Helix Expanded Carrier Screen

Item	Description
Test Name	Helix Expanded Carrier Screen
Test Type	Carrier Screening
Catalog Number	ECRF1
Procedure Code	H000325-7
Test Description	This panel evaluates 115 genes (females) and 99 genes (males) associated with a broad spectrum of autosomal recessive and X-linked inherited disorders where carrier frequency is ≥ 1 / 200. X-linked disorders are not included when the test is ordered for biological males.
Genes Tested	*ABCA3, ABCC8, ABCD1, ACADM, ACADVL, ACAT1, AGA, AGXT, AHI1, AIRE, ALDOB, ALPL, ANO10, ARSA, ARX, ASL, ASPA, ATP7B, BBS1, BBS2, BCKDHB, BLM, BTD, CBS, CC2D2A, CCDC88C, CEP290, CFTR, CHRNE, CLCN1, CLRN1, CNGB3, COL4A5, COL7A1, CPT2, CRYL1, CYP11A1, CYP21A2, CYP27A1, CYP27B1, DHCR7, DHDDS, DLD, DMD, DYNC2H1, ELP1, ERCC2, EVC2, F9, FAH, FANCC, FKRP, FKTN, FMO3, FMR1, G6PC1, GAA, GALC, GALT, GBA, GBE1, GJB2, GJB6, GLA, GNPTAB, GRIP1, HBA1, HBA2, HBB, HEXA, HOGA1, HPS1, HPS3, IDS, IDUA, L1CAM, LRP2, MCCC2, MCOLN1, MCPH1, MID1, MLC1, MMACHC, MMUT, MTM1, MVK, NAGA, NEB, NPHS1, NR0B1, OCA2, OTC, PAH, PCDH15, PKHD1, PLP1, PMM2, POLG, PRF1, RARS2, RNASEH2B, RS1, SCO2, SLC19A3, SLC22A5, SLC26A2, SLC26A4, SLC37A4, SLC6A8, SMN1, SMPD1, TF, TMEM216, USH2A, XPC*
Genetics Information	This test utilizes next-generation sequencing to detect single nucleotide variants, insertions and deletions up to 20 bp, and copy number variants in genes associated with autosomal recessive and X-linked inherited disorders.
Indications For Testing	Prenatal or preconception risk assessment for individuals who are pregnant or planning to conceive.
Clinical Descriptions	The genes on this panel are well understood in the literature, have clearly defined strong gene-disease associations with inherited conditions that present as profound, severe, or moderate as classified by ACMG, and have widely available prenatal diagnosis.
Interpretation	All detected variants are evaluated according to American College of Medical Genetics and Genomics recommendations. Variants are classified based on known, predicted, or possible pathogenicity and reported with interpretive comments detailing their potential or known significance.
Reclassification Of Variants	Helix reviews variant classifications annually when they arise in routine processes and upon request from providers. The timing of re-review depends on clinical risk. Providers can request a variant re-review by contacting Helix Customer Support. If a classification by Helix is updated, Helix identifies affected past patients and issues revised reports. Updated results are communicated to providers prior to results being uploaded to the EHR, and patients are notified through the EHR patient portal.

Item	Description
Variant Evaluation	Variant classification is performed using the guidelines set forth by the American College of Medical Genetics and Genomics and the Association for Molecular Pathology, with modifications as suggested by domain specific Expert Panels of the Clinical genome Resource (ClinGen) when available. Variant pathogenicity is categorized as benign, likely benign, variant of uncertain significance (VUS), likely pathogenic, or pathogenic. Distinct from this 5-tier system, FMR1 trinucleotide repeat expansions are classified by repeat size in accordance with ACMG technical standards as Normal, Intermediate, Premutation, or Full Mutation. Alleles are categorized as Normal (< 45 repeats), Intermediate (45-54 repeats), Premutation (55-200 repeats), or Full Mutation (> 200 repeats), consistent with current ACMG technical standards for Fragile X syndrome.
Turnaround Time - Standard	Typically 7 to 21 days
Turnaround Time - Requery (SOQO®)	Typically 7 to 21 days
Available In NY State	Yes
Test Classification	This test was developed, and its performance characteristics determined, by Fulgent Therapeutics LLC in a manner consistent with CLIA requirements. This test has not been cleared or approved by the US Food and Drug Administration.
Performing Laboratory Information	Fulgent Therapeutics LLC CLIA Laboratory Number: 05D2043189 Laboratory Hours of Operation: Monday-Friday (7AM-5PM PST) Saturday (8AM-3PM PST) Address: 4399 Santa Anita Ave., El Monte, CA 91731 Healthcare professionals may contact Helix directly to discuss results: Helix Customer Service: (844) 211-2070 Email: clinicalsupport@helix.com
Regulatory Information	CLIA Complexity: High Test Classification: Non-Waived/ Laboratory Developed Test
CLIA Category	Chemistry / Routine Chemistry

