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INVITAE CARRIER SCREEN RESULTS

Patient name:	DONOR 12967	**Sample type:**	Blood	**Report date:**	19-FEB-2023
DOB:	[redacted]	**Sample collection date:**	10-JAN-2023	**Invitae #:**	RQ4539976
Sex assigned at birth:	Male	**Sample accession date:**	11-JAN-2023	**Clinical team:**	Sarah O'Brien Jeffrey Olliffe
Gender:					
Patient ID (MRN):					

Reason for testing

Gamete donor

Test performed

Invitae Carrier Screen

RESULT: POSITIVE

This carrier test evaluated 514 gene(s) for genetic changes (variants) that are associated with an increased risk of having a child with a genetic condition. Knowledge of carrier status for one of these conditions may provide information that can be used to assist with family planning and/or preparation. Carrier screening is not intended for diagnostic purposes. To identify a potential genetic basis for a condition in the individual being tested, diagnostic testing for the gene(s) of interest is recommended.

This test shows the presence of clinically significant genetic change(s) in this individual in the gene(s) indicated below. No other clinically significant changes were identified in the remaining genes evaluated with this test.

RESULTS	GENE	VARIANT(S)	INHERITANCE	PARTNER TESTING RECOMMENDED
Carrier: Joubert syndrome and related disorders (RPGRIP1L-related)	RPGRIP1L	c.986_987del (p.Lys329Thrfs*9)	Autosomal recessive	Yes
Carrier: Metabolic crises with rhabdomyolysis, cardiac arrhythmias and neurodegeneration	TANGO2	Deletion (Exons 3-9)	Autosomal recessive	Yes

Next steps

- See the table above for recommendations regarding testing of this individual's reproductive partner.
- Even for genes that have a negative test result, there is always a small risk that an individual could still be a carrier. This is called “residual risk.” See the Carrier detection rates and residual risks document.
- Discussion with a physician and/or genetic counselor is recommended to further review the implications of this test result and to understand these results in the context of any family history of a genetic condition.
- All patients, regardless of result, may wish to consider additional screening for hemoglobinopathies by complete blood count (CBC) and hemoglobin electrophoresis, if this has not already been completed.
- Individuals can register their tests at **https://www.invitae.com/patients/** to access online results, educational resources, and next steps.

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Clinical summary

RESULT: CARRIER

Joubert syndrome and related disorders (RPGRIP1L-related)

A single Pathogenic variant, c.986_987del (p.Lys329Thrfs*9), was identified in RPGRIP1L.

What are Joubert syndrome and related disorders (RPGRIP1L-related)?

Joubert syndrome and related disorders (JSRD) is part of a spectrum of disorders called ciliopathies, which involve defects in the microscopic, finger-like projections (cilia) that are located on the surface of cells and that are involved in cell movement and signaling. Ciliopathies affect many parts of the body. JSRD can be caused by changes in several different genes.

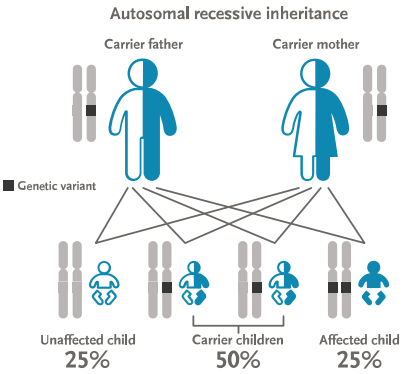
Historically, Joubert syndrome is characterized by specific brain malformations called the "molar tooth sign," low muscle tone (hypotonia), and developmental delay. Affected newborns can also have abnormally rapid breathing (tachypnea), temporary pauses in breathing (apnea), and difficulties controlling voluntary eye movement, particularly horizontal eye movement (oculomotor apraxia). Over time, breathing difficulties and visual acuity may improve. Individuals may develop difficulty coordinating movements (ataxia) and developmental delay during childhood. Many individuals have moderate intellectual disability. Other features include distinct facial appearance, extra fingers and toes (polydactyly), difficulty producing speech (speech apraxia), and seizures.

Joubert syndrome, Meckel syndrome, COACH syndrome, and orofaciodigital syndrome are part of the spectrum of conditions called JSRD. The term JSRD has been applied to individuals with characteristic Joubert syndrome features, in addition to other anomalies. This includes, but is not limited to, retinal dystrophy, a class of inherited eye conditions characterized by degeneration of the rods and cones (photoreceptors) which are the cells in the retina that respond to light, kidney disease, liver disease, neural tube defects, and congenital malformations affecting the skull and face (craniofacial malformations).

Symptoms and severity can vary, even between family members with the same genetic change. Prognosis depends on the severity of symptoms. Follow-up depends on each affected individual's specific situation, and discussion with a healthcare provider should be considered.

Next steps

Carrier testing for the reproductive partner is recommended.



If your partner tests positive:

In autosomal recessive inheritance, an individual must have disease-causing genetic changes in each copy of the RPGRIP1L gene to be affected. Carriers, who have a disease-causing genetic change in only one copy of the gene, typically do not have symptoms. When both reproductive partners are carriers of an autosomal recessive condition, there is a 25% chance for each child to have the condition.

If your partner tests negative:

A negative carrier test result reduces, but does not eliminate, the chance that a person may be a carrier. The risk that a person could still be a carrier, even after a negative test result, is called a residual risk. See the table below for your partner's hypothetical residual risk after testing negative for Joubert syndrome and related disorders (RPGRIP1L-related). These values are provided only as a guide, are based on the detection rate for the condition as tested at Invitae, and assume a negative family history, the absence of symptoms, and vary based on the ethnic background of an individual. For genes associated with both dominant and recessive inheritance, the numbers provided apply to the recessive condition(s) associated with the gene.

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DISORDER (INHERITANCE)	GENE	ETHNICITY	CARRIER FREQUENCY BEFORE SCREENING	CARRIER RESIDUAL RISK AFTER NEGATIVE RESULT
Joubert syndrome and related disorders (RPGRIP1L-related) (AR) NM_015272.2	RPGRIP1L	Pan-ethnic	1 in 259	1 in 5160

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RESULT: CARRIER

Metabolic crises with rhabdomyolysis, cardiac arrhythmias and neurodegeneration

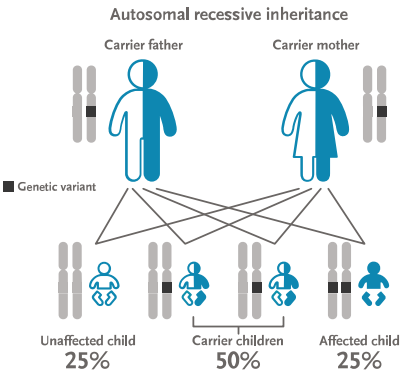
A single Pathogenic variant, Deletion (Exons 3-9), was identified in TANGO2.

What is metabolic crises with rhabdomyolysis, cardiac arrhythmias and neurodegeneration?

Metabolic crises with rhabdomyolysis, cardiac arrhythmias and neurodegeneration (MECRCN) is a condition that affects several body systems. Symptoms of MECRCN include early onset recurrent metabolic crises, breakdown of muscle tissue (rhabdomyolysis), seizures, neurodevelopmental disability, muscle weakness, structural brain abnormalities, and abnormal heart rhythm (arrhythmia). The first episode of rhabdomyolysis may present in infancy or childhood, and is typically preceded by weakness, developmental delay, and difficulty coordinating movements (ataxia). Low blood sugar (hypoglycemia), increased lactate in the blood (lactic acidemia), and a buildup of ammonia in the blood (hyperammonemia) are often observed during metabolic crises. Some affected individuals have impaired thyroid hormone production (hypothyroidism). Cardiac symptoms may be life-threatening. Follow-up depends on each affected individual's specific situation, and discussion with a healthcare provider should be considered.

Next steps

Carrier testing for the reproductive partner is recommended.



If your partner tests positive:

In autosomal recessive inheritance, an individual must have disease-causing genetic changes in each copy of the TANGO2 gene to be affected. Carriers, who have a disease-causing genetic change in only one copy of the gene, typically do not have symptoms. When both reproductive partners are carriers of an autosomal recessive condition, there is a 25% chance for each child to have the condition.

If your partner tests negative:

A negative carrier test result reduces, but does not eliminate, the chance that a person may be a carrier. The risk that a person could still be a carrier, even after a negative test result, is called a residual risk. See the table below for your partner's hypothetical residual risk after testing negative for metabolic crises with rhabdomyolysis, cardiac arrhythmias and neurodegeneration. These values are provided only as a guide, are based on the detection rate for the condition as tested at Invitae, and assume a negative family history, the absence of symptoms, and vary based on the ethnic background of an individual. For genes associated with both dominant and recessive inheritance, the numbers provided apply to the recessive condition(s) associated with the gene.

DISORDER (INHERITANCE)	GENE	ETHNICITY	CARRIER FREQUENCY BEFORE SCREENING	CARRIER RESIDUAL RISK AFTER NEGATIVE RESULT
Metabolic crises with rhabdomyolysis, cardiac arrhythmias and neurodegeneration (AR) NM_152906.6	TANGO2	Pan-ethnic	≤1 in 500	Reduced

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Results to note

HBA1/HBA2

- Additional copy(ies) detected (HBA1 copy number=3). Result not associated with alpha-thalassemia.
- Additional copy(ies) of the alpha-globin gene (HBA1/HBA2) do not impact alpha-thalassemia risk, however, co-inheritance of this genetic change with beta-thalassemia may worsen the clinical and hematological features of the latter condition. There is also a possibility for hematological changes or clinical symptoms in beta-thalassemia carriers when this alpha-globin gene change is present. Carrier testing of the reproductive partner for beta-thalassemia (HBB) may be considered.

SMN1

- Negative result. SMN1: 2 copies; c.*3+80T>G not detected.

Pseudodeficiency allele(s)

- Benign change, c.1685T>C (p.Ile562Thr), known to be a pseudodeficiency allele, identified in the GALC gene. Pseudodeficiency alleles are not known to be associated with disease, including Krabbe disease.
- The presence of a pseudodeficiency allele does not impact this individual's risk to be a carrier. Individuals with pseudodeficiency alleles may exhibit false positive results on related biochemical tests, including newborn screening. However, pseudodeficiency alleles are not known to cause disease, even when there are two copies of the variant (homozygous) or when in combination with another disease-causing variant (compound heterozygous). Carrier testing for the reproductive partner is not indicated based on this result.

Variant details

RPGRIP1L, Exon 8, c.986_987del (p.Lys329Thrfs*9), heterozygous, PATHOGENIC

- This sequence change creates a premature translational stop signal (p.Lys329Thrfs*9) in the RPGRIP1L gene. It is expected to result in an absent or disrupted protein product. Loss-of-function variants in RPGRIP1L are known to be pathogenic (PMID: 17558409).
- This variant is present in population databases (no rsID available, gnomAD 0.002%).
- This variant has not been reported in the literature in individuals affected with RPGRIP1L-related conditions.
- ClinVar contains an entry for this variant (Variation ID: 664390).
- For these reasons, this variant has been classified as Pathogenic.

TANGO2, Deletion (Exons 3-9), heterozygous, PATHOGENIC

- This variant is a gross deletion of the genomic region encompassing exon(s) 3-9 of the TANGO2 gene, which includes the termination codon. This deletion extends beyond the assayed region for this gene and therefore may encompass additional genes. While this deletion is not anticipated to lead to nonsense mediated decay, it is expected to alter mRNA translation or result in a truncated protein product.
- A similar copy number variant has been observed in individuals with rhabdomyolysis, metabolic crisis, and cardiac abnormalities (PMID: 26805781, 26805782).
- For these reasons, this variant has been classified as Pathogenic.

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Residual risk

No carrier test can detect 100% of carriers. There still remains a small risk of being a carrier after a negative test (residual risk). Residual risk values assume a negative family history and are inferred from published carrier frequencies and estimated detection rates based on testing technologies used at Invitae. You can view Invitae's complete Carrier detection rates and residual risks document (containing all carrier genes) online at **https://www.invitae.com/carrier-residual-risks/**. Additionally, the order-specific information for this report is available to download in the portal (under this order's documents) or can be requested by contacting Invitae Client Services. The complete Carrier detection rates and residual risks document will not be applicable for any genes with specimen-specific limitations in sequencing and/or deletion/duplication coverage. Please see the final bullet point in the Limitations section of this report to view if this specimen had any gene-specific coverage gaps.

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Genes analyzed

This table represents a complete list of genes analyzed for this individual, including the relevant gene transcript(s). If more than one transcript is listed for a single gene, variants were reported using the first transcript listed unless otherwise indicated in the report. An asterisk (*) indicates that this gene has a limitation. Please see the Limitations section for details. Results are negative, unless otherwise indicated in the report.

GENE	TRANSCRIPT
AAAS	NM_015665.5
ABCA12	NM_173076.2
ABCA3	NM_001089.2
ABCA4	NM_000350.2
ABCB11	NM_003742.2
ABCB4	NM_000443.3
ABCC2*	NM_000392.4
ABCC8	NM_000352.4
ACAD9	NM_014049.4
ACADM	NM_000016.5
ACADVL	NM_000018.3
ACAT1	NM_000019.3
ACOX1	NM_004035.6
ACSF3	NM_174917.4
ADA	NM_000022.2
ADAMTS2	NM_014244.4
ADAMTSL4	NM_019032.5
ADGRG1	NM_005682.6
ADGRV1	NM_032119.3
AGA	NM_000027.3
AGL	NM_000642.2
AGPS	NM_003659.3
AGXT	NM_000030.2
AHI1	NM_017651.4
AIPL1*	NM_014336.4
AIRE	NM_000383.3
ALDH3A2	NM_000382.2
ALDH7A1	NM_001182.4
ALDOB	NM_000035.3
ALG1	NM_019109.4
ALG6	NM_013339.3
ALMS1	NM_015120.4
ALPL	NM_000478.5
AMN*	NM_030943.3
AMT	NM_000481.3
ANO10*	NM_018075.3
AP1S1	NM_001283.3
AQP2	NM_000486.5
ARG1	NM_000045.3
ARL6	NM_177976.2
ARSA	NM_000487.5
ARSB	NM_000046.3
ASL	NM_000048.3
ASNS	NM_133436.3
ASPA	NM_000049.2
ASS1	NM_000050.4
ATM*	NM_000051.3
ATP6V1B1	NM_001692.3
ATP7B	NM_000053.3
ATP8B1*	NM_005603.4
BBS1	NM_024649.4
BBS10	NM_024685.3
BBS12	NM_152618.2
BBS2	NM_031885.3
BBS4	NM_033028.4
BBS5	NM_152384.2
BBS7	NM_176824.2
BBS9*	NM_198428.2
BCKDHA	NM_000709.3
BCKDHB	NM_183050.2
BCS1L	NM_004328.4
BLM	NM_000057.3
BLOC1S3	NM_212550.4
BLOC1S6	NM_012388.3
BMP1	NM_006129.4;NM_001199.3
BRIP1	NM_032043.2
BSND	NM_057176.2
BTD	NM_000060.3
CAD	NM_004341.4
CANT1	NM_138793.3
CAPN3	NM_000070.2
CASQ2	NM_001232.3
CBS	NM_000071.2
CC2D1A	NM_017721.5
CC2D2A	NM_001080522.2
CCDC103	NM_213607.2
CCDC39	NM_181426.1
CCDC88C	NM_001080414.3
CD3D	NM_000732.4
CD3E	NM_000733.3
CD40	NM_001250.5
CD59	NM_203330.2
CDH23	NM_022124.5
CEP152	NM_014985.3
CEP290	NM_025114.3
CERKL	NM_001030311.2
CFTR*	NM_000492.3
CHAT	NM_020549.4
CHRNE	NM_000080.3
CHRNG	NM_005199.4
CIITA	NM_000246.3
CLCN1	NM_000083.2
CLN3	NM_001042432.1
CLN5	NM_006493.2
CLN6	NM_017882.2
CLN8	NM_018941.3
CLRN1	NM_174878.2
CNGB3	NM_019098.4
COL11A2*	NM_080680.2
COL17A1	NM_000494.3
COL27A1	NM_032888.3
COL4A3	NM_000091.4
COL4A4	NM_000092.4
COL7A1	NM_000094.3
COX15	NM_004376.6
CPS1	NM_001875.4
CPT1A	NM_001876.3
CPT2	NM_000098.2

