# Donor 6665

# Genetic Testing Summary

Fairfax Cryobank recommends reviewing this genetic testing summary
with your healthcare provider to determine suitability.

Last Updated: 03/15/24

Donor Reported Ancestry: Indian                                    Jewish Ancestry: No

| Genetic Test* | Result | Comments/Donor's Residual Risk** |
|---|---|---|
| Chromosome analysis (karyotype) | Normal male karyotype | No evidence of clinically significant chromosome abnormalities |
| Hemoglobin evaluation | Normal hemoglobin fractionation and MCV/MCH results | Reduced risk to be a carrier for sickle cell anemia, beta thalassemia, alpha thalassemia trait (aa/-- and  a-/a-) and other hemoglobinopathies |
| Expanded Genetic Disease Carrier Screening Panel attached- 514 diseases by gene sequencing. | Negative for genes sequenced. | Residual risks for negative results can be seen here:<br><br>https://www.invitae.com/carrier-residual-risks/ |

*No single test can screen for all genetic disorders. A negative screening result significantly reduces, but cannot eliminate, the risk for these conditions in a pregnancy.

**Donor residual risk is the chance the donor is still a carrier after testing negative.

# INVITAE

| Patient name: | Donor 6665 | Sample type: | Blood | Report date: | 18-JUL-2023 |
| DOB: | ▓▓▓▓ | Sample collection date: | 10-JUL-2023 | Invitae #: | ▓▓▓▓ |
| Sex assigned at birth: | Male | Sample accession date: | 11-JUL-2023 | Clinical team: | ▓▓▓▓ |
| Gender: | | | | | |
| Patient ID (MRN): | | | | | |

**Reason for testing**

Gamete donor

**Test performed**

Invitae Comprehensive Carrier Screen without X-linked Disorders

- Primary Panel (CF, SMA)
- Add-on Comprehensive Carrier Screen without X-linked Disorders genes

