Donor 6942

Genetic Testing Summary

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

Last Updated: 09/26/23

Donor Reported Ancestry: Chinese   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: 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

INVITAE CARRIER SCREEN RESULTS

Patient name:	Donor 6942	**Sample type:**	Blood	**Report date:**	24-FEB-2023
DOB:	▓▓▓▓	**Sample collection date:**	13-FEB-2023	**Invitae #:**	▓▓▓▓
Sex assigned at birth:	Male	**Sample accession date:**	14-FEB-2023	**Clinical team:**	▓▓▓▓
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.

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	GENE	TRANSCRIPT	GENE	TRANSCRIPT
PAH	NM_000277.1	POR	NM_000941.2	SGSH	NM_000199.3
PANK2	NM_153638.2	POU1F1	NM_000306.3	SKIV2L	NM_006929.4
PC	NM_000920.3	PPT1	NM_000310.3	SLC12A1	NM_000338.2
PCBD1	NM_000281.3	PRCD	NM_001077620.2	SLC12A3	NM_000339.2
PCCA	NM_000282.3	PRDM5	NM_018699.3	SLC12A6	NM_133647.1
PCCB	NM_000532.4	PRF1	NM_001083116.1	SLC17A5	NM_012434.4
PCDH15	NM_033056.3	PROP1	NM_006261.4	SLC19A2	NM_006996.2
PCNT	NM_006031.5	PSAP	NM_002778.3	SLC19A3	NM_025243.3
PDHB	NM_000925.3	PTPRC*	NM_002838.4	SLC1A4	NM_003038.4
PEPD	NM_000285.3	PTS	NM_000317.2	SLC22A5	NM_003060.3
PET100	NM_001171155.1	PUS1	NM_025215.5	SLC25A13	NM_014251.2
PEX1*	NM_000466.2	PYGM	NM_005609.3	SLC25A15	NM_014252.3
PEX10	NM_153818.1	QDPR	NM_000320.2	SLC25A20	NM_000387.5
PEX12	NM_000286.2	RAB23	NM_183227.2	SLC26A2	NM_000112.3
PEX13	NM_002618.3	RAG1	NM_000448.2	SLC26A3	NM_000111.2
PEX16	NM_004813.2	RAG2	NM_000536.3	SLC26A4	NM_000441.1
PEX2	NM_000318.2	RAPSN	NM_005055.4	SLC27A4	NM_005094.3
PEX26	NM_017929.5	RARS2	NM_020320.3	SLC35A3	NM_012243.2
PEX5	NM_001131025.1	RDH12	NM_152443.2	SLC37A4	NM_001164277.1
PEX6	NM_000287.3	RLBP1	NM_000326.4	SLC38A8	NM_001080442.2
PEX7	NM_000288.3	RMRP	NR_003051.3	SLC39A4	NM_130849.3
PFKM	NM_000289.5	RNASEH2A	NM_006397.2	SLC45A2	NM_016180.4
PGM3	NM_001199917.1	RNASEH2B	NM_024570.3	SLC4A11	NM_032034.3
PHGDH	NM_006623.3	RNASEH2C	NM_032193.3	SLC5A5	NM_000453.2
PHKB	NM_000293.2;NM_00103183 5.2	RPE65	NM_000329.2	SLC7A7	NM_001126106.2
PHKG2	NM_000294.2	RPGRIP1L	NM_015272.2	SMARCAL1	NM_014140.3
PHYH	NM_006214.3	RTEL1	NM_001283009.1	SMN1*	NM_000344.3
PIGN	NM_176787.4	RXYLT1	NM_014254.2	SMPD1	NM_000543.4
PKHD1*	NM_138694.3	RYR1	NM_000540.2	SNAP29	NM_004782.3
PLA2G6	NM_003560.2	SACS	NM_014363.5	SPG11	NM_025137.3
PLEKHG5	NM_020631.4	SAMD9	NM_017654.3	SPR	NM_003124.4
PLOD1	NM_000302.3	SAMHD1	NM_015474.3	SRD5A2	NM_000348.3
PMM2	NM_000303.2	SCO2	NM_005138.2	ST3GAL5	NM_003896.3
PNPO	NM_018129.3	SEC23B	NM_006363.4	STAR	NM_000349.2
POLG	NM_002693.2	SEPSECS	NM_016955.3	STX11	NM_003764.3
POLH	NM_006502.2	SGCA	NM_000023.2	STXBP2	NM_006949.3
POMGNT1	NM_017739.3	SGCB	NM_000232.4	SUMF1	NM_182760.3
POMT1	NM_007171.3	SGCD	NM_000337.5	SUOX	NM_000456.2
POMT2	NM_013382.5	SGCG	NM_000231.2	SURF1	NM_003172.3

