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Patient Information
Patient Last, Patient First
DOB: Jan 01, 1900
Sex: ?
MR#: 000-000-000
FD Patient#: FT-PT21496

Partner Information
Not Tested

Physician:
Physician Last, Physician First

Phone:
Fax:

Laboratory:
Report Date:
{{DATESIGNED}}

Accession:
FT-1612508
Test#: FT-TS51816
Specimen Type: TBD
Collection Date:

Accession:
N/A

DRAFT RESULTS

No carrier mutations identified.

TEST PERFORMED

ASPiRA GenetiX Female Carrier Screening 327
(327 Gene Panel; full gene sequencing with deletion and duplication analysis)

INTERPRETATION:

Notes and Recommendations:

- Testing for a trinucleotide (CGG) repeat sequence in the FMR1 gene was performed to screen for your carrier status for Fragile X Syndrome. The repeat sizes detected were: 19 and 28 repeats. These results are within the normal range. Therefore, you are not considered a carrier for Fragile X Syndrome.
- Testing for copy number changes in the SMN1 gene was performed to screen for your carrier status for Spinal Muscular Atrophy. Two copies of the SMN1 gene were detected. These results are within the normal range for non-carriers. See Limitations section for more information.
- No carrier mutations were identified in the submitted specimen. A negative result does not rule out the possibility of a genetic predisposition nor does it rule out any pathogenic mutations in areas not assessed by this test or in regions that were covered at a level too low to reliably assess. Also, it does not rule out mutations that are of the sort not queried by this test; see Methods and Limitations for more information. Genetic counseling is recommended.
- This carrier screening test does not screen for all possible genetics conditions, nor for all possible mutations in every gene tested. Individuals with negative test results may still have up to a 3-4% risk to have a child with a birth defect due to genetic and/or environmental factors.

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GENES TESTED:

ASPiRA GenetiX Female Carrier Screening 327

327 genes tested (99.32% of coding bases at >20x). For more gene specific information and assistance with residual risk calculation, see SUPPLEMENTAL TABLE.

ABCB11	*ABCC8*	*ABCD1*	*ACAD9*	*ACADM*	*ACADS*
ACADVL	*ACAT1*	*ACOX1*	*ACSF3*	*ADA*	*ADAMTS2*
ADGRG1	*AGA*	*AGL*	*AGPS*	*AGXT*	*AIRE*
ALDH3A2	*ALDOB*	*ALG6*	*ALMS1*	*ALPL*	*AMT*
AQP2	*ARG1*	*ARSA*	*ARSB*	*ARSE*	*ASL*
ASNS	*ASPA*	*ASS1*	*ATM*	*ATP6V1B1*	*ATP7A*
ATP7B	*ATRX*	*BBS1*	*BBS10*	*BBS12*	*BBS2*
BCKDHA	*BCKDHB*	*BCS1L*	*BLM*	*BSND*	*BTD*
CAPN3	*CBS*	*CCDC103*	*CCDC151*	*CCDC39*	*CD40LG*
CDH23	*CEP290*	*CERKL*	*CFTR*	*CHM*	*CHRNE*
CHRNG	*CIITA*	*CLN3*	*CLN5*	*CLN6*	*CLN8*
CLRN1	*CNGB3*	*COL27A1*	*COL4A3*	*COL4A4*	*COL4A5*
COL7A1	*CPS1*	*CPT1A*	*CPT2*	*CRB1*	*CTNS*
CTSK	*CYBA*	*CYBB*	*CYP11B1*	*CYP11B2*	*CYP17A1*
CYP19A1	*CYP1B1*	*CYP21A2*	*CYP27A1*	*DBT*	*DCLRE1C*
DCX	*DHCR7*	*DHDDS*	*DLD*	*DMD*	*DNAH5*
DNAI1	*DNAI2*	*DNAL1*	*DPYD*	*DYSF*	*EDA*
EIF2AK3	*EIF2B5*	*EMD*	*ERCC6*	*ERCC8*	*ESCO2*
ETFA	*ETFB*	*ETFDH*	*ETHE1*	*EVC*	*EVC2*
EXOSC3	*EYS*	*F11*	*F8*	*F9*	*FAH*
FAM161A	*FANCA*	*FANCC*	*FANCG*	*FH*	*FKRP*
FKTN	*FMR1*	*G6PC*	*GAA*	*GALC*	*GALK1*
GALNS	*GALT*	*GAMT*	*GBA*	*GBE1*	*GCDH*
GFM1	*GJB1*	*GJB2*	*GJB6*	*GLA*	*GLB1*
GLDC	*GLE1*	*GNE*	*GNPTAB*	*GNPTG*	*GNS*
GP1BA	*GP9*	*GRHPR*	*GUSB*	*HADHA*	*HAX1*
HBA1	*HBA2*	*HBB*	*HEXA*	*HEXB*	*HFE2*
HGD	*HGSNAT*	*HLCS*	*HMGCL*	*HOGA1*	*HPS1*
HPS3	*HSD17B4*	*HSD3B2*	*HYAL1*	*HYLS1*	*IDS*
IDUA	*IKBKAP*	*IL2RG*	*IVD*	*KCNJ11*	*L1CAM*
LAMA2	*LAMA3*	*LAMB3*	*LAMC2*	*LCA5*	*LHX3*
LIFR	*LIPA*	*LOXHD1*	*LRPPRC*	*LYST*	*MAN2B1*
MCCC1	*MCCC2*	*MCOLN1*	*MED17*	*MEFV*	*MESP2*
MFSD8	*MKS1*	*MLC1*	*MMAA*	*MMAB*	*MMACHC*
MMADHC	*MPI*	*MPL*	*MPV17*	*MTM1*	*MTRR*
MTTP	*MUT*	*MYO7A*	*NAGLU*	*NAGS*	*NBN*
NDRG1	*NDUFAF5*	*NDUFS6*	*NEB*	*NPC1*	*NPC2*
NPHP1	*NPHS1*	*NPHS2*	*NR0B1*	*NR2E3*	*NTRK1*
OAT	*OCRL*	*OPA3*	*OTC*	*PAH*	*PC*
PCCA	*PCCB*	*PCDH15*	*PDHA1*	*PDHB*	*PEX1*
PEX10	*PEX12*	*PEX2*	*PEX6*	*PEX7*	*PFKM*

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PHGDH	*PKHD1*	*PLA2G6*	*PMM2*	*POLG*	*POMGNT1*
PPT1	*PROP1*	*PRPS1*	*PSAP*	*PTS*	*PUS1*
PYGM	*RAB23*	*RAG1*	*RAG2*	*RAPSN*	*RARS2*
RDH12	*RMRP*	*RPE65*	*RPGRIP1L*	*RS1*	*RTEL1*
SACS	*SAMHD1*	*SEPSECS*	*SGCA*	*SGCB*	*SGCD*
SGCG	*SGSH*	*SLC12A3*	*SLC12A6*	*SLC17A5*	*SLC22A5*
SLC25A13	*SLC25A15*	*SLC25A20*	*SLC26A2*	*SLC26A3*	*SLC26A4*
SLC35A3	*SLC37A4*	*SLC39A4*	*SLC4A11*	*SLC6A8*	*SLC7A7*
SMARCAL1	*SMN1*	*SMPD1*	*STAR*	*SUMF1*	*TAT*
TCIRG1	*TECPR2*	*TFR2*	*TGM1*	*TH*	*TMEM216*
TPP1	*TRIM32*	*TRMU*	*TSFM*	*TTC37*	*TTPA*
TYMP	*UGT1A1*	*USH1C*	*USH2A*	*VPS13A*	*VPS13B*
VPS45	*VRK1*	*VSX2*	*WAS*	*WNT10A*	*XPA*
XPC	*ZFYVE26*	*CRYL1*			

METHODS:

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, variants were detected in regions of at least 10x coverage. For this specimen, 99.37% and 99.32% of coding regions and splicing junctions of genes listed had been sequenced with coverage of at least 10x and 20x respectively 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. All the variants with quality score less than 500 (roughly 40x of coverage for a heterozygous variant) will be confirmed by Sanger sequencing. Only variants classified as pathogenic, likely-pathogenic are reported. 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 (see Gene Specific Limitations below). Putative deletions or duplications identified are confirmed by an orthogonal method (qPCR or MLPA). If included in the panel, FMR1 repeat analysis is performed by repeat-primed PCR (rpPCR) and amplicon length analysis. Methylation studies are not performed. Variants are classified using the ACMG Guidelines for Sequence Variant Interpretation (PubMed: 27993330) unless otherwise specified.

LIMITATIONS:

General Limitations

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. 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

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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 are two or more contiguous exons in size; 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 Limitations

Spinal Muscular Atrophy: SMN1
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. The 3-copy SMN1 state can be detected by this test and will be reported out if present.

Hemogolobin disorders: HBA1 and HBA2
The phase of heterozygous alterations in the HBA1 and HBA2 genes cannot be determined, but can be confirmed through parental testing.

Gaucher Disease: 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.

SIGNATURE:

{{SIGNATURE}}

DISCLAIMER:

This test was developed and its performance characteristics determined by Fulgent Genetics: 4978 Santa Anita Ave., Suite 205, Temple Cit, CA 91780 CAP#: 8042697 CLIA#: 05D204318. 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 844.277.4721 or aspirasupport@vermillion.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.