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GENE	TRANSCRIPT
CRB1	NM_201253.2
CRTAP	NM_006371.4
CTNS	NM_004937.2
CTSA	NM_000308.3
CTSC	NM_001814.5
CTSD	NM_001909.4
CTSK	NM_000396.3
CYBA	NM_000101.3
CYP11A1	NM_000781.2
CYP11B1	NM_000497.3
CYP11B2	NM_000498.3
CYP17A1	NM_000102.3
CYP19A1	NM_031226.2
CYP1B1	NM_000104.3
CYP21A2*	NM_000500.7
CYP27A1	NM_000784.3
CYP27B1	NM_000785.3
CYP7B1	NM_004820.3
DBT	NM_001918.3
DCAF17	NM_025000.3
DCLRE1C	NM_001033855.2
DDX11*	NM_030653.3
DFNB59	NM_001042702.3
DGAT1	NM_012079.5
DGUOK	NM_080916.2
DHCR7	NM_001360.2
DHDDS	NM_024887.3
DLD	NM_000108.4
DLL3	NM_016941.3
DNAH11	NM_001277115.1
DNAH5	NM_001369.2
DNAI1	NM_012144.3
DNAI2	NM_023036.4
DNMT3B	NM_006892.3
DOK7	NM_173660.4
DUOX2*	NM_014080.4
DYNC2H1	NM_001080463.1
DYSF	NM_003494.3
EIF2AK3	NM_004836.6

GENE	TRANSCRIPT
EIF2B1	NM_001414.3
EIF2B2	NM_014239.3
EIF2B3	NM_020365.4
EIF2B4	NM_015636.3
EIF2B5	NM_003907.2
ELP1	NM_003640.3
EPG5	NM_020964.2
ERCC2	NM_000400.3
ERCC6	NM_000124.3
ERCC8	NM_000082.3
ESCO2	NM_001017420.2
ETFA	NM_000126.3
ETFB	NM_001985.2
ETFDH	NM_004453.3
ETHE1	NM_014297.3
EVC	NM_153717.2
EVC2	NM_147127.4
EXOSC3	NM_016042.3
EYS*	NM_001142800.1
FAH*	NM_000137.2
FAM161A	NM_001201543.1
FANCA	NM_000135.2
FANCC	NM_000136.2
FANCD2*	NM_033084.3
FANCE	NM_021922.2
FANCG	NM_004629.1
FANCI	NM_001113378.1
FANCL*	NM_018062.3
FBP1	NM_000507.3
FBXO7	NM_012179.3
FH*	NM_000143.3
FKBP10	NM_021939.3
FKRP	NM_024301.4
FKTN	NM_001079802.1
FMO3	NM_006894.6
FOXN1	NM_003593.2
FOXRED1	NM_017547.3
FRAS1	NM_025074.6
FREM2	NM_207361.5

GENE	TRANSCRIPT
FUCA1	NM_000147.4
G6PC	NM_000151.3
G6PC3	NM_138387.3
GAA	NM_000152.3
GALC*	NM_000153.3
GALE*	NM_000403.3
GALK1	NM_000154.1
GALNS	NM_000512.4
GALNT3	NM_004482.3
GALT	NM_000155.3
GAMT	NM_000156.5
GATM	NM_001482.2
GBA*	NM_001005741.2
GBE1	NM_000158.3
GCDH	NM_000159.3
GCH1	NM_000161.2
GDF5	NM_000557.4
GFM1	NM_024996.5
GHR*	NM_000163.4
GJB2	NM_004004.5
GLB1	NM_000404.2
GLDC	NM_000170.2
GLE1	NM_001003722.1
GNE*	NM_001128227.2
GNPAT	NM_014236.3
GNPTAB	NM_024312.4
GNPTG	NM_032520.4
GNS	NM_002076.3
GORAB	NM_152281.2
GRHPR	NM_012203.1
GRIP1	NM_021150.3
GSS	NM_000178.2
GUCY2D	NM_000180.3
GUSB	NM_000181.3
HADH	NM_005327.4
HADHA	NM_000182.4
HADHB	NM_000183.2
HAMP	NM_021175.2
HAX1	NM_006118.3

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GENE	TRANSCRIPT
HBA1*	NM_000558.4
HBA2	NM_000517.4
HBB*	NM_000518.4
HEXA	NM_000520.4
HEXB	NM_000521.3
HGSNAT	NM_152419.2
HJV	NM_213653.3
HLCS	NM_000411.6
HMGCL	NM_000191.2
HMOX1	NM_002133.2
HOGA1	NM_138413.3
HPD	NM_002150.2
HPS1	NM_000195.4
HPS3	NM_032383.4
HPS4	NM_022081.5
HPS5	NM_181507.1
HPS6	NM_024747.5
HSD17B3	NM_000197.1
HSD17B4	NM_000414.3
HSD3B2	NM_000198.3
HYAL1	NM_153281.1
HYLS1	NM_145014.2
IDUA	NM_000203.4
IGHMBP2	NM_002180.2
IKBKB	NM_001556.2
IL7R	NM_002185.3
INVS	NM_014425.3
ITGA6	NM_000210.3
ITGB3	NM_000212.2
ITGB4	NM_001005731.2
IVD	NM_002225.3
JAK3	NM_000215.3
KCNJ1	NM_000220.4
KCNJ11	NM_000525.3
LAMA2	NM_000426.3
LAMA3	NM_000227.4
LAMB3	NM_000228.2
LAMC2	NM_005562.2
LARGE1	NM_004737.4
LCA5	NM_181714.3
LDLR	NM_000527.4
LDLRAP1	NM_015627.2
LHX3	NM_014564.4
LIFR*	NM_002310.5
LIG4	NM_002312.3
LIPA	NM_000235.3
LMBRD1	NM_018368.3
LOXHD1	NM_144612.6
LPL	NM_000237.2
LRAT	NM_004744.4
LRP2	NM_004525.2
LRPPRC	NM_133259.3
LYST	NM_000081.3
MAK	NM_001242957.2
MAN2B1	NM_000528.3
MANBA	NM_005908.3
MCEE	NM_032601.3
MCOLN1	NM_020533.2
MCPH1	NM_024596.4
MECR	NM_016011.3
MED17	NM_004268.4
MESP2	NM_001039958.1
MFSD8	NM_152778.2
MKKS	NM_018848.3
MKS1	NM_017777.3
MLC1*	NM_015166.3
MLYCD	NM_012213.2
MMAA	NM_172250.2
MMAB	NM_052845.3
MMACHC	NM_015506.2
MMADHC	NM_015702.2
MOCS1	NM_001358530.2
MOCS2A	NM_176806.3
MOCS2B	NM_004531.4
MPI	NM_002435.2
MPL	NM_005373.2
MPV17	NM_002437.4
MRE11	NM_005591.3
MTHFR*	NM_005957.4
MTR	NM_000254.2
MTRR	NM_002454.2
MTTP	NM_000253.3
MUSK	NM_005592.3
MUT	NM_000255.3
MVK	NM_000431.3
MYO15A	NM_016239.3
MYO7A	NM_000260.3
NAGA	NM_000262.2
NAGLU	NM_000263.3
NAGS	NM_153006.2
NBN	NM_002485.4
NCF2	NM_000433.3
NDRG1	NM_006096.3
NDUFAF2	NM_174889.4
NDUFAF5	NM_024120.4
NDUFS4	NM_002495.3
NDUFS6	NM_004553.4
NDUFS7	NM_024407.4
NDUFV1	NM_007103.3
NEB*	NM_001271208.1
NEU1	NM_000434.3
NGLY1	NM_018297.3
NPC1	NM_000271.4
NPC2	NM_006432.3
NPHP1	NM_000272.3
NPHS1	NM_004646.3
NPHS2	NM_014625.3
NR2E3	NM_014249.3
NSMCE3	NM_138704.3
NTRK1	NM_001012331.1
OAT*	NM_000274.3
OCA2	NM_000275.2
OPA3	NM_025136.3
OSTM1	NM_014028.3
OTOA*	NM_144672.3
OTOF	NM_194248.2;NM_194323.2
P3H1	NM_022356.3

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GENE	TRANSCRIPT
PAH	NM_000277.1
PANK2	NM_153638.2
PC	NM_000920.3
PCBD1	NM_000281.3
PCCA	NM_000282.3
PCCB	NM_000532.4
PCDH15	NM_033056.3
PCNT	NM_006031.5
PDHB	NM_000925.3
PEPD	NM_000285.3
PET100	NM_001171155.1
PEX1*	NM_000466.2
PEX10	NM_153818.1
PEX12	NM_000286.2
PEX13	NM_002618.3
PEX16	NM_004813.2
PEX2	NM_000318.2
PEX26	NM_017929.5
PEX5	NM_001131025.1
PEX6	NM_000287.3
PEX7	NM_000288.3
PFKM	NM_000289.5
PGM3	NM_001199917.1
PHGDH	NM_006623.3
PHKB	NM_000293.2;NM_001031835.2
PHKG2	NM_000294.2
PHYH	NM_006214.3
PIGN	NM_176787.4
PKHD1*	NM_138694.3
PLA2G6	NM_003560.2
PLEKHG5	NM_020631.4
PLOD1	NM_000302.3
PMM2	NM_000303.2
PNPO	NM_018129.3
POLG	NM_002693.2
POLH	NM_006502.2
POMGNT1	NM_017739.3
POMT1	NM_007171.3
POMT2	NM_013382.5

GENE	TRANSCRIPT
POR	NM_000941.2
POU1F1	NM_000306.3
PPT1	NM_000310.3
PRCD	NM_001077620.2
PRDM5	NM_018699.3
PRF1	NM_001083116.1
PROP1	NM_006261.4
PSAP	NM_002778.3
PTPRC*	NM_002838.4
PTS	NM_000317.2
PUS1	NM_025215.5
PYGM	NM_005609.3
QDPR	NM_000320.2
RAB23	NM_183227.2
RAG1	NM_000448.2
RAG2	NM_000536.3
RAPSN	NM_005055.4
RARS2	NM_020320.3
RDH12	NM_152443.2
RLBP1	NM_000326.4
RMRP	NR_003051.3
RNASEH2A	NM_006397.2
RNASEH2B	NM_024570.3
RNASEH2C	NM_032193.3
RPE65	NM_000329.2
RPGRIP1L	NM_015272.2
RTEL1	NM_001283009.1
RXYLT1	NM_014254.2
RYR1	NM_000540.2
SACS	NM_014363.5
SAMD9	NM_017654.3
SAMHD1	NM_015474.3
SCO2	NM_005138.2
SEC23B	NM_006363.4
SEPSECS	NM_016955.3
SGCA	NM_000023.2
SGCB	NM_000232.4
SGCD	NM_000337.5
SGCG	NM_000231.2

GENE	TRANSCRIPT
SGSH	NM_000199.3
SKIV2L	NM_006929.4
SLC12A1	NM_000338.2
SLC12A3	NM_000339.2
SLC12A6	NM_133647.1
SLC17A5	NM_012434.4
SLC19A2	NM_006996.2
SLC19A3	NM_025243.3
SLC1A4	NM_003038.4
SLC22A5	NM_003060.3
SLC25A13	NM_014251.2
SLC25A15	NM_014252.3
SLC25A20	NM_000387.5
SLC26A2	NM_000112.3
SLC26A3	NM_000111.2
SLC26A4	NM_000441.1
SLC27A4	NM_005094.3
SLC35A3	NM_012243.2
SLC37A4	NM_001164277.1
SLC38A8	NM_001080442.2
SLC39A4	NM_130849.3
SLC45A2	NM_016180.4
SLC4A11	NM_032034.3
SLC5A5	NM_000453.2
SLC7A7	NM_001126106.2
SMARCAL1	NM_014140.3
SMN1*	NM_000344.3
SMPD1	NM_000543.4
SNAP29	NM_004782.3
SPG11	NM_025137.3
SPR	NM_003124.4
SRD5A2	NM_000348.3
ST3GAL5	NM_003896.3
STAR	NM_000349.2
STX11	NM_003764.3
STXBP2	NM_006949.3
SUMF1	NM_182760.3
SUOX	NM_000456.2
SURF1	NM_003172.3

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GENE	TRANSCRIPT
SYNE4	NM_001039876.2
TANGO2	NM_152906.6
TAT	NM_000353.2
TBCD	NM_005993.4
TBCE*	NM_003193.4
TCIRG1	NM_006019.3
TCN2	NM_000355.3
TECPR2	NM_014844.3
TERT	NM_198253.2
TF	NM_001063.3
TFR2	NM_003227.3
TG*	NM_003235.4
TGM1	NM_000359.2
TH	NM_199292.2
TK2	NM_004614.4
TMC1	NM_138691.2
TMEM216	NM_001173990.2
TMEM67	NM_153704.5
TMPRSS3	NM_024022.2
TPO	NM_000547.5
TPP1	NM_000391.3
TREX1	NM_033629.4
TRIM32	NM_012210.3
TRIM37	NM_015294.4
TRMU	NM_018006.4
TSEN54	NM_207346.2
TSFM*	NM_001172696.1
TSHB	NM_000549.4
TSHR	NM_000369.2
TTC37	NM_014639.3
TTPA	NM_000370.3
TULP1	NM_003322.4
TYMP	NM_001953.4
TYR*	NM_000372.4
TYRP1	NM_000550.2
UBR1	NM_174916.2
UNC13D	NM_199242.2
USH1C*	NM_005709.3
USH2A	NM_206933.2

GENE	TRANSCRIPT
VDR	NM_001017535.1
VLDLR	NM_003383.4
VPS11	NM_021729.5
VPS13A*	NM_033305.2
VPS13B	NM_017890.4
VPS45	NM_007259.4
VPS53*	NM_001128159.2
VRK1	NM_003384.2
VSX2	NM_182894.2
WISP3	NM_003880.3
WNT10A	NM_025216.2
WRN*	NM_000553.4
XPA	NM_000380.3
XPC	NM_004628.4
ZBTB24	NM_014797.2
ZFYVE26	NM_015346.3
ZNF469	NM_001127464.2