## ⊖ 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.

# Results to note

**SMN1**

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

# 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.

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

| GENE | TRANSCRIPT | GENE | TRANSCRIPT | GENE | TRANSCRIPT |
|------|-----------|------|-----------|------|-----------|
| CRB1 | NM_201253.2 | EIF2B1 | NM_001414.3 | FUCA1 | NM_000147.4 |
| CRTAP | NM_006371.4 | EIF2B2 | NM_014239.3 | G6PC | NM_000151.3 |
| CTNS | NM_004937.2 | EIF2B3 | NM_020365.4 | G6PC3 | NM_138387.3 |
| CTSA | NM_000308.3 | EIF2B4 | NM_015636.3 | GAA | NM_000152.3 |
| CTSC | NM_001814.5 | EIF2B5 | NM_003907.2 | GALC* | NM_000153.3 |
| CTSD | NM_001909.4 | ELP1 | NM_003640.3 | GALE* | NM_000403.3 |
| CTSK | NM_000396.3 | EPG5 | NM_020964.2 | GALK1 | NM_000154.1 |
| CYBA | NM_000101.3 | ERCC2 | NM_000400.3 | GALNS | NM_000512.4 |
| CYP11A1 | NM_000781.2 | ERCC6 | NM_000124.3 | GALNT3 | NM_004482.3 |
| CYP11B1 | NM_000497.3 | ERCC8 | NM_000082.3 | GALT | NM_000155.3 |
| CYP11B2 | NM_000498.3 | ESCO2 | NM_001017420.2 | GAMT | NM_000156.5 |
| CYP17A1 | NM_000102.3 | ETFA | NM_000126.3 | GATM | NM_001482.2 |
| CYP19A1 | NM_031226.2 | ETFB | NM_001985.2 | GBA* | NM_001005741.2 |
| CYP1B1 | NM_000104.3 | ETFDH | NM_004453.3 | GBE1 | NM_000158.3 |
| CYP21A2* | NM_000500.7 | ETHE1 | NM_014297.3 | GCDH | NM_000159.3 |
| CYP27A1 | NM_000784.3 | EVC | NM_153717.2 | GCH1 | NM_000161.2 |
| CYP27B1 | NM_000785.3 | EVC2 | NM_147127.4 | GDF5 | NM_000557.4 |
| CYP7B1 | NM_004820.3 | EXOSC3 | NM_016042.3 | GFM1 | NM_024996.5 |
| DBT | NM_001918.3 | EYS* | NM_001142800.1 | GHR* | NM_000163.4 |
| DCAF17 | NM_025000.3 | FAH* | NM_000137.2 | GJB2 | NM_004004.5 |
| DCLRE1C | NM_001033855.2 | FAM161A | NM_001201543.1 | GLB1 | NM_000404.2 |
| DDX11* | NM_030653.3 | FANCA | NM_000135.2 | GLDC | NM_000170.2 |
| DFNB59 | NM_001042702.3 | FANCC | NM_000136.2 | GLE1 | NM_001003722.1 |
| DGAT1 | NM_012079.5 | FANCD2* | NM_033084.3 | GNE* | NM_001128227.2 |
| DGUOK | NM_080916.2 | FANCE | NM_021922.2 | GNPAT | NM_014236.3 |
| DHCR7 | NM_001360.2 | FANCG | NM_004629.1 | GNPTAB | NM_024312.4 |
| DHDDS | NM_024887.3 | FANCI | NM_001113378.1 | GNPTG | NM_032520.4 |
| DLD | NM_000108.4 | FANCL* | NM_018062.3 | GNS | NM_002076.3 |
| DLL3 | NM_016941.3 | FBP1 | NM_000507.3 | GORAB | NM_152281.2 |
| DNAH11 | NM_001277115.1 | FBXO7 | NM_012179.3 | GRHPR | NM_012203.1 |
| DNAH5 | NM_001369.2 | FH* | NM_000143.3 | GRIP1 | NM_021150.3 |
| DNAI1 | NM_012144.3 | FKBP10 | NM_021939.3 | GSS | NM_000178.2 |
| DNAI2 | NM_023036.4 | FKRP | NM_024301.4 | GUCY2D | NM_000180.3 |
| DNMT3B | NM_006892.3 | FKTN | NM_001079802.1 | GUSB | NM_000181.3 |
| DOK7 | NM_173660.4 | FMO3 | NM_006894.6 | HADH | NM_005327.4 |
| DUOX2* | NM_014080.4 | FOXN1 | NM_003593.2 | HADHA | NM_000182.4 |
| DYNC2H1 | NM_001080463.1 | FOXRED1 | NM_017547.3 | HADHB | NM_000183.2 |
| DYSF | NM_003494.3 | FRAS1 | NM_025074.6 | HAMP | NM_021175.2 |
| EIF2AK3 | NM_004836.6 | FREM2 | NM_207361.5 | HAX1 | NM_006118.3 |

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

| 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_00103183 5.2 |
| 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 |

| 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 |

# 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

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.

- GALE: Sequencing analysis for exons 10 includes only cds +/- 5 bp. DDX11: NM_030653.3:c.1763-1G>C variant only. DUOX2: Deletion/duplication and sequencing analysis is not offered for exons 6-7. PTPRC: Sequencing analysis is not offered for exons 3, 15. ABCC2: Deletion/duplication analysis is not offered for exons 24-25. OTOA: Deletion/duplication and sequencing analysis is not offered for exons 20-28. TBCE: Sequencing analysis for exons 2 includes only cds +/- 10 bp. PKHD1: Deletion/duplication analysis is not offered for exon 13. 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. 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. GHR: Deletion/duplication and sequencing analysis is not offered for exon 3. BBS9: Deletion/duplication analysis is not offered for exon 4. WRN: Deletion/duplication analysis is not offered for exons 10-11. Sequencing analysis for exons 8, 10-11 includes only cds +/- 10 bp. OAT: Deletion/duplication analysis is not offered for exon 2. 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. 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. ANO10: Sequencing analysis for exons 8 includes only cds +/- 0 bp. ATP8B1: Sequencing analysis for exons 19 includes only cds +/- 10 bp. 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. 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. AIPL1: Sequencing analysis for exons 2 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
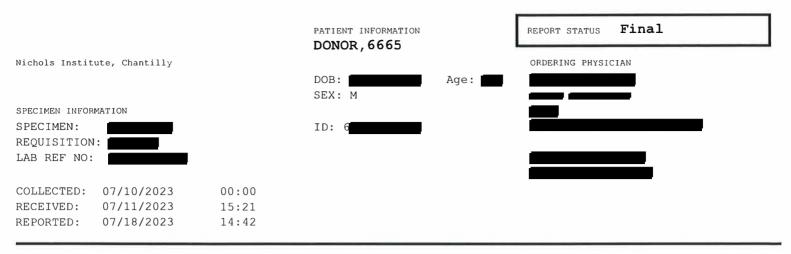
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includes only cds +/- 5 bp. AMN: Deletion/duplication analysis is not offered for exon 1. TYR: Deletion/duplication and sequencing analysis is not offered for exon 5. TG: Deletion/duplication analysis is not offered for exon 18. Sequencing analysis for exons 44 includes only cds +/- 0 bp. FANCL: Sequencing analysis for exons 4, 10 includes only cds +/- 10 bp. PEX1: Sequencing analysis for exons 16 includes only cds +/- 0 bp. USH1C: Deletion/duplication analysis is not offered for exons 5-6. ATM: Sequencing analysis for exons 6, 24, 43 includes only cds +/- 10 bp. FAH: Deletion/duplication analysis is not offered for exon 14. GALC: Deletion/duplication analysis is not offered for exon 6. MLC1: Sequencing analysis for exons 11 includes only cds +/- 10 bp.