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Supplemental Table

Gene	Mode	Condition	Ethnicity	Carrier Frequency	Detection Rate	Post-test Carrier Probability*	Residual Risk to have an affected child*
ABCB11	AR	Progressive Familial Intrahepatic Cholestasis	General Population	1 in 112	98%	1 in 5,551	1 in 22,204
ABCC8	AR	Familial hyperinsulinism, ABCC8-related	General Population	1 in 112	98%	1 in 5,551	1 in 22,204
			Ashkenazi Jewish Population	1 in 44	98%	1 in 2,151	1 in 8,604
			Finnish Population	1 in 25	98%	1 in 1,201	1 in 4,804
			Middle-Eastern Population	1 in 25	98%	1 in 1,201	1 in 4,804
ABCD1	XL	Adrenoleukodystrophy, X-linked	General Population	<1 in 500	99%	<1 in 49,901	<1 in 199,604
ACAD9	AR	Acyl-CoA dehydrogenase-9 (ACAD9) Deficiency	General Population	<1 in 500	98%	<1 in 24,951	<1 in 99,804
ACADM	AR	Medium-chain acyl-CoA dehydrogenase (MCAD) deficiency	General Population	1 in 69	98%	1 in 3,401	1 in 13,604
			Caucasian / European Population	1 in 52	99%	1 in 5,101	1 in 20,404
			East Asian Population	1 in 198	99%	1 in 19,701	1 in 78,804
			Native American Population	1 in 43	96%	1 in 1,051	1 in 4,204
ACADS	AR	Short-chain acyl-coA dehydrogenase (SCAD) Deficiency	General Population	1 in 85	99%	1 in 8,401	1 in 33,604
			African/African American Population	1 in 52	99%	1 in 5,101	1 in 20,404
			Caucasian / European Population	1 in 76	99%	1 in 7,501	1 in 30,004
			Middle-Eastern Population	1 in 52	99%	1 in 5,101	1 in 20,404
			South Asian/Indian Population	1 in 51	99%	1 in 5,001	1 in 20,004
ACADVL	AR	Very long-chain acyl-CoA dehydrogenase (VLCAD) deficiency	General Population	1 in 118	93%	1 in 1,672	1 in 6,690
			Middle-Eastern Population	1 in 74	93%	1 in 1,044	1 in 4,175
			Native American Population	1 in 61	93%	1 in 858	1 in 3,433
			South Asian/Indian Population	1 in 73	93%	1 in 1,030	1 in 4,118
ACAT1	AR	3-ketothiolase deficiency	General Population	<1 in 500	98%	<1 in 24,951	<1 in 99,804
ACOX1	AR	Peroxisomal acyl-CoA oxidase deficiency	General Population	<1 in 500	98%	<1 in 24,951	<1 in 99,804
ACSF3	AR	Combined malonic and methylmalonic aciduria	General Population	<1 in 500	98%	<1 in 24,951	<1 in 99,804
ADA	AR	Adenosine deaminase deficiency	General Population	1 in 224	93%	1 in 3,187	1 in 12,747
ADAMTS2	AR	Ehlers-Danlos syndrome, Dermatosparaxis type VIIC	General Population	<1 in 500	98%	<1 in 24,951	<1 in 99,804
			Ashkenazi Jewish Population	1 in 248	98%	1 in 12,351	1 in 49,404
ADGRG1	AR	Bilateral frontoparietal polymicrogyria	General Population	<1 in 500	98%	<1 in 24,951	<1 in 99,804
AGA	AR	Aspartylglucosaminuria	General Population	<1 in 500	98%	<1 in 24,951	<1 in 99,804
			Finnish Population	1 in 71	98%	1 in 3,501	1 in 14,004
AGL	AR	Glycogen storage disease type III	General Population	1 in 158	95%	1 in 3,141	1 in 12,564
			Faroese Population	1 in 28	95%	1 in 541	1 in 2,164
			Inuit Population	1 in 25	95%	1 in 481	1 in 1,924
			North African Jewish Population	1 in 37	95%	1 in 721	1 in 2,884
AGPS	AR	Rhizomelic chondrodysplasia punctata, type 3	General Population	<1 in 500	98%	<1 in 24,951	<1 in 99,804
AGXT	AR	Primary hyperoxaluria type 1	General Population	1 in 120	99%	1 in 11,901	1 in 47,604
			Caucasian / European Population	1 in 173	99%	1 in 17,201	1 in 68,804
AIRE	AR	Autoimmune polyendocrinopathy syndrome type I	General Population	1 in 150	98%	1 in 7,451	1 in 29,804
			Finnish Population	1 in 79	98%	1 in 3,901	1 in 15,604
ALDH3A2	AR	Sjögren-Larsson syndrome	General Population	1 in 250	98%	1 in 12,451	1 in 49,804
ALDOB	AR	Hereditary fructose intolerance	General Population	1 in 122	99%	1 in 12,101	1 in 48,404
			African/African American Population	1 in 250	99%	1 in 24,901	1 in 99,604
			Caucasian / European Population	1 in 67	99%	1 in 6,601	1 in 26,404
			Middle-Eastern Population	1 in 97	99%	1 in 9,601	1 in 38,404
ALG6	AR	Congenital disorder of glycosylation type Ic	General Population	<1 in 500	98%	<1 in 24,951	<1 in 99,804
ALMS1	AR	Alstrom syndrome	General Population	1 in 500	98%	1 in 24,951	1 in 99,804
ALPL	AR	Hypophosphatasia	General Population	1 in 158	95%	1 in 3,141	1 in 12,564
			Caucasian / European Population	1 in 274	95%	1 in 5,461	1 in 21,844
			Mennonite Population	1 in 25	95%	1 in 481	1 in 1,924
AMT	AR	Glycine encephalopathy, AMT-related	General Population	1 in 373	98%	1 in 18,601	1 in 74,404
			Finnish Population	1 in 117	98%	1 in 5,801	1 in 23,204

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AQP2	AR	Nephrogenic diabetes insipidus	General Population	<1 in 500	95%	<1 in 9,981	<1 in 39,924
			Finnish Population	1 in 169	95%	1 in 3,361	1 in 13,444
ARG1	AR	Arginase deficiency	General Population	1 in 296	98%	1 in 14,751	1 in 59,004
ARSA	AR	Metachromatic leukodystrophy	General Population	1 in 100	95%	1 in 1,981	1 in 7,924
			Caucasian / European Population	1 in 78	95%	1 in 1,541	1 in 6,164
ARSB	AR	Mucopolysaccharidosis type VI (Maroteaux-Lamy syndrome)	General Population	1 in 250	98%	1 in 12,451	1 in 49,804
			Western Australian Population	1 in 283	98%	1 in 14,101	1 in 56,404
ARSE	XL	Chondrodysplasia punctata type 1, X-linked	General Population	<1 in 500	98%	<1 in 24,951	<1 in 99,804
ASL	AR	Argininosuccinate lyase deficiency	General Population	1 in 132	90%	1 in 1,311	1 in 5,244
ASNS	AR	Asparagine synthetase deficiency	General Population	<1 in 500	98%	<1 in 24,951	<1 in 99,804
ASPA	AR	Canavan disease	General Population	1 in 300	97%	1 in 9,968	1 in 39,871
			Ashkenazi Jewish Population	1 in 55	96%	1 in 1,351	1 in 5,404
ASS1	AR	Citrullinemia	General Population	1 in 119	96%	1 in 2,951	1 in 11,804
			East Asian Population	1 in 132	96%	1 in 3,276	1 in 13,104
ATM	AR	Ataxia-telangiectasia	General Population	1 in 100	92%	1 in 1,239	1 in 4,954
ATP6V1B1	AR	Renal tubular acidosis with deafness	General Population	<1 in 500	98%	<1 in 24,951	<1 in 99,804
ATP7A	XL	Menkes disease	General Population	<1 in 500	99%	<1 in 49,901	<1 in 199,604
ATP7B	AR	Wilson disease	General Population	1 in 87	98%	1 in 4,301	1 in 17,204
			Caucasian / European Population	1 in 42	98%	1 in 2,051	1 in 8,204
ATRX	XL	Alpha thalassemia X-linked intellectual disability syndrome	General Population	<1 in 500	99%	<1 in 49,901	<1 in 199,604
BBS1	AR	Bardet-Biedl syndrome type 1	General Population	1 in 367	99%	1 in 36,601	1 in 146,404
BBS10	AR	Bardet-Biedl syndrome type 10	General Population	1 in 395	99%	1 in 39,401	1 in 157,604
BBS12	AR	Bardet-Biedl syndrome type 12	General Population	1 in 791	99%	1 in 79,001	1 in 316,004
BBS2	AR	Bardet-Biedl syndrome 2; Retinitis Pigmentosa 74	General Population	1 in 621	99%	1 in 62,001	1 in 248,004
			Ashkenazi Jewish Population	1 in 107	99%	1 in 10,601	1 in 42,404
BCKDHA	AR	Maple syrup urine disease type Ia	General Population	1 in 321	98%	1 in 16,001	1 in 64,004
			Mennonite Population	1 in 10	98%	1 in 451	1 in 1,804
BCKDHB	AR	Maple syrup urine disease type Ib	General Population	1 in 364	98%	1 in 18,151	1 in 72,604
			Ashkenazi Jewish Population	1 in 97	98%	1 in 4,801	1 in 19,204
BCS1L	AR	Björnstad syndrome; GRACILE syndrome; Mitochondrial complex III deficiency	General Population	<1 in 500	98%	<1 in 24,951	<1 in 99,804
BLM	AR	Bloom syndrome	General Population	1 in 800	87%	1 in 6,147	1 in 24,589
			Ashkenazi Jewish Population	1 in 134	99%	1 in 13,301	1 in 53,204
BSND	AR	Bartter syndrome	General Population	1 in 500	98%	1 in 24,951	1 in 99,804
BTD	AR	Biotinidase deficiency	General Population	1 in 124	99%	1 in 12,301	1 in 49,204
			Caucasian / European Population	1 in 71	99%	1 in 7,001	1 in 28,004
			Latino Population	1 in 136	99%	1 in 13,501	1 in 54,004
			Middle-Eastern Population	1 in 55	99%	1 in 5,401	1 in 21,604
CAPN3	AR	Limb-girdle muscular dystrophy type 2A	General Population	<1 in 500	98%	<1 in 24,951	<1 in 99,804
			Caucasian / European Population	1 in 103	98%	1 in 5,101	1 in 20,404
CBS	AR	Homocystinuria due to cystathionine beta-synthase deficiency	General Population	1 in 224	99%	1 in 22,301	1 in 89,204
			Caucasian / European Population	1 in 86	99%	1 in 8,501	1 in 34,004
			Middle-Eastern Population	1 in 21	99%	1 in 2,001	1 in 8,004
CCDC103	AR	Primary ciliary dyskinesia, type 17	General Population	1 in 316	98%	1 in 15,751	1 in 63,004
CCDC151	AR	Primary ciliary dyskinesia, type 30	General Population	1 in 365	98%	1 in 18,201	1 in 72,804
CCDC39	AR	Primary ciliary dyskinesia, type 14	General Population	1 in 211	98%	1 in 10,501	1 in 42,004
CD40LG	XL	Hyper IgM syndrome, X-linked	General Population	<1 in 500	98%	<1 in 24,951	<1 in 99,804
CDH23	AR/Digenic	Usher syndrome, type 1D	General Population	1 in 285	90%	1 in 2,841	1 in 11,364
CEP290	AR	Bardet-Biedl syndrome 14; Joubert syndrome 5; Leber congenital amaurosis 10; Meckel syndrome 4; Senior-Løken syndrome 6	General Population	1 in 190	98%	1 in 9,451	1 in 37,804