GENE	TRANSCRIPT	GENE	TRANSCRIPT
SYNE4	NM_001039876.2	VDR	NM_001017535.1
TANGO2	NM_152906.6	VLDLR	NM_003383.4
TAT	NM_000353.2	VPS11	NM_021729.5
TBCD	NM_005993.4	VPS13A*	NM_033305.2
TBCE*	NM_003193.4	VPS13B	NM_017890.4
TCIRG1	NM_006019.3	VPS45	NM_007259.4
TCN2	NM_000355.3	VPS53*	NM_001128159.2
TECPR2	NM_014844.3	VRK1	NM_003384.2
TERT	NM_198253.2	VSX2	NM_182894.2
TF	NM_001063.3	WISP3	NM_003880.3
TFR2	NM_003227.3	WNT10A	NM_025216.2
TG*	NM_003235.4	WRN*	NM_000553.4
TGM1	NM_000359.2	XPA	NM_000380.3
TH	NM_199292.2	XPC	NM_004628.4
TK2	NM_004614.4	ZBTB24	NM_014797.2
TMC1	NM_138691.2	ZFYVE26	NM_015346.3
TMEM216	NM_001173990.2	ZNF469	NM_001127464.2
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		

Methods

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

- This report only includes variants that have a clinically significant association with the conditions tested as of the report date. Variants of uncertain significance, benign variants, and likely benign variants are not included in this report. However, if additional evidence becomes available to indicate that the clinical significance of a variant has changed, Invitae may update this report and provide notification.

- A PMID is a unique identifier referring to a published, scientific paper. Search by PMID at http://www.ncbi.nlm.nih.gov/pubmed.

- An rsID is a unique identifier referring to a single genomic position, and is used to associate population frequency information with sequence changes at that position. Reported population frequencies are derived from a number of public sites that aggregate data from large-scale population sequencing projects, including ExAC (http://exac.broadinstitute.org), gnomAD (http://gnomad.broadinstitute.org), and dbSNP (http://ncbi.nlm.nih.gov/SNP).

Disclaimer

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

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.

