Genetic Carrier Screening Report

	Male ♂	Female ♀
Name	Mr. Dummy	Mrs. Dummy
Sample ID	XXX	XXX
Age	28 Years	26 Years
Ethnicity	Unknown	Unknown
Referring Clinician	NA	
Clinical History	Mr. Dummy and Mrs. Dummy are a prospective couple who intend to get married consanguineously. No significant personal and family history. Wants to know the genes common in them, with a potential to cause genetic disorders and rule out the possibility of autosomal recessive disorders. MLPA done- No Deletion and No Duplication detected in DMD gene.	
Consanguinity	Yes	

Sample Type	Blood	Sample Receipt Date	04.03.2024
Sample Collected	NA	Report Delivered	09.04.2024

Results and Interpretation

- Male and Female do not carry any common variants.
- The male was found to be a carrier for –
  - The *MYO3A* gene, c.3513_3525delTGAAGAGGAAACC, p.Glu1172Profs*17 in exon 30, that results in deafness, autosomal recessive 30, inherited in an autosomal recessive manner. The depth of the variant is 51X.
  - The *BBS2* gene, c.1527+1G>A, p.? in intron 12 that results in Bardet-Biedl syndrome 2, inherited in an autosomal recessive manner. The depth of the variant is 36X.
- The female was found to be a carrier for –
  - The *SHOC1* gene, c.1174+2T>C, p.? in intron 11 that results in spermatogenic failure 75, inherited in an autosomal recessive manner. The depth of the variant is 13X. Sanger validation of the *SHOC1* variant is recommended in the proband due to low allele read depth.
  - The *AGPAT2* gene, c.646A>T, p.Lys216Ter in exon 5 that results in lipodystrophy, congenital generalized, type 1, inherited in an autosomal recessive manner. The depth of the variant is 64X.
  - The *CDH23* gene, c.945+1G>A, p.? in intron 10 that results in deafness, autosomal recessive 12 and Usher syndrome, type 1D/ Usher syndrome, type 1D/F digenic, inherited in an autosomal recessive manner. The depth of the variant is 55X.

Variant Details

Disease	Male ♂	Female ♀
Deafness, autosomal recessive 30 [OMIM ID: 607101]	**CARRIER Gene: *MYO3A* Variant Location:**	

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	Chr10:26462700:CAAACCTGAAGAGG:C c.3513_3525delTGAAGAGGAAACC p.Glu1172Profs*17 Classification: Likely Pathogenic	***NONE***
Bardet-Biedl syndrome 2 [OMIM ID: 615981]	**CARRIER Gene: *BBS2* Variant Location: chr16:56533689:C>T c.1527+1G>A p.? Classification: Likely Pathogenic**	***NONE***
Spermatogenic failure 75 [OMIM ID: 619949]	***NONE***	**CARRIER Gene: *SHOC1* Variant Location: Chr9:114503754:A>G c.1174+2T>C p.? Classification: Likely Pathogenic**
Lipodystrophy, congenital generalized, type 1 [OMIM ID: 608594]	***NONE***	**CARRIER Gene: *AGPAT2* Variant Location: Chr9:139569202:T>A c.646A>T p.Lys216Ter Classification: Likely Pathogenic**
Deafness, autosomal recessive 12 [OMIM ID: 601386] / Usher syndrome, type 1D/ Usher syndrome, type 1D/F digenic [OMIM ID: 601067]	***NONE***	**CARRIER Gene: *CDH23* Variant Location: Chr10:73375374:G>A c.945+1G>A p.? Classification: Likely Pathogenic**

Note: Sanger validation of the SHOC1 variant is recommended in the proband due to low allele read depth.

Detailed Variant Interpretation

***MYO3A* Chr. 10:26462701 – Likely pathogenic:**

The frameshift deletion NM_017433.5(*MYO3A*):c.3513_3525delTGAAGAGGAAACC (p.Glu1172Profs*17) has not been reported previously as a pathogenic variant nor as a benign variant, to our knowledge. The p.Glu1172Profs*17 variant is not reported in any individuals in gnomAD All. The p.Glu1172Profs*17 variant is not reported in any individuals in 1kG All. This variant is predicted to cause loss of normal protein function through protein truncation causing a frameshift mutation. The frame shifted sequence continues 17 residues until a stop codon is reached. This variant is a frameshift variant which occurs in an exon of *MYO3A* upstream of where nonsense mediated decay is predicted to occur. There are 14 downstream pathogenic loss of function variants, with the furthest variant being 331 residues downstream of this variant. This indicates that the region is critical to protein function. The p.Glu1172Profs*17 variant is a loss of function variant in the gene *MYO3A*, which is intolerant of Loss of Function variants, as indicated by the presence of existing pathogenic loss of function variant NP_059129.3:p.Q45* and 17 others. For these reasons, this variant has been classified as **Likely Pathogenic.**