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CERKL	AR	Retinitis pigmentosa 26	General Population	1 in 148	98%	1 in 7,351	1 in 29,404
CFTR	AR	Cystic fibrosis	General Population	1 in 32	99%	1 in 3101	1 in 12,404
			African/African American Population	1 in 61	99%	1 in 6001	1 in 24,004
			Ashkenazi Jewish Population	1 in 24	99%	1 in 2301	1 in 9,204
			Caucasian / European Population	1 in 25	99%	1 in 2401	1 in 9,604
			East Asian Population	1 in 94	99%	1 in 9301	1 in 37,204
			Latino Population	1 in 58	99%	1 in 5701	1 in 22,804
CHM	XL	Choroideremia	General Population	<1 in 500	95%	<1 in 9,981	<1 in 39,924
CHRNE	AR	Congenital myasthenic syndrome, CHRNE-related	General Population	1 in 408	99%	1 in 40,701	1 in 162,804
CHRNG	AR	Multiple pterygium syndrome	General Population	<1 in 500	98%	<1 in 24,951	<1 in 99,804
CIITA	AR	Bare lymphocyte syndrome, type II	General Population	<1 in 500	98%	<1 in 24,951	<1 in 99,804
CLN3	AR	Neuronal ceroid lipofuscinosis, CLN3-related	General Population	1 in 230	98%	1 in 11,451	1 in 45,804
			Finnish Population	1 in 72	98%	1 in 3,551	1 in 14,204
CLN5	AR	Neuronal ceroid lipofuscinosis, CLN5-related	General Population	<1 in 500	95%	<1 in 9,981	<1 in 39,924
			Finnish Population	1 in 115	95%	1 in 2,281	1 in 9,124
CLN6	AR	Neuronal ceroid lipofuscinosis, CLN6-related	General Population	<1 in 500	92%	<1 in 6,239	<1 in 24,954
CLN8	AR	Neuronal ceroid lipofuscinosis, CLN8-related	General Population	<1 in 500	95%	<1 in 9,981	<1 in 39,924
			Finnish Population	1 in 135	95%	1 in 2,681	1 in 10,724
CLRN1	AR	Usher syndrome, type 3A	General Population	1 in 500	98%	1 in 24,951	1 in 99,804
			Ashkenazi Jewish Population	1 in 120	98%	1 in 5,951	1 in 23,804
			Finnish Population	1 in 70	98%	1 in 3,451	1 in 13,804
CNGB3	AR	Achromatopsia	General Population	1 in 87	99%	1 in 8,601	1 in 34,404
			Micronesian Population	1 in 2	99%	1 in 101	1 in 404
COL27A1	AR	Steel syndrome	General Population	<1 in 500	98%	<1 in 24,951	<1 in 99,804
COL4A3	AR	Alport syndrome, COL4A3-related	General Population	1 in 267	98%	1 in 13,301	1 in 53,204
			Ashkenazi Jewish Population	1 in 188	98%	1 in 9,351	1 in 37,404
COL4A4	AR	Alport syndrome, COL4A4-related	General Population	1 in 267	98%	1 in 13,301	1 in 53,204
COL4A5	XL	Alport syndrome, COL4A5-related	General Population	1 in 139	98%	1 in 6,901	1 in 27,604
COL7A1	AR	Dystrophic epidermolysis bullosa	General Population	1 in 196	97%	1 in 6,501	1 in 26,004
CPS1	AR	Carbamoylphosphate synthetase I deficiency	General Population	1 in 570	98%	1 in 28,451	1 in 113,804
CPT1A	AR	Carnitine palmitoyltransferase IA deficiency	General Population	1 in 354	90%	1 in 3,531	1 in 14,124
			Hutterite Population	1 in 16	90%	1 in 151	1 in 604
CPT2	AR	Carnitine palmitoyltransferase II deficiency	General Population	<1 in 500	95%	<1 in 9,981	<1 in 39,924
			Ashkenazi Jewish Population	1 in 51	95%	1 in 1,001	1 in 4,004
CRB1	AR	Leber congenital amaurosis 8; Retinitis pigmentosa 12	General Population	1 in 104	98%	1 in 5,151	1 in 20,604
CTNS	AR	Cystinosis	General Population	1 in 158	99%	1 in 15,701	1 in 62,804
			British Population	1 in 81	99%	1 in 8,001	1 in 32,004
CTSK	AR	Pycnodysostosis	General Population	<1 in 500	98%	<1 in 24,951	<1 in 99,804
CYBA	AR	Chronic granulomatous disease	General Population	1 in 224	99%	1 in 22,301	1 in 89,204
CYBB	XL	Chronic granulomatous disease, X-linked	General Population	<1 in 500	99%	<1 in 49,901	<1 in 199,604
CYP11B1	AR	Congenital adrenal hyperplasia due to 11-beta-hydroxylase deficiency	General Population	1 in 158	98%	1 in 7,851	1 in 31,404
			Morrocan Jewish Population	1 in 35	98%	1 in 1,701	1 in 6,804
CYP11B2	AR	Corticosterone methyloxidase deficiency	General Population	<1 in 500	98%	<1 in 24,951	<1 in 99,804
CYP17A1	AR	Congenital adrenal hyperplasia due to 17-alpha-hydroxylase deficiency	General Population	1 in 500	98%	1 in 24,951	1 in 99,804
CYP19A1	AR	Aromatase deficiency	General Population	<1 in 500	98%	<1 in 24,951	<1 in 99,804
CYP1B1	AR	Primary congenital glaucoma	General Population	1 in 50	99%	1 in 4,901	1 in 19,604
CYP21A2	AR	Congenital adrenal hyperplasia due to 21-hydroxylase deficiency	General Population	1 in 61	99%	1 in 6,001	1 in 24,004
			Inuit Population	1 in 9	99%	1 in 801	1 in 3,204
			Middle-Eastern Population	1 in 35	99%	1 in 3,401	1 in 13,604
CYP27A1	AR	Cerebrotendinous xanthomatosis	General Population	1 in 500	98%	1 in 24,951	1 in 99,804