Conditions for Helix Expanded Carrier Screen

- *(ABCA3)* Surfactant metabolism dysfunction, pulmonary 3
- *(ABCC8)* Familial hyperinsulinism
- *(ABCD1)* Adrenoleukodystrophy, X-linked
- *(ACADM)* Medium-chain acyl-CoA dehydrogenase (MCAD) deficiency
- *(ACADVL)* Very long-chain acyl-CoA dehydrogenase (VLCAD) deficiency
- *(ACAT1)* 3-ketothiolase deficiency
- *(AGA)* Aspartylglucosaminuria
- *(AGXT)* Primary hyperoxaluria type 1
- *(AHI1)* Joubert syndrome, AHI1-related
- *(AIRE)* Autoimmune polyendocrinopathy syndrome type I
- *(ALDOB)* Hereditary fructose intolerance
- *(ALPL)* Hypophosphatasia
- *(ANO10)* Spinocerebellar ataxia 10
- *(ARSA)* Metachromatic leukodystrophy
- *(ARX)* X-linked intellectual disability, ARX-related
- *(ASL)* Argininosuccinate lyase deficiency
- *(ASPA)* Canavan disease
- *(ATP7B)* Wilson disease
- *(BBS1)* Bardet-Biedl syndrome type 1
- *(BBS2)* BBS2-related ciliopathies
- *(BCKDHB)* Maple syrup urine disease type Ib
- *(BLM)* Bloom syndrome
- *(BTD)* Biotinidase deficiency
- *(CBS)* Homocystinuria due to cystathionine beta-synthase deficiency
- *(CC2D2A)* Joubert syndrome 9
- *(CCDC88C)* Congenital hydrocephalus 1
- *(CEP290)* CEP290-related Ciliopathies
- *(CFTR)* Cystic Fibrosis
- *(CHRNE)* Congenital myasthenic syndrome
- *(CLCN1)* Autosomal recessive congenital myotonia
- *(CLRN1)* Usher syndrome, type 3A
- *(CNGB3)* Achromatopsia
- *(COL4A5)* Alport syndrome, COL4A5-related
- *(COL7A1)* Dystrophic epidermolysis bullosa
- *(CPT2)* Carnitine palmitoyltransferase II deficiency
- *(CYP11A1)* Congenital adrenal insufficiency
- *(CYP21A2)* Congenital adrenal hyperplasia due to 21-hydroxylase deficiency
- *(CYP27A1)* Cerebrotendinous xanthomatosis
- *(CYP27B1)* Vitamin D-dependent rickets, type 1
- *(DHCR7)* Smith-Lemli-Opitz syndrome
- *(DHDDS)* Retinitis pigmentosa 59
- *(DLD)* Dihydrolipoamide dehydrogenase deficiency
- *(DMD)* Dystrophinopathies
- *(DYNC2H1)* Short-rib thoracic dysplasia 3 with or without polydactyly
- *(ELP1)* Familial Dysautonomia
- *(ERCC2)* ERCC2-related disorders
- *(EVC2)* EVC2-related bone growth disorders
- *(F9)* Hemophilia B
- *(FAH)* Tyrosinemia, type 1
- *(FANCC)* Fanconi anemia group C
- *(FKRP)* FKRP Alpha-dystroglycanopathies-
- *(FKTN)* FKTN Alpha-dystroglycanopathies
- *(FMO3)* Trimethylaminuria
- *(FMR1)* Fragile X Syndrome
- *(G6PC)* Glycogen storage disease, type Ia
- *(GAA)* Pompe disease
- *(GALC)* Krabbe disease
- *(GALT)* Galactosemia
- *(GBA)* Gaucher disease
- *(GBE1)* Glycogen storage disease IV
- *(GJB2, GJB6, CRYL1)* Nonsyndromic hearing loss 1A
- *(GLA)* Fabry disease
- *(GNPTAB)* Mucolipidosis II & III
- *(GRIP1)* Fraser syndrome
- *(HBA1)* Alpha thalassemia
- *(HBA2)* Alpha thalassemia
- *(HBB)* Sickle cell disease, Hemoglobin C disease, Beta thalassemia
- *(HEXA)* Tay-Sachs disease
- *(HOGA1)* Primary hyperoxaluria type III
- *(HPS1)* Hermansky-Pudlak syndrome 1
- *(HPS3)* Hermansky-Pudlak syndrome 3

