Genetic testing: prenatal and preconception carrier screening

These services may or may not be covered by your HealthPartners plan. Please see your plan documents for your specific coverage information. If there is a difference between this general information and your plan documents, your plan documents will be used to determine your coverage.

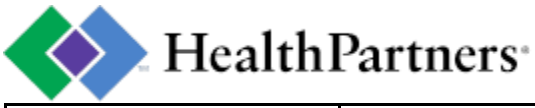
Administrative Process

Prior authorization is not required for prenatal and preconception carrier screening.

Policy Reference Table

If available, codes are listed below for informational purposes only, and do not guarantee member coverage or provider reimbursement. The list may not be all-inclusive.

Coverage Criteria Sections	Example Tests (Labs)	Common CPT Codes	Common ICD Codes
Expanded Carrier Screening Panels	Foresight Universal Panel Carrier Screen (Myriad Genetics)	81329, 81443	O09, Z13, Z31, Z34, Z36, Z84
	Inheritest 500 Plus Panel (Labcorp)	81443	
	GeneSeq Plus (Labcorp)	81336, 81405, 81408, 81479	
	QHerit Expanded Carrier Screen (Quest Diagnostics)	81243, 81443	
	Horizon 27 (27 disease Pan-ethnic Standard Panel (Natera)	81243, 81257, 81329, 81443	
	Genesys Carrier Panel (Genesys Diagnostics)	0400U	
Basic Carrier Screening Panels (Cystic Fibrosis, Spinal Muscular Atrophy, Fragile X, Hemoglobinopathies, not more than 14 genes)	Inheritest Core Panel (Labcorp)	81220, 81222, 81223, 81243, 81257, 81329, 81336, 81361	O09, Z13, Z31, Z34, Z36, Z84
	Inheritest 14-gene Panel (LabCorp)		
	Prenatal Carrier Panel (CFvantage, Fragile X, SMA) (Quest Diagnostics)		
	Foresight Fundamental Panel (Myriad)		
	UNITY Carrier Screen (BillionToOne)	0449U	
Cystic Fibrosis Carrier Screening			
CFTR Targeted Variant Analysis	CFTR One Known Familial Variant in a Nuclear Gene (GeneDx)	81221	O09, Z13, Z31, Z36, Z83.49
CFTR Sequencing Deletion/Duplication Analysis, or Mutation Panel	Cystic Fibrosis Complete Rare Variant Analysis, Entire Gene Sequence (Quest Diagnostics)	81223	
	Cystic Fibrosis Gene Deletion or Duplication (Quest Diagnostics)	81222	
	CFvantage Cystic Fibrosis Expanded Screen (Quest Diagnostics)	81220	