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			Morrocan Jewish Population	1 in 5	98%	1 in 201	1 in 804
DBT	AR	Maple syrup urine disease, type II	General Population	1 in 481	98%	1 in 24,001	1 in 96,004
DCLRE1C	AR	Severe combined immunodeficiency with sensitivity to ionizing radiation	General Population	<1 in 500	98%	<1 in 24,951	<1 in 99,804
DCX	XL	Lissencephaly, X-linked	General Population	<1 in 500	98%	<1 in 24,951	<1 in 99,804
DHCR7	AR	Smith-Lemli-Opitz syndrome	General Population	1 in 30	96%	1 in 726	1 in 2,904
			African/African American Population	1 in 138	96%	1 in 3,426	1 in 13,704
			Ashkenazi Jewish Population	1 in 36	96%	1 in 876	1 in 3,504
DHDDS	AR	Retinitis pigmentosa 59	General Population	1 in 296	98%	1 in 14,751	1 in 59,004
			Ashkenazi Jewish Population	1 in 118	98%	1 in 5,851	1 in 23,404
DLD	AR	Dihydrolipoamide dehydrogenase deficiency	General Population	1 in 500	98%	1 in 24,951	1 in 99,804
			Ashkenazi Jewish Population	1 in 107	98%	1 in 5,301	1 in 21,204
DMD	XL	Duchenne muscular dystrophy	General Population	<1 in 500	93%	<1 in 7,130	<1 in 28,518
DNAH5	AR	Primary ciliary dyskinesia, DNAH5-related	General Population	1 in 142	98%	1 in 7,051	1 in 28,204
DNAI1	AR	Primary ciliary dyskinesia, DNAI1-related	General Population	1 in 230	98%	1 in 11,451	1 in 45,804
DNAI2	AR	Primary ciliary dyskinesia, DNAI2-related	General Population	1 in 447	98%	1 in 22,301	1 in 89,204
DNAL1	AR	Primary ciliary dyskinesia, DNAL1-related	General Population	<1 in 500	98%	<1 in 24,951	<1 in 99,804
DPYD	AR	Dihydropyrimidine dehydrogenase deficiency	General Population	<1 in 500	98%	<1 in 24,951	<1 in 99,804
DYSF	AR	Limb-girdle muscular dystrophy type 2B	General Population	<1 in 500	95%	<1 in 9,981	<1 in 39,924
			Japanese Population	1 in 332	95%	1 in 6,621	1 in 26,484
			LIbyan Jewish Population	1 in 18	95%	1 in 341	1 in 1,364
EDA	XL	Hypohidrotic ectodermal dysplasia	General Population	<1 in 500	99%	<1 in 49,901	<1 in 199,604
EIF2AK3	AR	Wolcott-Rallison syndrome	General Population	<1 in 500	98%	<1 in 24,951	<1 in 99,804
EIF2B5	AR	Leukoencephalopathy with vanishing white matter	General Population	<1 in 500	98%	<1 in 24,951	<1 in 99,804
EMD	XL	Emery-Dreifuss muscular dystrophy	General Population	<1 in 500	99%	<1 in 49,901	<1 in 199,604
ERCC6	AR	Cockayne syndrome type B; De Sanctis-Cacchione syndrome	General Population	1 in 500	99%	1 in 49,901	1 in 199,604
			Japanese Population	1 in 74	99%	1 in 7,301	1 in 29,204
ERCC8	AR	Cockayne syndrome type A	General Population	1 in 822	98%	1 in 41,051	1 in 164,204
ESCO2	AR	Roberts syndrome	General Population	<1 in 500	99%	<1 in 49,901	<1 in 199,604
ETFA	AR	Glutaric aciduria IIA	General Population	1 in 500	98%	1 in 24,951	1 in 99,804
ETFB	AR	Glutaric aciduria IIB	General Population	1 in 500	98%	1 in 24,951	1 in 99,804
ETFDH	AR	Glutaric aciduria IIC	General Population	1 in 250	98%	1 in 12,451	1 in 49,804
			East Asian Population	1 in 74	98%	1 in 3,651	1 in 14,604
ETHE1	AR	Ethylmalonic encephalopathy	General Population	<1 in 500	98%	<1 in 24,951	<1 in 99,804
EVC	AR	Ellis-van Creveld syndrome, EVC-related; Weyers acrofacial dysostosis, EVC-related	General Population	1 in 142	98%	1 in 7,051	1 in 28,204
			Amish Population	1 in 7	98%	1 in 301	1 in 1,204
EVC2	AR	Ellis-van Creveld syndrome, EVC2-related; Weyers acrodental dysostosis, EVC2-related	General Population	1 in 240	98%	1 in 11,951	1 in 47,804
			Amish Population	1 in 7	98%	1 in 301	1 in 1,204
EXOSC3	AR	Pontocerebellar hypoplasia type 1B	General Population	<1 in 500	98%	<1 in 24,951	<1 in 99,804
EYS	AR	Retinitis pigmentosa 25	General Population	1 in 66	98%	1 in 3,251	1 in 13,004
F11	AR	Factor XI deficiency	General Population	1 in 500	98%	1 in 24,951	1 in 99,804
			Ashkenazi Jewish Population	1 in 11	98%	1 in 501	1 in 2,004
F8	XL	Hemophilia A	General Population	<1 in 500	48%	<1 in 49,901	<1 in 199,604
F9	XL	Hemophilia B	General Population	<1 in 500	99%	<1 in 49,901	<1 in 199,604
FAH	AR	Tyrosinemia, type 1	General Population	1 in 99	95%	1 in 1,961	1 in 7,844
			Ashkenazi Jewish Population	1 in 150	95%	1 in 2,981	1 in 11,924
			Finnish Population	1 in 122	95%	1 in 2,421	1 in 9,684
			French Canadian Population	1 in 66	95%	1 in 1,301	1 in 5,204
			South Asian/Indian Population	1 in 172	95%	1 in 3,421	1 in 13,684
FAM161A	AR	Retinitis pigmentosa 28	General Population	1 in 296	98%	1 in 14,751	1 in 59,004
FANCA	AR	Fanconi anemia group A	General Population	1 in 239	98%	1 in 11,901	1 in 47,604

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FANCC	AR	Fanconi anemia group C	General Population	1 in 535	99%	1 in 53,401	1 in 213,604
			Ashkenazi Jewish Population	1 in 99	99%	1 in 9,801	1 in 39,204
FANCG	AR	Fanconi anemia group G	General Population	1 in 632	90%	1 in 6,311	1 in 25,244
FH	AR	Fumarase deficiency	General Population	<1 in 500	90%	<1 in 4,991	<1 in 19,964
FKRP	AR	Muscular dystrophy-dystroglycanopathy, FKRP-related	General Population	1 in 158	98%	1 in 7,851	1 in 31,404
FKTN	AR	Muscular dystrophy-dystroglycanopathy, FKTN-related; Fukuyama congenital muscular dystrophy	General Population	<1 in 500	99%	<1 in 49,901	<1 in 199,604
			Ashkenazi Jewish Population	1 in 150	99%	1 in 14,901	1 in 59,604
			Japanese Population	1 in 82	99%	1 in 8,101	1 in 32,404
FMR1	XL	Fragile X syndrome	General Population	1 in 151	99%	1 in 15,001	1 in 60,004
			Ashkenazi Jewish Population	1 in 115	99%	1 in 11,401	1 in 45,604
G6PC	AR	Glycogen Storage disease, type 1a	General Population	1 in 177	95%	1 in 3,521	1 in 14,084
			Ashkenazi Jewish Population	1 in 64	95%	1 in 1,261	1 in 5,044
GAA	AR	Pompe disease	General Population	1 in 100	98%	1 in 4,951	1 in 19,804
			African/African American Population	1 in 60	98%	1 in 2,951	1 in 11,804
			East Asian Population	1 in 112	98%	1 in 5,551	1 in 22,204
GALC	AR	Krabbe disease	General Population	1 in 158	99%	1 in 15,701	1 in 62,804
			Israeli Druze Population	1 in 6	99%	1 in 501	1 in 2,004
GALK1	AR	Galactokinase deficiency	General Population	1 in 110	95%	1 in 2,181	1 in 8,724
			Irish Population	1 in 64	95%	1 in 1,261	1 in 5,044
GALNS	AR	Mucopolysaccharidosis IVA (Morquio syndrome A)	General Population	1 in 224	97%	1 in 7,434	1 in 29,737
GALT	AR	Galactosemia	General Population	1 in 110	95%	1 in 2,181	1 in 8,724
			African/African American Population	1 in 94	95%	1 in 1,861	1 in 7,444
GAMT	AR	Guanidinoacetate methyltransferase deficiency	General Population	1 in 371	99%	1 in 37,001	1 in 148,004
GBA	AR	Gaucher disease	General Population	1 in 77	99%	1 in 7,601	1 in 30,404
			African/African American Population	1 in 35	99%	1 in 3,401	1 in 13,604
			Ashkenazi Jewish Population	1 in 15	99%	1 in 1,401	1 in 5,604
GBE1	AR	Glycogen storage disease IV	General Population	1 in 387	99%	1 in 38,601	1 in 154,404
GCDH	AR	Glutaric aciduria, type I	General Population	1 in 87	98%	1 in 4,301	1 in 17,204
			Amish Population	1 in 9	98%	1 in 401	1 in 1,604
GFM1	AR	Combined oxidative phosphorylation deficiency, GFM1-related	General Population	<1 in 500	98%	<1 in 24,951	<1 in 99,804
GJB1	XL	Charcot-Marie-Tooth disease, X-linked type 1	General Population	1 in 667	90%	1 in 6,661	1 in 26,644
GJB2	AR	Nonsyndromic hearing loss, GJB2-related	General Population	1 in 42	99%	1 in 4,101	1 in 16,404
			African/African American Population	1 in 25	99%	1 in 2,401	1 in 9,604
			Ashkenazi Jewish Population	1 in 21	99%	1 in 2,001	1 in 8,004
			Caucasian / European Population	1 in 33	99%	1 in 3,201	1 in 12,804
			Latino Population	1 in 100	99%	1 in 9,901	1 in 39,604
			Middle-Eastern Population	1 in 83	99%	1 in 8,201	1 in 32,804
			South Asian/Indian Population	1 in 148	99%	1 in 14,701	1 in 58,804
GJB6	AR	Nonsyndromic hearing loss, GJB6-related	General Population	1 in 423	99%	1 in 42,201	1 in 168,804
GLA	XL	Fabry disease	General Population	<1 in 500	99%	<1 in 49,901	<1 in 199,604
GLB1	AR	Mucopolysaccharidosis type IVB (Morquio syndrome B); GM1-gangliosidosis	General Population	1 in 134	99%	1 in 13,301	1 in 53,204
			Maltese Population	1 in 30	99%	1 in 2,901	1 in 11,604
			Roma Population	1 in 50	99%	1 in 4,901	1 in 19,604
GLDC	AR	Glycine encephalopathy, GLDC-related	General Population	1 in 193	98%	1 in 9,601	1 in 38,404
			British Columbia Canadian Population	1 in 125	99%	1 in 12,401	1 in 49,604
			Finnish Population	1 in 117	99%	1 in 11,601	1 in 46,404
GLE1	AR	Lethal congenital contracture syndrome 1	General Population	<1 in 500	98%	<1 in 24,951	<1 in 99,804
			Finnish Population	1 in 80	98%	1 in 3,951	1 in 15,804
GNE	AR	Inclusion body myopathy type 2 (Nonaka myopathy)	General Population	<1 in 500	80%	<1 in 2,496	<1 in 9,984