- *(IDS)* Mucopolysaccharidosis type II (Hunter syndrome)
- *(IDUA)* Mucopolysaccharidosis, type I (Hurler syndrome)
- *(L1CAM)* L1 syndrome
- *(LRP2)* Donnai-Barrow syndrome
- *(MCCC2)* 3-Methylcrotonyl-CoA carboxylase 2 deficiency (3-MCC deficiency)
- *(MCOLN1)* Mucolipidosis IV
- *(MCPH1)* Primary microcephaly 1, recessive
- *(MID1)* Opitz GBBB syndrome, type I
- *(MLC1)* Megalencephalic leukoencephalopathy with subcortical cysts
- *(MMACHC)* Methylmalonic aciduria and homocystinuria, cblC type
- *(MTM1)* Myotubular myopathy, X-linked
- *(MUT)* Methylmalonic aciduria-methylmalonyl-CoA mutase deficiency
- *(MVK)* Mevalonate kinase deficiency
- *(NAGA)* Schindler disease types 1 and 3
- *(NEB)* Nemaline myopathy
- *(NPHS1)* Congenital nephrotic syndrome, type 1
- *(NR0B1)* Congenital adrenal hypoplasia, X-linked
- *(OCA2)* Oculocutaneous albinism type II
- *(OTC)* Ornithine transcarbamylase deficiency
- *(PAH)* Phenylalanine Hydroxylase deficiency (Phenylketonuria)
- *(PCDH15)* PCDH15-related sensory loss
- *(PKHD1)* Polycystic kidney disease, PKHD1-related
- *(PLP1)* PLP1-related disorders
- *(PMM2)* Congenital disorder of glycosylation type Ia
- *(POLG)* POLG-related disorders
- *(PRF1)* Hemophagocytic lymphohistiocytosis familial, 2
- *(RARS2)* Pontocerebellar hypoplasia type 6
- *(RNASEH2B)* Aicardi Goutieres syndrome 2
- *(RS1)* Juvenile retinoschisis, X-linked
- *(SCO2)* Mitochondrial complex IV deficiency
- *(SLC19A3)* Biotin-responsive basal ganglia disease
- *(SLC22A5)* Systemic primary carnitine deficiency
- *(SLC26A2)* SLC26A2-related disorders
- *(SLC26A4)* Pendred syndrome
- *(SLC37A4)* Glycogen storage disease, type Ib
- *(SLC6A8)* Creatine deficiency syndrome
- *(SMN1)* Spinal muscular atrophy
- *(SMPD1)* Niemann-Pick disease, type A/B
- *(TF)* Atransferrinemia
- *(TMEM216)* TMEM216-related ciliopathies
- *(USH2A)* Usher syndrome, type 2A
- *(XPC)* Xeroderma pigmentosum, group C