CFTR Intron 9 PolyT and TG Analysis (previously called Intron 8 poly-T/TG Analysis)	*CFTR* Intron 8 Poly-T Analysis (Quest Diagnostics)	81224	
Spinal Muscular Atrophy Carrier Screening			
SMN1 Targeted Variant Analysis	Spinal Muscular Atrophy - SMN1 Known Variant Testing (Nemours) Targeted Variant Analysis (SMN1) (LabCorp)	81337, 81403	O09, Z13, Z31, Z34, Z36, Z84
SMN1 Sequencing and/or Deletion/Duplication Analysis and *SMN2* Deletion/Duplication Analysis	Spinal Muscular Atrophy Carrier Test (Natera)	81329, 81336, 81401, 81405	
	Genomic Unity SMN1/2 Analysis (Variantyx Inc)	0236U	
Fragile X Syndrome Carrier Screening			
FMR1 Repeat Analysis for Carrier Screening	FMR1 CGG Repeat Analysis (Integrated Genetics)	81243, 81244	O09, Z13, Z31, Z34, Z36, Z84
	Fragile X Syndrome, Carrier (LabCorp)		
Hemoglobinopathy Carrier Screening			
HBA1, *HBA2*, or *HBB* Targeted Variant Analysis	Alpha-Globin Common Mutation Analysis (Quest Diagnostics)	81257, 81258	O09, Z13, Z31, Z34, Z36, Z84
	HBA1 One Known Familial Variant in a Nuclear Gene (GeneDx) HBA2 One Known Familial Variant in a Nuclear Gene t (GeneDx)		
	HBB One Known Familial Variant in a Nuclear Gene (GeneDx)	81361, 81362	
HBA1, *HBA2*, or *HBB* Sequencing and/or Deletion/Duplication Analysis	Alpha-Globin Gene Sequencing and Deletion/Duplication (Quest Diagnostics)	81259, 81269,81363, 81364	
	HBA1 Deletion/Duplication (GeneDx) HBA2 Deletion/Duplication (GeneDx)		
	Beta Globin Gene Dosage Analysis (Quest Diagnostics)		
	Beta-Globin Complete (Quest Diagnostics)		
Ashkenazi Jewish Carrier Panel Testing			
Ashkenazi Jewish Carrier Panel Testing	Ashkenazi Jewish Panel (11 Tests) (Quest Diagnostics)	81412	O09, Z13, Z31, Z34, Z36, Z84
Duchenne and Becker Muscular Dystrophy Carrier Screening			
DMD Targeted Variant Analysis	DMD One Known Familial Variant in a Nuclear Gene (GeneDx)	81479	O09, Z13, Z31, Z34, Z36, Z84
DMD Sequencing and/or Deletion/Duplication Analysis	Duchenne/Becker MD (DMD) Gene Sequencing (GeneDx)	81161, 81408	
	Duchenne/Becker MD (DMD) Del/Dup (GeneDx)		

	Genomic Unity DMD Gene Analysis (Variantyx)	0218U	

Coverage

Expanded Carrier Screening Panels

1. Expanded carrier screening panels may be considered **medically necessary** when:
   A. The member is considering pregnancy or is currently pregnant**, **and**
   B. The panel includes the genes *CFTR* and *SMN1.*
2. Expanded carrier screening panels are considered **investigational** for all other indications.

*Fragile X (81243) and spinal muscular atrophy (SMA) (81329) carrier screening may be billed along with 81443 if performed separately from the remainder of the panel per CPT Code Book Guidelines. If CPT code 81243 is billed along with 81443, the member should still meet the specific Fragile X syndrome criteria.
**American College of Medical Genetics and Genomics (ACMG) recommends follow-up screening for the partner of the member that is pregnant or considering pregnancy via analysis of the same gene that has the pathogenic or LP variant as identified in the member. Therefore, expanded carrier screening panels are not recommended to be completed by both reproductive partners in tandem.

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Basic Carrier Screening Panels (Cystic Fibrosis, Spinal Muscular Atrophy, Fragile X, Hemoglobinopathies, but not more than 14 genes)

1. Basic carrier screening panels (CFTR, SMN1/2, FMR1, HBB/HBA1/HBA2, but not more than 14 genes) may be considered **medically necessary** when:
   A. The member is considering pregnancy or is currently pregnant*, **and**
   B. The panel includes the genes CFTR and SMN1.
2. Basic carrier screening panels (CFTR, SMN1/2, FMR1, HBB/HBA1/HBA2, but not more than 14 genes) are considered **investigational** for all other indications.

*ACMG recommends follow-up screening for the partner of the member that is pregnant or considering pregnancy via analysis of the same gene that has the pathogenic or LP variant as identified in the member. Therefore, basic carrier screening panels are not recommended to be completed by both reproductive partners in tandem.

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Cystic Fibrosis Carrier Screening

CFTR Targeted Variant Analysis

1. Cystic fibrosis carrier screening via *CFTR* targeted variant analysis may be considered **medically necessary** when:
   A. The member or the member's reproductive partner is considering pregnancy or is currently pregnant, **and**
   B. The member has a close relative with a known pathogenic or likely pathogenic variant in *CFTR*.
2. Cystic fibrosis carrier screening via CFTR targeted mutation analysis for a known familial mutation (81221) is considered **investigational** for all other indications.

