INVITAE

# INVITAE CARRIER SCREEN RESULTS

| | | | | | |
|---|---|---|---|---|---|
| **Patient name:** | DONOR 14474 | **Sample type:** | Blood | **Report date:** | 28-APR-2023 |
| **DOB:** | [redacted] | **Sample collection date:** | 17-APR-2023 | **Invitae #:** | RQ4960593 |
| **Sex assigned at birth:** | Male | **Sample accession date:** | 18-APR-2023 | **Clinical team:** | Hallie Yoshimura<br>Jeffrey Olliffe |
| **Gender:** | | | | | |
| **Patient ID (MRN):** | | | | | |

**Reason for testing**

Gamete donor

**Test performed**

Invitae Carrier Screen

## RESULT: NEGATIVE

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 did not identify any genetic changes in the gene(s) analyzed that are currently recognized as clinically significant. This negative result reduces, but does not eliminate, the chance that this individual is a carrier for conditions caused by any of the genes tested. This individual may still be a carrier for a genetic condition that is not evaluated by this test.

## Next steps

- 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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## Results to note

### SMN1

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

### Pseudodeficiency allele(s)

- Benign change, c.2065G>A (p.Glu689Lys), known to be a pseudodeficiency allele, identified in the GAA gene. Pseudodeficiency alleles are not known to be associated with disease, including glycogen storage disease type II (Pompe 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.

## 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 table. 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 as needed based on stringent criteria using one of several validated orthogonal approaches (PubMed ID 30610921). Sequencing is performed by Invitae Corporation (1400 16th Street, San Francisco, CA 94103, #05D2040778). Confirmatory sequencing is performed by Invitae Corporation (1400 16th Street, San Francisco, CA 94103, #05D2040778).

  The following additional 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, cytosine-guanine-guanine (CGG) triplet repeats in the 5' untranslated region (5' UTR) of the FMR1 gene are detected by triplet repeat-primed PCR (RP-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.

- 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


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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.

## 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.
- 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. VPS13A: Deletion/duplication analysis is not offered for exons 2-3, 27-28. GALE: Sequencing analysis for exons 10 includes only cds +/- 5 bp. DDX11: NM_030653.3:c.1763-1G>C variant only. GNE: Sequencing analysis for exons 8 includes only cds +/- 10 bp. 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. PKHD1: Deletion/duplication analysis is not offered for exon 13. ABCC2: Deletion/duplication analysis is not offered for exons 24-25. OTOA: Deletion/duplication and sequencing analysis is not offered for exons 20-28. DUOX2: Deletion/duplication and sequencing analysis is not offered for exons 6-7. TBCE: Sequencing analysis for exons 2 includes only cds +/- 10 bp. PTPRC: Sequencing analysis is not offered for exons 3, 15. GHR: Deletion/duplication and sequencing analysis is not offered for exon 3. CFTR: Sequencing analysis for exons 7 includes only cds +/- 10 bp. EYS: Sequencing analysis for exons 30 includes only cds +/- 0 bp. 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. BBS9: Deletion/duplication analysis is not offered for exon 4. OAT: Deletion/duplication analysis is not offered for exon 2. 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. TSFM: Sequencing analysis is not offered for exon 5. VPS53: Sequencing analysis for exons 14 includes only cds +/- 5 bp. COL11A2: Deletion/duplication analysis is not offered for exon 36. 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, 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. ANO10: Sequencing analysis for exons 8 includes only cds +/- 0 bp. ATP8B1: Sequencing analysis for exons 19 includes only cds +/- 10 bp. 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. LIFR: Sequencing analysis for exons 3 includes only cds +/- 5 bp. AIPL1: Sequencing analysis for


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exons 2 includes only cds +/- 10 bp. TG: Deletion/duplication analysis is not offered for exon 18. Sequencing analysis for exons 44 includes only cds +/- 0 bp. TYR: Deletion/duplication and sequencing analysis is not offered for exon 5. FANCL: Sequencing analysis for exons 4, 10 includes only cds +/- 10 bp. USH1C: Deletion/duplication analysis is not offered for exons 5-6. AMN: Deletion/duplication analysis is not offered for exon 1. GALC: Deletion/duplication analysis is not offered for exon 6. MLC1: Sequencing analysis for exons 11 includes only cds +/- 10 bp. PEX1: Sequencing analysis for exons 16 includes only cds +/- 0 bp. ATM: Sequencing analysis for exons 6, 24, 43 includes only cds +/- 10 bp. FAH: Deletion/duplication analysis is not offered for exon 14.

## This report has been reviewed and approved by:

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INVITAE

# CARRIER DETECTION RATES AND RESIDUAL RISKS

This table is relevant to patient report RQ4960593
Issue date: 04/28/2023

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)<br>NM_000191.2 | HMGCL | Pan-ethnic | ≤1 in 500 | 99% | Reduced |
| 17-beta hydroxysteroid dehydrogenase 3 deficiency (AR)<br>NM_000197.1 | HSD17B3 | Pan-ethnic | ≤1 in 500 | 99% | Reduced |
| ABCA3-related conditions (AR)<br>NM_001089.2 | ABCA3 | Pan-ethnic | 1 in 277 | 99% | 1 in 27600 |
| ABCA4-related conditions (AR)<br>NM_000350.2 | ABCA4 | Pan-ethnic | 1 in 45 | 90% | 1 in 441 |
| ABCB4-related conditions (AR)<br>NM_000443.3 | ABCB4 | Pan-ethnic | 1 in 204 | 99% | 1 in 20300 |
| ABCB11-related conditions (AR)<br>NM_003742.2 | ABCB11 | Pan-ethnic | 1 in 100 | 99% | 1 in 9900 |
| ABCC8-related conditions (AR)<br>NM_000352.4<br>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)<br>NM_000253.3 | MTTP | Pan-ethnic | ≤1 in 500 | 99% | Reduced |
| Achromatopsia (CNGB3-related) (AR)<br>NM_019098.4 | CNGB3 | Pan-ethnic | 1 in 93 | 99% | 1 in 9200 |
| ACOX1-related conditions (AR)<br>NM_004035.6 | ACOX1 | Pan-ethnic | ≤1 in 500 | 99% | Reduced |
| Acrodermatitis enteropathica (AR)<br>NM_130849.3 | SLC39A4 | Pan-ethnic | 1 in 354 | 99% | 1 in 35300 |
| Adenosine deaminase deficiency (AR)<br>NM_000022.2 | ADA | Pan-ethnic | 1 in 224 | 92% | 1 in 2788 |
| ADGRV1-related conditions (AR)<br>NM_032119.3 | ADGRV1 | Pan-ethnic | 1 in 223 | 99% | 1 in 22200 |
| AHI1-related conditions (AR)<br>NM_017651.4 | AHI1 | Pan-ethnic | 1 in 447 | 99% | 1 in 44600 |
| Aicardi-Goutieres syndrome 2 (AR)<br>NM_024570.3 | RNASEH2B | Pan-ethnic | ≤1 in 500 | 99% | Reduced |
| Aicardi-Goutieres syndrome 3 (AR)<br>NM_032193.3 | RNASEH2C | Pan-ethnic | ≤1 in 500 | 99% | Reduced |
| Aicardi-Goutieres syndrome 4 (AR)<br>NM_006397.2 | RNASEH2A | Pan-ethnic | ≤1 in 500 | 99% | Reduced |
| Aicardi-Goutieres syndrome 5 (AR)<br>NM_015474.3 | SAMHD1 | Pan-ethnic | ≤1 in 500 | 99% | Reduced |
| AIPL1-related conditions (AR)<br>NM_014336.4 | AIPL1 * | Pan-ethnic | 1 in 408 | 99% | 1 in 40700 |
| Aldosterone synthase deficiency (AR)<br>NM_000498.3 | CYP11B2 | Pan-ethnic | ≤1 in 500 | 99% | Reduced |
| Alpha-mannosidosis (AR)<br>NM_000528.3 | MAN2B1 | Pan-ethnic | 1 in 354 | 99% | 1 in 35300 |
| Alpha-N-acetylgalactosaminidase deficiency (AR)<br>NM_000262.2 | NAGA | Pan-ethnic | ≤1 in 500 | 99% | Reduced |
| Alpha-thalassemia (AR)<br>NM_000558.4, NM_000517.4 | HBA1/<br>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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# CARRIER DETECTION RATES AND RESIDUAL RISKS