Genomic DNA was isolated from the submitted specimen indicated above (if cellular material was submitted). DNA was barcoded, and enriched for the coding exons of targeted genes using hybrid capture technology. Prepared DNA libraries were then sequenced using a Next Generation Sequencing technology. Following alignment to the human genome reference sequence (assembly GRCh37), variants were detected in regions of at least 10x coverage. For this specimen, 99.66% and 99.61% of coding regions and splicing junctions of genes listed had been sequenced with coverage of at least 10x and 20x, respectively, by NGS or by Sanger sequencing. The remaining regions did not have 10x coverage, and were not evaluated. Variants were interpreted manually using locus specific databases, literature searches, and other molecular biological principles. To minimize false positive results, any variants that do not meet internal quality standards are confirmed by Sanger sequencing. Variants classified as pathogenic, likely pathogenic, or risk allele which are located in the coding regions and nearby intronic regions (+/- 20bp) of the genes listed above are reported. Variants outside these intervals may be reported but are typically not guaranteed. When a single pathogenic or likely pathogenic variant is identified in a clinically relevant gene with autosomal recessive inheritance, the laboratory will attempt to ensure 100% coverage of coding sequences either through NGS or Sanger sequencing technologies ("fill-in"). All genes listed were evaluated for large deletions and/or duplications. However, single exon deletions or duplications will not be detected in this assay, nor will copy number alterations in regions of genes with significant pseudogenes. Putative deletions or duplications are analyzed using Fulgent Germline proprietary pipeline for this specimen. Bioinformatics: The Fulgent Germline v2019.2 pipeline was used to analyze this specimen.

These test results and variant interpretation are based on the proper identification of the submitted specimen, accuracy of any stated familial relationships, and use of the correct human reference sequences at the queried loci. In very rare instances, errors may result due to mix-up or co-mingling of specimens. Positive results do not imply that there are no other contributors, genetic or otherwise, to future pregnancies, and negative results do not rule out the genetic risk to a pregnancy. Official gene names change over time. Fulgent uses the most up to date gene names based on HUGO Gene Nomenclature Committee (https://www.genenames.org) recommendations. If the gene name on report does not match that of ordered gene, please contact the laboratory and details can be provided. Result interpretation is based on the available clinical and family history information for this individual, collected published information, and Alamut annotation available at the time of reporting. This assay is not designed or validated for the detection of low-level mosaicism or somatic mutations. This assay will not detect certain types of genomic aberrations such as translocations, inversions, or repeat expansions other than specified genes. DNA alterations in regulatory regions or deep intronic regions (greater than 20bp from an exon) may not be detected by this test. Unless otherwise indicated, no additional assays have been performed to evaluate genetic changes in this specimen. There are technical limitations on the ability of DNA sequencing to detect small insertions and deletions. Our laboratory uses a sensitive detection algorithm, however these types of alterations are not detected as reliably as single nucleotide variants. Rarely, due to systematic chemical, computational, or human error, DNA variants may be missed. Although next generation sequencing technologies and our bioinformatics analysis significantly reduce the confounding contribution of pseudogene sequences or other highly-homologous sequences, sometimes these may still interfere with the technical ability of the assay to identify pathogenic alterations in both sequencing and deletion/duplication analyses. Deletion/duplication analysis can identify alterations of genomic regions which include one whole gene (buccal swab specimens and whole blood specimens) and are two or more contiguous exons in size (whole blood specimens only); single exon deletions or duplications may occasionally be identified, but are not routinely detected by this test. When novel DNA duplications are identified, it is not possible to discern the genomic location or orientation of the duplicated segment, hence the effect of the duplication cannot be predicted. Where deletions are detected, it is not

always possible to determine whether the predicted product will remain in-frame or not. Unless otherwise indicated, deletion/duplication analysis has not been performed in regions that have been sequenced by Sanger.