Deafness, autosomal recessive 30 [OMIM ID: 607101]:

Deafness, autosomal recessive 30 (DFNB30) is caused by homozygous or compound heterozygous mutation in the myosin IIIA gene (*MYO3A*; 606808). Walsh et al. (2002) studied a family that traced its ancestry to the Jewish community of Mosul, Iraq. This community dated to 586 B.C. and was highly endogamous, with considerable emigration but little immigration, for more than 2,500 years. Most remaining Jewish residents of Mosul, including this family, migrated to Israel in 1950-1951. Three generations of the family had experienced bilateral progressive hearing loss, which first affected the high frequencies. Hearing loss began in the second decade, and by age 50, was severe in high and middle frequencies and moderate at low frequencies. Vision and balance of all affected individuals were normal. Inheritance of deafness in this family was likely recessive with age-dependent penetrance, although dominant inheritance could not be excluded.

***BBS2* Chr. 16:56533689 – Likely pathogenic:**

The splice donor variant NM_031885.5(*BBS2*):c.1527+1G>A has been reported to ClinVar as Likely pathogenic with a status of (2 stars) criteria provided, multiple submitters, no conflicts (Variation ID 1339082 as of 2024-03-07). The c.1527+1G>A variant is not reported in any individuals in gnomAD All. The c.1527+1G>A variant is not reported in any individuals in 1kG All. This variant mutates a splice-donor sequence, potentially resulting in the retention of large segments of intronic DNA by the mRNA and nonfunctional proteins. This variant results in the loss of an donor splice site for the clinically relevant transcript. This variant disrupts the donor splice site for an exon upstream from the penultimate exon junction and is therefore predicted to cause nonsense mediated decay. The c.1527+1G>A variant is a loss of function variant in the gene *BBS2*, which is intolerant of Loss of Function variants, as indicated by the presence of existing pathogenic loss of function variant NP_114091.4:p.L31Afs*49 and 60 others. For these reasons, this variant has been classified as **Likely Pathogenic.**

Bardet-Biedl syndrome 2 [OMIM ID: 615981]:

Bardet-Biedl syndrome 2 (*BBS2*) is caused by homozygous or compound heterozygous mutations in the *BBS2* gene (606151) on chromosome 16q13. *BBS2* is an autosomal recessive ciliopathy characterized by retinal degeneration, polydactyly, renal disease, hypogonadism, obesity, dysmorphic features, and variable degrees of cognitive impairment (Innes et al., 2010). Mutation in the *BBS2* gene is the third most frequent cause of BBS, accounting for approximately 8% of cases (Zaghloul and Katsanis, 2009).

***SHOC1* Chr. 9:114503754 – Likely pathogenic:**

The splice donor variant NM_001378211.1(*SHOC1*):c.1174+2T>C has not been reported previously as a pathogenic variant nor as a benign variant, to our knowledge. The c.1174+2T>C variant is not reported in any individuals in gnomAD All. The c.1174+2T>C variant is not reported in any individuals in 1kG All. This variant mutates a splice-donor sequence, potentially resulting in the retention of large segments of intronic DNA by the mRNA and nonfunctional proteins. This variant results in the loss of an donor splice site for the clinically relevant transcript. This variant disrupts the donor splice site for an exon upstream from the penultimate exon junction and is therefore predicted to cause nonsense mediated decay. The c.1174+2T>C variant is a loss of function variant in the gene *SHOC1*, which is intolerant of Loss of Function variants, as indicated by the presence of existing pathogenic loss of function variant NP_001365140.1:p.E426Vfs*25. For these reasons, this variant has been classified as **Likely Pathogenic.**

Spermatogenic failure 75 [OMIM ID: 619949]:

Spermatogenic failure 75 (SPGF75) is caused by homozygous or compound heterozygous mutation in the *SHOC1* gene (618038) on chromosome 9q31. Spermatogenic failure-75 (SPGF75) is characterized by male infertility due to nonobstructive azoospermia resulting from maturation arrest at the spermatocyte stage (Krausz et al., 2020; Yao et al., 2021). Yao et al. (2021) studied 3 Chinese men from 2 families with infertility due to NOA and mutation in the *SHOC1* gene. In family 1, both affected brothers had slightly reduced testicular volumes but normal examination otherwise, and normal reproductive hormone levels. Semen analysis in both showed normal volume but complete azoospermia. In family 2, a brother and sister were infertile. The 28-year-old brother had normal-volume testes and normal hormone levels, but semen analysis showed normal volume with complete azoospermia. The proband reported that his older sister had a 3-year history of infertility, but she and her medical records were unavailable. The authors also reported a 25-year-old sporadic infertile male, who had

slightly reduced testicular volumes, normal reproductive hormone levels, and NOA. PAS assay of seminiferous tubule cross-sections from the infertile men showed a reduced number of spermatocytes and no spermatids or spermatozoa. Immunohistochemical staining of their testicular biopsies demonstrated absence of PNA, an acrosomal marker of spermatids and spermatozoa, and the XY body, a marker for the pachytene stage, indicating arrest at the spermatocyte stage. Meiotic chromosomal spread analysis showed no signal for the synaptonemal complex or XY body, consistent with spermatogenic arrest at the zygotene (meiosis I prophase I) stage.

***AGPAT2* Chr. 9:139569202 – Likely pathogenic:**

The stop gained NM_006412.4(*AGPAT2*):c.646A>T (p.Lys216Ter) has been reported to ClinVar as Conflicting classifications of pathogenicity with a status of criteria provided, conflicting classifications (Variation ID 365922 as of 2024-03-07). This variant is predicted to cause loss of normal protein function through protein truncation. There are 3 downstream pathogenic loss of function variants, with the furthest variant being 13 residues downstream of this variant. This indicates that the region is critical to protein function. The p.Lys216Ter variant is a loss of function variant in the gene *AGPAT2*, which is intolerant of Loss of Function variants, as indicated by the presence of existing pathogenic loss of function variant NP_006403.2:p.L13* and 6 others. For these reasons, this variant has been classified as **Likely Pathogenic.**

Lipodystrophy, congenital generalized, type 1 [OMIM ID: 608594]:

Lipodystrophy, congenital generalized, type 1 (CGL1) is caused by homozygous or compound heterozygous mutation in the gene encoding 1-acylglycerol-3-phosphate O-acyltransferase-2 (*AGPAT2*; 603100) on chromosome 9q34. Congenital generalized lipodystrophy (CGL), or Berardinelli-Seip syndrome, is a rare autosomal recessive disease characterized by a near absence of adipose tissue from birth or early infancy and severe insulin resistance. Other clinical and biologic features include acanthosis nigricans, muscular hypertrophy, hepatomegaly, altered glucose tolerance or diabetes mellitus, and hypertriglyceridemia (Garg, 2004).

***CDH23* Chr. 10:73375374 – Likely pathogenic:**

The splice donor variant NM_022124.6(*CDH23*):c.945+1G>A has been reported to ClinVar as Pathogenic with a status of (2 stars) criteria provided, multiple submitters, no conflicts (Variation ID 585305 as of 2024-03-07). The c.945+1G>A variant is not reported in any individuals in gnomAD All. The c.945+1G>A variant is not reported in any individuals in 1kG All. This variant mutates a splice-donor sequence, potentially resulting in the retention of large segments of intronic DNA by the mRNA and nonfunctional proteins. This variant results in the loss of an donor splice site for the clinically relevant transcript. This variant disrupts the donor splice site for an exon upstream from the penultimate exon junction and is therefore predicted to cause nonsense mediated decay. The c.945+1G>A variant is a loss of function variant in the gene *CDH23*, which is intolerant of Loss of Function variants, as indicated by the presence of existing pathogenic loss of function variant NP_071407.4:p.M1Afs*3 and 253 others. For these reasons, this variant has been classified as **Likely Pathogenic.**

Deafness, autosomal recessive 12 [OMIM ID: 601386]:

Deafness, autosomal recessive 12 (DFNB12) is caused by homozygous or compound heterozygous mutation in the cadherin-23 gene (*CDH23*; 605516) on chromosome 10q22. Chaib et al. (1996) described a consanguineous Sunni family with profound prelingual sensorineural hearing impairment living in an isolated village in Syria. Wagatsuma et al. (2007) reported 5 unrelated Japanese families with DFNB12. All patients had a similar phenotype, with moderate to profound high-frequency progressive sensorineural hearing loss. The average hearing loss was 84.0 dB. Vestibular function was normal.