This table is relevant to patient report RQ4960593
Issue date: 04/28/2023

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

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Issue date: 04/28/2023

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

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# CARRIER DETECTION RATES AND RESIDUAL RISKS

This table is relevant to patient report RQ4960593
Issue date: 04/28/2023

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

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# CARRIER DETECTION RATES AND RESIDUAL RISKS

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

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

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|---|---|---|---|---|---|
| HGSNAT-related conditions (AR)<br>NM_152419.2 | HGSNAT | Pan-ethnic | ≤1 in 500 | 99% | Reduced |
| Holocarboxylase synthetase deficiency (AR)<br>NM_000411.6 | HLCS | Pan-ethnic | 1 in 224 | 99% | 1 in 22300 |
| Homocystinuria due to cobalamin E deficiency (AR)<br>NM_002454.2 | MTRR | Pan-ethnic | ≤1 in 500 | 99% | Reduced |
| Homocystinuria due to cobalamin G deficiency (AR)<br>NM_000254.2 | MTR | Pan-ethnic | ≤1 in 500 | 99% | Reduced |
| Homocystinuria due to cystathionine beta-synthase deficiency (AR)<br>NM_000071.2 | CBS | Pan-ethnic | 1 in 224 | 99% | 1 in 22300 |
| Homocystinuria due to MTHFR deficiency (AR)<br>NM_005957.4 | MTHFR * | Pan-ethnic | ≤1 in 500 | 99% | Reduced |
| HSD17B4-related conditions (AR)<br>NM_000414.3 | HSD17B4 | Pan-ethnic | 1 in 158 | 99% | 1 in 15700 |
| Hydrolethalus syndrome type 1 (AR)<br>NM_145014.2 | HYLS1 | Pan-ethnic | ≤1 in 500 | 99% | Reduced |
| Hyper-IgM immunodeficiency (CD40-related) (AR)<br>NM_001250.5 | CD40 | Pan-ethnic | ≤1 in 500 | 99% | Reduced |
| Hyperornithinemia-hyperammonemia-homocitrullinuria syndrome (AR)<br>NM_014252.3 | SLC25A15 | Pan-ethnic | ≤1 in 500 | 99% | Reduced |
| Hyperphosphatemic familial tumoral calcinosis (GALNT3-related) (AR)<br>NM_004482.3 | GALNT3 | Pan-ethnic | ≤1 in 500 | 99% | Reduced |
| Hypomyelinating leukodystrophy-12 (AR)<br>NM_021729.5 | VPS11 | Pan-ethnic | ≤1 in 500 | 99% | Reduced |
| Hypophosphatasia (AR)<br>NM_000478.5 | ALPL | Pan-ethnic | 1 in 150 | 95% | 1 in 2980 |
| Ichthyosis prematurity syndrome (AR)<br>NM_005094.3 | SLC27A4 | Pan-ethnic | ≤1 in 500 | 99% | Reduced |
| IGHMBP2-related conditions (AR)<br>NM_002180.2 | IGHMBP2 | Pan-ethnic | ≤1 in 500 | 99% | Reduced |
| IKBKB-related conditions (AR)<br>NM_001556.2 | IKBKB | Pan-ethnic | ≤1 in 500 | 99% | Reduced |
| Imerslund-Gräsbeck syndrome (AR)<br>NM_030943.3 | AMN * | Pan-ethnic | ≤1 in 500 | 99% | Reduced |
| Immunodeficiency-centromeric instability-facial anomalies syndrome 1 (AR)<br>NM_006892.3 | DNMT3B | Pan-ethnic | ≤1 in 500 | 99% | Reduced |
| Immunodeficiency-centromeric instability-facial anomalies syndrome 2 (AR)<br>NM_014797.2 | ZBTB24 | Pan-ethnic | ≤1 in 500 | 99% | Reduced |
| Isolated ectopia lentis (AR)<br>NM_019032.5 | ADAMTSL4 | Pan-ethnic | ≤1 in 500 | 99% | Reduced |
| Isovaleric acidemia (AR)<br>NM_002225.3 | IVD | Pan-ethnic | 1 in 250 | 99% | 1 in 24900 |
| ITGB3-related conditions (AR)<br>NM_000212.2 | ITGB3 | Pan-ethnic | ≤1 in 500 | 99% | Reduced |
| Johanson-Blizzard syndrome (AR)<br>NM_174916.2 | UBR1 | Pan-ethnic | 1 in 250 | 99% | 1 in 24900 |
| Joubert syndrome and related disorders (MKS1-related) (AR)<br>NM_017777.3 | MKS1 | Pan-ethnic | 1 in 260 | 95% | 1 in 5180 |
| Joubert syndrome and related disorders (RPGRIP1L-related) (AR)<br>NM_015272.2 | RPGRIP1L | Pan-ethnic | 1 in 259 | 95% | 1 in 5160 |
| Joubert syndrome and related disorders (TMEM216-related) (AR)<br>NM_001173990.2 | TMEM216 | Pan-ethnic | ≤1 in 500 | 99% | Reduced |
| Junctional epidermolysis bullosa (LAMC2-related) (AR)<br>NM_005562.2 | LAMC2 | Pan-ethnic | ≤1 in 500 | 99% | Reduced |

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# CARRIER DETECTION RATES AND RESIDUAL RISKS

This table is relevant to patient report RQ4960593
Issue date: 04/28/2023

| DISORDER (INHERITANCE) | GENE | ETHNICITY | CARRIER FREQUENCY | DETECTION RATE | RISK TO BE A CARRIER AFTER NEGATIVE RESULT |
|---|---|---|---|---|---|
| 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 |
| 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 |