Gene Specific Notes:
BTD: If detected, the variant NM_001370658.1:c.1270G>C (p.Asp424His) will not be reported as this variant is associated with low disease penetrance and is primarily associated with reduced enzyme activity when homozygous. CFTR: Analysis of the intron 8 polymorphic region (e.g. IVS8-5T allele) is only performed if the p.Arg117His (R117H) mutation is detected. Single exon deletion/duplication analysis is limited to deletions of previously reported exons: 1, 2, 3, 11, 19, 20, 21. CFTR variants primarily associated with CFTR-related isolated congenital bilateral absence of the vas deferens and CFTR-related pancreatitis are not included in this analysis. CFTR variants with insufficient evidence of being cystic fibrosis mutations will not be reported either. CRYL1: This gene is only included for whole gene deletions related to GJB2-related hearing loss. CYP21A2: Significant pseudogene interference and/or reciprocal exchanges between the CYP21A2 gene and its pseudogene, CYP21A1P, have been known to occur and may impact results. As such, the relevance of variants reported in this gene must be interpreted clinically in the context of the clinical findings, biochemical profile, and family history of each patient. CYP21A2 variants primarily associated with non-classic congenital adrenal hyperplasia (CAH) are not included in this analysis (PubMed: 23359698). The variants associated with non-classic disease, including but not limited to c.188A>T (p.His63Leu), c.844G>T (p.Val282Leu), c.1174G>A (p.Ala392Thr), and c.1360C>T (p.Pro454Ser) will not be reported. LR-PCR is not routinely ordered for NM_000500.9:c.955C>T (p.Gln319Ter). Individuals with c.955C>T (p.Gln319Ter) will be reported as a Possible Carrier indicating that the precise nature of the variant has not been determined by LR-PCR and that the variant may occur in the CYP21A2 wild-type gene or in the CYP21A1P pseudogene. The confirmation test is recommended if the second reproductive partner is tested positive for variants associated with classic CAH. DMD: Single exon deletion/duplication analysis is limited to exons with >1 patient reported in the UMD database (http://www.umd.be/DMD/W_DMD/index.html), accessed Dec 29,2020 and all out-of-frame exons after exon 3. This includes deletion of exon 1, and duplication of exon 2, and del/dup for exons 3, 6~8, 11, 12, 17~22, 43~46, 48, 50~56, 58~63, 65~70, 75, 76, and 78. Single-exon detection is limited to blood samples. FMR1: The exact size of alleles >200 CGG repeats cannot be determined; these alleles are pathogenic for X-Linked Fragile X Syndrome. Alleles with <10 repeats may fail to amplify; these alleles are benign. The repeat length for this gene may vary by +/- 1 repeat unit. Methylation is not analyzed. RP-PCR analysis of the FMR1 promoter is not routinely performed in males. Small degrees of size mosaicism, including gonadal mosaicism, may not be detected. GALT: The D2 "Duarte" allele is not included in this analysis. While this allele can cause positive newborn screening results, it is not known to cause clinical symptoms in any state (PubMed: 25473725, 30593450). GBA: The current testing method may not be able to reliably detect certain pathogenic variants in the GBA gene due to homologous recombination between the pseudogene and the functional gene. GJB6: This gene is only included for whole gene deletions related to GJB2-related hearing loss. SMN1: The current testing method detects sequencing variants in exon 7 and copy number variations in exons 7-8 of the SMN1 gene (NM_022874.2). Sequencing and deletion/duplication analysis are not performed on any other region in this gene. About 5%-8% of the population have two copies of SMN1 on a single chromosome and a deletion on the other chromosome, known as a [2+0] configuration (PubMed: 20301526). The current testing method cannot directly detect carriers with a [2+0] SMN1 configuration, but can detect linkage between the silent carrier allele and certain population-specific single nucleotide changes. As a result, a negative result for carrier testing greatly reduces but does not eliminate the chance that a person is a carrier. Only abnormal results will be reported.

Disclaimer:
This test was developed, performed, and its performance characteristics determined by Fulgent Therapeutics LLC (CAP# 8042697, CLIA# 05D2043189), 4399 Santa Anita Ave., El Monte, CA 91731. It has not been cleared or approved by the FDA. The laboratory is regulated under CLIA as qualified to perform high-complexity testing. This test is used for clinical purposes. It should not be regarded as investigational or for research. Since genetic variation, as well as systematic and technical factors, can affect the accuracy of testing, the results of testing should always be interpreted in the context of clinical and familial data. For assistance with interpretation of these results, healthcare professionals may contact us directly at 844-211-2070 or by email at clinicalsupport@helix.com. It is recommended that patients receive appropriate genetic counseling to explain the implications of the test result, including its residual risks, uncertainties and reproductive or medical options.

Targeted Genes & Methodology for Helix Expanded Carrier Screen

The following applies to the Helix Expanded Carrier Screen. Testing is performed to evaluate for the presence of variants in coding regions and extending to +/- 20 base pairs of adjacent intronic sequences on either side of the coding exons of the genes analyzed. In addition, the analysis will cover select non-coding variants, as listed below. Next-generation sequencing is performed to test for the presence of small variants and copy number variants in the genes analyzed. Confirmation of select reportable variants may be performed by alternate methodologies based on internal laboratory criteria.