## This report has been reviewed and approved by:

*Fatimah Nahhas*

Fatimah Nahhas-Alwan, PhD, FACMG
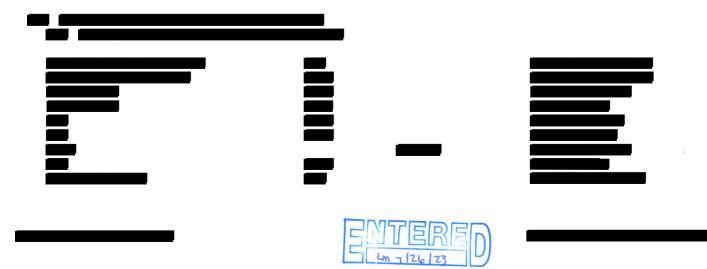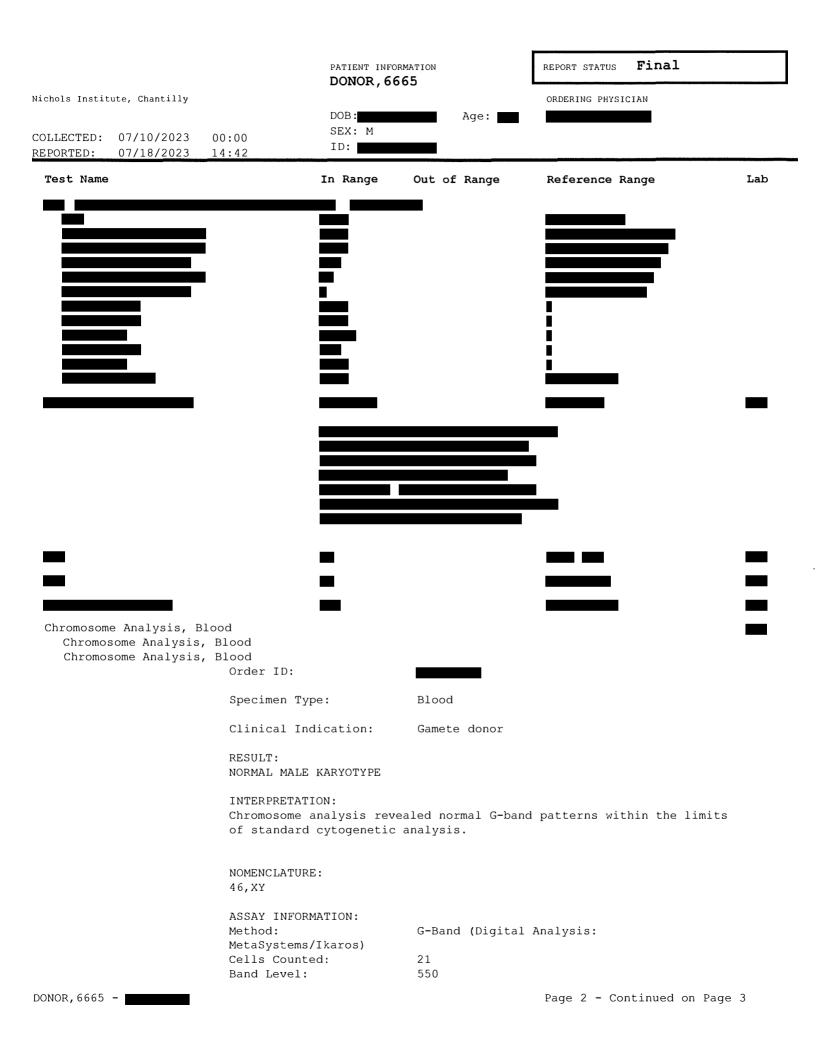Clinical Molecular Geneticist