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			Iranian Jewish Population	1 in 11	80%	1 in 51	1 in 204
GNPTAB	AR	Mucolipidosis III alpha/beta; Mucolipidosis II alpha/beta	General Population	<1 in 500	95%	<1 in 9,981	<1 in 39,924
GNPTG	AR	Mucolipidosis III gamma	General Population	<1 in 500	95%	<1 in 9,981	<1 in 39,924
GNS	AR	Mucopolysaccharidosis IIID (Sanfilippo syndrome D)	General Population	1 in 500	98%	1 in 24,951	1 in 99,804
GP1BA	AR	Bernard-Soulier syndrome type A1	General Population	1 in 500	98%	1 in 24,951	1 in 99,804
GP9	AR	Bernard-Soulier syndrome type C	General Population	1 in 500	98%	1 in 24,951	1 in 99,804
GRHPR	AR	Primary Hyperoxaluria type II	General Population	<1 in 500	99%	<1 in 49,901	<1 in 199,604
GUSB	AR	Mucopolysaccharidosis type VII	General Population	1 in 250	98%	1 in 12,451	1 in 49,804
HADHA	AR	Long-chain 3-hydroxyacyl-CoA dehydrogenase (LCHAD) deficiency; Trifunctional protein deficiency	General Population	<1 in 500	98%	<1 in 24,951	<1 in 99,804
			Finnish Population	1 in 124	98%	1 in 6,151	1 in 24,604
HAX1	AR	Severe Congenital Neutropenia, HAX1-related	General Population	1 in 224	98%	1 in 11,151	1 in 44,604
HBA1/HBA2	AR	Alpha thalassemia	General Population	1 in 20	90%	1 in 191	1 in 764
			African/African American Population	1 in 3	90%	1 in 21	1 in 84
			Ashkenazi Jewish Population	1 in 13	90%	1 in 121	1 in 484
			East Asian Population	1 in 8	90%	1 in 71	1 in 284
			Middle-Eastern Population	1 in 3	90%	1 in 21	1 in 84
			South Asian/Indian Population	1 in 5	90%	1 in 41	1 in 164
HBB	AR	Sickle cell disease; Beta thalassemia	General Population	1 in 158	95%	1 in 3,141	1 in 12,564
			African/African American Population	1 in 10	95%	1 in 181	1 in 724
			East Asian Population	1 in 50	95%	1 in 981	1 in 3,924
			Latino Population	1 in 128	95%	1 in 2,541	1 in 10,164
			Mediterranean Population	1 in 3	95%	1 in 41	1 in 164
			South Asian/Indian Population	1 in 25	95%	1 in 481	1 in 1,924
HEXA	AR	Tay-Sachs disease	General Population	1 in 300	99%	1 in 29,901	1 in 119,604
			Ashkenazi Jewish Population	1 in 27	99%	1 in 2,601	1 in 10,404
HEXB	AR	Sandhoff disease	General Population	1 in 600	98%	1 in 29,951	1 in 119,804
HFE2	AR	Hemochromatosis, type 2A	General Population	1 in 500	99%	1 in 49,901	1 in 199,604
HGD	AR	Alkaptonuria	General Population	1 in 250	90%	1 in 2,491	1 in 9,964
HGSNAT	AR	Mucopolysaccharidosis type IIIC (Sanfilippo syndrome C)	General Population	1 in 434	98%	1 in 21,651	1 in 86,604
			Caucasian / European Population	1 in 345	98%	1 in 17,201	1 in 68,804
HLCS	AR	Holocarboxylase synthetase deficiency	General Population	1 in 500	98%	1 in 24,951	1 in 99,804
HMGCL	AR	3-hydroxy-3-methylglutaryl-CoA lyase deficiency	General Population	<1 in 500	98%	<1 in 24,951	<1 in 99,804
HOGA1	AR	Primary hyperoxaluria type III	General Population	1 in 184	99%	1 in 18,301	1 in 73,204
HPS1	AR	Hermansky-Pudlak syndrome 1	General Population	1 in 354	98%	1 in 17,651	1 in 70,604
			Puerto Rican Population	1 in 21	98%	1 in 1,001	1 in 4,004
HPS3	AR	Hermansky-Pudlak syndrome 3	General Population	1 in 354	98%	1 in 17,651	1 in 70,604
HSD17B4	AR	D-bifunctional protein deficiency	General Population	1 in 158	98%	1 in 7,851	1 in 31,404
HSD3B2	AR	Congenital adrenal hyperplasia due to 3-beta-hydroxysteroid dehydrogenase 2 deficiency	General Population	<1 in 500	98%	<1 in 24,951	<1 in 99,804
HYAL1	AR	Mucopolysaccharidosis type IX	General Population	<1 in 500	98%	<1 in 24,951	<1 in 99,804
HYLS1	AR	Hydrolethalus syndrome	General Population	<1 in 500	98%	<1 in 24,951	<1 in 99,804
			Finnish Population	1 in 50	98%	1 in 2,451	1 in 9,804
IDS	XL	Mucopolysaccharidosis type II (Hunter syndrome)	General Population	<1 in 500	91%	<1 in 5,545	<1 in 22,182
IDUA	AR	Mucopolysaccharidosis, type I (Hurler syndrome)	General Population	<1 in 500	95%	<1 in 9,981	<1 in 39,924
			Caucasian / European Population	1 in 153	95%	1 in 3,041	1 in 12,164
IKBKAP	AR	Familial dysautonomia	General Population	1 in 300	99%	1 in 29,901	1 in 119,604
			Ashkenazi Jewish Population	1 in 31	99%	1 in 3,001	1 in 12,004
IL2RG	XL	Severe combined immunodeficiency, X-linked	General Population	<1 in 500	99%	<1 in 49,901	<1 in 199,604
IVD	AR	Isovaleric acidemia	General Population	1 in 167	90%	1 in 1,661	1 in 6,644
			African/African American Population	1 in 100	90%	1 in 991	1 in 3,964