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Methods

- Genomic DNA obtained from the submitted sample is enriched for targeted regions using a hybridization-based protocol, and sequenced using Illumina technology. Unless otherwise indicated, all targeted regions are sequenced with ≥50x depth or are supplemented with additional analysis. Reads are aligned to a reference sequence (GRCh37), and sequence changes are identified and interpreted in the context of a single clinically relevant transcript, indicated in the Genes analyzed section. Enrichment and analysis focus on the coding sequence of the indicated transcripts, 20bp of flanking intronic sequence, and other specific genomic regions demonstrated to be causative of disease at the time of assay design. Promoters, untranslated regions, and other non-coding regions are not otherwise interrogated. Exonic deletions and duplications are called using an in-house algorithm that determines copy number at each target by comparing the read depth for each target in the proband sequence with both mean read-depth and read-depth distribution, obtained from a set of clinical samples. Markers across the X and Y chromosomes are analyzed for quality control purposes and may detect deviations from the expected sex chromosome complement. Such deviations may be included in the report in accordance with internal guidelines. Invitae utilizes a classification methodology to identify next-generation sequencing (NGS)-detected variants that require orthogonal confirmation (Lincoln, et al. J Mol Diagn. 2019 Mar;21(2):318-329.). Confirmation of the presence and location of reportable variants is performed based on stringent criteria established by Invitae (1400 16th Street, San Francisco, CA 94103, #05D2040778), as needed, using one of several validated orthogonal approaches (PubMed ID 30610921). The following analyses are performed if relevant to the requisition. For GBA the reference genome has been modified to mask the sites of polymorphic paralog sequence variants (PSVs) in both the gene and pseudogene. For CYP21A2 and GBA, if one or more reportable variants, gene conversion, or fusion event is identified via our NGS pipeline (see Limitations), these variants are confirmed by PacBio sequencing of an amplicon generated by long-range PCR and subsequent short-range PCR. In some cases, it may not be possible to disambiguate between the gene and pseudogene. For GJB2, the reportable range includes large upstream deletions overlapping GJB6. For HBA1/2, the reference genome has been modified to force some sequencing reads derived from HBA1 to align to HBA2, and variant calling algorithms are modified to support an expectation of 4 alleles in these regions. HBA1/2 copy number calling is performed by a custom hypothesis testing algorithm which generates diplotype calls. If sequence data for a sample does not support a unique high confidence match from among hypotheses tested, that sample is flagged for manual review. Copy number variation is only reported for coding sequence of HBA1 and HBA2 and the HS-40 region. This assay does not distinguish among the -α3.7 subtypes, and all -α3.7 variants are called as HBA1 deletions. This assay may not detect overlapping copy gain and copy loss events when the breakpoints of those events are similar. For FMR1, triplet repeats are detected by PCR with fluorescently labeled primers followed by capillary electrophoresis. Reference ranges: Normal: <45 CGG repeats, intermediate: 45-54 CGG repeats, premutation: 55-200 CGG repeats, full mutation: >200 CGG repeats. For alleles with 55-90 triplet repeats, the region surrounding the FMR1 repeat is amplified by PCR. The PCR amplicons are then processed through PacBio SMRTBell library prep and sequenced using PacBio long read technology. The number of AGG interruptions within the 55-90 triplet repeat is read directly from the resulting DNA sequences. Technical component of confirmatory sequencing is performed by Invitae Corporation (1400 16th Street, San Francisco, CA 94103, #05D2040778).
- This report only includes variants that have a clinically significant association with the conditions tested as of the report date. Variants of uncertain significance, benign variants, and likely benign variants are not included in this report. However, if additional evidence becomes available to indicate that the clinical significance of a variant has changed, Invitae may update this report and provide notification.
- A PMID is a unique identifier referring to a published, scientific paper. Search by PMID at **http://www.ncbi.nlm.nih.gov/pubmed**.
- An rsID is a unique identifier referring to a single genomic position, and is used to associate population frequency information with sequence changes at that position. Reported population frequencies are derived from a number of public sites that aggregate data from large-scale population sequencing projects, including ExAC (**http://exac.broadinstitute.org**), gnomAD (**http://gnomad.broadinstitute.org**), and dbSNP (**http://ncbi.nlm.nih.gov/SNP**).

Disclaimer

DNA studies do not constitute a definitive test for the selected condition(s) in all individuals. It should be realized that there are possible sources of error. Errors can result from trace contamination, rare technical errors, rare genetic variants that interfere with analysis, recent scientific developments, and alternative classification systems. This test should be one of many aspects used by the healthcare provider to help with a diagnosis and treatment plan, but it is not a diagnosis itself. This test was developed and its performance characteristics determined by Invitae. It has not been cleared or approved by the FDA. The laboratory is regulated under the Clinical Laboratory Improvement Act (CLIA) as qualified to perform high-complexity clinical tests (CLIA ID: 05D2040778). This test is used for clinical purposes. It should not be regarded as investigational or for research.

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Limitations

- Based on validation study results, this assay achieves >99% analytical sensitivity and specificity for single nucleotide variants, insertions and deletions <15bp in length, and exon-level deletions and duplications. Invitae's methods also detect insertions and deletions larger than 15bp but smaller than a full exon but sensitivity for these may be marginally reduced. Invitae's deletion/duplication analysis determines copy number at a single exon resolution at virtually all targeted exons. However, in rare situations, single-exon copy number events may not be analyzed due to inherent sequence properties or isolated reduction in data quality. Certain types of variants, such as structural rearrangements (e.g. inversions, gene conversion events, translocations, etc.) or variants embedded in sequence with complex architecture (e.g. short tandem repeats or segmental duplications), may not be detected. Additionally, it may not be possible to fully resolve certain details about variants, such as mosaicism, phasing, or mapping ambiguity. Unless explicitly guaranteed, sequence changes in the promoter, non-coding exons, and other non-coding regions are not covered by this assay. Please consult the test definition on our website for details regarding regions or types of variants that are covered or excluded for this test. This report reflects the analysis of an extracted genomic DNA sample. While this test is intended to reflect the analysis of extracted genomic DNA from a referred patient, in very rare cases the analyzed DNA may not represent that individual's constitutional genome, such as in the case of a circulating hematolymphoid neoplasm, bone marrow transplant, blood transfusion, chimerism, culture artifact or maternal cell contamination.
- TBCE: Sequencing analysis for exons 2 includes only cds +/- 10 bp. CFTR: Sequencing analysis for exons 7 includes only cds +/- 10 bp. SMN1: Systematic exon numbering is used for all genes, including SMN1, and for this reason the exon typically referred to as exon 7 in the literature (PMID: 8838816) is referred to as exon 8 in this report. This assay unambiguously detects SMN1 exon 8 copy number. The presence of the g.27134T>G variant (also known as c.*3+80T>G) is reported if SMN1 copy number = 2. SMN1 or SMN2: NM_000344.3:c.*3+80T>G variant only. AMN: Deletion/duplication analysis is not offered for exon 1. MLC1: Sequencing analysis for exons 11 includes only cds +/- 10 bp. BBS9: Deletion/duplication analysis is not offered for exon 4. CYP21A2: Analysis includes the most common variants (c.92C>T(p.Pro31Leu), c.293-13C>G (intronic), c.332_339delGAGACTAC (p.Gly111Valfs*21), c.518T>A (p.Ile173Asn), c.710T>A (p.Ile237Asn), c.713T>A (p.Val238Glu), c.719T>A (p.Met240Lys), c.844G>T (p.Val282Leu), c.923dupT (p.Leu308Phefs*6), c.955C>T (p.Gln319*), c.1069C>T(p.Arg357Trp), c.1360C>T (p.Pro454Ser) and the 30Kb deletion) as well as select rare HGMD variants only (list available upon request). Full gene duplications are reported only in the presence of a pathogenic variant(s). When a duplication and a pathogenic variant(s) is identified, phase (cis/trans) cannot be determined. Full gene deletion analysis is not offered. Sensitivity to detect these variants, if they result from complex gene conversion/fusion events, may be reduced. PTPRC: Sequencing analysis is not offered for exons 3, 15. COL11A2: Deletion/duplication analysis is not offered for exon 36. USH1C: Deletion/duplication analysis is not offered for exons 5-6. VPS53: Sequencing analysis for exons 14 includes only cds +/- 5 bp. DUOX2: Deletion/duplication and sequencing analysis is not offered for exons 6-7. FANCD2: Deletion/duplication analysis is not offered for exons 14-17, 22 and sequencing analysis is not offered for exons 15-17. Sequencing analysis for exons 6, 14, 18, 20, 23, 25, 34 includes only cds +/- 10 bp. EYS: Sequencing analysis for exons 30 includes only cds +/- 0 bp. GNE: Sequencing analysis for exons 8 includes only cds +/- 10 bp. HBB: Sequencing analysis is not offered for exons 2-3. LIFR: Sequencing analysis for exons 3 includes only cds +/- 5 bp. OAT: Deletion/duplication analysis is not offered for exon 2. PEX1: Sequencing analysis for exons 16 includes only cds +/- 0 bp. PKHD1: Deletion/duplication analysis is not offered for exon 13. TSFM: Sequencing analysis is not offered for exon 5. GALE: Sequencing analysis for exons 10 includes only cds +/- 5 bp. TYR: Deletion/duplication and sequencing analysis is not offered for exon 5. ANO10: Sequencing analysis for exons 8 includes only cds +/- 0 bp. FANCL: Sequencing analysis for exons 4, 10 includes only cds +/- 10 bp. GHR: Deletion/duplication and sequencing analysis is not offered for exon 3. FAH: Deletion/duplication analysis is not offered for exon 14. FH: Sequencing analysis for exons 9 includes only cds +/- 10 bp. WRN: Deletion/duplication analysis is not offered for exons 10-11. Sequencing analysis for exons 8, 10-11 includes only cds +/- 10 bp. VPS13A: Deletion/duplication analysis is not offered for exons 2-3, 27-28. NEB: Deletion/duplication analysis is not offered for exons 82-105. NEB variants in this region with no evidence towards pathogenicity are not included in this report, but are available upon request. ATM: Sequencing analysis for exons 6, 24, 43 includes only cds +/- 10 bp. ABCC2: Deletion/duplication analysis is not offered for exons 24-25. OTOA: Deletion/duplication and sequencing analysis is not offered for exons 20-28. TG: Deletion/duplication analysis is not offered for exon 18. Sequencing analysis for exons 44 includes only cds +/- 0 bp. ATP8B1: Sequencing analysis for exons 19 includes only cds +/- 10 bp. AIPL1: Sequencing analysis for exons 2 includes only cds +/- 10 bp. DDX11: NM_030653.3:c.1763-1G>C variant only. GALC: Deletion/duplication analysis is not offered for exon 6. GBA: c.84dupG (p.Leu29Alafs*18), c.115+1G>A (Splice donor), c.222_224delTAC (p.Thr75del), c.475C>T (p.Arg159Trp), c.595_596delCT (p.Leu199Aspfs*62), c.680A>G (p.Asn227Ser), c.721G>A (p.Gly241Arg), c.754T>A (p.Phe252Ile), c.1226A>G (p.Asn409Ser), c.1246G>A (p.Gly416Ser), c.1263_1317del (p.Leu422Profs*4), c.1297G>T (p.Val433Leu), c.1342G>C (p.Asp448His), c.1343A>T (p.Asp448Val), c.1448T>C (p.Leu483Pro), c.1504C>T (p.Arg502Cys), c.1505G>A (p.Arg502His), c.1603C>T (p.Arg535Cys), c.1604G>A (p.Arg535His) variants only. Rarely, sensitivity to detect these variants may be reduced. When sensitivity is reduced, zygosity may be reported as "unknown". HBA1/2: This assay is designed to detect deletions and duplications of HBA1 and/or HBA2, resulting from the -alpha20.5, --MED, --SEA, --FIL/--THAI, -alpha3.7, -alpha4.2, anti3.7 and anti4.2. Sensitivity to detect other copy number variants may be reduced. Detection of overlapping deletion and duplication events will be limited to combinations of events with significantly differing boundaries. In addition, deletion of the enhancer element HS-40 and the sequence variant,

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Constant Spring (NM_000517.4:c.427T>C), can be identified by this assay. MTHFR: The NM_005957.4:c.665C>T (p.Ala222Val) (aka 677C>T) and c.1286A>C (p.Glu429Ala) (aka 1298A>C) variants are not reported in our primary report.

This report has been reviewed and approved by:

Matteo Vatta, Ph.D., FACMG
Clinical Molecular Geneticist

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CARRIER DETECTION RATES AND RESIDUAL RISKS

This table is relevant to patient report RQ4539976
Issue date: 02/19/2023

This table displays residual risks after a negative result for each of the genes and corresponding disorders. The values provided assume a negative family history and the absence of symptoms for each disorder. For genes associated with both dominant and recessive inheritance, the numbers in this table apply to the recessive condition(s) associated with the gene, unless otherwise noted. Residual risk values are provided for disorders when carrier frequency is greater than 1 in 500. For disorders with carrier frequency equal to, or less than, 1 in 500, residual risk is considered to be reduced substantially. When provided, residual risk values are inferred from published carrier frequencies, and estimated detection rates are based on testing technologies used at Invitae. Residual risks are provided only as a guide for assessing approximate risk given a negative result; values may vary based on the ethnic background(s) of an individual. For any genes marked with an asterisk*, refer to the Limitations section of the patient report for detailed coverage information. In the case of a sample-specific limitation, "N/A" indicates that a residual risk value could not be calculated. AR = autosomal recessive, XL = X-linked, AD = autosomal dominant.

