma, parathyroid carcinoma, malignant pheochromocytoma or paraganglioma); duplication/deletion analysis panel, must include analyses for SDHB, SDHC, SDHD, and VHL
81440	Nuclear encoded mitochondrial genes (eg, neurologic or myopathic phenotypes), genomic sequence panel, must include analysis of at least 100 genes, including: BCS1L, C10orf2, COQ2, COX10, DGUOK, MPV17, OPA1, PDSS2, POLG, POLG2, RRM2B, SCO1, SCO2, SLC25A4, SUCLA2, SUCLG1, TAZ, TK2, and TYMP
81443	Genetic testing for severe inherited conditions (eg, cystic fibrosis, Ashkenazi Jewish-associated disorders [eg, Bloom syndrome, Canavan disease, Fanconi anemia type C, mucolipidosis type VI, Gaucher disease, Tay-Sachs disease], beta hemoglobinopathies, phenylketonuria, galactosemia), genomic sequence analysis panel, must include sequencing of at least 15 genes (eg, ACADM, ARSA, ASPA, ATP7B, BCKDHA, BCKDHB, BLM, CFTR, DHCR7, FANCC, G6PC, GAA, GALT, GBA, GBE1, HBB, HEXA, IKBKAP, MCOLN1, PAH)
81479	Unlisted molecular pathology procedure
0335U	Rare diseases (constitutional/heritable disorders), whole genome sequence analysis, including small sequence changes, copy number variants, deletions, duplications, mobile element insertions, uniparental disomy (UPD), inversions, aneuploidy, mitochondrial genome sequence analysis with heteroplasmy and large deletions, short tandem repeat (STR) gene expansions, fetal sample, identification, and categorization of genetic variants
0336U	Rare diseases (constitutional/heritable disorders), whole genome sequence analysis, including small sequence changes, copy number variants, deletions, duplications, mobile element insertions, uniparental disomy (UPD), inversions, aneuploidy, mitochondrial genome sequence analysis with heteroplasmy and large deletions, short tandem repeat

CPT	DESCRIPTION
	(STR) gene expansions, blood or saliva, identification and categorization of genetic variants, each comparator genome (eg, parent)
0400U	Obstetrics (expanded carrier screening), 145 genes by next-generation sequencing, fragment analysis and multiplex ligation-dependent probe amplification, DNA, reported as carrier positive or negative

Reviewed by / Approval Signatures

Population Health & Clinical Quality Committee (PHCQC): Clinton White, MD
Senior Medical Director: Jim Romero, MD
Medical Director: Kresta Antillon, MD
Date Approved: 05-28-2025

References

1. HSD, LOD#32, Prenatal Genetic Screening for Cystic Fibrosis, Spinal Muscular Atrophy (SMA) and Fetal Chromosomal Aneuploidy Dated November 17, 2024, [Cited 4/21/2025]
2. Hayes, Expanded Carrier Screening, Precision Medicine Insights, April 18, 2024 [Cited 4/21/2025]
3. UpToDate, Preconception and prenatal panethnic expanded carrier screening, Literature review current through: Feb 2024. Version 28 This topic last updated: Nov 27, 2024. [Cited 4/21/2025]
4. American College of Obstetricians and Gynecologists (ACOG)
    a. Carrier screening in the age of genomic medicine. Committee Opinion No. 690. Dated March, 2017, Reaffirmed 2023: *Accessed 4/21/2025*
    b. Hemoglobinopathies in Pregnancy. Practice Bulletin No. 78. Dated January, 2007. Reaffirmed September 2024 . Available at URL address: HERE [Cited 4/22/2025]
    c. Committee on Genetics. Committee Opinion No. 691: Mar 2017. Reaffirmed 2023. Carrier Screening for Genetic Conditions. Available at URL address: HERE *Accessed 4/22/2025*
    d. Committee on Obstetrics. Practice Bulletin No. 197. Inherited thrombophilias in pregnancy. 2018 Jul. Accessed Nov 10, 2023. Available at URL address: HERE *Accessed 4/22/2025*
    e. Committee on Practice Bulletins-Obstetrics. Practice Bulletin No. 196. Thromboembolism in pregnancy. 2018 Jul. Accessed Nov 10, 2023. Available at URL address: HERE *Accessed 4/22/2025*
    f. The Society for Maternal/Fetal Medicine. Practice Bulletin No. 162. Prenatal diagnostic testing for genetic disorders. 2016 May. Accessed Nov 10, 2023. Available at URL address: HERE *Accessed 4/22/2025*
    g. Practice Bulletin No. 226 (Oct 2020, replaces Practice Bulletin 163) Screening for fetal chromosomal abnormalities. Accessed Nov 10, 2023. Available at URL address: HERE *Accessed 4/22/2025*
5. MCG,
    a. Deafness and Hearing Loss, Nonsyndromic - Gene and Gene Panel Testing, (A-0823), 29th edition. Last Update: 1/25/2025. [Cited 04/22/2025]
    b. Autosomal Recessive and X-Linked Disease Carrier Screening - Expanded Gene Panels, (A-0768), 29th Edition, Last Update: 1/25/2025. [Cited 04/22/2025]
6. UHC, Carrier Testing Panels for Genetic Diseases, Policy Number: 2025T0586P Effective Date: January 1, 2025, [Cited 4/22/2025]
7. Aetna, Genetic Testing Number: 0140, Last Review: 04/03/2025, Effective: 05/31/1996, Next Review: 02/12/2026, [Cited 4/22/2025]
8. Humana, Genetic Testing for Carrier Screening, Effective Date: 08/29/2024 Revision Date: 08/29/2024 Review Date: 08/29/2024 Policy Number: HUM-0463-039, [Cited 4/22/2025]
9. Cigna, Genetic Testing for Reproductive Carrier, Screening and Prenatal Diagnosis, Policy # 0514, Effective date: 11/1/2024, Next Review Date: 10/15/2025, [Cited 4/22/2025]