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|---|---|---|---|---|---|
| Medium/short-chain 3-hydroxyacyl-CoA dehydrogenase deficiency (AR)<br>NM_005327.4 | HADH | Pan-ethnic | ≤1 in 500 | 99% | Reduced |
| MEDNIK syndrome (AR)<br>NM_001283.3 | AP1S1 | Pan-ethnic | ≤1 in 500 | 99% | Reduced |
| Megalencephalic leukoencephalopathy with subcortical cysts 1 (AR)<br>NM_015166.3 | MLC1 * | Pan-ethnic | ≤1 in 500 | 99% | Reduced |
| Metabolic crises with rhabdomyolysis, cardiac arrhythmias and neurodegeneration (AR)<br>NM_152906.6 | TANGO2 | Pan-ethnic | ≤1 in 500 | 99% | Reduced |
| Metachromatic leukodystrophy (ARSA-related) (AR)<br>NM_000487.5 | ARSA | Pan-ethnic | 1 in 100 | 95% | 1 in 1980 |
| Methylmalonic acidemia (MCEE-related) (AR)<br>NM_032601.3 | MCEE | Pan-ethnic | ≤1 in 500 | 99% | Reduced |
| Methylmalonic acidemia (MMAA-related) (AR)<br>NM_172250.2 | MMAA | Pan-ethnic | 1 in 316 | 97% | 1 in 10500 |
| Methylmalonic acidemia (MMAB-related) (AR)<br>NM_052845.3 | MMAB | Pan-ethnic | 1 in 456 | 98% | 1 in 22750 |
| Methylmalonic acidemia (MUT-related) (AR)<br>NM_000255.3 | MUT | Pan-ethnic | 1 in 204 | 96% | 1 in 5075 |
| MFSD8-related conditions (AR)<br>NM_152778.2 | MFSD8 | Pan-ethnic | ≤1 in 500 | 99% | Reduced |
| Microcephalic osteodysplastic primordial dwarfism type II (AR)<br>NM_006031.5 | PCNT | Pan-ethnic | ≤1 in 500 | 99% | Reduced |
| Microcephaly, postnatal progressive, with seizures and brain atrophy (AR)<br>NM_004268.4 | MED17 | Pan-ethnic | ≤1 in 500 | 99% | Reduced |
| Mitochondrial complex I deficiency 1 (AR)<br>NM_002495.3 | NDUFS4 | Pan-ethnic | ≤1 in 500 | 99% | Reduced |
| Mitochondrial complex I deficiency 3 (AR)<br>NM_024407.4 | NDUFS7 | Pan-ethnic | 1 in 387 | 99% | 1 in 38600 |
| Mitochondrial complex I deficiency 4 (AR)<br>NM_007103.3 | NDUFV1 | Pan-ethnic | 1 in 387 | 99% | 1 in 38600 |
| Mitochondrial complex I deficiency 9 (AR)<br>NM_004553.4 | NDUFS6 | Pan-ethnic | ≤1 in 500 | 99% | Reduced |
| Mitochondrial complex I deficiency 10 (AR)<br>NM_174889.4 | NDUFAF2 | Pan-ethnic | 1 in 387 | 99% | 1 in 38600 |
| Mitochondrial complex I deficiency 16 (AR)<br>NM_024120.4 | NDUFAF5 | Pan-ethnic | ≤1 in 500 | 99% | Reduced |
| Mitochondrial complex I deficiency 19 (AR)<br>NM_017547.3 | FOXRED1 | Pan-ethnic | 1 in 376 | 99% | 1 in 37500 |
| Mitochondrial complex I deficiency 20/ACAD9 deficiency (AR)<br>NM_014049.4 | ACAD9 | Pan-ethnic | ≤1 in 500 | 99% | Reduced |
| Mitochondrial complex IV deficiency 6 (AR)<br>NM_004376.6 | COX15 | Pan-ethnic | ≤1 in 500 | 99% | Reduced |
| Mitochondrial complex IV deficiency 12 (AR)<br>NM_001171155.1 | PET100 | Pan-ethnic | 1 in 387 | 99% | 1 in 38600 |
| Mitochondrial complex IV deficiency / Leigh syndrome, French Canadian type (AR)<br>NM_133259.3 | LRPPRC | Pan-ethnic | ≤1 in 500 | 99% | Reduced |
| Mitochondrial DNA depletion syndrome-2 (AR)<br>NM_004614.4 | TK2 | Pan-ethnic | ≤1 in 500 | 99% | Reduced |
| Mitochondrial neurogastrointestinal encephalomyopathy (AR)<br>NM_001953.4 | TYMP | Pan-ethnic | ≤1 in 500 | 99% | Reduced |
| Mitochondrial trifunctional protein deficiency (HADHB-related) (AR)<br>NM_000183.2 | HADHB | Pan-ethnic | ≤1 in 500 | 99% | Reduced |
| MKKS-related conditions (AR)<br>NM_018848.3 | MKKS | Pan-ethnic | ≤1 in 500 | 99% | Reduced |

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|---|---|---|---|---|---|
| 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 |
| 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 |

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|---|---|---|---|---|---|
| Myotonia congenita (AR)<br>NM_000083.2 | CLCN1 | Pan-ethnic | 1 in 112 | 99% | 1 in 11100 |
| N-acetylglutamate synthase deficiency (AR)<br>NM_153006.2 | NAGS | Pan-ethnic | ≤1 in 500 | 99% | Reduced |
| Nemaline myopathy 2 (AR)<br>NM_001271208.1 | NEB * | Pan-ethnic | 1 in 158 | 95% | 1 in 3140 |
| Nephrogenic diabetes insipidus (AQP2-related) (AR)<br>NM_000486.5 | AQP2 | Pan-ethnic | ≤1 in 500 | 99% | Reduced |
| Nephronophthisis (INVS-related) (AR)<br>NM_014425.3 | INVS | Pan-ethnic | ≤1 in 500 | 99% | Reduced |
| Nephronophthisis (NPHP1-related) (AR)<br>NM_000272.3 | NPHP1 | Pan-ethnic | ≤1 in 500 | 99% | Reduced |
| Neuronal ceroid lipofuscinosis type 1 (AR)<br>NM_000310.3 | PPT1 | Pan-ethnic | 1 in 199 | 98% | 1 in 9900 |
| Neuronal ceroid lipofuscinosis type 2 (AR)<br>NM_000391.3 | TPP1 | Pan-ethnic | 1 in 250 | 97% | 1 in 8300 |
| Neuronal ceroid lipofuscinosis type 5 (AR)<br>NM_006493.2 | CLN5 | Pan-ethnic | ≤1 in 500 | 99% | Reduced |
| Neuronal ceroid lipofuscinosis type 6 (AR)<br>NM_017882.2 | CLN6 | Pan-ethnic | ≤1 in 500 | 99% | Reduced |
| Neuronal ceroid lipofuscinosis type 8 (AR)<br>NM_018941.3 | CLN8 | Pan-ethnic | ≤1 in 500 | 99% | Reduced |
| Neuronal ceroid lipofuscinosis type 10 (AR)<br>NM_001909.4 | CTSD | Pan-ethnic | ≤1 in 500 | 99% | Reduced |
| Niemann-Pick disease type C (NPC1-related) (AR)<br>NM_000271.4 | NPC1 | Pan-ethnic | 1 in 183 | 99% | 1 in 18200 |
| Niemann-Pick disease type C (NPC2-related) (AR)<br>NM_006432.3 | NPC2 | Pan-ethnic | ≤1 in 500 | 99% | Reduced |
| Niemann-Pick disease types A and B (AR)<br>NM_000543.4 | SMPD1 | Pan-ethnic | 1 in 250 | 95% | 1 in 4980 |
| Nijmegen breakage syndrome (AR)<br>NM_002485.4 | NBN | Pan-ethnic | ≤1 in 500 | 99% | Reduced |
| Nonsyndromic deafness (LOXHD1-related) (AR)<br>NM_144612.6 | LOXHD1 | Pan-ethnic | ≤1 in 500 | 99% | Reduced |
| Nonsyndromic deafness (MYO15A-related) (AR)<br>NM_016239.3 | MYO15A | Pan-ethnic | ≤1 in 500 | 99% | Reduced |
| Nonsyndromic deafness (OTOA-related) (AR)<br>NM_144672.3 | OTOA * | Pan-ethnic | ≤1 in 500 | 88% | Reduced |
| Nonsyndromic deafness (SYNE4-related) (AR)<br>NM_001039876.2 | SYNE4 | Pan-ethnic | ≤1 in 500 | 99% | Reduced |
| Nonsyndromic deafness (TMC1-related) (AR)<br>NM_138691.2 | TMC1 | Pan-ethnic | ≤1 in 500 | 99% | Reduced |
| Nonsyndromic deafness (TMPRSS3-related) (AR)<br>NM_024022.2 | TMPRSS3 | Pan-ethnic | ≤1 in 500 | 99% | Reduced |
| Nonsyndromic intellectual disability (CC2D1A-related) (AR)<br>NM_017721.5 | CC2D1A | Pan-ethnic | ≤1 in 500 | 99% | Reduced |
| NR2E3-related conditions (AR)<br>NM_014249.3 | NR2E3 | Pan-ethnic | ≤1 in 500 | 99% | Reduced |
| NSMCE3 deficiency (AR)<br>NM_138704.3 | NSMCE3 | Pan-ethnic | ≤1 in 500 | 99% | Reduced |
| Oculocutaneous albinism type 2 (AR)<br>NM_000275.2 | OCA2 | Pan-ethnic | 1 in 95 | 99% | 1 in 9400 |
| Oculocutaneous albinism type 3 (AR)<br>NM_000550.2 | TYRP1 | Pan-ethnic | ≤1 in 500 | 99% | Reduced |
| Oculocutaneous albinism type 4 (AR)<br>NM_016180.4 | SLC45A2 | Pan-ethnic | 1 in 158 | 99% | 1 in 15700 |
| Oculocutaneous albinism types 1A and 1B (AR)<br>NM_000372.4 | TYR * | Pan-ethnic | 1 in 100 | 97% | 1 in 3300 |
| OPA3-related conditions (AR)<br>NM_025136.3 | OPA3 | Pan-ethnic | ≤1 in 500 | 99% | Reduced |
| Osteogenesis imperfecta (BMP1-related) (AR)<br>NM_006129.4 | BMP1 | Pan-ethnic | ≤1 in 500 | 99% | Reduced |