DISORDER (INHERITANCE)	GENE	ETHNICITY	CARRIER FREQUENCY	DETECTION RATE	RISK TO BE A CARRIER AFTER NEGATIVE RESULT
3-hydroxy-3-methylglutaryl-CoA lyase deficiency (AR) NM_000191.2	HMGCL	Pan-ethnic	≤1 in 500	99%	Reduced
17-beta hydroxysteroid dehydrogenase 3 deficiency (AR) NM_000197.1	HSD17B3	Pan-ethnic	≤1 in 500	99%	Reduced
ABCA3-related conditions (AR) NM_001089.2	ABCA3	Pan-ethnic	1 in 277	99%	1 in 27600
ABCA4-related conditions (AR) NM_000350.2	ABCA4	Pan-ethnic	1 in 45	90%	1 in 441
ABCB4-related conditions (AR) NM_000443.3	ABCB4	Pan-ethnic	1 in 204	99%	1 in 20300
ABCB11-related conditions (AR) NM_003742.2	ABCB11	Pan-ethnic	1 in 100	99%	1 in 9900
ABCC8-related conditions (AR) NM_000352.4 When the mother is a noncarrier, but the father is a carrier, there is a residual risk for focal disease (1 in 540 for the Ashkenazi Jewish population; undetermined in other ethnic groups)	ABCC8	Pan-ethnic	1 in 177	99%	1 in 17600
Abetalipoproteinemia (AR) NM_000253.3	MTTP	Pan-ethnic	≤1 in 500	99%	Reduced
Achromatopsia (CNGB3-related) (AR) NM_019098.4	CNGB3	Pan-ethnic	1 in 93	99%	1 in 9200
ACOX1-related conditions (AR) NM_004035.6	ACOX1	Pan-ethnic	≤1 in 500	99%	Reduced
Acrodermatitis enteropathica (AR) NM_130849.3	SLC39A4	Pan-ethnic	1 in 354	99%	1 in 35300
Adenosine deaminase deficiency (AR) NM_000022.2	ADA	Pan-ethnic	1 in 224	92%	1 in 2788
ADGRV1-related conditions (AR) NM_032119.3	ADGRV1	Pan-ethnic	1 in 223	99%	1 in 22200
AHI1-related conditions (AR) NM_017651.4	AHI1	Pan-ethnic	1 in 447	99%	1 in 44600
Aicardi-Goutieres syndrome 2 (AR) NM_024570.3	RNASEH2B	Pan-ethnic	≤1 in 500	99%	Reduced
Aicardi-Goutieres syndrome 3 (AR) NM_032193.3	RNASEH2C	Pan-ethnic	≤1 in 500	99%	Reduced
Aicardi-Goutieres syndrome 4 (AR) NM_006397.2	RNASEH2A	Pan-ethnic	≤1 in 500	99%	Reduced
Aicardi-Goutieres syndrome 5 (AR) NM_015474.3	SAMHD1	Pan-ethnic	≤1 in 500	99%	Reduced
AIPL1-related conditions (AR) NM_014336.4	AIPL1 *	Pan-ethnic	1 in 408	99%	1 in 40700
Aldosterone synthase deficiency (AR) NM_000498.3	CYP11B2	Pan-ethnic	≤1 in 500	99%	Reduced
Alpha-mannosidosis (AR) NM_000528.3	MAN2B1	Pan-ethnic	1 in 354	99%	1 in 35300
Alpha-N-acetylgalactosaminidase deficiency (AR) NM_000262.2	NAGA	Pan-ethnic	≤1 in 500	99%	Reduced
Alpha-thalassemia (AR) NM_000558.4, NM_000517.4	HBA1/ HBA2 *	African-American	1 in 30	90%	1 in 291
		Asian	1 in 20	90%	1 in 191
		Caucasian	≤1 in 500	90%	Reduced

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DISORDER (INHERITANCE)	GENE	ETHNICITY	CARRIER FREQUENCY	DETECTION RATE	RISK TO BE A CARRIER AFTER NEGATIVE RESULT
		Pan-ethnic	1 in 25	90%	1 in 241
Alport syndrome (COL4A3-related) (AR) NM_000091.4	COL4A3	Pan-ethnic	1 in 354	99%	1 in 35300
Alport syndrome (COL4A4-related) (AR) NM_000092.4	COL4A4	Pan-ethnic	1 in 353	99%	1 in 35200
Alström syndrome (AR) NM_015120.4	ALMS1	Pan-ethnic	≤1 in 500	99%	Reduced
Arginase deficiency (AR) NM_000045.3	ARG1	Pan-ethnic	1 in 274	99%	1 in 27300
Argininosuccinate lyase deficiency (AR) NM_000048.3	ASL	Pan-ethnic	1 in 133	90%	1 in 1321
ARL6-related conditions (AR) NM_177976.2	ARL6	Pan-ethnic	≤1 in 500	99%	Reduced
Aromatase deficiency (AR) NM_031226.2	CYP19A1	Pan-ethnic	≤1 in 500	99%	Reduced
Asparagine synthetase deficiency (AR) NM_133436.3	ASNS	Pan-ethnic	≤1 in 500	99%	Reduced
Aspartylglucosaminuria (AR) NM_000027.3	AGA	Pan-ethnic	≤1 in 500	99%	Reduced
Ataxia with vitamin E deficiency (AR) NM_000370.3	TTPA	Pan-ethnic	≤1 in 500	90%	Reduced
Ataxia-telangiectasia-like disorder (AR) NM_005591.3	MRE11	Pan-ethnic	≤1 in 500	99%	Reduced
ATM-related conditions (AR) NM_000051.3	ATM *	Pan-ethnic	1 in 100	99%	1 in 9900
ATP8B1-related conditions (AR) NM_005603.4	ATP8B1 *	Pan-ethnic	1 in 112	99%	1 in 11100
Atransferrinemia (AR) NM_001063.3	TF	Pan-ethnic	≤1 in 500	99%	Reduced
Autoimmune polyendocrinopathy with candidiasis and ectodermal dysplasia (AR) NM_000383.3	AIRE	Pan-ethnic	1 in 150	99%	1 in 14900
Autosomal recessive congenital ichthyosis (ABCA12-related) (AR) NM_173076.2	ABCA12	Pan-ethnic	≤1 in 500	99%	Reduced
Autosomal recessive congenital ichthyosis (TGM1-related) (AR) NM_000359.2	TGM1	Pan-ethnic	1 in 224	95%	1 in 4460
Autosomal recessive spastic ataxia of Charlevoix-Saguenay (AR) NM_014363.5	SACS	Pan-ethnic	≤1 in 500	99%	Reduced
Bardet-Biedl syndrome (BBS7-related) (AR) NM_176824.2	BBS7	Pan-ethnic	≤1 in 500	99%	Reduced
Bardet-Biedl syndrome (BBS9-related) (AR) NM_198428.2	BBS9 *	Pan-ethnic	≤1 in 500	99%	Reduced
Bardet-Biedl syndrome (BBS10-related) (AR) NM_024685.3	BBS10	Pan-ethnic	1 in 354	99%	1 in 35300
Bardet-Biedl syndrome (BBS12-related) (AR) NM_152618.2	BBS12	Pan-ethnic	≤1 in 500	99%	Reduced
Bartter syndrome type 1 (AR) NM_000338.2	SLC12A1	Pan-ethnic	1 in 224	99%	1 in 22300
Bartter syndrome type 2 (AR) NM_000220.4	KCNJ1	Pan-ethnic	≤1 in 500	99%	Reduced
BBS1-related conditions (AR) NM_024649.4	BBS1	Pan-ethnic	1 in 330	99%	1 in 32900
BBS2-related conditions (AR) NM_031885.3	BBS2	Pan-ethnic	≤1 in 500	99%	Reduced
BBS4-related conditions (AR) NM_033028.4	BBS4	Pan-ethnic	≤1 in 500	99%	Reduced
BBS5-related conditions (AR) NM_152384.2	BBS5	Pan-ethnic	≤1 in 500	99%	Reduced
BCS1L-related conditions (AR) NM_004328.4	BCS1L	Pan-ethnic	≤1 in 500	99%	Reduced

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CARRIER DETECTION RATES AND RESIDUAL RISKS

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DISORDER (INHERITANCE)	GENE	ETHNICITY	CARRIER FREQUENCY	DETECTION RATE	RISK TO BE A CARRIER AFTER NEGATIVE RESULT
Beta-ketothiolase deficiency (AR) NM_000019.3	ACAT1	Pan-ethnic	≤1 in 500	99%	Reduced
Beta-mannosidosis (AR) NM_005908.3	MANBA	Pan-ethnic	≤1 in 500	99%	Reduced
Biopterin-deficient hyperphenylalaninemia (PCBD1-related) (AR) NM_000281.3	PCBD1	Pan-ethnic	≤1 in 500	99%	Reduced
Biopterin-deficient hyperphenylalaninemia (PTS-related) (AR) NM_000317.2	PTS	Pan-ethnic	1 in 433	99%	1 in 43200
Biopterin-deficient hyperphenylalaninemia (QDPR-related) (AR) NM_000320.2	QDPR	Pan-ethnic	≤1 in 500	99%	Reduced
Biotin-responsive basal ganglia disease (AR) NM_025243.3	SLC19A3	Pan-ethnic	≤1 in 500	99%	Reduced
Biotinidase deficiency (AR) NM_000060.3	BTD	Pan-ethnic	1 in 125	99%	1 in 12400
Bloom syndrome (AR) NM_000057.3	BLM	Pan-ethnic	≤1 in 500	99%	Reduced
BRIP1-related conditions (AR) NM_032043.2	BRIP1	Pan-ethnic	≤1 in 500	99%	Reduced
Brittle cornea syndrome (PRDM5-related) (AR) NM_018699.3	PRDM5	Pan-ethnic	≤1 in 500	99%	Reduced
Brittle cornea syndrome (ZNF469-related) (AR) NM_001127464.2	ZNF469	Pan-ethnic	≤1 in 500	99%	Reduced
BSND-related conditions (AR) NM_057176.2	BSND	Pan-ethnic	≤1 in 500	99%	Reduced
Canavan disease (AR) NM_000049.2	ASPA	Pan-ethnic	1 in 159	99%	1 in 15800
Carbamoyl phosphate synthetase I deficiency (AR) NM_001875.4	CPS1	Pan-ethnic	≤1 in 500	99%	Reduced
Cardioencephalomyopathy (AR) NM_005138.2	SCO2	Pan-ethnic	1 in 387	99%	1 in 38600
Carnitine palmitoyltransferase I deficiency (AR) NM_001876.3	CPT1A	Pan-ethnic	≤1 in 500	99%	Reduced
Carnitine palmitoyltransferase II deficiency (AR) NM_000098.2	CPT2	Pan-ethnic	1 in 182	99%	1 in 18100
Carnitine-acylcarnitine translocase deficiency (AR) NM_000387.5	SLC25A20	Pan-ethnic	≤1 in 500	99%	Reduced
Carpenter syndrome (RAB23-related) (AR) NM_183227.2	RAB23	Pan-ethnic	≤1 in 500	99%	Reduced
Cartilage-hair hypoplasia-anauxetic dysplasia spectrum disorders (AR) NR_003051.3	RMRP	Pan-ethnic	≤1 in 500	99%	Reduced
Catecholaminergic polymorphic ventricular tachycardia (CASQ2-related) (AR) NM_001232.3	CASQ2	Pan-ethnic	1 in 224	99%	1 in 22300
CC2D2A-related conditions (AR) NM_001080522.2	CC2D2A	Pan-ethnic	1 in 426	99%	1 in 42500
CDH23-related conditions (AR) NM_022124.5	CDH23	Pan-ethnic	1 in 202	95%	1 in 4020
CEP290-related conditions (AR) NM_025114.3	CEP290	Pan-ethnic	1 in 185	99%	1 in 18400
Cerebellar ataxia, intellectual disability, and dysequilibrium syndrome 1 (AR) NM_003383.4	VLDLR	Pan-ethnic	≤1 in 500	99%	Reduced
Cerebral dysgenesis, neuropathy, ichthyosis, and keratoderma (AR) NM_004782.3	SNAP29	Pan-ethnic	≤1 in 500	99%	Reduced
Cerebrotendinous xanthomatosis (AR) NM_000784.3	CYP27A1	Pan-ethnic	1 in 112	98%	1 in 5550
CERKL-related conditions (AR) NM_001030311.2	CERKL	Pan-ethnic	1 in 137	99%	1 in 13600

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DISORDER (INHERITANCE)	GENE	ETHNICITY	CARRIER FREQUENCY	DETECTION RATE	RISK TO BE A CARRIER AFTER NEGATIVE RESULT
CFTR-related conditions (AR) NM_000492.3	CFTR *	Pan-ethnic - classic CF	1 in 45	99%	1 in 4400
		Pan-ethnic - classic CF and CFTR-related disorders	1 in 9	99%	1 in 800
Charcot-Marie-Tooth disease type 4D (AR) NM_006096.3	NDRG1	Pan-ethnic	≤1 in 500	99%	Reduced
Chediak-Higashi syndrome (AR) NM_000081.3	LYST	Pan-ethnic	≤1 in 500	99%	Reduced
Childhood-onset dystonia with optic atrophy and basal ganglia abnormalities (AR) NM_016011.3	MECR	Pan-ethnic	≤1 in 500	99%	Reduced
Chorea-acanthocytosis (AR) NM_033305.2	VPS13A *	Pan-ethnic	≤1 in 500	97%	Reduced
Chronic granulomatous disease (CYBA-related) (AR) NM_000101.3	CYBA	Pan-ethnic	≤1 in 500	99%	Reduced
Chronic granulomatous disease (NCF2-related) (AR) NM_000433.3	NCF2	Pan-ethnic	≤1 in 500	99%	Reduced
Citrin deficiency (AR) NM_014251.2	SLC25A13	Pan-ethnic	1 in 313	99%	1 in 31200
Citrullinemia type 1 (AR) NM_000050.4	ASS1	Pan-ethnic	1 in 120	96%	1 in 2975
CLN3-related conditions (AR) NM_001042432.1	CLN3	Pan-ethnic	1 in 230	99%	1 in 22900
CLRN1-related conditions (AR) NM_174878.2	CLRN1	Pan-ethnic	≤1 in 500	99%	Reduced
Cobalamin C deficiency (AR) NM_015506.2	MMACHC	Pan-ethnic	1 in 123	99%	1 in 12200
Cobalamin D deficiency (AR) NM_015702.2	MMADHC	Pan-ethnic	≤1 in 500	99%	Reduced
Cobalamin F deficiency (AR) NM_018368.3	LMBRD1	Pan-ethnic	≤1 in 500	99%	Reduced
Cockayne syndrome A (AR) NM_000082.3	ERCC8	Pan-ethnic	≤1 in 500	99%	Reduced
Cockayne syndrome B (AR) NM_000124.3	ERCC6	Pan-ethnic	1 in 377	99%	1 in 37600
Cohen syndrome (AR) NM_017890.4	VPS13B	Pan-ethnic	≤1 in 500	99%	Reduced
COL11A2-related conditions (AR) NM_080680.2	COL11A2 *	Pan-ethnic	≤1 in 500	99%	Reduced
COL17A1-related conditions (AR) NM_000494.3	COL17A1	Pan-ethnic	≤1 in 500	99%	Reduced
Combined malonic and methylmalonic aciduria (AR) NM_174917.4	ACSF3	Pan-ethnic	1 in 87	99%	1 in 8600
Combined oxidative phosphorylation deficiency 1 (AR) NM_024996.5	GFM1	Pan-ethnic	≤1 in 500	99%	Reduced
Combined oxidative phosphorylation deficiency 3 (AR) NM_001172696.1	TSFM *	Pan-ethnic	≤1 in 500	93%	Reduced
Combined pituitary hormone deficiency (LHX3-related) (AR) NM_014564.4	LHX3	Pan-ethnic	≤1 in 500	99%	Reduced
Combined pituitary hormone deficiency (POU1F1-related) (AR) NM_000306.3	POU1F1	Pan-ethnic	≤1 in 500	99%	Reduced
Combined pituitary hormone deficiency (PROP1-related) (AR) NM_006261.4	PROP1	Pan-ethnic	1 in 45	98%	1 in 2200
Congenital adrenal hyperplasia due to 3-beta-hydroxysteroid dehydrogenase deficiency (AR) NM_000198.3	HSD3B2	Pan-ethnic	≤1 in 500	99%	Reduced
Congenital adrenal hyperplasia due to 21-hydroxylase deficiency (AR) NM_000500.7	CYP21A2 *	Pan-ethnic	1 in 61	92%	1 in 751
Congenital adrenal insufficiency (AR) NM_000781.2	CYP11A1	Pan-ethnic	≤1 in 500	99%	Reduced