- VPS53: Sequencing analysis for exons 14 includes only cds +/- 5 bp. FH: Sequencing analysis for exons 9 includes only cds +/- 10 bp. GALC: Deletion/duplication analysis is not offered for exon 6. GNE: Sequencing analysis for exons 8 includes only cds +/- 10 bp. AIPL1: Sequencing analysis for exons 2 includes only cds +/- 10 bp. FANCL: Sequencing analysis for exons 4, 10 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. EYS: Sequencing analysis for exons 30 includes only cds +/- 0 bp. 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. GHR: Deletion/duplication and sequencing analysis is not offered for exon 3. 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. ABCC2: Deletion/duplication analysis is not offered for exons 24-25. LIFR: Sequencing analysis for exons 3 includes only cds +/- 5 bp. MLC1: Sequencing analysis for exons 11 includes only cds +/- 10 bp. 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. 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. PEX1: Sequencing analysis for exons 16 includes only cds +/- 0 bp. PKHD1: Deletion/duplication analysis is not offered for exon 13. FAH: Deletion/duplication analysis is not offered for exon 14. USH1C: Deletion/duplication analysis is not offered for exons 5-6. DUOX2: Deletion/duplication and sequencing analysis is not offered for exons 6-7. FANCD2: Deletion/duplication analysis is not offered for exons 14-17, 22 and sequencing analysis is not offered for exons 15-17. Sequencing analysis for exons 6, 14, 18, 20, 23, 25, 34 includes only cds +/- 10 bp. GALE: Sequencing analysis for exons 10 includes only cds +/- 5 bp. BBS9: Deletion/duplication analysis is not offered for exon 4. ATM: Sequencing analysis for exons 6, 24, 43 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. TSFM: Sequencing analysis is not offered for exon 5. VPS13A: Deletion/duplication analysis is not offered for exons 2-3, 27-28. ATP8B1: Sequencing analysis for exons 19 includes only cds +/- 10 bp. OTOA: Deletion/duplication and sequencing analysis is not offered for exons 20-28. TYR: Deletion/duplication and sequencing analysis is not offered for exon 5. PTPRC: Sequencing analysis is not offered for exons 3, 15. DDX11: NM_030653.3:c.1763-1G>C variant only. COL11A2: Deletion/duplication analysis is not offered for exon 36. ANO10: Sequencing analysis for exons 8 includes only cds +/- 0 bp. TBCE: Sequencing analysis for exons 2 includes only cds +/- 10 bp. SMN1: Systematic exon numbering is used for all genes, including SMN1, and for this reason the exon typically referred to as exon 7 in the literature (PMID: 8838816) is referred to as exon 8 in this report. This assay unambiguously detects SMN1 exon 8 copy number. The presence of the g.27134T>G variant (also known as c.*3+80T>G) is reported if SMN1 copy number = 2. SMN1 or SMN2: NM_000344.3:c.*3+80T>G variant only. AMN: Deletion/duplication analysis is not offered for exon 1. CFTR: Sequencing analysis for exons 7 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
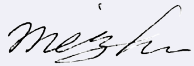
(p.Arg535His) variants only. Rarely, sensitivity to detect these variants may be reduced. When sensitivity is reduced, zygosity may be reported as "unknown". OAT: Deletion/duplication analysis is not offered for exon 2.

This report has been reviewed and approved by:

Mei Zhu

Mei Zhu, Ph.D., FACMG
Clinical Molecular Geneticist

6942,DONOR ⚠

DOB: ▓▓▓▓▓   Age: ▓▓   Specimen: ▓▓▓▓▓   Collected: 02/13/2023 13:00   Client #: 9595
Sex: M   Fasting: Requisition: ▓▓▓▓▓   Received: 02/14/2023 13:31
Phone: Lab Reference ▓▓▓▓▓   Reported: 02/23/2023 11:09
Patient ID: 6942-▓▓▓   Report Status: FINAL / SEE REPORT

⚠ Hemoglobinopathy Evaluation (FINAL)

Lab: AMD

Analyte	Value		
Hemoglobinopathy Evaluation			(FINAL)
Red Blood Cell Count	5.38	Reference Range: 4.20-5.80 Mill/uL	(FINAL)
HEMOGLOBIN	16.9	Reference Range: 13.2-17.1 g/dL	(FINAL)
Hematocrit			(FINAL)
⚠ **Hematocrit**	51.3 H	Reference Range: 38.5-50.0 %	(FINAL)
MCV	95.4	Reference Range: 80.0-100.0 fL	(FINAL)
MCH	31.4	Reference Range: 27.0-33.0 pg	(FINAL)
RDW	11.7	Reference Range: 11.0-15.0 %	(FINAL)
Hemoglobinopathy Evaluation			(FINAL)
Hemoglobin A	97.5	Reference Range: >96.0 %	(FINAL)
Hemoglobin F	0.0	Reference Range: <2.0 %	(FINAL)
Hemoglobin A2 (Quant)	2.5	Reference Range: 2.2-3.2 %	(FINAL)

Interpretation (FINAL)

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.

Chromosome Analysis, Blood   (FINAL)

Analyte	Value

Chromosome Analysis, Blood   (FINAL)

Order ID: 23-66365

Specimen Type: Blood

Clinical Indication: Donor screening

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:
MetaSystems/Ikaros)
Cells Counted: 20
Band Level: 550
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.

Debra Boles, Ph.D., FACMG, Technical Director, Cytogenetics and
Genomics, 703-802-7156

Electronic Signature: 2/23/2023 10:25 AM

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

Performing Sites