EmblemHealth

Carrier Screening for Parents or Prospective Parents

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Definitions

Carrier screening	Used to identify people who carry one copy of a gene mutation that, when present in two copies, causes a genetic disorder. The testing is offered to individuals who have a family history of a genetic disorder and to people in certain ethnic groups with an increased risk of specific genetic conditions
Gene mutation	A change from the normal gene sequence of which underlies a detrimental clinical presentation associated with disease
First-degree relative	Parents, siblings, and children
Second-degree relative	Grandparents, aunts, uncles, half-siblings, nieces, nephews and grandchildren.
Third-degree relative	Great-grandparents, great-aunts, great-uncles, grand-niece, grand-nephew, first cousin, great-grandchildren
Hereditary (aka germline) gene mutations	Mutations inherited from a parent and present throughout an individual's life in virtually every cell in the body. When an egg and a sperm (germ) cell unite, the resulting fertilized egg cell receives DNA from both parents. If this DNA has a mutation, the child that grows from the fertilized egg will have the mutation in each of his or her cells
X-linked disorder	Disease caused by a gene mutation located on the X-sex. X-linked disorders are expressed in all males with the defective gene but only females with mutated genes on both X chromosomes express the disease
Autosomal dominant	A gene mutation located on a numbered or non-sex chromosome which expresses a disease condition when present as part of a heterozygotic gene pair
Autosomal recessive	A gene mutation located on a numbered or non-sex chromosome which expresses a disease condition only when present in homozygous pairs. Carriers are not at risk of developing the disease

Guideline

Carrier screening for parents or prospective parents is considered medically necessary. Pre/post-test genetic counseling is strongly recommended:

A. Preconceptional or prenatal genetic testing is considered medically necessary when any of the following criteria based on family history is met:

1. Parents or prospective parents have a previously affected child with the genetic disease
2. One or both parents or prospective parents have 1st or 2nd degree relative who is affected or they have a first degree relative with an affected child
3. One parent or prospective parent is at high risk for a genetic disorder with a late onset presentation or is a known carrier for an autosomal recessive condition
4. One or both prospective parents have an autosomal dominant disorder
5. Prospective parents have a history of unexplained stillbirth or repeated 1st trimester miscarriages (≥ 3)

B. Specific genetic testing is considered medically necessary when any of the above criteria are met and all of the following criteria are met:

1. The genetic disorder has been established in the scientific literature to be reliably associated with the disease and is associated high morbidity in the homozygous or compound heterozygous state
2. Alternate biochemical or other clinical tests are not available, provide an indeterminate result or are less effective than genetic testing

C. The following test-specific policies are considered medically necessary:

1. Ashkenazi Jewish genetic disorders (JGDs) — to determine carrier status (i.e., [list may not be all-inclusive], Ashkenazi panel for Bloom syndrome, Canavan disease, cystic fibrosis, familial dysautonomia, familial hyperinsulinemia, Fanconi anemia Type C, Gaucher disease, glycogen storage disease [GSD] Type 1a, Joubert syndrome, maple-syrup urine disease [MSUD], mucolipidosis Type IV [MLIV], Niemann-Pick disease Type A, Tay-Sachs disease [TSD], Usher syndrome *)

   *** Screening for Tay-Sachs disease should be offered when considering pregnancy or during pregnancy if either member of a couple is of Ashkenazi Jewish, French Canadian, or Cajun descent. Those with a family history consistent with Tay-Sachs disease also should be offered screening.**
2. Cystic fibrosis (CF) — to determine carrier status for up to 23 CFTR gene mutations
3. Fragile X syndrome (no restrictions)
4. Hemoglobinopathies — to determine carrier status if any of the following is present:
   a. Mother is planning a pregnancy or currently pregnant and at least one parent is in an at-risk population (African, Southeast Asian and/or Mediterranean ancestry)
   b. Mother has a family history of hemoglobinopathy
5. The other partner is a known carrier or affected with a hemoglobinopathy (e.g., Sickle cell disease)
6. Spinal muscular atrophy (no restrictions)

Limitations/Exclusions

A. Coverage is limited to once per lifetime, as repeat screening is not considered medically necessary.

B. Coverage for CF testing is limited to analysis of the 23 the most common CFTR gene mutations.

C. If one parent/prospective parent screens negative for a disease associated with an autosomal recessive disorder then testing the other parent is not considered medically necessary

D. Coverage is contingent on the following:

1. Test targets analysis to the number of genes associated with medically necessary indications listed above (applicable to "D" below)
2. Test result must impact medical management of a current pregnancy and/or the reproductive choices of the (if request is for conditions other than those listed above)
3. Test must has proven validity in the medical community for the identification of a specific genetically linked inheritable disease

E. Next generation sequencing (i.e., rapid sequencing of large numbers of DNA segments, up to and including entire genomes) is not considered medically necessary. Expanded panels (e.g., nonstandard universal-type genetic tests) typically screen for diseases present with increased frequency in specific populations; however, they also screen for diseases outside the carrier risk (diminishing the clinical utility of the test) and thus presenting considerable ethical and genetic counseling challenges.