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Congenital chronic diarrhea (DGAT1-related) (AR) NM_012079.5	DGAT1	Pan-ethnic	≤1 in 500	99%	Reduced
Congenital disorder of glycosylation (SLC35A3-related) (AR) NM_012243.2	SLC35A3	Pan-ethnic	≤1 in 500	99%	Reduced
Congenital disorder of glycosylation type Ia (AR) NM_000303.2	PMM2	Pan-ethnic	1 in 190	99%	1 in 18900
Congenital disorder of glycosylation type Ib (AR) NM_002435.2	MPI	Pan-ethnic	≤1 in 500	99%	Reduced
Congenital disorder of glycosylation type Ic (AR) NM_013339.3	ALG6	Pan-ethnic	≤1 in 500	99%	Reduced
Congenital disorder of glycosylation type Ik (AR) NM_019109.4	ALG1	Pan-ethnic	≤1 in 500	99%	Reduced
Congenital disorder of glycosylation type Iv (AR) NM_018297.3	NGLY1	Pan-ethnic	≤1 in 500	99%	Reduced
Congenital dyserythropoietic anemia type II (AR) NM_006363.4	SEC23B	Pan-ethnic	≤1 in 500	99%	Reduced
Congenital hydrocephalus-1 (AR) NM_001080414.3	CCDC88C	Pan-ethnic	≤1 in 500	99%	Reduced
Congenital hypothyroidism (TSHB-related) (AR) NM_000549.4	TSHB	Pan-ethnic	≤1 in 500	99%	Reduced
Congenital insensitivity to pain with anhidrosis (AR) NM_001012331.1	NTRK1	Pan-ethnic	≤1 in 500	99%	Reduced
Congenital myasthenic syndrome (CHAT-related) (AR) NM_020549.4	CHAT	Pan-ethnic	≤1 in 500	99%	Reduced
Congenital myasthenic syndrome (CHRNE-related) (AR) NM_000080.3	CHRNE	Pan-ethnic	1 in 200	99%	1 in 19900
Congenital nephrotic syndrome type 1 (AR) NM_004646.3	NPHS1	Pan-ethnic	≤1 in 500	99%	Reduced
Congenital nephrotic syndrome type 2 (AR) NM_014625.3	NPHS2	Pan-ethnic	≤1 in 500	99%	Reduced
Congenital secretory chloride diarrhea (AR) NM_000111.2	SLC26A3	Pan-ethnic	≤1 in 500	99%	Reduced
Corneal dystrophy and perceptive deafness (AR) NM_032034.3	SLC4A11	Pan-ethnic	≤1 in 500	99%	Reduced
CRB1-related conditions (AR) NM_201253.2	CRB1	Pan-ethnic	1 in 112	99%	1 in 11100
CTSC-related conditions (AR) NM_001814.5	CTSC	Pan-ethnic	1 in 250	99%	1 in 24900
CYP1B1-related conditions (AR) NM_000104.3	CYP1B1	Pan-ethnic	1 in 79	99%	1 in 7800
CYP7B1-related conditions (AR) NM_004820.3	CYP7B1	Pan-ethnic	≤1 in 500	99%	Reduced
CYP11B1-related conditions (AR) NM_000497.3	CYP11B1	Pan-ethnic	1 in 194	99%	1 in 19300
CYP17A1-related conditions (AR) NM_000102.3	CYP17A1	Pan-ethnic	≤1 in 500	99%	Reduced
Cystinosis (AR) NM_004937.2	CTNS	Pan-ethnic	1 in 158	99%	1 in 15700
Cytochrome P450 oxidoreductase deficiency (AR) NM_000941.2	POR	Pan-ethnic	1 in 158	99%	1 in 15700
Desbuquois dysplasia type 1 (AR) NM_138793.3	CANT1	Pan-ethnic	≤1 in 500	99%	Reduced
Developmental and epileptic encephalopathy (CAD-related) (AR) NM_004341.4	CAD	Pan-ethnic	≤1 in 500	99%	Reduced
DGUOK-related conditions (AR) NM_080916.2	DGUOK	Pan-ethnic	≤1 in 500	99%	Reduced
DHDDS-related conditions (AR) NM_024887.3	DHDDS	Pan-ethnic	≤1 in 500	99%	Reduced
Dihydrolipoamide dehydrogenase deficiency (AR) NM_000108.4	DLD	Pan-ethnic	≤1 in 500	99%	Reduced

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Distal renal tubular acidosis with deafness (ATP6V1B1-related) (AR) NM_001692.3	ATP6V1B1	Pan-ethnic	≤1 in 500	99%	Reduced
DOK7-related conditions (AR) NM_173660.4	DOK7	Pan-ethnic	1 in 115	99%	1 in 11400
Donnai-Barrow syndrome (AR) NM_004525.2	LRP2	Pan-ethnic	≤1 in 500	99%	Reduced
Dubin-Johnson syndrome (AR) NM_000392.4	ABCC2 *	Pan-ethnic	≤1 in 500	99%	Reduced
DUOX2-related conditions (AR) NM_014080.4	DUOX2 *	Pan-ethnic	1 in 58	91%	1 in 634
DYNC2H1-related conditions (AR) NM_001080463.1	DYNC2H1	Pan-ethnic	1 in 224	99%	1 in 22300
DYSF-related conditions (AR) NM_003494.3	DYSF	Pan-ethnic	1 in 311	99%	1 in 31000
Dyskeratosis congenita spectrum disorders (RTEL1-related) (AR) NM_001283009.1	RTEL1	Pan-ethnic	≤1 in 500	99%	Reduced
Dyskeratosis congenita spectrum disorders (TERT-related) (AR) NM_198253.2	TERT	Pan-ethnic	≤1 in 500	99%	Reduced
Dystrophic epidermolysis bullosa (AR) NM_000094.3	COL7A1	Pan-ethnic	1 in 370	97%	1 in 12300
Ehlers-Danlos syndrome, dermatosparaxis type (AR) NM_014244.4	ADAMTS2	Pan-ethnic	≤1 in 500	99%	Reduced
Ehlers-Danlos syndrome, kyphoscoliotic type (AR) NM_000302.3	PLOD1	Pan-ethnic	1 in 150	99%	1 in 14900
Ellis-van Creveld syndrome (EVC-related) (AR) NM_153717.2	EVC	Pan-ethnic	1 in 220	99%	1 in 21900
Epidermolysis bullosa with pyloric atresia (ITGB4-related) (AR) NM_001005731.2	ITGB4	Pan-ethnic	1 in 393	99%	1 in 39200
Epimerase deficiency galactosemia (AR) NM_000403.3	GALE *	Pan-ethnic	≤1 in 500	99%	Reduced
ERCC2-related conditions (AR) NM_000400.3	ERCC2	Pan-ethnic	≤1 in 500	99%	Reduced
Ethylmalonic encephalopathy (AR) NM_014297.3	ETHE1	Pan-ethnic	≤1 in 500	99%	Reduced
EVC2-related conditions (AR) NM_147127.4	EVC2	Pan-ethnic	1 in 199	99%	1 in 19800
Familial chylomicronemia syndrome (AR) NM_000237.2	LPL	Pan-ethnic	≤1 in 500	99%	Reduced
Familial dysautonomia (AR) NM_003640.3	ELP1	Pan-ethnic	≤1 in 500	99%	Reduced
Familial hemophagocytic lymphohistiocytosis type 2 (AR) NM_001083116.1	PRF1	Pan-ethnic	1 in 177	99%	1 in 17600
Familial hemophagocytic lymphohistiocytosis type 3 (AR) NM_199242.2	UNC13D	Pan-ethnic	1 in 177	93%	1 in 2515
Familial hemophagocytic lymphohistiocytosis type 4 (AR) NM_003764.3	STX11	Pan-ethnic	1 in 224	99%	1 in 22300
Familial hemophagocytic lymphohistiocytosis type 5 (AR) NM_006949.3	STXBP2	Pan-ethnic	1 in 224	99%	1 in 22300
Familial hypercholesterolemia (LDLR-related) (AD) NM_000527.4	LDLR	Pan-ethnic	1 in 250	99%	1 in 24900
Familial hypercholesterolemia (LDLRAP1-related) (AR) NM_015627.2	LDLRAP1	Pan-ethnic	≤1 in 500	99%	Reduced
Fanconi anemia type A (AR) NM_000135.2	FANCA	Pan-ethnic	1 in 345	99%	1 in 34400
Fanconi anemia type C (AR) NM_000136.2	FANCC	Pan-ethnic	1 in 417	99%	1 in 41600
Fanconi anemia type D2 (AR) NM_033084.3	FANCD2 *	Pan-ethnic	≤1 in 500	94%	Reduced

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Fanconi anemia type E (AR) NM_021922.2	FANCE	Pan-ethnic	≤1 in 500	99%	Reduced
Fanconi anemia type G (AR) NM_004629.1	FANCG	Pan-ethnic	≤1 in 500	99%	Reduced
Fanconi anemia type I (AR) NM_001113378.1	FANCI	Pan-ethnic	≤1 in 500	99%	Reduced
Fanconi anemia type L (AR) NM_018062.3	FANCL *	Pan-ethnic	≤1 in 500	99%	Reduced
FH-related conditions (AR) NM_000143.3	FH *	Pan-ethnic	≤1 in 500	99%	Reduced
FKBP10-related conditions (AR) NM_021939.3	FKBP10	Pan-ethnic	≤1 in 500	99%	Reduced
Foveal hypoplasia (SLC38A8-related) (AR) NM_001080442.2	SLC38A8	Pan-ethnic	≤1 in 500	99%	Reduced
FOXN1-related conditions (AR) NM_003593.2	FOXN1	Pan-ethnic	≤1 in 500	99%	Reduced
Fraser syndrome (FRAS1-related) (AR) NM_025074.6	FRAS1	Pan-ethnic	1 in 316	99%	1 in 31500
Fraser syndrome (FREM2-related) (AR) NM_207361.5	FREM2	Pan-ethnic	≤1 in 500	99%	Reduced
Fraser syndrome (GRIP1-related) (AR) NM_021150.3	GRIP1	Pan-ethnic	1 in 447	99%	1 in 44600
Fructose-1,6-bisphosphatase deficiency (AR) NM_000507.3	FBP1	Pan-ethnic	≤1 in 500	99%	Reduced
Fucosidosis (AR) NM_000147.4	FUCA1	Pan-ethnic	≤1 in 500	99%	Reduced
Galactokinase deficiency galactosemia (AR) NM_000154.1	GALK1	Pan-ethnic	1 in 122	99%	1 in 12100
Galactosemia (GALT-related) (AR) NM_000155.3	GALT	Pan-ethnic	1 in 100	99%	1 in 9900
Galactosialidosis (AR) NM_000308.3	CTSA	Pan-ethnic	≤1 in 500	99%	Reduced
GATM-related conditions (AR) NM_001482.2	GATM	Pan-ethnic	≤1 in 500	99%	Reduced
GBA-related conditions including Gaucher disease (AR) NM_001005741.2	GBA *	Ashkenazi Jewish	1 in 15	94%	1 in 234
		Pan-ethnic	1 in 158	72%	1 in 561
GBE1-related conditions (AR) NM_000158.3	GBE1	Pan-ethnic	1 in 387	99%	1 in 38600
GCH1-related conditions (AR) NM_000161.2	GCH1	Pan-ethnic	≤1 in 500	99%	Reduced
GDF5-related conditions (AR) NM_000557.4	GDF5	Pan-ethnic	≤1 in 500	99%	Reduced
Geroderma osteodysplastica (AR) NM_152281.2	GORAB	Pan-ethnic	≤1 in 500	99%	Reduced
GHR-related conditions (AR) NM_000163.4	GHR *	Pan-ethnic	≤1 in 500	98%	Reduced
Gitelman syndrome (AR) NM_000339.2	SLC12A3	Pan-ethnic	1 in 100	99%	1 in 9900
GJB2-related conditions (AR) NM_004004.5	GJB2	Pan-ethnic	1 in 50	99%	1 in 4900
GLB1-related conditions (AR) NM_000404.2	GLB1	Pan-ethnic	1 in 158	99%	1 in 15700
GLE1-related conditions (AR) NM_001003722.1	GLE1	Pan-ethnic	≤1 in 500	99%	Reduced
Glutaric acidemia type I (AR) NM_000159.3	GCDH	Pan-ethnic	1 in 87	99%	1 in 8600
Glutaric acidemia type IIA (AR) NM_000126.3	ETFA	Pan-ethnic	≤1 in 500	99%	Reduced
Glutaric acidemia type IIB (AR) NM_001985.2	ETFB	Pan-ethnic	≤1 in 500	99%	Reduced
Glutaric acidemia type IIC (AR) NM_004453.3	ETFDH	Pan-ethnic	1 in 250	99%	1 in 24900

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Glutathione synthetase deficiency (AR) NM_000178.2	GSS	Pan-ethnic	≤1 in 500	99%	Reduced
Glycine encephalopathy (AMT-related) (AR) NM_000481.3	AMT	Pan-ethnic	1 in 325	99%	1 in 32400
Glycine encephalopathy (GLDC-related) (AR) NM_000170.2	GLDC	Pan-ethnic	1 in 165	99%	1 in 16400
Glycogen storage disease type Ia (AR) NM_000151.3	G6PC	Pan-ethnic	1 in 177	95%	1 in 3520
Glycogen storage disease type II (Pompe disease) (AR) NM_000152.3	GAA	Pan-ethnic	1 in 100	99%	1 in 9900
Glycogen storage disease type III (AR) NM_000642.2	AGL	Pan-ethnic	1 in 159	95%	1 in 3160
Glycogen storage disease type IXb (AR) NM_000293.2	PHKB	Pan-ethnic	≤1 in 500	99%	Reduced
Glycogen storage disease type IXc (AR) NM_000294.2	PHKG2	Pan-ethnic	≤1 in 500	99%	Reduced
Glycogen storage disease type V (AR) NM_005609.3	PYGM	Pan-ethnic	1 in 171	99%	1 in 17000
Glycogen storage disease type VII (AR) NM_000289.5	PFKM	Pan-ethnic	≤1 in 500	99%	Reduced
GM3 synthase deficiency (AR) NM_003896.3	ST3GAL5	Pan-ethnic	≤1 in 500	99%	Reduced
GNE-related conditions (AR) NM_001128227.2	GNE *	Pan-ethnic	1 in 179	99%	1 in 17800
GNPTAB-related conditions (AR) NM_024312.4	GNPTAB	Pan-ethnic	1 in 200	99%	1 in 19900
Guanidinoacetate methyltransferase deficiency (AR) NM_000156.5	GAMT	Pan-ethnic	≤1 in 500	99%	Reduced
GUCY2D-related conditions (AR) NM_000180.3	GUCY2D	Pan-ethnic	1 in 204	99%	1 in 20300
Gyrate atrophy of the choroid and retina (AR) NM_000274.3	OAT *	Pan-ethnic	≤1 in 500	99%	Reduced
HADHA-related conditions (AR) NM_000182.4	HADHA	Pan-ethnic	1 in 350	99%	1 in 34900
HBB-related hemoglobinopathies (AR) NM_000518.4	HBB *	Pan-ethnic	1 in 49	99%	1 in 4800
Heme oxygenase 1 deficiency (AR) NM_002133.2	HMOX1	Pan-ethnic	≤1 in 500	99%	Reduced
Hemolytic anemia, CD59-mediated (AR) NM_203330.2	CD59	Pan-ethnic	≤1 in 500	99%	Reduced
Hereditary fructose intolerance (AR) NM_000035.3	ALDOB	Pan-ethnic	1 in 122	99%	1 in 12100
Hereditary hemochromatosis type 2 (HAMP-related) (AR) NM_021175.2	HAMP	Pan-ethnic	≤1 in 500	99%	Reduced
Hereditary hemochromatosis type 2 (HJV-related) (AR) NM_213653.3	HJV	Pan-ethnic	≤1 in 500	99%	Reduced
Hereditary hemochromatosis type 3 (AR) NM_003227.3	TFR2	Pan-ethnic	≤1 in 500	99%	Reduced
Hermansky-Pudlak syndrome type 1 (AR) NM_000195.4	HPS1	Pan-ethnic	≤1 in 500	99%	Reduced
Hermansky-Pudlak syndrome type 3 (AR) NM_032383.4	HPS3	Pan-ethnic	≤1 in 500	99%	Reduced
Hermansky-Pudlak syndrome type 4 (AR) NM_022081.5	HPS4	Pan-ethnic	≤1 in 500	99%	Reduced
Hermansky-Pudlak syndrome type 5 (AR) NM_181507.1	HPS5	Pan-ethnic	≤1 in 500	99%	Reduced
Hermansky-Pudlak syndrome type 6 (AR) NM_024747.5	HPS6	Pan-ethnic	≤1 in 500	99%	Reduced
Hermansky-Pudlak syndrome type 8 (AR) NM_212550.4	BLOC1S3	Pan-ethnic	≤1 in 500	99%	Reduced
Hermansky-Pudlak syndrome type 9 (AR) NM_012388.3	BLOC1S6	Pan-ethnic	≤1 in 500	99%	Reduced