Usher syndrome, type 1D/ Usher syndrome, type 1D/F digenic [OMIM ID: 601067]:

Usher syndrome type ID (USH1D) is caused by homozygous or compound heterozygous mutation in the gene encoding cadherin-23 (*CDH23*; 605516) on chromosome 10q22. The same gene is the site of mutation in a form of nonsyndromic autosomal recessive deafness, DFNB12 (601386). Usher syndrome type I is an autosomal recessive condition characterized by profound congenital hearing impairment with unintelligible speech, early retinitis pigmentosa (usually evident within the first decade), and constant vestibular dysfunction. Type I is distinguished from type II (276901) on the basis of severity of

hearing loss and the extent of vestibular involvement. Type I patients are profoundly deaf, whereas type II patients are 'hard of hearing.' Vestibular function is defective in type I patients, whereas type II patients have normal vestibular function (Moller et al., 1989). Patients with type III (USH3; 276902) have progressive hearing loss.

Recommendations:

- Genetic counseling is recommended to discuss the potential clinical implications of this result.
- **Clinical/ Genotype-phenotype correlation is strongly recommended.**
- **Sanger validation of the identified variant(s) in the couple and segregation analysis in the family members and close relatives is recommended.**
- **Re-analysis of the exome sequencing data can be done if additional phenotype is provided.**
- If the clinician suspects copy number variations as a cause of the patient's phenotype, then additional testing with chromosomal microarray is recommended with better sensitivity and specificity for the detection of copy number variants.
- Prenatal genetic testing or pre-implantation genetic diagnosis is not recommended for the variants reported as variants of uncertain significance (VUS).
- If the above results do not correlate completely with patient phenotype, additional testing is advised based on the clinician's recommendation.

Test Information and Methodology

Redcliffe's Carrier screening test is a panel of over ~570 genes that are associated with disorders inherited in an autosomal recessive (some X-linked recessive) manner and are mostly severe, childhood onset diseases. Carrier screen is intended for an individual at a reproductive age as a preconception or prenatal screen to determine if he/she carries one or more mutations for diseases. These mutations were selected based on current American College of Medical Genetics (ACMG) and American College of Obstetrics and Gynecology (ACOG) recommendations, as well as a thorough review of scientific literature and assessment of their clinical utility. This test is not intended for diagnostic testing of children suspected of having any of the diseases in the panel. Rare false negatives may occur in the setting of bone marrow transplantation, blood transfusion, and genetic variants such as other point mutations and deletions.

Methodology

Sequencing of the protein coding regions of approximately 30Mb of the human exome (targeting approximately 99% of regions in CCDS and RefSeq) was performed using Illumina NovaSeq platform at a mean depth of 80-100X and % of bases covered at 20X depth >90% in the target region. The individual's DNA was extracted and fragmented, with fragments from the coding regions of the selected gene panel targeted for amplification and sequencing. Reads from the sequence output were aligned to the human reference genome (GRCh37) using the Burrows-Wheeler Aligner (BWA). Duplicate reads identification and removal, base quality recalibration and re-alignment of reads based on indels were done using inbuilt DRAGEN bio-IT pipeline. Variants to the reference were called using the Genomic Analysis Tool Kit (GATK). The variants were annotated and filtered using the Golden Helix VarSeq and Varsome analysis workflow implementing the ACMG guidelines for interpretation of sequence variants. This includes comparison against the gnomAD population catalogue of variants in 123,136 exomes, the 1000 Genomes Project Consortium's publication of 2,500 genomes, the NCBI ClinVar database of clinical assertions on variant's pathogenicity and multiple lines of computational evidence on conservation and functional impact. All variants with minor allele frequency (MAF) of less than 1% in gnomAD database, and disease-causing variants reported in HGMD, in ClinVar are considered. The investigation for relevant variants is focused on coding exons and flanking +/-10 intronic nucleotides of genes with clear gene-phenotype evidence (based on OMIM information). All potential modes of inheritance patterns are considered. In addition, provided family history and clinical information are used to evaluate identified variants with respect to their pathogenicity and causality. This test has not been cleared or approved by the U.S. Food and Drug Administration (FDA). The FDA has determined that such clearance or approval is not necessary.