PATIENT INFORMATION
**DONOR,6665**

REPORT STATUS  **Final**

DOB: ███████  Age: ███

SEX: M

ID: 6███████

ORDERING PHYSICIAN
███████████
████ ███████
███
███████████████████
███████████████
███████████████

COMMENTS:    =INDIA

| Test Name | In Range | Out of Range | Reference Range | |
|---|---|---|---|---|
| Hemoglobinopathy Evaluation | | | | |
| Red Blood Cell Count | 5.40 | | 4.20-5.80 Mill/uL | |
| HEMOGLOBIN | 15.1 | | 13.2-17.1 g/dL | |
| Hematocrit | | | | |
| Hematocrit | 48.2 | | 38.5-50.0 % | |
| MCV | 89.3 | | 80.0-100.0 fL | |
| MCH | 28.0 | | 27.0-33.0 pg | |
| RDW | 13.1 | | 11.0-15.0 % | |
| Hemoglobin A | 97.1 | | >96.0 % | |
| Hemoglobin F | 0.0 | | <2.0 % | |
| Hemoglobin A2 (Quant) | 2.9 | | 2.2-3.2 % | |
| Interpretation | | | | |

NORMAL PATTERN

There is a normal pattern of hemoglobins and
normal levels of Hb A2 and Hb F are present.
No variant hemoglobins are observed. This is
consistent with A/A phenotype.
If iron deficiency coexists with a mild/silent beta
thalassemia trait Hb A2 may be in the normal range.
Rare variant hemoglobins have no separation from
hemoglobin A by capillary zone electrophoresis (CZE) or
high-performance liquid chromatography (HPLC).
If clinically indicated, Thalassemia and
Hemoglobinopathy Comprehensive (TC 17365) should be
considered.

ENTERED
Lm 7/26/23

Nichols Institute, Chantilly

COLLECTED: 07/10/2023 00:00
REPORTED: 07/18/2023 14:42

DOB: ███████ Age: ██

SEX: M

ID: ███████

ORDERING PHYSICIAN

| Test Name | In Range | Out of Range | Reference Range | Lab |
|---|---|---|---|---|

Chromosome Analysis, Blood
   Chromosome Analysis, Blood
   Chromosome Analysis, Blood

Order ID: ███████

Specimen Type: Blood

Clinical Indication: Gamete donor

RESULT:
NORMAL MALE KARYOTYPE

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

NOMENCLATURE:
46,XY

ASSAY INFORMATION:
Method: G-Band (Digital Analysis:
MetaSystems/Ikaros)
Cells Counted: 21
Band Level: 550

Nichols Institute, Chantilly

ORDERING PHYSICIAN

COLLECTED: 07/10/2023 00:00
REPORTED: 07/18/2023 14:42

DOB: ▉  Age: ▉
SEX: M
ID: 6▉

| Test Name | In Range | Out of Range | Reference Range | Lab |
|---|---|---|---|---|

Chromosome Analysis, Blood (Continued)
  Chromosome Analysis, Blood (Continued)

       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.

Steven A. Schonberg, Ph.D., FACMG, Technical Director, Cytogenetics
and Genomics, 703-802-7156

Electronic Signature:       7/18/2023 1:57 PM

For additional information, please refer to
http://education.questdiagnostics.com/faq/chromsblood
(This link is being provided for informational/
educational purposes only).

----------------------------------------------------------------------------

**Performing Laboratory Information:**

AMD    Quest Diagnostics Nichols Institute 14225 Newbrook Drive Chantilly VA  20151 Laboratory Director: Patrick W Mason, MD PhD