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HGSNAT-related conditions (AR) NM_152419.2	HGSNAT	Pan-ethnic	≤1 in 500	99%	Reduced
Holocarboxylase synthetase deficiency (AR) NM_000411.6	HLCS	Pan-ethnic	1 in 224	99%	1 in 22300
Homocystinuria due to cobalamin E deficiency (AR) NM_002454.2	MTRR	Pan-ethnic	≤1 in 500	99%	Reduced
Homocystinuria due to cobalamin G deficiency (AR) NM_000254.2	MTR	Pan-ethnic	≤1 in 500	99%	Reduced
Homocystinuria due to cystathionine beta-synthase deficiency (AR) NM_000071.2	CBS	Pan-ethnic	1 in 224	99%	1 in 22300
Homocystinuria due to MTHFR deficiency (AR) NM_005957.4	MTHFR *	Pan-ethnic	≤1 in 500	99%	Reduced
HSD17B4-related conditions (AR) NM_000414.3	HSD17B4	Pan-ethnic	1 in 158	99%	1 in 15700
Hydrolethalus syndrome type 1 (AR) NM_145014.2	HYLS1	Pan-ethnic	≤1 in 500	99%	Reduced
Hyper-IgM immunodeficiency (CD40-related) (AR) NM_001250.5	CD40	Pan-ethnic	≤1 in 500	99%	Reduced
Hyperornithinemia-hyperammonemia-homocitrullinuria syndrome (AR) NM_014252.3	SLC25A15	Pan-ethnic	≤1 in 500	99%	Reduced
Hyperphosphatemic familial tumoral calcinosis (GALNT3-related) (AR) NM_004482.3	GALNT3	Pan-ethnic	≤1 in 500	99%	Reduced
Hypomyelinating leukodystrophy-12 (AR) NM_021729.5	VPS11	Pan-ethnic	≤1 in 500	99%	Reduced
Hypophosphatasia (AR) NM_000478.5	ALPL	Pan-ethnic	1 in 150	95%	1 in 2980
Ichthyosis prematurity syndrome (AR) NM_005094.3	SLC27A4	Pan-ethnic	≤1 in 500	99%	Reduced
IGHMBP2-related conditions (AR) NM_002180.2	IGHMBP2	Pan-ethnic	≤1 in 500	99%	Reduced
IKBKB-related conditions (AR) NM_001556.2	IKBKB	Pan-ethnic	≤1 in 500	99%	Reduced
Imerslund-Gräsbeck syndrome (AR) NM_030943.3	AMN *	Pan-ethnic	≤1 in 500	99%	Reduced
Immunodeficiency-centromeric instability-facial anomalies syndrome 1 (AR) NM_006892.3	DNMT3B	Pan-ethnic	≤1 in 500	99%	Reduced
Immunodeficiency-centromeric instability-facial anomalies syndrome 2 (AR) NM_014797.2	ZBTB24	Pan-ethnic	≤1 in 500	99%	Reduced
Isolated ectopia lentis (AR) NM_019032.5	ADAMTSL4	Pan-ethnic	≤1 in 500	99%	Reduced
Isovaleric acidemia (AR) NM_002225.3	IVD	Pan-ethnic	1 in 250	99%	1 in 24900
ITGB3-related conditions (AR) NM_000212.2	ITGB3	Pan-ethnic	≤1 in 500	99%	Reduced
Johanson-Blizzard syndrome (AR) NM_174916.2	UBR1	Pan-ethnic	1 in 250	99%	1 in 24900
Joubert syndrome and related disorders (MKS1-related) (AR) NM_017777.3	MKS1	Pan-ethnic	1 in 260	95%	1 in 5180
Joubert syndrome and related disorders (TMEM216-related) (AR) NM_001173990.2	TMEM216	Pan-ethnic	≤1 in 500	99%	Reduced
Junctional epidermolysis bullosa (LAMC2-related) (AR) NM_005562.2	LAMC2	Pan-ethnic	≤1 in 500	99%	Reduced
Junctional epidermolysis bullosa with pyloric atresia (ITGA6-related) (AR) NM_000210.3	ITGA6	Pan-ethnic	≤1 in 500	99%	Reduced
KCNJ11-related conditions (AR) NM_000525.3	KCNJ11	Pan-ethnic	≤1 in 500	99%	Reduced

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Krabbe disease (AR) NM_000153.3	GALC *	Pan-ethnic	1 in 158	99%	1 in 15700
LAMA2-related muscular dystrophy (AR) NM_000426.3	LAMA2	Pan-ethnic	1 in 87	99%	1 in 8600
LAMA3-related conditions (AR) NM_000227.4	LAMA3	Pan-ethnic	≤1 in 500	99%	Reduced
LAMB3-related conditions (AR) NM_000228.2	LAMB3	Pan-ethnic	1 in 317	99%	1 in 31600
Leber congenital amaurosis 5 (AR) NM_181714.3	LCA5	Pan-ethnic	≤1 in 500	97%	Reduced
Leukoencephalopathy with vanishing white matter (EIF2B1-related) (AR) NM_001414.3	EIF2B1	Pan-ethnic	≤1 in 500	99%	Reduced
Leukoencephalopathy with vanishing white matter (EIF2B2-related) (AR) NM_014239.3	EIF2B2	Pan-ethnic	≤1 in 500	99%	Reduced
Leukoencephalopathy with vanishing white matter (EIF2B3-related) (AR) NM_020365.4	EIF2B3	Pan-ethnic	≤1 in 500	99%	Reduced
Leukoencephalopathy with vanishing white matter (EIF2B4-related) (AR) NM_015636.3	EIF2B4	Pan-ethnic	≤1 in 500	99%	Reduced
Leukoencephalopathy with vanishing white matter (EIF2B5-related) (AR) NM_003907.2	EIF2B5	Pan-ethnic	≤1 in 500	99%	Reduced
LIG4 syndrome (AR) NM_002312.3	LIG4	Pan-ethnic	≤1 in 500	99%	Reduced
Limb-girdle muscular dystrophy (CAPN3-related) (AR) NM_000070.2	CAPN3	Pan-ethnic	1 in 134	99%	1 in 13300
Limb-girdle muscular dystrophy type 2C (AR) NM_000231.2	SGCG	Pan-ethnic	≤1 in 500	99%	Reduced
Limb-girdle muscular dystrophy type 2D (AR) NM_000023.2	SGCA	Pan-ethnic	≤1 in 500	99%	Reduced
Limb-girdle muscular dystrophy type 2E (AR) NM_000232.4	SGCB	Pan-ethnic	≤1 in 500	92%	Reduced
Limb-girdle muscular dystrophy type 2F (AR) NM_000337.5	SGCD	Pan-ethnic	≤1 in 500	99%	Reduced
Lipoid congenital adrenal hyperplasia (AR) NM_000349.2	STAR	Pan-ethnic	≤1 in 500	99%	Reduced
LRAT-related conditions (AR) NM_004744.4	LRAT	Pan-ethnic	1 in 296	99%	1 in 29500
Lysinuric protein intolerance (AR) NM_001126106.2	SLC7A7	Pan-ethnic	≤1 in 500	99%	Reduced
Lysosomal acid lipase deficiency (AR) NM_000235.3	LIPA	Pan-ethnic	1 in 359	94%	1 in 5967
Major histocompatibility complex class II deficiency (CIITA-related) (AR) NM_000246.3	CIITA	Pan-ethnic	≤1 in 500	99%	Reduced
Malonyl-CoA decarboxylase deficiency (AR) NM_012213.2	MLYCD	Pan-ethnic	≤1 in 500	99%	Reduced
Maple syrup urine disease type 1A (AR) NM_000709.3	BCKDHA	Pan-ethnic	1 in 373	99%	1 in 37200
Maple syrup urine disease type 1B (AR) NM_183050.2	BCKDHB	Pan-ethnic	1 in 346	99%	1 in 34500
Maple syrup urine disease type 2 (AR) NM_001918.3	DBT	Pan-ethnic	≤1 in 500	99%	Reduced
Medium-chain acyl-CoA dehydrogenase deficiency (AR) NM_000016.5	ACADM	Pan-ethnic	1 in 66	99%	1 in 6500
Medium/short-chain 3-hydroxyacyl-CoA dehydrogenase deficiency (AR) NM_005327.4	HADH	Pan-ethnic	≤1 in 500	99%	Reduced
MEDNIK syndrome (AR) NM_001283.3	AP1S1	Pan-ethnic	≤1 in 500	99%	Reduced

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Megalencephalic leukoencephalopathy with subcortical cysts 1 (AR) NM_015166.3	MLC1 *	Pan-ethnic	≤1 in 500	99%	Reduced
Metachromatic leukodystrophy (ARSA-related) (AR) NM_000487.5	ARSA	Pan-ethnic	1 in 100	95%	1 in 1980
Methylmalonic acidemia (MCEE-related) (AR) NM_032601.3	MCEE	Pan-ethnic	≤1 in 500	99%	Reduced
Methylmalonic acidemia (MMAA-related) (AR) NM_172250.2	MMAA	Pan-ethnic	1 in 316	97%	1 in 10500
Methylmalonic acidemia (MMAB-related) (AR) NM_052845.3	MMAB	Pan-ethnic	1 in 456	98%	1 in 22750
Methylmalonic acidemia (MUT-related) (AR) NM_000255.3	MUT	Pan-ethnic	1 in 204	96%	1 in 5075
MFSD8-related conditions (AR) NM_152778.2	MFSD8	Pan-ethnic	≤1 in 500	99%	Reduced
Microcephalic osteodysplastic primordial dwarfism type II (AR) NM_006031.5	PCNT	Pan-ethnic	≤1 in 500	99%	Reduced
Microcephaly, postnatal progressive, with seizures and brain atrophy (AR) NM_004268.4	MED17	Pan-ethnic	≤1 in 500	99%	Reduced
Mitochondrial complex I deficiency 1 (AR) NM_002495.3	NDUFS4	Pan-ethnic	≤1 in 500	99%	Reduced
Mitochondrial complex I deficiency 3 (AR) NM_024407.4	NDUFS7	Pan-ethnic	1 in 387	99%	1 in 38600
Mitochondrial complex I deficiency 4 (AR) NM_007103.3	NDUFV1	Pan-ethnic	1 in 387	99%	1 in 38600
Mitochondrial complex I deficiency 9 (AR) NM_004553.4	NDUFS6	Pan-ethnic	≤1 in 500	99%	Reduced
Mitochondrial complex I deficiency 10 (AR) NM_174889.4	NDUFAF2	Pan-ethnic	1 in 387	99%	1 in 38600
Mitochondrial complex I deficiency 16 (AR) NM_024120.4	NDUFAF5	Pan-ethnic	≤1 in 500	99%	Reduced
Mitochondrial complex I deficiency 19 (AR) NM_017547.3	FOXRED1	Pan-ethnic	1 in 376	99%	1 in 37500
Mitochondrial complex I deficiency 20/ACAD9 deficiency (AR) NM_014049.4	ACAD9	Pan-ethnic	≤1 in 500	99%	Reduced
Mitochondrial complex IV deficiency 6 (AR) NM_004376.6	COX15	Pan-ethnic	≤1 in 500	99%	Reduced
Mitochondrial complex IV deficiency 12 (AR) NM_001171155.1	PET100	Pan-ethnic	1 in 387	99%	1 in 38600
Mitochondrial complex IV deficiency / Leigh syndrome, French Canadian type (AR) NM_133259.3	LRPPRC	Pan-ethnic	≤1 in 500	99%	Reduced
Mitochondrial DNA depletion syndrome-2 (AR) NM_004614.4	TK2	Pan-ethnic	≤1 in 500	99%	Reduced
Mitochondrial neurogastrointestinal encephalomyopathy (AR) NM_001953.4	TYMP	Pan-ethnic	≤1 in 500	99%	Reduced
Mitochondrial trifunctional protein deficiency (HADHB-related) (AR) NM_000183.2	HADHB	Pan-ethnic	≤1 in 500	99%	Reduced
MKKS-related conditions (AR) NM_018848.3	MKKS	Pan-ethnic	≤1 in 500	99%	Reduced
Molybdenum cofactor deficiency (MOCS1-related) (AR) NM_001358530.2	MOCS1	Pan-ethnic	1 in 226	99%	1 in 22500
Molybdenum cofactor deficiency (MOCS2-related) (AR) NM_004531.4	MOCS2B	Pan-ethnic	≤1 in 500	99%	Reduced
Molybdenum cofactor deficiency (MOCS2-related) (AR) NM_176806.3	MOCS2A	Pan-ethnic	≤1 in 500	99%	Reduced
MPL-related conditions (AR) NM_005373.2	MPL	Pan-ethnic	≤1 in 500	99%	Reduced

