UHA
HEALTH INSURANCE

# Genetic Testing: Carrier Screening and Preimplantation Diagnosis

## *I. Policy*

University Health Alliance (UHA) will reimburse for carrier screening for inheritable genetic diseases and preimplantation genetic diagnosis (PGD) testing when they are determined to be medically necessary and when they meet the medical criteria guidelines (subject to limitations and exclusions) indicated below.

## *II. Criteria/Guidelines*

A. Carrier screening for inheritable genetic diseases is covered (subject to Limitations and Administrative Guidelines) only in members who are fertile and contemplating having children.

B. Carrier screening for inheritable genetic diseases is covered (subject to Limitations and Administrative Guidelines) when one of the following criteria is met:

1. One or both individuals have a first- or second-degree relative who is affected; or
2. One individual is known to be a carrier; or
3. One or both individuals are members of a population known to have a carrier rate that exceeds a threshold considered appropriate for testing for a particular condition (Limitations III.A); and
4. The natural history of the disease is well understood and there is a reasonable likelihood that the disease is one with high morbidity in the homozygous or compound heterozygous state; and
5. Alternative biochemical or other clinical tests to definitively diagnose carrier status are not available or, if available, provide an indeterminate result or are individually less efficacious than genetic testing; and
6. The genetic test has adequate clinical validity to guide clinical decision making and for which residual risk is understood (see Limitations Section III.B); and
7. An association of the marker with the disorder has been established.

*NOTE: First-degree relatives include biological parent, brother, sister, or child; second-degree relatives include biologic grandparent, aunt, uncle, niece, nephew, grandchildren, and half-sibling*.

C. Preimplantation genetic diagnosis (PGD) testing is covered (subject to Limitations/Exclusions and Administrative Guidelines) as an adjunct to IVF, in couples meeting one of the following criteria:

1. Couples who are known carriers of a genetic mutation which causes a potentially lethal or severely disabling condition with limited treatment options meeting one of the following criteria:
    a. Both partners are known carriers of the same single autosomal recessive disorder;
    b. One partner is a known carrier of an autosomal recessive disorder and the couple have previously produced offspring affected by that disorder;
    c. One partner is a known carrier of a single gene autosomal dominant disorder; or
    d. One partner is a known carrier of a single X-linked disorder.
2. Couples with balanced or unbalanced chromosomal translocation with an elevated risk of a chromosomal abnormality which causes a potentially lethal or severely disabling condition with limited treatment options.

D. Laboratories performing clinical tests must be certified under the Clinical Laboratory Improvement Amendments (CLIA) of 1988.

***NOTE:***

*This UHA payment policy is a guide to coverage, the need for prior authorization and other administrative directives. It is not meant to provide instruction in the practice of medicine and it should not deter a provider from expressing his/her judgment.*

*Even though this payment policy may indicate that a particular service or supply is considered covered, specific provider contract terms and/or member individual benefit plans may apply, and this policy is not a guarantee of payment. UHA reserves the right to apply this payment policy to all UHA companies and subsidiaries.*

*UHA understands that opinions about and approaches to clinical problems may vary. Questions concerning medical necessity (see Hawaii Revised Statutes §432E-1.4) are welcome. A provider may request that UHA reconsider the application of the medical necessity criteria in light of any supporting documentation.*

## *III. Limitations/Exclusions*

A. If there is no family history of risk based or ethnic predilection for a disease, carrier screening is not covered when the carrier rate is less than 1% in the general population.

B. Carrier screening will only be covered for adults who are fertile and contemplating having children.

C. Expanded carrier screening panels are not covered as they are not known to be effective in improving long-term health outcomes

D. PGD does not meet payment determination criteria in the following situations:

1. When performed in couples undergoing IVF who have failed prior IVF cycles, solely to increase the chances of live birth rates
2. For aneuploidy screening in IVF performed solely because of advanced maternal age
3. When used to determine gender selection or "family balancing"
4. To determine the human leukocyte antigen (HLA) or other marker status of an embryo as a potential future stem cell donor
5. For testing of late onset disease or disease disposition

E. PGD services are not related to or dependant upon the member's IVF benefit (see IVF policy for details of benefits).

## *IV. Administrative Guidelines*

A. Prior authorization is required.

B. To request prior authorization, please submit via UHA's online portal. If a login has not been established, you may contact UHA at 808-532-4000 to establish one.