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|---|---|---|---|---|---|
| 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 |
| 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 |

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|---|---|---|---|---|---|
| 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 |
| 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 |

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# CARRIER DETECTION RATES AND RESIDUAL RISKS

This table is relevant to patient report RQ4960593
Issue date: 04/28/2023

| DISORDER (INHERITANCE) | GENE | ETHNICITY | CARRIER FREQUENCY | DETECTION RATE | RISK TO BE A CARRIER AFTER NEGATIVE RESULT |
|---|---|---|---|---|---|
| Sandhoff disease (AR)<br>NM_000521.3 | HEXB | Pan-ethnic | 1 in 180 | 99% | 1 in 17900 |
| Schimke immuno-osseous dysplasia (AR)<br>NM_014140.3 | SMARCAL1 | Pan-ethnic | ≤1 in 500 | 99% | Reduced |
| Seckel syndrome (CEP152-related) (AR)<br>NM_014985.3 | CEP152 | Pan-ethnic | ≤1 in 500 | 99% | Reduced |
| Sepiapterin reductase deficiency (AR)<br>NM_003124.4 | SPR | Pan-ethnic | ≤1 in 500 | 99% | Reduced |
| Severe combined immunodeficiency due to CD3-delta deficiency (AR)<br>NM_000732.4 | CD3D | Pan-ethnic | ≤1 in 500 | 99% | Reduced |
| Severe combined immunodeficiency due to CD3-epsilon deficiency (AR)<br>NM_000733.3 | CD3E | Pan-ethnic | ≤1 in 500 | 99% | Reduced |
| Severe combined immunodeficiency due to CD45 deficiency (AR)<br>NM_002838.4 | PTPRC * | Pan-ethnic | ≤1 in 500 | 99% | Reduced |
| Severe combined immunodeficiency due to DCLRE1C (Artemis) deficiency (AR)<br>NM_001033855.2 | DCLRE1C | Pan-ethnic | ≤1 in 500 | 99% | Reduced |
| Severe combined immunodeficiency due to IL7R-alpha deficiency (AR)<br>NM_002185.3 | IL7R | Pan-ethnic | 1 in 348 | 99% | 1 in 34700 |
| Severe combined immunodeficiency due to JAK3 deficiency (AR)<br>NM_000215.3 | JAK3 | Pan-ethnic | 1 in 455 | 99% | 1 in 45400 |
| Severe combined immunodeficiency due to RAG1 deficiency (AR)<br>NM_000448.2 | RAG1 | Pan-ethnic | 1 in 301 | 99% | 1 in 30000 |
| Severe combined immunodeficiency due to RAG2 deficiency (AR)<br>NM_000536.3 | RAG2 | Pan-ethnic | ≤1 in 500 | 99% | Reduced |
| Severe congenital neutropenia due to G6PC3 deficiency (AR)<br>NM_138387.3 | G6PC3 | Pan-ethnic | ≤1 in 500 | 99% | Reduced |
| Severe congenital neutropenia due to HAX1 deficiency (AR)<br>NM_006118.3 | HAX1 | Pan-ethnic | ≤1 in 500 | 99% | Reduced |
| Severe congenital neutropenia due to VPS45 deficiency (AR)<br>NM_007259.4 | VPS45 | Pan-ethnic | ≤1 in 500 | 99% | Reduced |
| Sialic acid storage diseases (AR)<br>NM_012434.4 | SLC17A5 | Pan-ethnic | ≤1 in 500 | 99% | Reduced |
| Sialidosis (AR)<br>NM_000434.3 | NEU1 | Pan-ethnic | ≤1 in 500 | 99% | Reduced |
| Sjögren-Larsson syndrome (AR)<br>NM_000382.2 | ALDH3A2 | Pan-ethnic | ≤1 in 500 | 99% | Reduced |
| SLC12A6-related conditions (AR)<br>NM_133647.1 | SLC12A6 | Pan-ethnic | ≤1 in 500 | 99% | Reduced |
| SLC26A2-related conditions (AR)<br>NM_000112.3 | SLC26A2 | Pan-ethnic | 1 in 158 | 95% | 1 in 3140 |
| SLC26A4-related conditions (AR)<br>NM_000441.1 | SLC26A4 | Pan-ethnic | 1 in 80 | 99% | 1 in 7900 |
| SLC37A4-related conditions (AR)<br>NM_001164277.1 | SLC37A4 | Pan-ethnic | 1 in 354 | 95% | 1 in 7060 |
| Smith-Lemli-Opitz syndrome (AR)<br>NM_001360.2 | DHCR7 | Pan-ethnic | 1 in 71 | 99% | 1 in 7000 |
| Spastic paraplegia type 15 (AR)<br>NM_015346.3 | ZFYVE26 | Pan-ethnic | ≤1 in 500 | 99% | Reduced |
| Spastic paraplegia type 49 (AR)<br>NM_014844.3 | TECPR2 | Pan-ethnic | ≤1 in 500 | 99% | Reduced |
| Spastic tetraplegia, thin corpus callosum, and progressive microcephaly (AR)<br>NM_003038.4 | SLC1A4 | Pan-ethnic | ≤1 in 500 | 99% | Reduced |