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			Caucasian / European Population	1 in 115	90%	1 in 1,141	1 in 4,564
			East Asian Population	1 in 407	90%	1 in 4,061	1 in 16,244
KCNJ11	AR	Congenital hyperinsulinism; Permanent neonatal diabetes mellitus	General Population	1 in 423	99%	1 in 42,201	1 in 168,804
			Caucasian / European Population	1 in 232	99%	1 in 23,101	1 in 92,404
L1CAM	XL	L1 syndrome	General Population	<1 in 500	99%	<1 in 49,901	<1 in 199,604
LAMA2	AR	Muscular dystrophy, LAMA2-related	General Population	<1 in 500	99%	<1 in 24,951	<1 in 99,804
			Caucasian / European Population	1 in 125	99%	1 in 12,401	1 in 49,604
LAMA3	AR	Junctional epidermolysis bullosa, LAMA3-related; Laryngo-onycho-cutaneous syndrome	General Population	1 in 781	98%	1 in 39,001	1 in 156,004
LAMB3	AR	Junctional epidermolysis bullosa, LAMB3-related	General Population	1 in 781	98%	1 in 39,001	1 in 156,004
LAMC2	AR	Junctional epidermolysis bullosa, LAMC2-related	General Population	1 in 781	98%	1 in 39,001	1 in 156,004
LCA5	AR	Leber congenital amaurosis 5	General Population	1 in 500	98%	1 in 24,951	1 in 99,804
LHX3	AR	Combined pituitary hormone deficiency 3	General Population	1 in 45	98%	1 in 2,201	1 in 8,804
LIFR	AR	Stuve-Wiedemann syndrome	General Population	<1 in 500	98%	<1 in 24,951	<1 in 99,804
LIPA	AR	Lysosomal acid lipase deficiency	General Population	<1 in 500	99%	<1 in 24,951	<1 in 99,804
			Caucasian / European Population	1 in 112	99%	1 in 11,101	1 in 44,404
LOXHD1	AR	Nonsyndromic hearing loss, LOXHD1-related	General Population	1 in 500	98%	1 in 24,951	1 in 99,804
			Ashkenazi Jewish Population	1 in 180	98%	1 in 8,951	1 in 35,804
LRPPRC	AR	Leigh syndrome with Complex IV deficiency	General Population	1 in 447	98%	1 in 22,301	1 in 89,204
			Faroese Population	1 in 21	98%	1 in 1,001	1 in 4,004
			French Canadian Population	1 in 22	98%	1 in 1,051	1 in 4,204
LYST	AR	Chediak-Higashi syndrome	General Population	<1 in 500	90%	<1 in 4,991	<1 in 19,964
MAN2B1	AR	Alpha-mannosidosis	General Population	1 in 354	99%	1 in 35,301	1 in 141,204
			Caucasian / European Population	1 in 274	99%	1 in 27,301	1 in 109,204
MCCC1	AR	3-Methylcrotonyl-CoA carboxylase 1 deficiency (3-MCC deficiency)	General Population	1 in 95	98%	1 in 4,701	1 in 18,804
MCCC2	AR	3-Methylcrotonyl-CoA carboxylase 2 deficiency (3-MCC deficiency)	General Population	1 in 95	98%	1 in 4,701	1 in 18,804
MCOLN1	AR	Mucolipidosis IV	General Population	1 in 300	99%	1 in 29,901	1 in 119,604
			Ashkenazi Jewish Population	1 in 100	99%	1 in 9,901	1 in 39,604
MED17	AR	Postnatal progressive microcephaly with seizures and brain atrophy	General Population	<1 in 500	99%	<1 in 49,901	<1 in 199,604
MEFV	AR	Familial Mediterranean fever	General Population	1 in 20	99%	1 in 1,901	1 in 7,604
			Mediterranean Population	1 in 7	90%	1 in 61	1 in 244
MESP2	AR	Spondylocostal dysostosis	General Population	<1 in 500	98%	<1 in 24,951	<1 in 99,804
MFSD8	AR	Neuronal ceroid lipofuscinosis, MFSD8-related	General Population	<1 in 500	95%	<1 in 9,981	<1 in 39,924
MKS1	AR	Joubert syndrome 28; Meckel syndrome 1; Bardet-Biedl syndrome 13	General Population	1 in 260	98%	1 in 12,951	1 in 51,804
			Finnish Population	1 in 47	98%	1 in 2,301	1 in 9,204
MLC1	AR	Megalencephalic leukoencephalopathy with subcortical cysts	General Population	<1 in 500	97%	<1 in 16,634	<1 in 66,537
MMAA	AR	Methylmalonic aciduria, cblA type	General Population	1 in 301	97%	1 in 10,001	1 in 40,004
MMAB	AR	Methylmalonic aciduria, cblB type	General Population	1 in 435	98%	1 in 21,701	1 in 86,804
MMACHC	AR	Methylmalonic aciduria and homocystinuria, cblC type	General Population	1 in 134	90%	1 in 1,331	1 in 5,324
MMADHC	AR	Methylmalonic aciduria and homocystinuria, cblD type	General Population	<1 in 500	98%	<1 in 24,951	<1 in 99,804
MPI	AR	Congenital disorder of glycosylation type Ib	General Population	<1 in 500	98%	<1 in 24,951	<1 in 99,804
MPL	AR	Congenital amegakaryocytic thrombocytopenia	General Population	1 in 102	98%	1 in 5,051	1 in 20,204
			Ashkenazi Jewish Population	1 in 55	98%	1 in 2,701	1 in 10,804
MPV17	AR	Hepatocerebral mitochondrial DNA depletion syndrome, MPV17-related	General Population	<1 in 500	96%	<1 in 12,476	<1 in 49,904
			Native American Population	1 in 20	96%	1 in 476	1 in 1,904
MTM1	XL	Myotubular myopathy, X-linked	General Population	<1 in 500	98%	<1 in 24,951	<1 in 99,804

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MTRR	AR	Homocystinuria-megaloblastic anemia, cobalamin E type	General Population	<1 in 500	98%	<1 in 24,951	<1 in 99,804
MTTP	AR	Abetalipoproteinemia	General Population	<1 in 500	98%	<1 in 24,951	<1 in 99,804
			Ashkenazi Jewish Population	1 in 180	98%	1 in 8,951	1 in 35,804
MUT	AR	Methylmalonic acidemia, MUT-related	General Population	1 in 195	96%	1 in 4,851	1 in 19,404
			East Asian Population	1 in 53	96%	1 in 1,301	1 in 5,204
			Middle-Eastern Population	1 in 52	96%	1 in 1,276	1 in 5,104
MYO7A	AR	Non-syndromic hearing loss, MYO7A-related; Usher syndrome, type 1B	General Population	1 in 206	98%	1 in 10,251	1 in 41,004
			East Asian Population	1 in 62	98%	1 in 3,051	1 in 12,204
NAGLU	AR	Mucopolysaccharidosis type IIIB (Sanfilippo syndrome B)	General Population	<1 in 500	99%	<1 in 49,901	<1 in 199,604
			Caucasian / European Population	1 in 346	99%	1 in 34,501	1 in 138,004
			East Asian Population	1 in 298	99%	1 in 29,701	1 in 118,804
NAGS	AR	N-acetylglutamate synthase deficiency	General Population	<1 in 500	98%	<1 in 24,951	<1 in 99,804
NBN	AR	Nijmegen breakage syndrome	General Population	1 in 158	99%	1 in 15,701	1 in 62,804
NDRG1	AR	Charcot-Marie-Tooth disease, type 4D	General Population	1 in 22	98%	1 in 1,051	1 in 4,204
NDUFAF5	AR	Mitochondrial complex I deficiency (Leigh syndrome), NDUFAF5-related	General Population	1 in 447	98%	1 in 22,301	1 in 89,204
			Ashkenazi Jewish Population	1 in 290	98%	1 in 14,451	1 in 57,804
NDUFS6	AR	Mitochondrial complex I deficiency (Leigh syndrome), NDUFS6-related	General Population	<1 in 500	98%	<1 in 24,951	<1 in 99,804
NEB	AR	Nemaline myopathy	General Population	1 in 112	98%	1 in 5,551	1 in 22,204
			Amish Population	1 in 11	98%	1 in 501	1 in 2,004
			Ashkenazi Jewish Population	1 in 108	98%	1 in 5,351	1 in 21,404
			Finnish Population	1 in 112	98%	1 in 5,551	1 in 22,204
NPC1	AR	Niemann-Pick disease, type C1	General Population	1 in 194	90%	1 in 1,931	1 in 7,724
NPC2	AR	Niemann-Pick disease, type C2	General Population	1 in 194	99%	1 in 19,301	1 in 77,204
NPHP1	AR	Joubert syndrome 4; Senior-Løken syndrome 1; Nephronophthisis	General Population	1 in 480	98%	1 in 23,951	1 in 95,804
			Finnish Population	1 in 124	98%	1 in 6,151	1 in 24,604
NPHS1	AR	Congenital nephrotic syndrome, type 1	General Population	1 in 289	98%	1 in 14,401	1 in 57,604
			Finnish Population	1 in 50	98%	1 in 2,451	1 in 9,804
NPHS2	AR	Congenital nephrotic syndrome, type 2	General Population	1 in 289	98%	1 in 14,401	1 in 57,604
			Finnish Population	1 in 50	98%	1 in 2,451	1 in 9,804
NR0B1	XL	Congenital adrenal hypoplasia, X-linked	General Population	<1 in 500	99%	<1 in 49,901	<1 in 199,604
NR2E3	AR	Enhanced S-cone syndrome; Retinitis pigmentosa 37	General Population	1 in 209	98%	1 in 10,401	1 in 41,604
NTRK1	AR	Congenital insensitivity to pain with anhidrosis	General Population	<1 in 500	99%	<1 in 49,901	<1 in 199,604
OAT	AR	Gyrate atrophy of choroid and retina	General Population	<1 in 500	98%	<1 in 24,951	<1 in 99,804
OCRL	XL	Dent disease 2; Lowe syndrome	General Population	<1 in 500	95%	<1 in 9,981	<1 in 39,924
OPA3	AR	Costeff syndrome	General Population	<1 in 500	98%	<1 in 24,951	<1 in 99,804
			Iraqi Jewish Population	1 in 50	98%	1 in 2,451	1 in 9,804
OTC	XL	Ornithine transcarbamylase deficiency	General Population	<1 in 500	90%	<1 in 4,991	<1 in 19,964
PAH	AR	Phenylalanine hydroxylase deficiency (Phenylketonuria)	General Population	1 in 93	99%	1 in 9,201	1 in 36,804
			Caucasian / European Population	1 in 63	99%	1 in 6,201	1 in 24,804
			Middle-Eastern Population	1 in 74	99%	1 in 7,301	1 in 29,204
			South East Asian	1 in 59	99%	1 in 5,801	1 in 23,204
PC	AR	Pyruvate carboxylase deficiency	General Population	1 in 250	95%	1 in 4,981	1 in 19,924
PCCA	AR	Propionic acidemia, PCCA-related	General Population	1 in 224	96%	1 in 5,576	1 in 22,304
			Native American Population	1 in 85	96%	1 in 2,101	1 in 8,404
PCCB	AR	Propionic acidemia, PCCB-related	General Population	1 in 224	99%	1 in 22,301	1 in 89,204
			Native American Population	1 in 85	99%	1 in 8,401	1 in 33,604