Test Limitations and Disclaimer

Mr. Dummy and Mrs. Dummy XXX and XXX

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Genetic testing is based upon information, developments and testing techniques that are known today. Future research may reveal changes in the interpretation of previously obtained genetic testing results.

Certain genes may not be covered completely, and few mutations may be missed. We sequence coding exons for each given transcript, plus ~10 bp of flanking non -coding DNA for each exon. Unless specifically indicated, test reports contain no information on about other portions of the gene, such as regulatory domains, deep intronic regions, uncharacterized alternative exons, chromosomal rearrangements, repeat expansions, epigenetic effects, and mitochondrial genome variants. Also, this analysis cannot detect single and multi-exon deletions and duplications.

A negative finding does not rule out a genetic diagnosis. These results should be used in the context of available clinical findings and should not be used as the sole basis for treatment. As with all medical laboratories testing, there is a small chance that the laboratory could report inaccurate information. For example, the laboratory could report that a given genotype is present when in fact it is not. Any kind of laboratory error may lead to incorrect decisions regarding medical treatment and/or diet and fitness recommendations. If a laboratory error has occurred or is suspected, a health care professional may wish to pursue further evaluation and/or other testing. Further testing may be pursued to verify any results for any reason.

This test has not been cleared or approved by the FDA. FDA does not require this test to go through premarket FDA review. Rare circumstances such as but not limited to including poor sample quality, sample mix-up, trace contamination, and other unforeseen technical incidents may prevent Redcliffe Life Sciences from releasing a result.

A consult with a genetic counsellor is recommended to properly review and explain these results to the tested individual and or medical professional.

Appendix:

List of Genes Analysed in Carrier Screening					
AAAS	***CEP290***	***FANCI***	***LAMA3***	***PEPD***	***SMARCAL1***
ABCA12	***CERKL***	***FANCL***	***LAMB3***	***PET100***	***SMN1***
ABCA3	***CFTR***	***FBP1***	***LAMC2***	***PEX1***	***SMPD1***
ABCA4	***CHAT***	***FBXO7***	***LARGE1***	***PEX10***	***SNAP29***
ABCB11	***CHM***	***FH***	***LCA5***	***PEX12***	***SPG11***
ABCB4	***CHRNE***	***FHL1***	***LDLR***	***PEX13***	***SPR***
ABCC2	***CHRNG***	***FKBP10***	***LDLRAP1***	***PEX16***	***SRD5A2***
ABCC6	***CIITA***	***FKRP***	***LHX3***	***PEX2***	***ST3GAL5***
ABCC8	***CLCN1***	***FKTN***	***LIFR***	***PEX26***	***STAR***
ABCD1	***CLN3***	***FMO3***	***LIG4***	***PEX5***	***STX11***

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ACAD9	CLN5	FMR1	LIPA	PEX6	STXBP2
ACADM	CLN6	FOXN1	LMBRD1	PEX7	SUMF1
ACADS	CLN8	FOXRED1	LOXHD1	PFKM	SUOX
ACADVL	CLRN1	FRAS1	LPL	PGM3	SURF1
ACAT1	CNGB3	FREM2	LRAT	PHGDH	SYNE4
ACOX1	COL11A2	FUCA1	LRP2	PHKB	TANGO2
ACSF3	COL17A1	FXN	LRPPRC	PHKG2	TAT
ADA	COL27A1	G6PC1	LYST	PHYH	TBCD
ADA2	COL4A3	G6PC3	MAK	PIGN	TBCE
ADAMTS2	COL4A4	GAA	MAN2B1	PKHD1	TCIRG1
ADAMTSL4	COL4A5	GALC	MANBA	PLA2G6	TCN2
ADGRG1	COL7A1	GALE	MCCC1	PLEKHG5	TECPR2
ADGRV1	COX15	GALK1	MCCC2	PLOD1	TERT
ADSL	CPS1	GALNS	MCEE	PLP1	TF
AFF2	CPT1A	GALNT3	MCOLN1	PMM2	TFR2
AGA	CPT2	GALT	MCPH1	PNPO	TG
AGL	CRB1	GAMT	MECP2	POLG	TGM1
AGPS	CRTAP	GATM	MECR	POLH	TH
AGXT	CTNS	GBA	MED17	POMGNT1	TK2
AHI1	CTSA	GBE1	MEFV	POMT1	TMC1
AIPL1	CTSC	GCDH	MESP2	POMT2	TMEM216