Publication History

05-22-2024 **Original effective date**. Reviewed by PHP Medical Policy Committee on 04/10/2024. New policy for Prenatal Genetic Screening for ALOB and to support LOD#101. PA not requirement for Fragile X: (CPT codes: 81243, 81244, 81171, 81172); Spinal Muscular atrophy: (CPT codes: 81336, 81337, 81329); Cystic fibrosis (CF): (CPT code 81220, 81221, 81222, 81223, 81224); Huntington Disease:

(Code 81274); Muscular dystrophies (CPT code 81312, 81234, 81239) or Hemoglobinopathies (CPT Codes 81257, 81259, 81361).
Carrier Screening (CPT codes 0218U and 0449U) will require PA for ALOB, effective 04-01-2024- this was requested in MPM 7.1

8-21-24: Update -pre policy release- MPC met on 07-26-2024 and decided that codes listed on the RTM policies managed by Laboratory Benefit Management (LBM) will be removed from our current MPMs. Effective 08-23-24, LBM will manage the following (2) carrier screening codes: 0121U and 0122U, as part of the LBM policy 2162, LBM Laboratory Procedures Reimbursement**,** which is listed in the PHP Administrative Claims Edits Guide under Appendix A, LBM Program Policy, located at: https://onbaseext.phs.org/PEL/DisplayDocument?ContentID=OB_000000018213
Please note, 0121U and 0122U are both currently set to deny as not appropriate, per this policy.
Update 01/08/2025: Clerical error correction in exclusion section to change Foresight Carrier Screens to Foresight Universal Carrier Screens.

05-28-2025 **Annual Review:** Reviewed by PHP Medical Policy Committee on 04/23/2025. Policy for Prenatal Genetic Screening will continue as status quo, following homegrown criteria to meet LOD #32 (formerly 101), and LOD 42, Biomarkers. Updates to language have been made to clarify guidance for Biomarker Coverage outlined in the Healthcare Authority (HCA) Letter of Direction (LOD) #42. No change in current coverage criteria. Added Test 0449U, BillionToOne with configuration to deny as Investigational and Experimental (I & E) due to indications from ACOG showing this as not supported at this time.

This Medical Policy is intended to represent clinical guidelines describing medical appropriateness and is developed to assist Presbyterian Health Plan and Presbyterian Insurance Company, Inc. (Presbyterian) Health Services staff and Presbyterian medical directors in determination of coverage. The Medical Policy is not a treatment guide and should not be used as such.
For those instances where a member does not meet the criteria described in these guidelines, additional information supporting medical necessity is welcome and may be utilized by the medical director in reviewing the case. Please note that all Presbyterian Medical Policies are available online at: ***Click here for Medical Policies***

Web links:
At any time during your visit to this policy and find the source material web links has been updated, retired, or superseded, PHP is not responsible for the continued viability of websites listed in this policy.

When PHP follows a particular guideline such as LCDs, NCDs, MCG, NCCN etc., for the purposes of determining coverage; it is expected providers maintain or have access to appropriate documentation when requested to support coverage. See the References section to view the source materials used to develop this resource document.