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MPV17-related conditions (AR) NM_002437.4	MPV17	Pan-ethnic	≤1 in 500	99%	Reduced
Mucolipidosis type III gamma (AR) NM_032520.4	GNPTG	Pan-ethnic	≤1 in 500	99%	Reduced
Mucolipidosis type IV (AR) NM_020533.2	MCOLN1	Pan-ethnic	≤1 in 500	99%	Reduced
Mucopolysaccharidosis type I (AR) NM_000203.4	IDUA	Pan-ethnic	1 in 148	97%	1 in 4900
Mucopolysaccharidosis type IIIA (AR) NM_000199.3	SGSH	Pan-ethnic	1 in 215	99%	1 in 21400
Mucopolysaccharidosis type IIIB (AR) NM_000263.3	NAGLU	Pan-ethnic	1 in 224	99%	1 in 22300
Mucopolysaccharidosis type IIID (AR) NM_002076.3	GNS	Pan-ethnic	≤1 in 500	99%	Reduced
Mucopolysaccharidosis type IVA (AR) NM_000512.4	GALNS	Pan-ethnic	1 in 224	99%	1 in 22300
Mucopolysaccharidosis type IX (AR) NM_153281.1	HYAL1	Pan-ethnic	≤1 in 500	99%	Reduced
Mucopolysaccharidosis type VI (AR) NM_000046.3	ARSB	Pan-ethnic	1 in 250	99%	1 in 24900
Mucopolysaccharidosis type VII (AR) NM_000181.3	GUSB	Pan-ethnic	1 in 250	99%	1 in 24900
Mulibrey nanism (AR) NM_015294.4	TRIM37	Pan-ethnic	≤1 in 500	99%	Reduced
Multiple pterygium syndrome (AR) NM_005199.4	CHRNG	Pan-ethnic	≤1 in 500	99%	Reduced
Multiple sulfatase deficiency (AR) NM_182760.3	SUMF1	Pan-ethnic	≤1 in 500	99%	Reduced
Muscular dystrophy-dystroglycanopathy (FKRP-related) (AR) NM_024301.4	FKRP	Pan-ethnic	1 in 158	99%	1 in 15700
Muscular dystrophy-dystroglycanopathy (FKTN-related) (AR) NM_001079802.1	FKTN	Pan-ethnic	≤1 in 500	99%	Reduced
Muscular dystrophy-dystroglycanopathy (LARGE1-related) (AR) NM_004737.4	LARGE1	Pan-ethnic	≤1 in 500	99%	Reduced
Muscular dystrophy-dystroglycanopathy (POMT1-related) (AR) NM_007171.3	POMT1	Pan-ethnic	1 in 268	99%	1 in 26700
Muscular dystrophy-dystroglycanopathy (POMT2-related) (AR) NM_013382.5	POMT2	Pan-ethnic	1 in 371	99%	1 in 37000
Muscular dystrophy-dystroglycanopathy (RXYLT1-related) (AR) NM_014254.2	RXYLT1	Pan-ethnic	≤1 in 500	99%	Reduced
MUSK-related conditions (AR) NM_005592.3	MUSK	Pan-ethnic	1 in 447	99%	1 in 44600
MVK-related conditions (AR) NM_000431.3	MVK	Pan-ethnic	≤1 in 500	99%	Reduced
MYO7A-related conditions (AR) NM_000260.3	MYO7A	Pan-ethnic	1 in 200	95%	1 in 3980
Myopathy, lactic acidosis, and sideroblastic anemia 1 (AR) NM_025215.5	PUS1	Pan-ethnic	≤1 in 500	99%	Reduced
Myotonia congenita (AR) NM_000083.2	CLCN1	Pan-ethnic	1 in 112	99%	1 in 11100
N-acetylglutamate synthase deficiency (AR) NM_153006.2	NAGS	Pan-ethnic	≤1 in 500	99%	Reduced
Nemaline myopathy 2 (AR) NM_001271208.1	NEB *	Pan-ethnic	1 in 158	95%	1 in 3140
Nephrogenic diabetes insipidus (AQP2-related) (AR) NM_000486.5	AQP2	Pan-ethnic	≤1 in 500	99%	Reduced

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Nephronophthisis (INVS-related) (AR) NM_014425.3	INVS	Pan-ethnic	≤1 in 500	99%	Reduced
Nephronophthisis (NPHP1-related) (AR) NM_000272.3	NPHP1	Pan-ethnic	≤1 in 500	99%	Reduced
Neuronal ceroid lipofuscinosis type 1 (AR) NM_000310.3	PPT1	Pan-ethnic	1 in 199	98%	1 in 9900
Neuronal ceroid lipofuscinosis type 2 (AR) NM_000391.3	TPP1	Pan-ethnic	1 in 250	97%	1 in 8300
Neuronal ceroid lipofuscinosis type 5 (AR) NM_006493.2	CLN5	Pan-ethnic	≤1 in 500	99%	Reduced
Neuronal ceroid lipofuscinosis type 6 (AR) NM_017882.2	CLN6	Pan-ethnic	≤1 in 500	99%	Reduced
Neuronal ceroid lipofuscinosis type 8 (AR) NM_018941.3	CLN8	Pan-ethnic	≤1 in 500	99%	Reduced
Neuronal ceroid lipofuscinosis type 10 (AR) NM_001909.4	CTSD	Pan-ethnic	≤1 in 500	99%	Reduced
Niemann-Pick disease type C (NPC1-related) (AR) NM_000271.4	NPC1	Pan-ethnic	1 in 183	99%	1 in 18200
Niemann-Pick disease type C (NPC2-related) (AR) NM_006432.3	NPC2	Pan-ethnic	≤1 in 500	99%	Reduced
Niemann-Pick disease types A and B (AR) NM_000543.4	SMPD1	Pan-ethnic	1 in 250	95%	1 in 4980
Nijmegen breakage syndrome (AR) NM_002485.4	NBN	Pan-ethnic	≤1 in 500	99%	Reduced
Nonsyndromic deafness (LOXHD1-related) (AR) NM_144612.6	LOXHD1	Pan-ethnic	≤1 in 500	99%	Reduced
Nonsyndromic deafness (MYO15A-related) (AR) NM_016239.3	MYO15A	Pan-ethnic	≤1 in 500	99%	Reduced
Nonsyndromic deafness (OTOA-related) (AR) NM_144672.3	OTOA *	Pan-ethnic	≤1 in 500	88%	Reduced
Nonsyndromic deafness (SYNE4-related) (AR) NM_001039876.2	SYNE4	Pan-ethnic	≤1 in 500	99%	Reduced
Nonsyndromic deafness (TMC1-related) (AR) NM_138691.2	TMC1	Pan-ethnic	≤1 in 500	99%	Reduced
Nonsyndromic deafness (TMPRSS3-related) (AR) NM_024022.2	TMPRSS3	Pan-ethnic	≤1 in 500	99%	Reduced
Nonsyndromic intellectual disability (CC2D1A-related) (AR) NM_017721.5	CC2D1A	Pan-ethnic	≤1 in 500	99%	Reduced
NR2E3-related conditions (AR) NM_014249.3	NR2E3	Pan-ethnic	≤1 in 500	99%	Reduced
NSMCE3 deficiency (AR) NM_138704.3	NSMCE3	Pan-ethnic	≤1 in 500	99%	Reduced
Oculocutaneous albinism type 2 (AR) NM_000275.2	OCA2	Pan-ethnic	1 in 95	99%	1 in 9400
Oculocutaneous albinism type 3 (AR) NM_000550.2	TYRP1	Pan-ethnic	≤1 in 500	99%	Reduced
Oculocutaneous albinism type 4 (AR) NM_016180.4	SLC45A2	Pan-ethnic	1 in 158	99%	1 in 15700
Oculocutaneous albinism types 1A and 1B (AR) NM_000372.4	TYR *	Pan-ethnic	1 in 100	97%	1 in 3300
OPA3-related conditions (AR) NM_025136.3	OPA3	Pan-ethnic	≤1 in 500	99%	Reduced
Osteogenesis imperfecta (BMP1-related) (AR) NM_006129.4	BMP1	Pan-ethnic	≤1 in 500	99%	Reduced
Osteogenesis imperfecta (CRTAP-related) (AR) NM_006371.4	CRTAP	Pan-ethnic	≤1 in 500	99%	Reduced
Osteogenesis imperfecta (P3H1-related) (AR) NM_022356.3	P3H1	Pan-ethnic	≤1 in 500	99%	Reduced
Osteopetrosis (TCIRG1-related) (AR) NM_006019.3	TCIRG1	Pan-ethnic	1 in 317	99%	1 in 31600
OSTM1 deficiency associated osteopetrosis (AR) NM_014028.3	OSTM1	Pan-ethnic	≤1 in 500	99%	Reduced

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OTOF-related conditions (AR) NM_194248.2	OTOF	Pan-ethnic	≤1 in 500	99%	Reduced
Pantothenate kinase-associated neurodegeneration (AR) NM_153638.2	PANK2	Pan-ethnic	1 in 289	99%	1 in 28800
Parkinson disease 15 (AR) NM_012179.3	FBXO7	Pan-ethnic	≤1 in 500	99%	Reduced
PCDH15-related conditions (AR) NM_033056.3	PCDH15	Pan-ethnic	1 in 400	99%	1 in 39900
PEX5-related conditions (AR) NM_001131025.1	PEX5	Pan-ethnic	≤1 in 500	99%	Reduced
PEX7-related conditions (AR) NM_000288.3	PEX7	Pan-ethnic	1 in 157	99%	1 in 15600
PGM3-congenital disorder of glycosylation (AR) NM_001199917.1	PGM3	Pan-ethnic	≤1 in 500	99%	Reduced
Phenylalanine hydroxylase deficiency (AR) NM_000277.1	PAH	Pan-ethnic	1 in 58	99%	1 in 5700
Phosphoglycerate dehydrogenase deficiency (AR) NM_006623.3	PHGDH	Pan-ethnic	≤1 in 500	99%	Reduced
PIGN-congenital disorder of glycosylation (AR) NM_176787.4	PIGN	Pan-ethnic	≤1 in 500	99%	Reduced
PJVK-related conditions (AR) NM_001042702.3	DFNB59	Pan-ethnic	≤1 in 500	99%	Reduced
PLA2G6-related conditions (AR) NM_003560.2	PLA2G6	Pan-ethnic	≤1 in 500	99%	Reduced
PLEKHG5-related conditions (AR) NM_020631.4	PLEKHG5	Pan-ethnic	≤1 in 500	99%	Reduced
POLG-related conditions (AR) NM_002693.2	POLG	Pan-ethnic	1 in 113	95%	1 in 2240
Polycystic kidney disease (PKHD1-related) (AR) NM_138694.3	PKHD1 *	Pan-ethnic	1 in 70	99%	1 in 6900
Polymicrogyria (ADGRG1-related) (AR) NM_005682.6	ADGRG1	Pan-ethnic	≤1 in 500	99%	Reduced
POMGNT1-related conditions (AR) NM_017739.3	POMGNT1	Pan-ethnic	≤1 in 500	99%	Reduced
Pontocerebellar hypoplasia (TSEN54-related) (AR) NM_207346.2	TSEN54	Pan-ethnic	≤1 in 500	99%	Reduced
Pontocerebellar hypoplasia type 1B (AR) NM_016042.3	EXOSC3	Pan-ethnic	≤1 in 500	99%	Reduced
Pontocerebellar hypoplasia type 2D (AR) NM_016955.3	SEPSECS	Pan-ethnic	≤1 in 500	99%	Reduced
Pontocerebellar hypoplasia type 6 (AR) NM_020320.3	RARS2	Pan-ethnic	≤1 in 500	99%	Reduced
Primary carnitine deficiency (AR) NM_003060.3	SLC22A5	Pan-ethnic	1 in 71	99%	1 in 7000
Primary ciliary dyskinesia (CCDC39-related) (AR) NM_181426.1	CCDC39	Pan-ethnic	1 in 211	99%	1 in 21000
Primary ciliary dyskinesia (CCDC103-related) (AR) NM_213607.2	CCDC103	Pan-ethnic	1 in 316	99%	1 in 31500
Primary ciliary dyskinesia (DNAH5-related) (AR) NM_001369.2	DNAH5	Pan-ethnic	1 in 109	99%	1 in 10800
Primary ciliary dyskinesia (DNAH11-related) (AR) NM_001277115.1	DNAH11	Pan-ethnic	1 in 211	99%	1 in 21000
Primary ciliary dyskinesia (DNAI1-related) (AR) NM_012144.3	DNAI1	Pan-ethnic	1 in 250	99%	1 in 24900
Primary ciliary dyskinesia (DNAI2-related) (AR) NM_023036.4	DNAI2	Pan-ethnic	1 in 354	99%	1 in 35300
Primary hyperoxaluria type 1 (AR) NM_000030.2	AGXT	Pan-ethnic	1 in 135	99%	1 in 13400
Primary hyperoxaluria type 2 (AR) NM_012203.1	GRHPR	Pan-ethnic	≤1 in 500	99%	Reduced
Primary hyperoxaluria type 3 (AR) NM_138413.3	HOGA1	Pan-ethnic	1 in 354	99%	1 in 35300

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Primary microcephaly (MCPH1-related) (AR) NM_024596.4	MCPH1	Pan-ethnic	≤1 in 500	99%	Reduced
Progressive early-onset encepahlopathy with brain atrophy and thin corpus callosum (PEBAT) (AR) NM_005993.4	TBCD	Pan-ethnic	≤1 in 500	99%	Reduced
Progressive pseudorheumatoid dysplasia (AR) NM_003880.3	WISP3	Pan-ethnic	≤1 in 500	99%	Reduced
Prolidase deficiency (AR) NM_000285.3	PEPD	Pan-ethnic	≤1 in 500	99%	Reduced
Propionic acidemia (PCCA-related) (AR) NM_000282.3	PCCA	Pan-ethnic	1 in 224	96%	1 in 5575
Propionic acidemia (PCCB-related) (AR) NM_000532.4	PCCB	Pan-ethnic	1 in 224	99%	1 in 22300
PSAP-related conditions (AR) NM_002778.3	PSAP	Pan-ethnic	≤1 in 500	99%	Reduced
Pycnodysostosis (AR) NM_000396.3	CTSK	Pan-ethnic	1 in 438	99%	1 in 43700
Pyridoxal 5'-phosphate-dependent epilepsy (AR) NM_018129.3	PNPO	Pan-ethnic	≤1 in 500	99%	Reduced
Pyridoxine-dependent epilepsy (ALDH7A1-related) (AR) NM_001182.4	ALDH7A1	Pan-ethnic	1 in 127	99%	1 in 12600
Pyruvate carboxylase deficiency (AR) NM_000920.3	PC	Pan-ethnic	1 in 250	95%	1 in 4980
Pyruvate dehydrogenase complex deficiency (PDHB-related) (AR) NM_000925.3	PDHB	Pan-ethnic	≤1 in 500	99%	Reduced
RAPSN-related conditions (AR) NM_005055.4	RAPSN	Pan-ethnic	1 in 283	99%	1 in 28200
RDH12-related conditions (AR) NM_152443.2	RDH12	Pan-ethnic	1 in 460	99%	1 in 45900
Refsum disease (PHYH-related) (AR) NM_006214.3	PHYH	Pan-ethnic	≤1 in 500	99%	Reduced
Retinitis pigmentosa 25 (AR) NM_001142800.1	EYS *	Pan-ethnic	1 in 129	99%	1 in 12800
Retinitis pigmentosa 28 (AR) NM_001201543.1	FAM161A	Pan-ethnic	1 in 289	99%	1 in 28800
Retinitis pigmentosa 36 (AR) NM_001077620.2	PRCD	Pan-ethnic	1 in 296	99%	1 in 29500
Retinitis pigmentosa 62 (AR) NM_001242957.2	MAK	Pan-ethnic	1 in 274	99%	1 in 27300
Rhizomelic chondrodysplasia punctata type 2 (AR) NM_014236.3	GNPAT	Pan-ethnic	≤1 in 500	99%	Reduced
Rhizomelic chondrodysplasia punctata type 3 (AR) NM_003659.3	AGPS	Pan-ethnic	≤1 in 500	99%	Reduced
RLBP1-related conditions (AR) NM_000326.4	RLBP1	Pan-ethnic	1 in 296	99%	1 in 29500
Roberts syndrome (AR) NM_001017420.2	ESCO2	Pan-ethnic	≤1 in 500	99%	Reduced
RPE65-related conditions (AR) NM_000329.2	RPE65	Pan-ethnic	1 in 228	99%	1 in 22700
RYR1-related conditions (AR) NM_000540.2	RYR1	Pan-ethnic	≤1 in 500	99%	Reduced
SAMD9-related conditions (AR) NM_017654.3	SAMD9	Pan-ethnic	≤1 in 500	99%	Reduced
Sandhoff disease (AR) NM_000521.3	HEXB	Pan-ethnic	1 in 180	99%	1 in 17900
Schimke immuno-osseous dysplasia (AR) NM_014140.3	SMARCAL1	Pan-ethnic	≤1 in 500	99%	Reduced
Seckel syndrome (CEP152-related) (AR) NM_014985.3	CEP152	Pan-ethnic	≤1 in 500	99%	Reduced
Sepiapterin reductase deficiency (AR) NM_003124.4	SPR	Pan-ethnic	≤1 in 500	99%	Reduced