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CFTR Sequencing, Deletion/Duplication Analysis, or Mutation Panel

1. Cystic fibrosis carrier screening via CFTR sequencing, deletion/duplication analysis, or a mutation panel using at a minimum the ACMG-100 variant panel, may be considered **medically necessary** when:
   A. The member or the member's reproductive partner is considering pregnancy or is currently pregnant, **or**
   B. The member's reproductive partner is a known carrier for cystic fibrosis.
2. Cystic fibrosis carrier screening via *CFTR* sequencing, deletion/duplication analysis, or a mutation panel using at a minimum the ACMG-100 variant panel, is considered **investigational** for all other indications.

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CFTR Intron 9 PolyT and TG Analysis (previously called Intron 8 polyT/TG Analysis)

1. Analysis of the CFTR intron 9 polyT and TG regions for cystic fibrosis carrier screening may be considered **medically necessary** when:
   A. The member or the member's reproductive partner is considering pregnancy or is currently pregnant, **and**
   B. The member is known to have an R117H variant in the CFTR gene.
2. Analysis of the CFTR intron 9 polyT and TG regions for cystic fibrosis carrier screening is considered **investigational** for all other indications.

Note: Refer to **Genetic testing for multisystem inherited disorders, intellectual disability and developmental delay** policy for coverage criteria for genetic testing to establish a diagnosis of cystic fibrosis.

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Spinal Muscular Atrophy Carrier Screening

SMN1 Targeted Variant Analysis

1. Spinal muscular atrophy (SMA) carrier screening via *SMN1* targeted variant analysis may be considered **medically necessary** when:
   - A. The member or the member's reproductive partner is considering pregnancy or is currently pregnant, **and**
   - B. The member has a close relative with a known pathogenic or likely pathogenic variant in *SMN1*.
2. Spinal muscular atrophy (SMA) carrier screening via *SMN1* targeted variant analysis is considered **investigational** for all other indications.

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SMN1 Sequencing and/or Deletion/Duplication and *SMN2* Deletion/Duplication Analysis

1. Spinal muscular atrophy (SMA) carrier screening via *SMN1* sequencing and/or deletion/duplication analysis and *SMN2* deletion/duplication analysis is considered **medically necessary** when:
   - A. The member or member's reproductive partner is considering pregnancy or is currently pregnant, **or**
   - B. The member's reproductive partner is a known carrier for spinal muscular atrophy.
2. Spinal muscular atrophy (SMA) carrier screening via *SMN1* sequencing and/or deletion/duplication analysis and *SMN2* deletion/duplication analysis is considered **investigational** for all other indications.

Note: Refer to **Genetic testing for epilepsy, neuromuscular, and neurodegenerative disorders** policy for coverage criteria for genetic testing to establish a diagnosis of spinal muscular atrophy (SMA).

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Fragile X Syndrome Carrier Screening

FMR1 Repeat Analysis for Carrier Screening

1. Fragile X carrier screening via *FMR1* CGG-trinucleotide repeat analysis may be considered **medically necessary** when:
   - A. The member has been diagnosed with premature ovarian insufficiency or elevated follicle-stimulating hormone level before age 40 years, **or**
   - B. The member is considering a pregnancy or is currently pregnant, **and**
     - i. The member has one of the following:
       - a) Close relative with Fragile X syndrome (i.e., close relative has more than 200 CGG repeats in the *FMR1* gene), **or**
       - b) Close relative who is a known carrier for Fragile X syndrome (i.e., close relative has between 55-200 CGG repeats in the *FMR1* gene), **or**
       - c) Close relative with unexplained intellectual disability, developmental delay, or autism spectrum disorder, **or**
       - d) Close relative diagnosed with premature ovarian insufficiency or elevated follicle-stimulating hormone level before age 40 years.
2. Fragile X carrier screening via *FMR1* CGG-trinucleotide repeat analysis is considered **investigational** for all other indications.

Note: Refer to **Genetic testing for multisystem inherited disorders, intellectual disability and developmental delay** policy for coverage criteria for genetic testing to establish a diagnosis of fragile X syndrome. Additionally, if FMR repeat analysis (81243) is billed along with an additional carrier screen panel code (81443), the member should still meet the above Fragile X syndrome criteria.