This list is current from December 2025 to the present. This document is intended to highlight additional evaluations for variants of high clinical interest as well as technical limitations. For questions regarding genes, reference transcripts, or specific regions covered, contact Helix Customer Service at (844) 211-2070.

Genomic Build: GRCh37
Catalog Number: ECRF1

Gene	Transcript	Technical Limitations
ABCA3	NM_001089.3	
ABCC8	NM_000352.6	
ABCD1	NM_000033.4	
ACADM	NM_000016.6	
ACADVL	NM_000018.4	
ACAT1	NM_000019.4	
AGA	NM_000027.4	
AGXT	NM_000030.3	
AHI1	NM_017651.4	
AIRE	NM_000383.4	
ALDOB	NM_000035.4	
ALPL	NM_000478.6	
ANO10	NM_018075.5	
ARSA	NM_000487.6	
ARX	NM_139058.3	
ASL	NM_000048.4	
ASPA	NM_000049.4	
ATP7B	NM_000053.4	
BBS1	NM_024649.5	
BBS2	NM_031885.5	
BCKDHB	NM_183050.4	
BLM	NM_000057.4	
BTD	NM_001370658.1	The variant NM_001370658.1:c.1270G>C (p.Asp424His) is not reported.
CBS	NM_000071.3	

Gene	Transcript	Technical Limitations
CC2D2A	NM_001080522.2	
CCDC88C	NM_001080414.4	
CEP290	NM_025114.4	
CFTR	NM_000492.4	Analysis of the intron 8 polymorphic region (e.g. IVS8-5T allele) is only performed if the p.Arg117His (R117H) mutation is detected. Single exon deletion/duplication analysis is limited to deletions of previously reported exons: 1, 2, 3, 11, 19, 20, 21. CFTR variants primarily associated with CFTR-related isolated congenital bilateral absence of the vas deferens and CFTR-related pancreatitis are not included in this analysis. CFTR variants with insufficient evidence of being cystic fibrosis mutations will not be reported either.
CHRNE	NM_000080.4	
CLCN1	NM_000083.3	
CLRN1	NM_174878.3	
CNGB3	NM_019098.5	
COL4A5	NM_000495.5; NM_033380.3	
COL7A1	NM_000094.4	
CPT2	NM_000098.3	
CRYL1	NM_015974.3	Only whole gene deletions related to GJB2-related hearing loss are reported.
CYP11A1	NM_000781.3	
CYP21A2	NM_000500.9	Significant pseudogene interference and/or reciprocal exchanges between the CYP21A2 gene and its pseudogene, CYP21A1P, have been known to occur and may impact results. As such, the relevance of variants reported in this gene must be interpreted clinically in the context of the clinical findings, biochemical profile, and family history of each patient. CYP21A2 variants primarily associated with non-classic congenital adrenal hyperplasia (CAH) are not included in this analysis (PubMed: 23359698). The variants associated with non-classic disease, including but not limited to c.188A>T (p.His63Leu), c.844G>T (p.Val282Leu), c.1174G>A (p.Ala392Thr), and c.1360C>T (p.Pro454Ser) will not be reported. LR-PCR is not routinely ordered for NM_000500.9:c.955C>T (p.Gln319Ter). Individuals with c.955C>T (p.Gln319Ter) will be reported as a Possible Carrier indicating that the precise nature of the variant has not been determined by LR-PCR and that the variant may occur in the CYP21A2 wild-type gene or in the CYP21A1P pseudogene. The confirmation test is recommended if the second reproductive partner is tested positive for variants associated with classic CAH.
CYP27A1	NM_000784.4	
CYP27B1	NM_000785.4	
DHCR7	NM_001360.3	
DHDDS	NM_024887.3	