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AIRE	*CTSD*	*GCH1*	*MFSD8*	*POR*	*TMEM67*
ALDH3A2	*CTSK*	*GDF5*	*MID1*	*POU1F1*	*TMPRSS3*
ALDH7A1	*CYBA*	*GFM1*	*MKKS*	*PPT1*	*TNXB*
ALDOB	*CYBB*	*GHR*	*MKS1*	*PRCD*	*TPO*
ALG1	*CYP11A1*	*GJB1*	*MLC1*	*PRDM5*	*TPP1*
ALG13	*CYP11B1*	*GJB2*	*MLYCD*	*PRF1*	*TREX1*
ALG6	*CYP11B2*	*GJB6*	*MMAA*	*PROP1*	*TRIM32*
ALMS1	*CYP17A1*	*GLA*	*MMAB*	*PRPS1*	*TRIM37*
ALPL	*CYP19A1*	*GLB1*	*MMACHC*	*PSAP*	*TRMU*
AMN	*CYP1B1*	*GLDC*	*MMADHC*	*PTPRC*	*TSEN54*
AMT	*CYP21A2*	*GLE1*	*MMUT*	*PTS*	*TSFM*
ANO10	*CYP27A1*	*GNE*	*MOCS1*	*PUS1*	*TSHB*
AP1S1	*CYP27B1*	*GNPAT*	*MOCS2*	*PYGM*	*TSHR*
AQP2	*CYP7B1*	*GNPTAB*	*MPI*	*QDPR*	*TTPA*
AR	*DBT*	*GNPTG*	*MPL*	*RAB23*	*TULP1*
ARG1	*DCAF17*	*GNS*	*MPV17*	*RAG1*	*TYMP*
ARL6	*DCLRE1C*	*GORAB*	*MRE11*	*RAG2*	*TYR*
ARSA	*DDX11*	*GRHPR*	*MTHFR*	*RAPSN*	*TYRP1*
ARSB	*DGAT1*	*GRIP1*	*MTM1*	*RARS2*	*UBR1*
ARX	*DGUOK*	*GSS*	*MTR*	*RDH12*	*UNC13D*
ASL	*DHCR7*	*GUCY2D*	*MTRR*	*RLBP1*	*USH1C*

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ASNS	***DHDDS***	***GUSB***	***MTTP***	***RMRP***	***USH2A***
ASPA	***DKC1***	***HADH***	***MUSK***	***RNASEH2A***	***VDR***
ASS1	***DLD***	***HADHA***	***MVK***	***RNASEH2B***	***VLDLR***
ATM	***DLL3***	***HADHB***	***MYO15A***	***RNASEH2C***	***VPS11***
ATP6V1B1	***DMD***	***HAMP***	***MYO7A***	***RP2***	***VPS13A***
ATP7A	***DNAH11***	***HAX1***	***NAGA***	***RPE65***	***VPS13B***
ATP7B	***DNAH5***	***HBA1***	***NAGLU***	***RPGR***	***VPS45***
ATP8B1	***DNAI1***	***HBA2***	***NAGS***	***RPGRIP1L***	***VPS53***
ATRX	***DNAI2***	***HBB***	***NBN***	***RS1***	***VRK1***
AVPR2	***DNMT3B***	***HCFC1***	***NCF2***	***RTEL1***	***VSX2***
BBS1	***DOK7***	***HEXA***	***NDRG1***	***RXYLT1***	***WAS***
BBS10	***DPYD***	***HEXB***	***NDUFAF2***	***RYR1***	***WNT10A***
BBS12	***DUOX2***	***HFE***	***NDUFAF5***	***SACS***	***WRN***
BBS2	***DYNC2H1***	***HGSNAT***	***NDUFS4***	***SAMD9***	***XPA***
BBS4	***DYSF***	***HJV***	***NDUFS6***	***SAMHD1***	***XPC***
BBS5	***EDA***	***HLCS***	***NDUFS7***	***SCO2***	***ZBTB24***
BBS7	***EIF2AK3***	***HMGCL***	***NDUFV1***	***SEC23B***	***ZFYVE26***
BBS9	***EIF2B1***	***HMOX1***	***NEB***	***SEPSECS***	***ZNF469***
BCHE	***EIF2B2***	***HOGA1***	***NEU1***	***SERPINA1***	***AGPAT2***
BCKDHA	***EIF2B3***	***HPD***	***NGLY1***	***SGCA***	***SHOC1***
BCKDHB	***EIF2B4***	***HPRT1***	***NPC1***	***SGCB***	***MYO3A***