F. Home testing (e.g., direct-to-consumer; aka home-testing kits) or self-referral testing (e.g., genetic tests ordered by members via telephone or Internet) is not considered medically necessary, as there is no evidence of efficacy.

Revision History

Sept. 10, 2021	Added 4 autosomal recessive conditions (familial hyperinsulinemia, Joubert syndrome, maple syrup urine disease, and Usher syndrome) to Ashkenazi Jewish genetic disorders (eff. 12/10/2021), and noted that the list of examples may not be all-inclusive. Added note pertaining to Tay-Sachs disease regarding screening members of Ashkenazi Jewish, French Canadian, or Cajun descent, as well as those with a family history consistent with Tay-Sachs disease.
Sept. 14, 2018	Added to Limitations/Exclusions that if one parent screens negative for an autosomal recessive disorder then testing the other parent is not medically necessary.
Apr. 13, 2018	Removed SMA restrictions.
Oct. 13, 2017	Removed Fragile X restrictions.
Jan.1, 2016	Added hemoglobinopathy and SMA criteria for clarification.

Applicable Procedure Codes

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, deletions, and mobile element insertions (Not covered for Medicaid)
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) (eff. 4/1/2024) (Not covered for Medicaid)
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
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)
81242	FANCC (Fanconi anemia, complementation group C) (eg, Fanconi anemia, type C) gene analysis, common variant (eg, IVS4+4A>T)
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 methylation status)
81250	G6PC (glucose-6-phosphatase, catalytic subunit) (eg, Glycogen storage disease, type 1a, von Gierke disease) gene analysis, common variants (eg, R83C, Q347X)
81251	GBA (glucosidase, beta, acid) (eg, Gaucher disease) gene analysis, common variants (eg, N370S, 84GG, L444P, IVS2+1G>A)
81255	HEXA (hexosaminidase A [alpha polypeptide]) (eg, Tay-Sachs disease) gene analysis, common variants (eg, 1278insTATC, 1421+1G>C, G269S)
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)
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)
81271	HTT (huntingtin) (eg, Huntington disease) gene analysis; evaluation to detect abnormal (eg, expanded) alleles
81274	HTT (huntingtin) (eg, Huntington disease) gene analysis; characterization of alleles (eg, expanded size)
81290	MCOLN1 (mucolipin 1) (eg, Mucolipidosis, type IV) gene analysis, common variants (eg, IVS3-2A>G, del6.4kb)
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
81330	SMPD1(sphingomyelin phosphodiesterase 1, acid lysosomal) (eg, Niemann-Pick disease, Type A) gene analysis, common variants (eg, R496L, L302P, fsP330)
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)
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) LINC00518 (long intergenic non-protein coding RNA 518) (eg, melanoma), expression analysis PRAME (preferentially expressed antigen in melanoma) (eg, melanoma), expression analysis

81402	Molecular pathology procedure, Level 3 (eg, >10 SNPs, 2-10 methylated variants, or 2-10 somatic variants [typically using non-sequencing target variant analysis], immunoglobulin and T-cell receptor gene rearrangements, duplication/deletion variants of 1 exon, loss of heterozygosity [LOH], uniparental disomy [UPD])
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)
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) CPOX (coproporphyrinogen oxidase) (eg, hereditary coproporphyria), full gene sequence CTRC (chymotrypsin C) (eg, hereditary pancreatitis), full gene sequence PKLR (pyruvate kinase, liver and RBC) (eg, pyruvate kinase deficiency), full gene sequence
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, cytogenomic array analysis for neoplasia) ANOS1 (anosmin-1) (eg, Kallmann syndrome 1), full gene sequence HMBS (hydroxymethylbilane synthase) (eg, acute intermittent porphyria), full gene sequence PPOX (protoporphyrinogen oxidase) (eg, variegate porphyria), full gene sequence
81407	Molecular pathology procedure, Level 8 (eg, analysis of 26-50 exons by DNA sequence analysis, mutation scanning or duplication/deletion variants of >50 exons, sequence analysis of multiple genes on one platform)
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
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
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
81599	Unlisted multianalyte assay with algorithmic analysis