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# CARRIER DETECTION RATES AND RESIDUAL RISKS

This table is relevant to patient report RQ4960593
Issue date: 04/28/2023

| DISORDER (INHERITANCE) | GENE | ETHNICITY | CARRIER FREQUENCY | DETECTION RATE | RISK TO BE A CARRIER AFTER NEGATIVE RESULT |
|---|---|---|---|---|---|
| SPG11-related conditions (AR)<br>NM_025137.3 | SPG11 | Pan-ethnic | 1 in 141 | 99% | 1 in 14000 |
| Spinal muscular atrophy (AR)<br>NM_000344.3<br>Carrier residual risks listed are for 2 copy SMN1 results.<br>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 |
| Spinocerebellar ataxia (ANO10-related) (AR)<br>NM_018075.3 | ANO10 * | Pan-ethnic | ≤1 in 500 | 99% | Reduced |
| Spondylocostal dysostosis (DLL3-related) (AR)<br>NM_016941.3 | DLL3 | Pan-ethnic | 1 in 350 | 99% | 1 in 34900 |
| Spondylocostal dysostosis (MESP2-related) (AR)<br>NM_001039958.1 | MESP2 | Pan-ethnic | 1 in 224 | 99% | 1 in 22300 |
| Steel syndrome (AR)<br>NM_032888.3 | COL27A1 | Pan-ethnic | ≤1 in 500 | 99% | Reduced |
| Steroid 5-alpha-reductase deficiency (AR)<br>NM_000348.3 | SRD5A2 | Pan-ethnic | ≤1 in 500 | 99% | Reduced |
| Stüve-Wiedemann syndrome (AR)<br>NM_002310.5 | LIFR * | Pan-ethnic | ≤1 in 500 | 99% | Reduced |
| Sulfite oxidase deficiency (AR)<br>NM_000456.2 | SUOX | Pan-ethnic | ≤1 in 500 | 99% | Reduced |
| SURF1-related conditions (AR)<br>NM_003172.3 | SURF1 | Pan-ethnic | 1 in 128 | 99% | 1 in 12700 |
| Tay-Sachs disease (AR)<br>NM_000520.4 | HEXA | Pan-ethnic | 1 in 250 | 99% | 1 in 24900 |
| TBCE-related conditions (AR)<br>NM_003193.4 | TBCE * | Pan-ethnic | ≤1 in 500 | 99% | Reduced |
| Thiamine-responsive megaloblastic anemia (AR)<br>NM_006996.2 | SLC19A2 | Pan-ethnic | ≤1 in 500 | 99% | Reduced |
| Thyroid dyshormonogenesis (SLC5A5-related) (AR)<br>NM_000453.2 | SLC5A5 | Pan-ethnic | ≤1 in 500 | 99% | Reduced |
| Thyroid dyshormonogenesis (TG-related) (AR)<br>NM_003235.4 | TG * | Pan-ethnic | ≤1 in 500 | 99% | Reduced |
| Thyroid dyshormonogenesis (TPO-related) (AR)<br>NM_000547.5 | TPO | Pan-ethnic | 1 in 129 | 99% | 1 in 12800 |
| TMEM67-related conditions (AR)<br>NM_153704.5 | TMEM67 | Pan-ethnic | 1 in 316 | 99% | 1 in 31500 |
| Transcobalamin II deficiency (AR)<br>NM_000355.3 | TCN2 | Pan-ethnic | ≤1 in 500 | 99% | Reduced |
| Transient infantile liver failure (AR)<br>NM_018006.4 | TRMU | Pan-ethnic | ≤1 in 500 | 99% | Reduced |
| TREX1-related conditions (AR)<br>NM_033629.4 | TREX1 | Pan-ethnic | ≤1 in 500 | 99% | Reduced |
| Trichohepatoenteric syndrome (SKIV2L-related) (AR)<br>NM_006929.4 | SKIV2L | Pan-ethnic | ≤1 in 500 | 99% | Reduced |
| Trichohepatoenteric syndrome (TTC37-related) (AR)<br>NM_014639.3 | TTC37 | Pan-ethnic | ≤1 in 500 | 99% | Reduced |
| TRIM32-related conditions (AR)<br>NM_012210.3 | TRIM32 | Pan-ethnic | 1 in 408 | 99% | 1 in 40700 |
| Trimethylaminuria (AR)<br>NM_006894.6 | FMO3 | Pan-ethnic | ≤1 in 500 | 99% | Reduced |
| Triple A syndrome (AR)<br>NM_015665.5 | AAAS | Pan-ethnic | ≤1 in 500 | 99% | Reduced |
| TSHR-related conditions (AR)<br>NM_000369.2 | TSHR | Pan-ethnic | 1 in 158 | 99% | 1 in 15700 |
| TULP1-related conditions (AR)<br>NM_003322.4 | TULP1 | Pan-ethnic | 1 in 296 | 99% | 1 in 29500 |
| Tyrosine hydroxylase deficiency (AR)<br>NM_199292.2 | TH | Pan-ethnic | ≤1 in 500 | 99% | Reduced |
| Tyrosinemia type I (AR)<br>NM_000137.2 | FAH * | Pan-ethnic | 1 in 125 | 95% | 1 in 2480 |