Gene	Transcript	Technical Limitations
DLD	NM_000108.5	
DMD	NM_004006.2	Single exon deletion/duplication analysis is limited to exons with > 1 patient reported in the UMD database (http://www.umd.be/DMD/W_DMD/index.html), accessed Dec 29,2020 and all out-of-frame exons after exon 3. This includes deletion of exon 1, and duplication of exon 2, and del/dup for exons 3, 6~8, 11, 12, 17~22, 43~46, 48, 50~56, 58~63, 65~70, 75, 76, and 78. Single-exon detection is limited to blood samples.
DYNC2H1	NM_001080463.2	
ELP1	NM_003640.5	
ERCC2	NM_000400.4	
EVC2	NM_147127.5	
F9	NM_000133.4	
FAH	NM_000137.4	
FANCC	NM_000136.3	
FKRP	NM_024301.5	
FKTN	NM_001079802.2	
FMO3	NM_006894.6	
FMR1	NM_002024.6	The exact size of alleles >200 CGG repeats cannot be determined; these alleles are pathogenic for X-Linked Fragile X Syndrome. Alleles with <10 repeats may fail to amplify; these alleles are benign. The repeat length for this gene may vary by +/- 1 repeat unit. Methylation is not analyzed. RP-PCR analysis of the FMR1 promoter is not routinely performed in males. Small degrees of size mosaicism, including gonadal mosaicism, may not be detected.
GAA	NM_000152.5	
GALC	NM_000153.4	
GALT	NM_000155.4	The D2 "Duarte" allele is not included in this analysis.
GBA	NM_001005741.3	The current testing method may not be able to reliably detect certain pathogenic variants in the GBA gene due to homologous recombination between the pseudogene and the functional gene.
GBE1	NM_000158.4	
GJB2	NM_004004.6	
GJB6	NM_001110219.3	Only whole gene deletions related to GJB2-related hearing loss reported.
GLA	NM_000169.2	
GNPTAB	NM_024312.5	
GRIP1	NM_021150.4	
HBA1	NM_000558.5	
HBA2	NM_000517.6	

Targeted Genes & Methodology for Helix Expanded Carrier Screen

Gene	Transcript	Technical Limitations
HBB	NM_000518.5	
HEXA	NM_000520.6	
HOGA1	NM_138413.4	
HPS1	NM_000195.5	
HPS3	NM_032383.5	
IDS	NM_000202.8	
IDUA	NM_000203.5	
L1CAM	NM_000425.5	
LRP2	NM_004525.3	
MCCC2	NM_022132.5	
MCOLN1	NM_020533.3	
MCPH1	NM_024596.5	
MID1	NM_000381.4	
MLC1	NM_015166.4	
MMACHC	NM_015506.3	
MTM1	NM_000252.3	
MVK	NM_000431.4	
NAGA	NM_000262.3	
NEB	NM_001271208.2	
NPHS1	NM_004646.4	
NR0B1	NM_000475.5	
OCA2	NM_000275.3	
OTC	NM_000531.6	
PAH	NM_000277.3	
PCDH15	NM_033056.3	
PKHD1	NM_138694.4	
PLP1	NM_000533.5	
PMM2	NM_000303.3	
POLG	NM_002693.3	
PRF1	NM_001083116.3	
RARS2	NM_020320.5	
RNASEH2B	NM_024570.4	
RS1	NM_000330.4	

Gene	Transcript	Technical Limitations
SCO2	NM_005138.3	
SLC19A3	NM_025243.4	
SLC22A5	NM_003060.4	
SLC26A2	NM_000112.4	
SLC26A4	NM_000441.2	
SLC37A4	NM_001164277.1	
SLC6A8	NM_005629.4	
SMN1	NM_000344.3	The current testing method detects sequencing variants in exon 7 and copy number variations in exons 7-8 of the SMN1 gene (NM_022874.2). Sequencing and deletion/duplication analysis are not performed on any other region in this gene. About 5%-8% of the population have two copies of SMN1 on a single chromosome and a deletion on the other chromosome, known as a [2+0] configuration (PubMed: 20301526). The current testing method cannot directly detect carriers with a [2+0] SMN1 configuration, but can detect linkage between the silent carrier allele and certain population-specific single nucleotide changes. As a result, a negative result for carrier testing greatly reduces but does not eliminate the chance that a person is a carrier. Only abnormal results will be reported.
SMPD1	NM_000543.5	
TF	NM_001063.4	
TMEM216	NM_001173990.3	
USH2A	NM_206933.4	
XPC	NM_004628.5	