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PCDH15	AR/Digenic	Non-syndromic hearing loss, PCDH15-related; Usher syndrome, type 1F	General Population	1 in 395	98%	1 in 19,701	1 in 78,804
			Ashkenazi Jewish Population	1 in 72	98%	1 in 3,551	1 in 14,204
PDHA1	XL	Pyruvate dehydrogenase E1-alpha deficiency	General Population	<1 in 500	98%	<1 in 24,951	<1 in 99,804
PDHB	AR	Pyruvate dehydrogenase E1-beta deficiency	General Population	<1 in 500	98%	<1 in 24,951	<1 in 99,804
PEX1	AR	Zellweger syndrome, PEX1-related	General Population	1 in 147	95%	1 in 2,921	1 in 11,684
PEX10	AR	Zellweger syndrome, PEX10-related	General Population	1 in 500	95%	1 in 9,981	1 in 39,924
			Japanese Population	1 in 354	95%	1 in 7,061	1 in 28,244
PEX12	AR	Zellweger syndrome, PEX12-related	General Population	1 in 373	95%	1 in 7,441	1 in 29,764
PEX2	AR	Zellweger syndrome, PEX2-related	General Population	1 in 500	95%	1 in 9,981	1 in 39,924
			Ashkenazi Jewish Population	1 in 123	95%	1 in 2,441	1 in 9,764
PEX6	AR	Zellweger syndrome, PEX6-related	General Population	1 in 280	95%	1 in 5,581	1 in 22,324
PEX7	AR	Rhizomelic chondrodysplasia punctata, type 1	General Population	1 in 158	99%	1 in 15,701	1 in 62,804
PFKM	AR	Glycogen storage disease VII	General Population	<1 in 500	98%	<1 in 24,951	<1 in 99,804
PHGDH	AR	Phosphoglycerate dehydrogenase deficiency	General Population	<1 in 500	98%	<1 in 24,951	<1 in 99,804
			Ashkenazi Jewish Population	1 in 280	98%	1 in 13,951	1 in 55,804
PKHD1	AR	Polycystic kidney disease, PKHD1-related	General Population	1 in 70	98%	1 in 3,451	1 in 13,804
			Ashkenazi Jewish Population	1 in 107	98%	1 in 5,301	1 in 21,204
PLA2G6	AR	Infantile neuroaxonal dystrophy	General Population	1 in 500	97%	1 in 16,634	1 in 66,537
PMM2	AR	Congenital disorder of glycosylation type 1a	General Population	<1 in 500	99%	<1 in 49,901	<1 in 199,604
			Ashkenazi Jewish Population	1 in 57	99%	1 in 5,601	1 in 22,404
			Caucasian / European Population	1 in 71	99%	1 in 7,001	1 in 28,004
POLG	AR	Progressive external ophthalmoplegia; Alpers-Huttenlocher syndrome; Ataxia neuropathy spectrum; Myocerebrohepatopathy syndrome	General Population	1 in 113	95%	1 in 2,241	1 in 8,964
POMGNT1	AR	Muscular dystrophy-dystroglycanopathy; Retinitis pigmentosa 76	General Population	1 in 462	98%	1 in 23,051	1 in 92,204
			Finnish Population	1 in 111	98%	1 in 5,501	1 in 22,004
PPT1	AR	Neuronal ceroid lipofuscinosis, PPT1-related	General Population	1 in 368	98%	1 in 18,351	1 in 73,404
			Caucasian / European Population	1 in 488	98%	1 in 24,351	1 in 97,404
			Finnish Population	1 in 75	98%	1 in 3,701	1 in 14,804
PROP1	AR	Combined pituitary hormone deficiency 2	General Population	1 in 45	98%	1 in 2,201	1 in 8,804
PRPS1	XL	Arts syndrome; Rosenberg-Chutorian syndrome; Phosphoribosylpyrophosphate synthetase superactivity; Non-syndromic hearing loss, PRPS1-related	General Population	<1 in 500	98%	<1 in 24,951	<1 in 99,804
PSAP	AR	Metachromatic leukodystrophy due to saposin-b deficiency	General Population	<1 in 500	98%	<1 in 24,951	<1 in 99,804
PTS	AR	Tetrahydrobiopterin deficiency	General Population	1 in 354	96%	1 in 8,826	1 in 35,304
PUS1	AR	Mitochondrial myopathy and sideroblastic anemia 1	General Population	<1 in 500	98%	<1 in 24,951	<1 in 99,804
PYGM	AR	Glycogen storage disease type V	General Population	<1 in 500	99%	<1 in 49,901	<1 in 199,604
			Caucasian / European Population	1 in 206	99%	1 in 20,501	1 in 82,004
RAB23	AR	Carpenter syndrome	General Population	<1 in 500	98%	<1 in 24,951	<1 in 99,804
RAG1	AR	Omenn syndrome, RAG1-related	General Population	1 in 137	98%	1 in 6,801	1 in 27,204
RAG2	AR	Omenn syndrome, RAG2-related	General Population	1 in 137	98%	1 in 6,801	1 in 27,204
RAPSN	AR	Congenital myasthenic syndrome, RAPSN-related; Fetal akinesia deformation sequence	General Population	<1 in 500	99%	<1 in 49,901	<1 in 199,604
RARS2	AR	Pontocerebellar hypoplasia type 6	General Population	<1 in 500	98%	<1 in 24,951	<1 in 99,804
RDH12	AR	Leber congenital amaurosis type 13	General Population	<1 in 500	98%	<1 in 24,951	<1 in 99,804
			Caucasian / European Population	1 in 456	98%	1 in 22,751	1 in 91,004
RMRP	AR	Anauxetic dysplasia; Cartilage-hair hypoplasia; Metaphyseal dysplasia without hypotrichosis	General Population	<1 in 500	99%	<1 in 49,901	<1 in 199,604
			Amish Population	1 in 16	99%	1 in 1,501	1 in 6,004
			Finnish Population	1 in 76	99%	1 in 7,501	1 in 30,004

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RPE65	AR	Leber congenital amaurosis 2; Retinitis pigmentosa 20	General Population	1 in 228	98%	1 in 11,351	1 in 45,404
RPGRIP1L	AR	Meckel syndrome 5; Joubert syndrome 7; COACH syndrome	General Population	1 in 259	98%	1 in 12,901	1 in 51,604
RS1	XL	Juvenile retinoschisis, X-linked	General Population	<1 in 500	96%	<1 in 12,476	<1 in 49,904
RTEL1	AR	Dyskeratosis congenita type 5	General Population	1 in 500	99%	1 in 49,901	1 in 199,604
			Ashkenazi Jewish Population	1 in 203	99%	1 in 20,201	1 in 80,804
SACS	AR	Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay	General Population	<1 in 500	95%	<1 in 9,981	<1 in 39,924
			French Canadian Population	1 in 19	95%	1 in 361	1 in 1,444
SAMHD1	AR	Aicardi-Goutieres syndrome	General Population	<1 in 500	95%	<1 in 9,981	<1 in 39,924
SEPSECS	AR	Pontocerebellar hypoplasia, type 2D	General Population	<1 in 500	98%	<1 in 24,951	<1 in 99,804
SGCA	AR	Limb-girdle muscular dystrophy, type 2D	General Population	<1 in 500	98%	<1 in 24,951	<1 in 99,804
			Caucasian / European Population	1 in 288	98%	1 in 14,351	1 in 57,404
			Finnish Population	1 in 150	98%	1 in 7,451	1 in 29,804
SGCB	AR	Limb-girdle muscular dystrophy, type 2E	General Population	1 in 500	98%	1 in 24,951	1 in 99,804
			Caucasian / European Population	1 in 406	98%	1 in 20,251	1 in 81,004
SGCD	AR	Limb-girdle muscular dystrophy, type 2F	General Population	<1 in 500	98%	<1 in 24,951	<1 in 99,804
SGCG	AR	Limb-girdle muscular dystrophy, type 2C	General Population	1 in 381	98%	1 in 19,001	1 in 76,004
			Moroccan Population	1 in 250	98%	1 in 12,451	1 in 49,804
			Roma / Gypsy Population	1 in 96	98%	1 in 4,751	1 in 19,004
SGSH	AR	Mucopolysaccharidosis IIIA (Sanfilippo syndrome A)	General Population	1 in 454	98%	1 in 22,651	1 in 90,604
			Caucasian / European Population	1 in 253	98%	1 in 12,601	1 in 50,404
SLC12A3	AR	Gitelman syndrome	General Population	1 in 100	98%	1 in 4,951	1 in 19,804
SLC12A6	AR	Andermann syndrome	General Population	<1 in 500	98%	<1 in 24,951	<1 in 99,804
			French Canadian Population	1 in 23	99%	1 in 2,201	1 in 8,804
SLC17A5	AR	Sialic acid storage disorder	General Population	<1 in 500	91%	<1 in 5,545	<1 in 22,182
			Finnish Population	1 in 100	91%	1 in 1,101	1 in 4,404
SLC22A5	AR	Systemic primary carnitine deficiency	General Population	1 in 129	76%	1 in 534	1 in 2,137
			African/African American Population	1 in 86	76%	1 in 355	1 in 1,421
			East Asian Population	1 in 77	76%	1 in 318	1 in 1,271
			Faroese Population	1 in 9	76%	1 in 34	1 in 137
			Pacific Islander Population	1 in 37	76%	1 in 151	1 in 604
			South Asian/Indian Population	1 in 51	76%	1 in 209	1 in 837
SLC25A13	AR	Citrin deficiency	General Population	<1 in 500	95%	<1 in 9,981	<1 in 39,924
			East Asian Population	1 in 65	95%	1 in 1,281	1 in 5,124
SLC25A15	AR	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome (Triple H syndrome)	General Population	<1 in 500	99%	<1 in 49,901	<1 in 199,604
			French Canadian Population	1 in 37	99%	1 in 3,601	1 in 14,404
SLC25A20	AR	Carnitine-acylcarnitine translocase deficiency	General Population	<1 in 500	98%	<1 in 24,951	<1 in 99,804
SLC26A2	AR	Achondrogenesis, type IB; Atelosteogenesis II; Diastrophic dysplasia; Multiple epiphyseal dysplasia	General Population	1 in 158	90%	1 in 1,571	1 in 6,284
			Finnish Population	1 in 50	90%	1 in 491	1 in 1,964
SLC26A3	AR	Congenital secretory chloride diarrhea	General Population	<1 in 500	98%	<1 in 24,951	<1 in 99,804
			Middle-Eastern Population	1 in 57	98%	1 in 2,801	1 in 11,204
SLC26A4	AR	Pendred syndrome	General Population	1 in 80	98%	1 in 3,951	1 in 15,804
			African/African American Population	1 in 76	98%	1 in 3,751	1 in 15,004
			Caucasian / European Population	1 in 88	98%	1 in 4,351	1 in 17,404
			East Asian Population	1 in 74	98%	1 in 3,651	1 in 14,604
SLC35A3	AR	Arthrogryposis, mental retardation, and seizures	General Population	<1 in 500	98%	<1 in 24,951	<1 in 99,804
			Ashkenazi Jewish Population	1 in 453	98%	1 in 22,601	1 in 90,404
SLC37A4	AR	Glycogen storage disease, type Ib	General Population	1 in 158	95%	1 in 3,141	1 in 12,564
			Ashkenazi Jewish Population	1 in 71	95%	1 in 1,401	1 in 5,604
SLC39A4	AR	Acrodermatitis enteropathica	General Population	<1 in 500	98%	<1 in 24,951	<1 in 99,804