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# CARRIER DETECTION RATES AND RESIDUAL RISKS

This table is relevant to patient report RQ4960593
Issue date: 04/28/2023

| DISORDER (INHERITANCE) | GENE | ETHNICITY | CARRIER FREQUENCY | DETECTION RATE | RISK TO BE A CARRIER AFTER NEGATIVE RESULT |
|---|---|---|---|---|---|
| Tyrosinemia type II (AR)<br>NM_000353.2 | TAT | Pan-ethnic | 1 in 250 | 99% | 1 in 24900 |
| Tyrosinemia type III (AR)<br>NM_002150.2 | HPD | Pan-ethnic | ≤1 in 500 | 99% | Reduced |
| USH1C-related conditions (AR)<br>NM_005709.3 | USH1C * | Pan-ethnic | 1 in 353 | 90% | 1 in 3521 |
| USH2A-related conditions (AR)<br>NM_206933.2 | USH2A | Pan-ethnic | 1 in 112 | 99% | 1 in 11100 |
| Very long-chain acyl-CoA dehydrogenase deficiency (AR)<br>NM_000018.3 | ACADVL | Pan-ethnic | 1 in 100 | 99% | 1 in 9900 |
| Vici syndrome (AR)<br>NM_020964.2 | EPG5 | Pan-ethnic | ≤1 in 500 | 99% | Reduced |
| Vitamin D-dependent rickets type 1A (AR)<br>NM_000785.3 | CYP27B1 | Pan-ethnic | ≤1 in 500 | 99% | Reduced |
| Vitamin D-dependent rickets type 2A (AR)<br>NM_001017535.1 | VDR | Pan-ethnic | ≤1 in 500 | 99% | Reduced |
| VPS53-related conditions (AR)<br>NM_001128159.2 | VPS53 * | Pan-ethnic | ≤1 in 500 | 99% | Reduced |
| VRK1-related conditions (AR)<br>NM_003384.2 | VRK1 | Pan-ethnic | ≤1 in 500 | 99% | Reduced |
| VSX2-related conditions (AR)<br>NM_182894.2 | VSX2 | Pan-ethnic | ≤1 in 500 | 99% | Reduced |
| Warsaw syndrome (AR)<br>NM_030653.3 | DDX11 * | Pan-ethnic | ≤1 in 500 | 15% | Reduced |
| Werner syndrome (AR)<br>NM_000553.4 | WRN * | Pan-ethnic | 1 in 224 | 99% | 1 in 22300 |
| Wilson disease (AR)<br>NM_000053.3 | ATP7B | Pan-ethnic | 1 in 90 | 98% | 1 in 4450 |
| WNT10A-related conditions (AR)<br>NM_025216.2 | WNT10A | Pan-ethnic | 1 in 305 | 99% | 1 in 30400 |
| Wolcott-Rallison syndrome (AR)<br>NM_004836.6 | EIF2AK3 | Pan-ethnic | ≤1 in 500 | 99% | Reduced |
| Woodhouse-Sakati syndrome (AR)<br>NM_025000.3 | DCAF17 | Pan-ethnic | ≤1 in 500 | 99% | Reduced |
| Xeroderma pigmentosum complementation group A (AR)<br>NM_000380.3 | XPA | Pan-ethnic | ≤1 in 500 | 99% | Reduced |
| Xeroderma pigmentosum complementation group C (AR)<br>NM_004628.4 | XPC | Pan-ethnic | ≤1 in 500 | 99% | Reduced |
| Xeroderma pigmentosum, variant type (AR)<br>NM_006502.2 | POLH | Pan-ethnic | ≤1 in 500 | 99% | Reduced |
| Zellweger spectrum disorder (PEX1-related) (AR)<br>NM_000466.2 | PEX1 * | Pan-ethnic | 1 in 144 | 99% | 1 in 14300 |
| Zellweger spectrum disorder (PEX2-related) (AR)<br>NM_000318.2 | PEX2 | Pan-ethnic | ≤1 in 500 | 99% | Reduced |
| Zellweger spectrum disorder (PEX6-related) (AR)<br>NM_000287.3 | PEX6 | Pan-ethnic | 1 in 294 | 99% | 1 in 29300 |
| Zellweger spectrum disorder (PEX10-related) (AR)<br>NM_153818.1 | PEX10 | Pan-ethnic | ≤1 in 500 | 94% | Reduced |
| Zellweger spectrum disorder (PEX12-related) (AR)<br>NM_000286.2 | PEX12 | Pan-ethnic | 1 in 409 | 99% | 1 in 40800 |
| Zellweger spectrum disorder (PEX13-related) (AR)<br>NM_002618.3 | PEX13 | Pan-ethnic | ≤1 in 500 | 99% | Reduced |
| Zellweger spectrum disorder (PEX16-related) (AR)<br>NM_004813.2 | PEX16 | Pan-ethnic | ≤1 in 500 | 99% | Reduced |
| Zellweger spectrum disorder (PEX26-related) (AR)<br>NM_017929.5 | PEX26 | Pan-ethnic | ≤1 in 500 | 99% | Reduced |

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INVITAE TEST RESULTS
Variant list for:
Patient Name: DONOR 14474
Date of Birth: [redacted]
Invitae RQ: RQ4960593
Report release Date: 04.28.2023

This file provides a list of variants identified in the reportable range of the requisitioned or tested gene(s). The assigned variant classification reflects the interpretation at the time of clinical report release; classifications may change over time. THE DATA IN THE TABLE ARE FOR RESEARCH USE AND CLINICAL DECISIONS SHOULD NOT BE MADE SOLELY BASED ON RESULTS IN THIS SUPPLEMENT FILE. Additional professional evaluation and clinical confirmatory testing is recommended before using the information on the listed variants in this table to guide medical management. Variants may not have been clinically interpreted or orthogonally confirmed; the table may include variants that are potential sequencing artifacts (false positives); and variant-associated information may be inaccurate. Protein effects may not be included for all variants. Variants within a gene with known paralog interference and unknown zygosity may be located within the named gene, its pseudogene copy, or in both locations. Population frequencies are reported based on the gnomAD version used by the laboratory at the time of clinical report release. A population frequency of "None" may indicate that the variant was absent from the gnomAD data set, which could be due to nomenclature differences or variant filtering methods between the laboratory and gnomAD. Due to sequencing complexities, this may not be a comprehensive list of all variants within reportable range. Some types of variants, such as structural rearrangements (e.g. inversions, gene conversion events, translocations, etc.), variants embedded in sequence with complex architecture (e.g. short tandem repeats or segmental duplications) and others may not be detected or included in this Supplement. The transcript provided in this table may not be the only clinically relevant transcript or the transcript that the laboratory uses for reporting purposes. THE DATA IS PROVIDED TO YOU ON AN "AS IS" AND "AS AVAILABLE" BASIS WITHOUT WARRANTY OF ANY KIND EITHER EXPRESS OR IMPLIED. TO THE FULLEST EXTENT PERMISSIBLE PURSUANT TO APPLICABLE LAW, LABCORP GENETICS DISCLAIMS ALL WARRANTIES, EXPRESS OR IMPLIED, INCLUDING BUT NOT LIMITED TO ANY IMPLIED WARRANTIES OF MERCHANTABILITY, FITNESS FOR A PARTICULAR PURPOSE AND ACCURACY. LABCORP GENETICS EXPRESSLY DISCLAIMS ANY LIABILITY, WHETHER BASED IN CONTRACT, TORT, STRICT LIABILITY OR OTHERWISE, FOR ANY DIRECT, INDIRECT, INCIDENTAL, CONSEQUENTIAL OR SPECIAL DAMAGES ARISING OUT OF OR IN ANY WAY CONNECTED WITH ACCESS TO OR USE OF THE DATA, EVEN IF ADVISED OF THE POSSIBILITY OF SUCH DAMAGES, INCLUDING BUT NOT LIMITED TO ERRORS OR OMISSIONS IN THE DATA.

| Report Status | Gene | Variant | Protein Effect | Zygosity | Population Frequency | Classification |
|---|---|---|---|---|---|---|
| supplement | RYR1 | NM_000540.2:c.10218C>T | Silent | heterozygous | 21% | Benign |
| supplement | RYR1 | NM_000540.2:c.10687-10C>T | Intronic | heterozygous | 22% | Benign |
| supplement | RYR1 | NM_000540.2:c.1077T>C | Silent | homozygous | 99% | Benign |
| supplement | RYR1 | NM_000540.2:c.1668G>A | Silent | heterozygous | 70% | Benign |
| supplement | RYR1 | NM_000540.2:c.2286C>T | Silent | heterozygous | 70% | Benign |
| supplement | RYR1 | NM_000540.2:c.2943G>A | Silent | homozygous | 68% | Benign |
| supplement | RYR1 | NM_000540.2:c.594A>G | Silent | homozygous | 68% | Benign |
| supplement | RYR1 | NM_000540.2:c.7500G>A | Silent | heterozygous | 23% | Benign |
| supplement | RYR1 | NM_000540.2:c.7527G>A | Silent | heterozygous | 23% | Benign |
| supplement | RYR1 | NM_000540.2:c.7835+5A>G | Intronic | heterozygous | 45% | Benign |