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Hemoglobinopathy Carrier Screening

HBA1, *HBA2*, or *HBB* Targeted Variant Analysis

1. Hemoglobinopathy carrier screening via *HBA1*, *HBA2*, or *HBB* targeted variant analysis may be considered **medically necessary** when:
   - A. The member or the member's reproductive partner is considering pregnancy or is currently pregnant, **and**
     - i. The member has a close relative with a known pathogenic or likely pathogenic variant in *HBA1*, *HBA2*, or *HBB.*
2. Hemoglobinopathy carrier screening via *HBA1*, *HBA2*, or *HBB* targeted variant analysis is considered **investigational** for all other indications.

Note: If a member's reproductive partner is known to be a carrier of a hemoglobinopathy, via genetic testing results and/or hematologic screening results, the more appropriate test for the member is likely HBA1, HBA2, or HBB Sequencing and/or Deletion/Duplication Analysis.

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HBA1, *HBA2*, or *HBB* Sequencing and/or Deletion/Duplication Analysis

1. Hemoglobinopathy carrier screening via *HBA1*, *HBA2*, or *HBB* sequencing and/or deletion/duplication analysis may be considered **medically necessary** when:

A. The member or the member's reproductive partner is considering pregnancy or is currently pregnant.

2. Hemoglobinopathy carrier screening via *HBA1*, *HBA2*, or *HBB* sequencing and/or duplication analysis is considered **investigational** for all other indications, including fetal hemoglobin testing via circulating fetal DNA.

Note: Refer to **Genetic testing: hematologic disorders (non-cancerous)** for coverage criteria for genetic testing to establish a diagnosis of a hemoglobinopathy.

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Ashkenazi Jewish Carrier Panel Testing

1. Ashkenazi Jewish carrier panel testing may be considered **medically necessary** when:
   A. The member or the member's reproductive partner is considering pregnancy or is currently pregnant, **and**
   B. The member is of Ashkenazi Jewish ancestry, **and**
   C. The panel includes, at a minimum, screening for carrier status for genetic conditions associated with the following genetic conditions, as recommended by the American College of Obstetricians and Gynecologists (ACOG):
      i. Tay Sachs disease (HEXA)
      ii. Canavan disease (ASPA)
      iii. Cystic fibrosis (CFTR)
      iv. Familial dysautonomia (ELP1)
      v. Bloom syndrome (BLM)
      vi. Fanconi anemia (FANCC)
      vii. Niemann-Pick disease (SMPD1)
      viii. Gaucher disease (GBA)
      ix. Mucolipidosis IV (MCOLN1)
      x. Glycogen storage disease type I (G6PC1)
      xi. Joubert syndrome (*TMEM216*)
      xii. Maple syrup urine disease (*BCKDHB*)
      xiii. Usher syndrome types 1F and III (*PDCH15* and *CLRN1*).
2. Ashkenazi Jewish carrier panel testing is considered **investigational** for all other indications.

Note: If only one partner is of Ashkenazi Jewish ancestry, then testing of that partner is considered medically necessary. Testing of the other partner is considered medically necessary only if the result of testing of the Ashkenazi Jewish partner is positive.

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Duchenne and Becker Muscular Dystrophy Carrier Screening

DMD Targeted Variant Analysis

1. Duchenne and Becker muscular dystrophy carrier screening via *DMD* targeted variant analysis may be considered **medically necessary** when:
   A. The member is considering pregnancy or is currently pregnant, **and**
   B. The member has a close relative with a known pathogenic or likely pathogenic variant in *DMD*.
2. Duchenne and Becker muscular dystrophy carrier screening via *DMD* targeted variant analysis is considered **investigational** for all other indications.