| CPT Code | Description |
|---|---|
| 81200 | ASPA (aspartoacylase) (eg, Canavan disease) gene analysis, common variants (e.g., E285A, Y231X) |
| 81221 | CFTR (cystic fibrosis transmembrane conductance regulator) (e.g., cystic fibrosis) gene analysis; known familial variants |
| 81222 | CFTR (cystic fibrosis transmembrane conductance regulator) (e.g., cystic fibrosis) gene analysis; duplication/deletion variants |

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| 81223 | CFTR (cystic fibrosis transmembrane conductance regulator) (e.g., cystic fibrosis) gene analysis; full gene sequence |
| 81224 | CFTR (cystic fibrosis transmembrane conductance regulator) (e.g., cystic fibrosis) gene analysis; intron 8 poly-T analysis (e.g., male infertility) |
| 81240 | F2 (prothrombin, coagulation factor II) (e.g., hereditary hypercoagulability) gene analysis, 20210G>A variant |
| 81241 | F5 (coagulation factor V) (e.g., hereditary hypercoagulability) gene analysis, Leiden variant |
| 81243 | FMR1 (fragile X mental retardation 1) (e.g., fragile X mental retardation) gene analysis; evaluation to detect abnormal (e.g., expanded) alleles |
| 81244 | FMR1 (fragile X mental retardation 1) (eg, fragile X mental retardation) gene analysis; characterization of alleles (e.g., expanded size and methylation status) |
| 81251 | GBA (glucosidase, beta, acid) (e.g., Gaucher disease) gene analysis, common variants (e.g., N370S, 84GG, L444P, IVS2+1G>A) |
| 81255 | HEXA (hexosaminidase A [alpha polypeptide]) (eg, Tay-Sachs disease) gene analysis, common variants (e.g., 1278insTATC, 1421+1G>C, G269S) |
| 81256 | HFE (hemochromatosis) (e.g., hereditary hemochromatosis) gene analysis, common variants (e.g., C282Y, H63D) |
| 81257 | HBA1/HBA2 (alpha globin 1 and alpha globin 2) (e.g., alpha thalassemia, Hb Bart hydrops fetalis syndrome, HbH disease), gene analysis; common deletions or variant (e.g., Southeast Asian, Thai, Filipino, Mediterranean, alpha3.7, alpha4.2, alpha20.5, Constant Spring) |
| 81260 | IKBKAP (inhibitor of kappa light polypeptide gene enhancer in B-cells, kinase complex-associated protein) (e.g., familial dysautonomia) gene analysis, common variants (e.g., 2507+6T>C, R696P) |
| 81412 | Ashkenazi Jewish associated disorders (e.g., 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 |
| 81470 | X-linked intellectual disability (XLID) (e.g., syndromic and non-syndromic XLID); genomic sequence analysis panel, must include sequencing of at least 60 genes, including ARX, ATRX, CDKL5, FGD1, FMR1, HUWE1, IL1RAPL, KDM5C, L1CAM, MECP2, MED12, MID1, OCRL, RPS6KA3, and SLC16A2 |
| 81471 | X-linked intellectual disability (XLID) (e.g., syndromic and non-syndromic XLID); duplication/deletion gene analysis, must include analysis of at least 60 genes, including ARX, ATRX, CDKL5, FGD1, FMR1, HUWE1, IL1RAPL, KDM5C, L1CAM, MECP2, MED12, MID1, OCRL, RPS6KA3, and SLC16A2 |
| 89290 | Biopsy, oocyte polar body or embryo blastomere, microtechnique (for pre-implantation genetic diagnosis); less than or equal to 5 embryos |
| 89291 | Biopsy, oocyte polar body or embryo blastomere, microtechnique (for pre-implantation genetic diagnosis); greater than 5 embryos |

| HCPCS Code | Description |
|---|---|
| S3845 | Genetic testing for alpha-thalassemia |
| S3846 | Genetic testing for hemoglobin E beta-thalassemia |
| S3850 | Genetic testing for sickle cell anemia |

## *V. Policy History*

**Policy Number:** MPP-0023-120301

**Current Effective Date:** 03/01/2019

**Original Document Effective Date:** 03/01/2012

**Previous Revision Dates:** 09/01/2016, 02/12/2018

**PAC Approved Date:** 03/01/2012

**Previous Policy Title:** Genetic Testing – Preimplantation Genetic Diagnosis