| | | | | | | |
|---|---|---|---|---|---|---|
| supplement | RYR1 | NM_000540.2:c.7863C>T | Silent | heterozygous | 30% | Benign |
| supplement | RYR1 | NM_000540.2:c.7977G>A | Silent | heterozygous | 47% | Benign |
| supplement | RYR1 | NM_000540.2:c.8118T>C | Silent | heterozygous | 47% | Benign |
| supplement | RYR1 | NM_000540.2:c.8190T>C | Silent | heterozygous | 47% | Benign |
| supplement | RYR1 | NM_000540.2:c.8337G>A | Silent | heterozygous | 46% | Benign |
| supplement | RYR1 | NM_000540.2:c.8376G>A | Silent | heterozygous | 1.10% | Benign |
| supplement | RYR1 | NM_000540.2:c.8589T>C | Silent | heterozygous | 46% | Benign |
| supplement | RYR1 | NM_000540.2:c.8693-10G>C | Intronic | heterozygous | 46% | Benign |
| supplement | RYR1 | NM_000540.2:c.8816+17T>A | Intronic | heterozygous | 45% | Benign |
| supplement | RYR1 | NM_000540.2:c.9186A>G | Silent | heterozygous | 46% | Benign |

| Patient Information | Specimen Information | Client Information |
|---|---|---|
| **14474, DONOR**<br><br>**DOB:** [redacted] **AGE:** [redacted]<br>Gender: M Fasting: U<br>Phone: 858.732.8500<br>Patient ID: 14474<br>Health ID: 8573032228361864 | Specimen: EN373458P<br>Requisition: 0001419<br><br>Collected: 04/17/2023 / 09:50 PDT<br>Received: 04/18/2023 / 02:57 PDT<br>Reported: 04/28/2023 / 11:49 PDT | Client #: 92037397 MAIL000<br>OLLIFFE, JEFFREY F<br>SEATTLE SPERM BANK<br>Attn: STE B214<br>8950 VILLA LA JOLLA DR<br>LA JOLLA, CA 92037-1714 |

**COMMENTS:** FASTING:UNKNOWN

# Cytogenetic Report

## CHROMOSOME ANALYSIS, BLOOD - 14596 Lab:EZ

**CHROMOSOME ANALYSIS, BLOOD**

Order ID: 23-169723
Specimen Type: Blood
Clinical Indication: Sperm Donor

**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.

Reha M. Toydemir, MD, PhD, FACMG

Electronic Signature: 4/28/2023 2:04 PM



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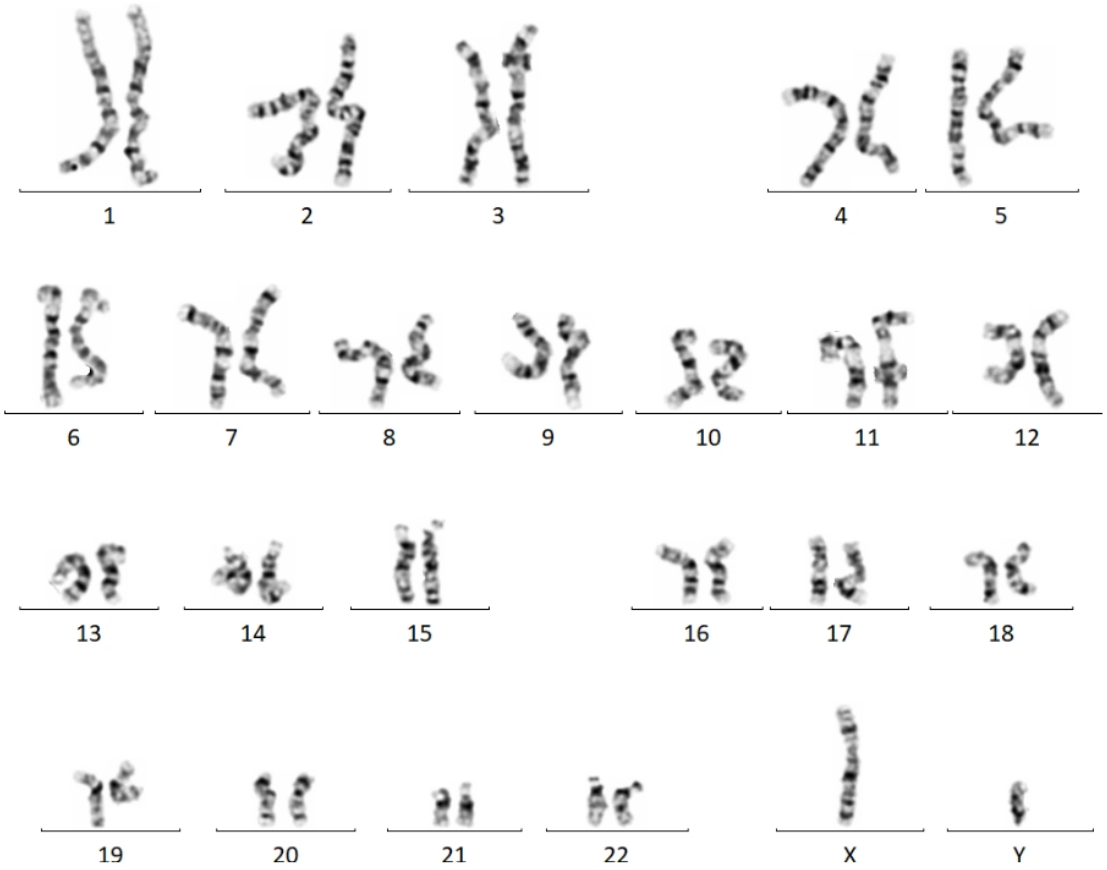
| Patient Information | Specimen Information | Client Information |
|---|---|---|
| **14474, DONOR**<br><br>**DOB:** [redacted] **AGE:** [redacted]<br>Gender: M Fasting: U<br>Patient ID: 14474<br>Health ID: 8573032228361864 | Specimen: EN373458P<br>Collected: 04/17/2023 / 09:50 PDT<br>Received: 04/18/2023 / 02:57 PDT<br>Reported: 04/28/2023 / 11:49 PDT | Client #: 92037397<br>OLLIFFE, JEFFREY F |




**PERFORMING SITE:**
EZ QUEST DIAGNOSTICS/NICHOLS SJC, 33608 ORTEGA HWY, SAN JUAN CAPISTRANO, CA 92675-2042 Laboratory Director: IRINA MARAMICA,MD,PHD,MBA, CLIA: 05D0643352



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**14474, Donor**
Patient ID:
Specimen ID: **107-253-2486-0**

DOB: [redacted]
Age: [redacted]
Sex: **Male**

**Patient Report**
Account Number: **46857540**
Ordering Physician: **J OLLIFFE**

labcorp

Date Collected: **04/17/2023** Date Received: **04/18/2023** Date Reported: **04/20/2023** Fasting: **Not Given**