Applicable ICD-10 Diagnosis Codes

D56.0	Alpha thalassemia
D56.4	Hereditary persistence of fetal hemoglobin [HPFH]
D56.5	Hemoglobin E-beta thalassemia
D56.8	Other thalassemias
D57.00	Hb-SS disease with crisis, unspecified
D57.01	Hb-SS disease with acute chest syndrome
D57.02	Hb-SS disease with splenic sequestration
D57.04	Hb-SS disease with dactylitis
D57.1	Sickle-cell disease without crisis
D57.20	Sickle-cell/Hb-C disease without crisis
D57.211	Sickle-cell/Hb-C disease with acute chest syndrome

D57.212	Sickle-cell/Hb-C disease with splenic sequestration
D57.214	Sickle-cell/Hb-C disease with dactylitis
D57.219	Sickle-cell/Hb-C disease with crisis, unspecified
D57.3	Sickle-cell trait
D57.40	Sickle-cell thalassemia without crisis
D57.411	Sickle-cell thalassemia with acute chest syndrome
D57.412	Sickle-cell thalassemia with splenic sequestration
D57.414	Sickle-cell thalassemia, unspecified, with dactylitis
D57.419	Sickle-cell thalassemia with crisis, unspecified
D57.434	Sickle-cell thalassemia beta zero with dactylitis
D57.454	Sickle-cell thalassemia beta plus with dactylitis
D57.80	Other sickle-cell disorders without crisis
D57.811	Other sickle-cell disorders with acute chest syndrome
D57.812	Other sickle-cell disorders with splenic sequestration
D57.814	Other sickle-cell disorders with dactylitis
D57.819	Other sickle-cell disorders with crisis, unspecified
D58.2	Other hemoglobinopathies
D61.02	Shwachman-Diamond syndrome
D61.09	Other constitutional aplastic anemia
D80.6	Antibody deficiency with near-normal immunoglobulins or with hyperimmunoglobulinemia
D82.1	Di George's syndrome
D89.84	IgG4-related disease
E23.6	Other disorders of pituitary gland
E28.8	Other ovarian dysfunction
E71.0	Maple-syrup-urine disease
E74.00	Glycogen storage disease, unspecified
E74.01	von Gierke disease
E74.02	Pompe disease
E74.03	Cori disease
E74.04	McArdle disease
E74.05	Lysosome-associated membrane protein 2 [LAMP2] deficiency
E74.09	Other glycogen storage disease
E74.4	Disorders of pyruvate metabolism and gluconeogenesis
E75.00	GM2 gangliosidosis, unspecified
E75.01	Sandhoff disease
E75.02	Tay-Sachs disease
E75.09	Other GM2 gangliosidosis

E75.10	Unspecified gangliosidosis
E75.11	Mucolipidosis IV
E75.19	Other gangliosidosis
E75.21	Fabry (-Anderson) disease
E75.22	Gaucher disease
E75.23	Krabbe disease
E75.240	Niemann-Pick disease type A
E75.241	Niemann-Pick disease type B
E75.242	Niemann-Pick disease type C
E75.243	Niemann-Pick disease type D
E75.248	Other Niemann-Pick disease
E75.249	Niemann-Pick disease, unspecified
E75.25	Metachromatic leukodystrophy
E75.27	Pelizaeus-Merzbacher disease
E75.28	Canavan disease
E75.29	Other sphingolipidosis
E75.3	Sphingolipidosis, unspecified
E75.4	Neuronal ceroid lipofuscinosis
E77.0	Defects in post-translational modification of lysosomal enzymes
E77.1	Defects in glycoprotein degradation
E77.8	Other disorders of glycoprotein metabolism
E77.9	Disorder of glycoprotein metabolism, unspecified
E79.81	Aicardi-Goutieres syndrome
E79.82	Hereditary xanthinuria
E79.89	Other specified disorders of purine and pyrimidine metabolism
E84.0	Cystic fibrosis with pulmonary manifestations
E84.11	Meconium ileus in cystic fibrosis
E84.19	Cystic fibrosis with other intestinal manifestations
E84.9	Cystic fibrosis, unspecified
E88.A	Wasting disease (syndrome) due to underlying condition
G11.0	Congenital nonprogressive ataxia
G11.1	Early-onset cerebellar ataxia
G11.2	Late-onset cerebellar ataxia
G11.5	Hypomyelination - hypogonadotropic hypogonadism - hypodontia
G11.6	Leukodystrophy with vanishing white matter disease
G12.1	Other inherited spinal muscular atrophy
G12.8	Other spinal muscular atrophies and related syndromes