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CARRIER DETECTION RATES AND RESIDUAL RISKS

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Issue date: 02/19/2023

DISORDER (INHERITANCE)	GENE	ETHNICITY	CARRIER FREQUENCY	DETECTION RATE	RISK TO BE A CARRIER AFTER NEGATIVE RESULT
Severe combined immunodeficiency due to CD3-delta deficiency (AR) NM_000732.4	CD3D	Pan-ethnic	≤1 in 500	99%	Reduced
Severe combined immunodeficiency due to CD3-epsilon deficiency (AR) NM_000733.3	CD3E	Pan-ethnic	≤1 in 500	99%	Reduced
Severe combined immunodeficiency due to CD45 deficiency (AR) NM_002838.4	PTPRC *	Pan-ethnic	≤1 in 500	99%	Reduced
Severe combined immunodeficiency due to DCLRE1C (Artemis) deficiency (AR) NM_001033855.2	DCLRE1C	Pan-ethnic	≤1 in 500	99%	Reduced
Severe combined immunodeficiency due to IL7R-alpha deficiency (AR) NM_002185.3	IL7R	Pan-ethnic	1 in 348	99%	1 in 34700
Severe combined immunodeficiency due to JAK3 deficiency (AR) NM_000215.3	JAK3	Pan-ethnic	1 in 455	99%	1 in 45400
Severe combined immunodeficiency due to RAG1 deficiency (AR) NM_000448.2	RAG1	Pan-ethnic	1 in 301	99%	1 in 30000
Severe combined immunodeficiency due to RAG2 deficiency (AR) NM_000536.3	RAG2	Pan-ethnic	≤1 in 500	99%	Reduced
Severe congenital neutropenia due to G6PC3 deficiency (AR) NM_138387.3	G6PC3	Pan-ethnic	≤1 in 500	99%	Reduced
Severe congenital neutropenia due to HAX1 deficiency (AR) NM_006118.3	HAX1	Pan-ethnic	≤1 in 500	99%	Reduced
Severe congenital neutropenia due to VPS45 deficiency (AR) NM_007259.4	VPS45	Pan-ethnic	≤1 in 500	99%	Reduced
Sialic acid storage diseases (AR) NM_012434.4	SLC17A5	Pan-ethnic	≤1 in 500	99%	Reduced
Sialidosis (AR) NM_000434.3	NEU1	Pan-ethnic	≤1 in 500	99%	Reduced
Sjögren-Larsson syndrome (AR) NM_000382.2	ALDH3A2	Pan-ethnic	≤1 in 500	99%	Reduced
SLC12A6-related conditions (AR) NM_133647.1	SLC12A6	Pan-ethnic	≤1 in 500	99%	Reduced
SLC26A2-related conditions (AR) NM_000112.3	SLC26A2	Pan-ethnic	1 in 158	95%	1 in 3140
SLC26A4-related conditions (AR) NM_000441.1	SLC26A4	Pan-ethnic	1 in 80	99%	1 in 7900
SLC37A4-related conditions (AR) NM_001164277.1	SLC37A4	Pan-ethnic	1 in 354	95%	1 in 7060
Smith-Lemli-Opitz syndrome (AR) NM_001360.2	DHCR7	Pan-ethnic	1 in 71	99%	1 in 7000
Spastic paraplegia type 15 (AR) NM_015346.3	ZFYVE26	Pan-ethnic	≤1 in 500	99%	Reduced
Spastic paraplegia type 49 (AR) NM_014844.3	TECPR2	Pan-ethnic	≤1 in 500	99%	Reduced
Spastic tetraplegia, thin corpus callosum, and progressive microcephaly (AR) NM_003038.4	SLC1A4	Pan-ethnic	≤1 in 500	99%	Reduced
SPG11-related conditions (AR) NM_025137.3	SPG11	Pan-ethnic	1 in 141	99%	1 in 14000
Spinal muscular atrophy (AR) NM_000344.3 Carrier residual risks listed are for 2 copy SMN1 results. Carrier residual risk for >2 copies are 5- to 10-fold lower.	SMN1 *	African-American	1 in 59	83%	1 in 342
		Ashkenazi Jewish	1 in 62	94%	1 in 1017
		Asian	1 in 50	93%	1 in 701
		Caucasian	1 in 45	95%	1 in 880
		Hispanic	1 in 48	94%	1 in 784
		Pan-ethnic	1 in 49	94%	1 in 800

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Spinocerebellar ataxia (ANO10-related) (AR) NM_018075.3	ANO10 *	Pan-ethnic	≤1 in 500	99%	Reduced
Spondylocostal dysostosis (DLL3-related) (AR) NM_016941.3	DLL3	Pan-ethnic	1 in 350	99%	1 in 34900
Spondylocostal dysostosis (MESP2-related) (AR) NM_001039958.1	MESP2	Pan-ethnic	1 in 224	99%	1 in 22300
Steel syndrome (AR) NM_032888.3	COL27A1	Pan-ethnic	≤1 in 500	99%	Reduced
Steroid 5-alpha-reductase deficiency (AR) NM_000348.3	SRD5A2	Pan-ethnic	≤1 in 500	99%	Reduced
Stüve-Wiedemann syndrome (AR) NM_002310.5	LIFR *	Pan-ethnic	≤1 in 500	99%	Reduced
Sulfite oxidase deficiency (AR) NM_000456.2	SUOX	Pan-ethnic	≤1 in 500	99%	Reduced
SURF1-related conditions (AR) NM_003172.3	SURF1	Pan-ethnic	1 in 128	99%	1 in 12700
Tay-Sachs disease (AR) NM_000520.4	HEXA	Pan-ethnic	1 in 250	99%	1 in 24900
TBCE-related conditions (AR) NM_003193.4	TBCE *	Pan-ethnic	≤1 in 500	99%	Reduced
Thiamine-responsive megaloblastic anemia (AR) NM_006996.2	SLC19A2	Pan-ethnic	≤1 in 500	99%	Reduced
Thyroid dyshormonogenesis (SLC5A5-related) (AR) NM_000453.2	SLC5A5	Pan-ethnic	≤1 in 500	99%	Reduced
Thyroid dyshormonogenesis (TG-related) (AR) NM_003235.4	TG *	Pan-ethnic	≤1 in 500	99%	Reduced
Thyroid dyshormonogenesis (TPO-related) (AR) NM_000547.5	TPO	Pan-ethnic	1 in 129	99%	1 in 12800
TMEM67-related conditions (AR) NM_153704.5	TMEM67	Pan-ethnic	1 in 316	99%	1 in 31500
Transcobalamin II deficiency (AR) NM_000355.3	TCN2	Pan-ethnic	≤1 in 500	99%	Reduced
Transient infantile liver failure (AR) NM_018006.4	TRMU	Pan-ethnic	≤1 in 500	99%	Reduced
TREX1-related conditions (AR) NM_033629.4	TREX1	Pan-ethnic	≤1 in 500	99%	Reduced
Trichohepatoenteric syndrome (SKIV2L-related) (AR) NM_006929.4	SKIV2L	Pan-ethnic	≤1 in 500	99%	Reduced
Trichohepatoenteric syndrome (TTC37-related) (AR) NM_014639.3	TTC37	Pan-ethnic	≤1 in 500	99%	Reduced
TRIM32-related conditions (AR) NM_012210.3	TRIM32	Pan-ethnic	1 in 408	99%	1 in 40700
Trimethylaminuria (AR) NM_006894.6	FMO3	Pan-ethnic	≤1 in 500	99%	Reduced
Triple A syndrome (AR) NM_015665.5	AAAS	Pan-ethnic	≤1 in 500	99%	Reduced
TSHR-related conditions (AR) NM_000369.2	TSHR	Pan-ethnic	1 in 158	99%	1 in 15700
TULP1-related conditions (AR) NM_003322.4	TULP1	Pan-ethnic	1 in 296	99%	1 in 29500
Tyrosine hydroxylase deficiency (AR) NM_199292.2	TH	Pan-ethnic	≤1 in 500	99%	Reduced
Tyrosinemia type I (AR) NM_000137.2	FAH *	Pan-ethnic	1 in 125	95%	1 in 2480
Tyrosinemia type II (AR) NM_000353.2	TAT	Pan-ethnic	1 in 250	99%	1 in 24900
Tyrosinemia type III (AR) NM_002150.2	HPD	Pan-ethnic	≤1 in 500	99%	Reduced
USH1C-related conditions (AR) NM_005709.3	USH1C *	Pan-ethnic	1 in 353	90%	1 in 3521
USH2A-related conditions (AR) NM_206933.2	USH2A	Pan-ethnic	1 in 112	99%	1 in 11100

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DISORDER (INHERITANCE)	GENE	ETHNICITY	CARRIER FREQUENCY	DETECTION RATE	RISK TO BE A CARRIER AFTER NEGATIVE RESULT
Very long-chain acyl-CoA dehydrogenase deficiency (AR) NM_000018.3	ACADVL	Pan-ethnic	1 in 100	99%	1 in 9900
Vici syndrome (AR) NM_020964.2	EPG5	Pan-ethnic	≤1 in 500	99%	Reduced
Vitamin D-dependent rickets type 1A (AR) NM_000785.3	CYP27B1	Pan-ethnic	≤1 in 500	99%	Reduced
Vitamin D-dependent rickets type 2A (AR) NM_001017535.1	VDR	Pan-ethnic	≤1 in 500	99%	Reduced
VPS53-related conditions (AR) NM_001128159.2	VPS53 *	Pan-ethnic	≤1 in 500	99%	Reduced
VRK1-related conditions (AR) NM_003384.2	VRK1	Pan-ethnic	≤1 in 500	99%	Reduced
VSX2-related conditions (AR) NM_182894.2	VSX2	Pan-ethnic	≤1 in 500	99%	Reduced
Warsaw syndrome (AR) NM_030653.3	DDX11 *	Pan-ethnic	≤1 in 500	15%	Reduced
Werner syndrome (AR) NM_000553.4	WRN *	Pan-ethnic	1 in 224	99%	1 in 22300
Wilson disease (AR) NM_000053.3	ATP7B	Pan-ethnic	1 in 90	98%	1 in 4450
WNT10A-related conditions (AR) NM_025216.2	WNT10A	Pan-ethnic	1 in 305	99%	1 in 30400
Wolcott-Rallison syndrome (AR) NM_004836.6	EIF2AK3	Pan-ethnic	≤1 in 500	99%	Reduced
Woodhouse-Sakati syndrome (AR) NM_025000.3	DCAF17	Pan-ethnic	≤1 in 500	99%	Reduced
Xeroderma pigmentosum complementation group A (AR) NM_000380.3	XPA	Pan-ethnic	≤1 in 500	99%	Reduced
Xeroderma pigmentosum complementation group C (AR) NM_004628.4	XPC	Pan-ethnic	≤1 in 500	99%	Reduced
Xeroderma pigmentosum, variant type (AR) NM_006502.2	POLH	Pan-ethnic	≤1 in 500	99%	Reduced
Zellweger spectrum disorder (PEX1-related) (AR) NM_000466.2	PEX1 *	Pan-ethnic	1 in 144	99%	1 in 14300
Zellweger spectrum disorder (PEX2-related) (AR) NM_000318.2	PEX2	Pan-ethnic	≤1 in 500	99%	Reduced
Zellweger spectrum disorder (PEX6-related) (AR) NM_000287.3	PEX6	Pan-ethnic	1 in 294	99%	1 in 29300
Zellweger spectrum disorder (PEX10-related) (AR) NM_153818.1	PEX10	Pan-ethnic	≤1 in 500	94%	Reduced
Zellweger spectrum disorder (PEX12-related) (AR) NM_000286.2	PEX12	Pan-ethnic	1 in 409	99%	1 in 40800
Zellweger spectrum disorder (PEX13-related) (AR) NM_002618.3	PEX13	Pan-ethnic	≤1 in 500	99%	Reduced
Zellweger spectrum disorder (PEX16-related) (AR) NM_004813.2	PEX16	Pan-ethnic	≤1 in 500	99%	Reduced
Zellweger spectrum disorder (PEX26-related) (AR) NM_017929.5	PEX26	Pan-ethnic	≤1 in 500	99%	Reduced

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12967,DONOR

DOB [redacted]
Sex: M
Phone: (206) 588-1484
Patient ID: 12967

Age [redacted]
Fasting: U

Specimen: OW535739U
Requisition: 0000473
Report Status: FINAL / SEE REPORT

Collected: 01/10/2023 00:00
Received: 01/11/2023 07:14
Reported: 01/20/2023 15:14

Client #: 98105026
OLLIFFE,JEFFREY F
SEATTLE SPERM BANK
4915 25TH AVE NE STE 204W
SEATTLE, WA 98105-5668
Phone: (206) 588-1484
Fax: (206) 466-4696

FASTING:UNKNOWN

CHROMOSOME ANALYSIS, BLOOD (FINAL)

Lab: EZ

Analyte	Value	
CHROMOSOME ANALYSIS, BLOOD (29770-5)	**See Below**	FINAL

```
Order ID:                 23-14728

Specimen Type:            Blood

Clinical Indication:      Rule out chromosome abnormality

RESULT:
NORMAL MALE KARYOTYPE

INTERPRETATION:
Chromosome analysis revealed normal G-band patterns within
the limits of standard cytogenetic analysis.

Please expect the results of any other concurrent study in a
separate report.


NOMENCLATURE:
46,XY

ASSAY INFORMATION:
Method:                   G-Band (Digital Analysis: MetaSyst
Cells Counted:            20
Band Level:               450
Cells Analyzed:           5
Cells Karyotyped:         5

This test does not address genetic disorders that cannot be
detected by standard cytogenetic methods or rare events such
as low level mosaicism or subtle rearrangements.

Morteza Hemmat, PhD, FACMG     (800) NICHOLS-4307

Electronic Signature:             1/20/2023 5:25 PM
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Performing Sites

EZ Quest Diagnostics/Nichols SJC-San Juan Capistrano,, 33608 Ortega Hwy, San Juan Capistrano, CA 92675-2042 Laboratory Director: Irina Maramica MD,PhD,MBA

Key

Priority Out of Range | Out of Range | (PEND) Pending Result | (PRE) Preliminary Result | (FINAL) Final Result | (RE) Reissued Result

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