Ordered Items: **LP+12AC+CBC/D/Plt+UA+Rh+ABO...**

Date Collected: **04/17/2023**

## LP+12AC+CBC/D/Plt+UA+Rh+ABO...

| Test | Current Result and Flag | Previous Result and Date | Units | Reference Interval |
|---|---|---|---|---|
| Glucose [01] | 86 | | mg/dL | 70-99 |
| Uric Acid [01] | 4.1 | | mg/dL | 3.8-8.4 |
| | | Therapeutic target for gout patients: <6.0<br>**Please note reference interval change** | | |
| BUN [01] | 8 | | mg/dL | 6-20 |
| Creatinine [01] | 0.76 | | mg/dL | 0.76-1.27 |
| eGFR | 120 | | mL/min/1.73 | >59 |
| Calcium [01] | 9.5 | | mg/dL | 8.7-10.2 |
| Protein, Total [01] | 7.9 | | g/dL | 6.0-8.5 |
| Albumin [01] | 4.9 | | g/dL | 4.0-5.0 |
| Bilirubin, Total [01] | 0.5 | | mg/dL | 0.0-1.2 |
| Alkaline Phosphatase [01] | 81 | | IU/L | 44-121 |
| LDH [01] | 154 | | IU/L | 121-224 |
| AST (SGOT) [01] | 14 | | IU/L | 0-40 |
| ALT (SGPT) [01] | 14 | | IU/L | 0-44 |
| Cholesterol, Total [01] | 116 | | mg/dL | 100-199 |
| Triglycerides [01] | 48 | | mg/dL | 0-149 |
| HDL Cholesterol [01] | 44 | | mg/dL | >39 |
| LDL Chol Calc (NIH) | 61 | | mg/dL | 0-99 |
| LDL/HDL Ratio | 1.4 | | ratio | 0.0-3.6 |
| Please Note: [01] | | | | |

LDL/HDL Ratio

| | Men | Women |
|---|---|---|
| 1/2 Avg.Risk | 1.0 | 1.5 |
| Avg.Risk | 3.6 | 3.2 |
| 2X Avg.Risk | 6.2 | 5.0 |
| 3X Avg.Risk | 8.0 | 6.1 |

| | | | | |
|---|---|---|---|---|
| Hgb Fractionation by CE: [02] | | | | |
| Hgb F [02] | 0.0 | | % | 0.0-2.0 |
| Hgb A [02] | 97.5 | | % | 96.4-98.8 |
| Hgb A2 [02] | 2.5 | | % | 1.8-3.2 |
| Hgb S [02] | 0.0 | | % | 0.0 |
| Interpretation: [02] | | | | |

Normal hemoglobin present; no hemoglobin variant or beta thalassemia identified.
Note: Alpha thalassemia may not be detected by the Hgb Fractionation Cascade panel. If alpha thalassemia is suspected, Labcorp offers Alpha-Thalassemia DNA Analysis (#511172).

| | | | | |
|---|---|---|---|---|
| ABO Grouping [01] | O | | | |
| Rh Factor [01] | Positive | | | |

Please note: Prior records for this patient's ABO / Rh type are not



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Date Collected: **04/17/2023**

## LP+12AC+CBC/D/Plt+UA+Rh+ABO... (Cont.)

available for additional verification.

| | | | | |
|---|---|---|---|---|
| .[01] | | | | |
| CBC, Platelet Ct, and Diff[01] | | | | |
| WBC[01] | 7.6 | | x10E3/uL | 3.4-10.8 |
| RBC[01] | 4.80 | | x10E6/uL | 4.14-5.80 |
| Hemoglobin[01] | 14.4 | | g/dL | 13.0-17.7 |
| Hematocrit[01] | 41.9 | | % | 37.5-51.0 |
| MCV[01] | 87 | | fL | 79-97 |
| MCH[01] | 30.0 | | pg | 26.6-33.0 |
| MCHC[01] | 34.4 | | g/dL | 31.5-35.7 |
| RDW[01] | 12.4 | | % | 11.6-15.4 |
| Platelets[01] | 345 | | x10E3/uL | 150-450 |
| Neutrophils[01] | 61 | | % | Not Estab. |
| Lymphs[01] | 26 | | % | Not Estab. |
| Monocytes[01] | 11 | | % | Not Estab. |
| Eos[01] | 2 | | % | Not Estab. |
| Basos[01] | 0 | | % | Not Estab. |
| Neutrophils (Absolute)[01] | 4.6 | | x10E3/uL | 1.4-7.0 |
| Lymphs (Absolute)[01] | 1.9 | | x10E3/uL | 0.7-3.1 |
| Monocytes(Absolute)[01] | 0.8 | | x10E3/uL | 0.1-0.9 |
| Eos (Absolute)[01] | 0.2 | | x10E3/uL | 0.0-0.4 |
| Baso (Absolute)[01] | 0.0 | | x10E3/uL | 0.0-0.2 |
| Immature Granulocytes[01] | 0 | | % | Not Estab. |
| Immature Grans (Abs)[01] | 0.0 | | x10E3/uL | 0.0-0.1 |
| .[01] | | | | |
| Urinalysis Gross Exam[01] | | | | |
| Specific Gravity[01] | 1.006 | | | 1.005-1.030 |
| pH[01] | 7.0 | | | 5.0-7.5 |
| Urine-Color[01] | Yellow | | | Yellow |
| Appearance[01] | Clear | | | Clear |
| WBC Esterase[01] | Negative | | | Negative |
| Protein[01] | Negative | | | Negative/Trace |
| Glucose[01] | Negative | | | Negative |
| Ketones[01] | Negative | | | Negative |
| ▶ **Occult Blood**[01] | **1+** | **Abnormal** | | Negative |
| Bilirubin[01] | Negative | | | Negative |
| Urobilinogen,Semi-Qn[01] | 0.2 | | mg/dL | 0.2-1.0 |
| Nitrite, Urine[01] | Negative | | | Negative |
| Microscopic Examination[01] | See below: | | | |
| Microscopic was indicated and was performed. | | | | |
| WBC[01] | None seen | | /hpf | 0 - 5 |
| RBC[01] | None seen | | /hpf | 0 - 2 |
| Epithelial Cells (non renal)[01] | None seen | | /hpf | 0 - 10 |
| Casts[01] | None seen | | /lpf | None seen |



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## LP+12AC+CBC/D/Plt+UA+Rh+ABO... (Cont.)

| | | |
|---|---|---|
| Bacteria [01] | None seen | None seen/Few |

**Disclaimer**
The Previous Result is listed for the most recent test performed by Labcorp in the past 5 years where there is sufficient patient demographic data to match the result to the patient. Results from certain tests are excluded from the Previous Result display.

**Icon Legend**
▲ Out of Reference Range ■ Critical or Alert

**Performing Labs**
01: SO - Labcorp San Diego 13112 Evening Creek Dr So Ste 200, San Diego, CA, 92128-4108 Dir: Jenny Galloway, MD
02: BN - Labcorp Burlington 1447 York Court, Burlington, NC, 27215-3361 Dir: Sanjai Nagendra, MD
For Inquiries, the physician may contact Branch: 206-861-7000 Lab: 858-668-3700

Patient Details
**14474, Donor**
**8950 VILLA LA JOLLA DR STE B214,STE, LA JOLLA, CA, 920371708**

Phone: **858-732-8500**
Date of Birth: [redacted]
Age: [redacted]
Sex: **Male**
Patient ID:
Alternate Patient ID:

Physician Details
**J OLLIFFE**
**Seattle Sperm Bank**
**4915 25th Ave Ne Ste 204, SEATTLE, WA, 98105**

Phone: **206-588-1484**
Account Number: **46857540**
Physician ID:
NPI: **1306838271**

Specimen Details
Specimen ID: **107-253-2486-0**
Control ID: **L2302491902**
Alternate Control Number: **L2302491902**
Date Collected: **04/17/2023 0950 Local**
Date Received: **04/18/2023 0000 ET**
Date Entered: **04/18/2023 0158 ET**
Date Reported: **04/20/2023 1008 ET**



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