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SLC4A11	AR	Corneal endothelial dystrophy	General Population	<1 in 500	98%	<1 in 24,951	<1 in 99,804
SLC6A8	XL	Creatine deficiency syndrome	General Population	<1 in 500	98%	<1 in 24,951	<1 in 99,804
SLC7A7	AR	Lysinuric protein intolerance	General Population	<1 in 500	95%	<1 in 9,981	<1 in 39,924
			Finnish Population	1 in 122	95%	1 in 2,421	1 in 9,684
			Japanese Population	1 in 119	95%	1 in 2,361	1 in 9,444
SMARCAL1	AR	Schimke immunoosseous dysplasia	General Population	1 in 500	90%	1 in 4,991	1 in 19,964
SMN1	AR	Spinal muscular atrophy	General Population	1 in 54	91%	1 in 590	1 in 2,360
			African/African American Population	1 in 72	71%	1 in 246	1 in 983
			Ashkenazi Jewish Population	1 in 67	91%	1 in 734	1 in 2,937
			Caucasian / European Population	1 in 47	95%	1 in 921	1 in 3,684
			East Asian Population	1 in 59	93%	1 in 830	1 in 3,318
			Latino Population	1 in 68	90%	1 in 671	1 in 2,684
SMPD1	AR	Niemann-Pick disease, type A/B	General Population	1 in 250	95%	1 in 4,981	1 in 19,924
			Ashkenazi Jewish Population	1 in 115	95%	1 in 2,281	1 in 9,124
			Latino Population	1 in 106	95%	1 in 2,101	1 in 8,404
STAR	AR	Lipoid congenital adrenal hyperplasia	General Population	<1 in 500	98%	<1 in 24,951	<1 in 99,804
SUMF1	AR	Multiple sulfatase deficiency	General Population	1 in 500	98%	1 in 24,951	1 in 99,804
			Ashkenazi Jewish Population	1 in 320	98%	1 in 15,951	1 in 63,804
TAT	AR	Tyrosinemia, type II	General Population	1 in 250	98%	1 in 12,451	1 in 49,804
TCIRG1	AR	Osteopetrosis, TCIRG1-related	General Population	1 in 250	98%	1 in 12,451	1 in 49,804
TECPR2	AR	Spastic paraplegia 49	General Population	<1 in 500	98%	<1 in 24,951	<1 in 99,804
TFR2	AR	Hemochromatosis, type 3	General Population	<1 in 500	98%	<1 in 24,951	<1 in 99,804
TGM1	AR	Congenital ichthyosis	General Population	1 in 224	95%	1 in 4,461	1 in 17,844
TH	AR	Segawa syndrome	General Population	1 in 224	98%	1 in 11,151	1 in 44,604
TMEM216	AR	Joubert syndrome 2; Meckel syndrome 2	General Population	1 in 141	98%	1 in 7,001	1 in 28,004
			Ashkenazi Jewish Population	1 in 92	98%	1 in 4,551	1 in 18,204
TPP1	AR	Neuronal ceroid lipofuscinosis, TPP1-related	General Population	1 in 252	97%	1 in 8,368	1 in 33,471
			French Canadian Population	1 in 53	97%	1 in 1,734	1 in 6,937
TRIM32	AR	Limb-girdle muscular dystrophy, type 2H; Bardet-Biedl syndrome 11	General Population	<1 in 500	98%	<1 in 24,951	<1 in 99,804
			Hutterite Population	1 in 12	98%	1 in 551	1 in 2,204
TRMU	AR	Liver failure, acute infantile	General Population	<1 in 500	98%	<1 in 24,951	<1 in 99,804
			Yemeni Jewish Population	1 in 34	98%	1 in 1,651	1 in 6,604
TSFM	AR	Combined oxidative phosphorylation deficiency, TSFM-related	General Population	<1 in 500	98%	<1 in 24,951	<1 in 99,804
			Finnish Population	1 in 80	98%	1 in 3,951	1 in 15,804
TTC37	AR	Trichohepatoenteric syndrome	General Population	1 in 500	98%	1 in 24,951	1 in 99,804
TTPA	AR	Ataxia with isolated vitamin E deficiency	General Population	<1 in 500	98%	<1 in 24,951	<1 in 99,804
			Caucasian / European Population	1 in 267	90%	1 in 2,661	1 in 10,644
TYMP	AR	Mitochondrial neurogastrointestinal encephalopathy (MNGIE) disease	General Population	<1 in 500	98%	<1 in 24,951	<1 in 99,804
UGT1A1	AR	Crigler-Najjar syndrome	General Population	<1 in 500	98%	<1 in 24,951	<1 in 99,804
USH1C	AR	Non-syndromic hearing loss, USH1C-related; Usher syndrome, type IC	General Population	1 in 353	90%	1 in 3,521	1 in 14,084
			French Canadian Population	1 in 227	90%	1 in 2,261	1 in 9,044
USH2A	AR	Usher syndrome, type 2A	General Population	1 in 126	96%	1 in 3,126	1 in 12,504
			Caucasian / European Population	1 in 73	96%	1 in 1,801	1 in 7,204
VPS13A	AR	Choreoacanthocytosis	General Population	<1 in 500	98%	<1 in 24,951	<1 in 99,804
VPS13B	AR	Cohen syndrome	General Population	<1 in 500	98%	<1 in 24,951	<1 in 99,804
VPS45	AR	Severe congenital neutropenia, VPS45-related	General Population	1 in 224	98%	1 in 11,151	1 in 44,604
VRK1	AR	Pontocerebellar hypoplasia type 1A	General Population	<1 in 500	98%	<1 in 24,951	<1 in 99,804
VSX2	AR	Microphthalmia with or without coloboma	General Population	1 in 91	98%	1 in 4,501	1 in 18,004
WAS	XL	Wiskott-Aldrich syndrome; Thrombocytopenia, X-linked; Severe Congenital Neutropenia, WAS-related	General Population	<1 in 500	99%	<1 in 49,901	<1 in 199,604

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Supplemental Table

Gene	Mode	Condition	Ethnicity	Carrier Frequency	Detection Rate	Post-test Carrier Probability*	Residual Risk to have an affected child*
WNT10A	AR	Schopf-Schulz-Passarge syndrome; Odontoonychodermal dysplasia	General Population	<1 in 500	99%	<1 in 49,901	<1 in 199,604
XPA	AR	Xeroderma pigmentosum, group A	General Population	1 in 500	99%	1 in 49,901	1 in 199,604
			Japanese Population	1 in 74	99%	1 in 7,301	1 in 29,204
XPC	AR	Xeroderma pigmentosum, group C	General Population	1 in 500	99%	1 in 49,901	1 in 199,604
ZFYVE26	AR	Spastic paraplegia 15	General Population	<1 in 500	98%	<1 in 24,951	<1 in 99,804

* For genes that have tested negative
Abbreviations: AR, autosomal recessive; XL, X-linked