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DMD Sequencing and/or Deletion/Duplication Analysis

1. Duchenne and Becker muscular dystrophy carrier screening via *DMD* sequencing and/or deletion/duplication analysis may be considered **medically necessary** when:
   A. The member is considering pregnancy or is currently pregnant, **and**
   B. The member has a first- or second-degree relative diagnosed with Duchenne or Becker muscular dystrophy.
2. Duchenne and Becker muscular dystrophy carrier screening via *DMD* sequencing and/or deletion/duplication analysis is considered i**nvestigational** for all other indications.

Note: Refer to **Genetic testing for epilepsy, neuromuscular, and neurodegenerative disorders** for coverage criteria for genetic testing to establish a diagnosis of Duchenne or Becker muscular dystrophy.

Definitions

1. Close relatives include first, second, and third-degree relatives on the same side of the family:
   A. **First-degree relatives** are parents, siblings, and children
   B. **Second-degree relatives** are grandparents, aunts, uncles, nieces, nephews, grandchildren, and half siblings
   C. **Third-degree relatives** are great grandparents, great aunts, great uncles, great grandchildren, and first cousins.

Products

This information is for most, but not all, HealthPartners plans. Please read your plan documents to see if your plan has limits or will not cover some items. If there is a difference between this general information and your plan documents, your plan documents will be used to determine your coverage. These coverage criteria do not apply to Medicare Products. For more information regarding Medicare coverage criteria or for a copy of a Medicare coverage policy, contact Member Services at 952-883-7272 or 1-877-778-8384.

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References

1. Deignan JL, Astbury C, Cutting GR, et al. CFTR variant testing: a technical standard of the American College of Medical Genetics and Genomics (ACMG). Genet Med. 2020;22(8):1288-1295. doi:10.1038/s41436-020-0822-5
2. Committee Opinion No. 690. Obstet Gynecol. 2017;129(3):e35-e40. doi:10.1097/AOG.0000000000001951
3. Committee Opinion No. 691. Obstet Gynecol. 2017 (Reaffirmed 2023);129(3):1-15. doi:10.1097/AOG.0000000000001948
4. Gregg AR, Aarabi M, Klugman S, et al. Screening for autosomal recessive and X-linked conditions during pregnancy and preconception: a practice resource of the American College of Medical Genetics and Genomics (ACMG) [published online ahead of print, 2021 Jul 20] [published correction appears in Genet Med. 2021 Aug 27;:]. Genet Med. 2021;10.1038/s41436-021-01203-z. doi:10.1038/s41436-021-01203-z
5. Darras BT, Urion DK, Ghosh PS. Dystrophinopathies. 2000 Sep 5 [Updated 2022 Jan 20]. In: Adam MP, Ardinger HH, Pagon RA, et al., editors. GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2022. Available from: https://www.ncbi.nlm.nih.gov/books/NBK1119/
6. Fratter C, Dalgleish R, Allen SK, et al. EMQN best practice guidelines for genetic testing in dystrophinopathies. Eur J Hum Genet. 2020;28(9):1141-1159. doi:10.1038/s41431-020-0643-7
7. Sherman S, Pletcher BA, Driscoll DA. Fragile X syndrome: diagnostic and carrier testing. Genet Med. 2005;7(8):584-587. doi:10.1097/01.gim.0000182468.22666.dd
8. Committee opinion no. 605: primary ovarian insufficiency in adolescents and young women. Obstet Gynecol. 2014 (Reaffirmed 2021);124(1):193-197. doi:10.1097/00125817-200103000-0001001.AOG.0000451757.51964.98
9. Deignan JL, Gregg AR, Grody WW, et al. Updated recommendations for *CFTR* carrier screening: a position statement of the American College of Medical Genetics and Genomics (ACMG). Genet Med. 2023;25(8):100867.
10. ACOG with the assistance of Ahizechukwu C. Eke, MD, MPH, PhD; Manisha Gandhi, MD; Anjali J. Kaimal, MD, MAS; Michelle Moniz, MD, MSc; and Andrea Shields, MD, MS. ACOG Practice Advisory: Hemoglobinopathies in Pregnancy. August 2022. Reaffirmed September 2023. https://www.acog.org/clinical/clinical-guidance/practice-advisory/articles/2022/08/hemoglobinopathies-in-pregnancy