G12.9	Spinal muscular atrophy, unspecified
G90.1	Familial dysautonomia [Riley-Day]
L10.4	Pemphigus erythematosus
N96	Recurrent pregnancy loss
Q04.3	Other reduction deformities of brain
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
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
Q92.0	Whole chromosome trisomy, nonmosaicism (meiotic nondisjunction)
Q92.1	Whole chromosome trisomy, mosaicism (mitotic nondisjunction)
Q92.2	Partial trisomy
Q92.7	Triploidy and polyploidy
Q92.8	Other specified trisomies and partial trisomies of autosomes
Q92.9	Trisomy and partial trisomy of autosomes, unspecified
Q95.0	Balanced translocation and insertion in normal individual
Q95.2	Balanced autosomal rearrangement in abnormal individual
Q95.3	Balanced sex/autosomal rearrangement in abnormal individual
Q96.0	Karyotype 45, X
Q96.1	Karyotype 46, X iso (Xq)
Q96.2	Karyotype 46, X with abnormal sex chromosome, except iso (Xq)
Q96.3	Mosaicism, 45, X/46, XX or XY
Q96.4	Mosaicism, 45, X/other cell line(s) with abnormal sex chromosome
Q97.0	Karyotype 47, XXX
Q97.1	Female with more than three X chromosomes

Q97.2	Mosaicism, lines with various numbers of X chromosomes
Q97.3	Female with 46, XY karyotype
Q97.8	Other specified sex chromosome abnormalities, female phenotype
Q97.9	Sex chromosome abnormality, female phenotype, unspecified
Q98.0	Klinefelter syndrome karyotype 47, XXY
Q98.1	Klinefelter syndrome, male with more than two X chromosomes
Q98.3	Other male with 46, XX karyotype
Q98.4	Klinefelter syndrome, unspecified
Q98.5	Karyotype 47, XYY
Q98.6	Male with structurally abnormal sex chromosome
Q98.7	Male with sex chromosome mosaicism
Q98.8	Other specified sex chromosome abnormalities, male phenotype
Q98.9	Sex chromosome abnormality, male phenotype, unspecified
Q99.0	Chimera 46, XX/46, XY
Q99.1	46, XX true hermaphrodite
Q99.2	Fragile X chromosome
Q99.8	Other specified chromosome abnormalities
Q99.9	Chromosomal abnormality, unspecified
Z14.1	Cystic fibrosis carrier
Z14.8	Genetic carrier of other disease
Z15.89	Genetic susceptibility to other disease
Z31.430	Encounter of female for testing for genetic disease carrier status for procreative management
Z31.438	Encounter for other genetic testing of female for procreative management
Z31.440	Encounter of male for testing for genetic disease carrier status for procreative management
Z31.441	Encounter for testing of male partner of patient with recurrent pregnancy loss
Z31.5	Encounter for procreative genetic counselingEncounter for genetic counseling
Z36.0	Encounter for antenatal screening for chromosomal anomalies
Z36.1	Encounter for antenatal screening for raised alphafetoprotein level
Z36.2	Encounter for other antenatal screening follow-up
Z36.3	Encounter for antenatal screening for malformations
Z36.4	Encounter for antenatal screening for fetal growth retardation
Z36.5	Encounter for antenatal screening for isoimmunization
Z36.81	Encounter for antenatal screening for hydrops fetalis
Z36.82	Encounter for antenatal screening for nuchal translucency
Z36.83	Encounter for fetal screening for congenital cardiac abnormalities
Z36.84	Encounter for antenatal screening for fetal lung maturity
Z36.85	Encounter for antenatal screening for Streptococcus B

Z36.86	Encounter for antenatal screening for cervical length
Z36.87	Encounter for antenatal screening for uncertain dates
Z36.88	Encounter for antenatal screening for fetal macrosomia
Z36.89	Encounter for other specified antenatal screening
Z36.8A	Encounter for antenatal screening for other genetic defects
Z84.81	Family history of carrier of genetic disease

References

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