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BCS1L	*EIF2B5*	*HPS1*	*NPC2*	*SGCD*	
BLM	*ELP1*	*HPS3*	*NPHP1*	*SGCG*	
BLOC1S3	*EMD*	*HPS4*	*NPHS1*	*SGSH*	
BLOC1S6	*EPG5*	*HPS5*	*NPHS2*	*SLC12A1*	
BMP1	*ERCC2*	*HPS6*	*NR0B1*	*SLC12A3*	
BRIP1	*ERCC6*	*HSD17B10*	*NR2E3*	*SLC12A6*	
BSND	*ERCC8*	*HSD17B3*	*NSMCE3*	*SLC17A5*	
BTD	*ESCO2*	*HSD17B4*	*NTRK1*	*SLC19A2*	
BTK	*ETFA*	*HSD3B2*	*OAT*	*SLC19A3*	
CA3	*ETFB*	*HYAL1*	*OCA2*	*SLC1A4*	
CAD	*ETFDH*	*HYLS1*	*OCRL*	*SLC22A5*	
CANT1	*ETHE1*	*IDS*	*OPA3*	*SLC25A13*	
CAPN3	*EVC*	*IDUA*	*OSTM1*	*SLC25A15*	
CASQ2	*EVC2*	*IGHMBP2*	*OTC*	*SLC25A20*	
CBS	*EXOSC3*	*IKBKB*	*OTOA*	*SLC26A2*	
CC2D1A	*EYS*	*IL2RG*	*OTOF*	*SLC26A3*	
CC2D2A	*F11*	*IL7R*	*P3H1*	*SLC26A4*	
CCDC103	*F8*	*INVS*	*PAH*	*SLC27A4*	
CCDC39	*F9*	*ITGA6*	*PANK2*	*SLC35A3*	
CCDC88C	*FAH*	*ITGB3*	*PC*	*SLC37A4*	
CD3D	*FAM161A*	*ITGB4*	*PCBD1*	*SLC38A8*	

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CD3E	***FANCA***	***IVD***	***PCCA***	***SLC39A4***	
CD40	***FANCB***	***JAK3***	***PCCB***	***SLC45A2***	
CD40LG	***FANCC***	***KCNJ1***	***PCDH15***	***SLC4A11***	
CD59	***FANCD2***	***KCNJ11***	***PCNT***	***SLC5A5***	
CDH23	***FANCE***	***L1CAM***	***PDHA1***	***SLC6A8***	
CEP152	***FANCG***	***LAMA2***	***PDHB***	***SLC7A7***	

Residual Risk after Negative test results

In the case of a negative test result (not a carrier), there is a residual risk that the patient may have a mutation that is not part of the test panel. Included in the table below are the residual risk estimates for the carrier conditions in the carrier screening test. Population carrier rate, carrier detection rate and residual risk are shown for conditions and specific populations for which the data is known. For some other conditions and populations that are not shown, the prevalence is rare, the mutation detection rate is unknown and residual risk is not calculable.

Disease Information & Residual Risk					
Gene	**Disease**	**Ethnicity**	**Detection Rate**	**Carrier Frequency**	**Residual Risk**
ACADM	**Acyl-CoA dehydrogenase, medium chain, deficiency of**	**European Caucasian**	**>80%**	**1 in 50**	**< 1 in 250**
		Saudi Arabian	**95%**	**1 in 68**	**< 1 in 1300**
ACADVL	**Acyl-CoA dehydrogenase, very long-chain, deficiency of**	**General Population**	**> 18%**	**< 1 in 87**	**< 1 in 100**
ADA	**Severe combined immunodeficiency due to ADA deficiency**	**General Population**	**5%**	**1 in 500**	**< 1 in 525**

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Disease Information & Residual Risk					
Gene	**Disease**	**Ethnicity**	**Detection Rate**	**Carrier Frequency**	**Residual Risk**
AGA	**Aspartylglucosaminuria**	**Finnish**	**98%**	**1 in 69**	**< 1 in 3000**
AGL	**Glycogen storage disease IIIa, IIb**	**Caucasian** **North African Jewish**	**19% all GSD** **III, 51% GSD** **IIIb** **>99%**	**1 in 159** **1 in 37**	**1 in 196 all GSD III, <** **1 in 300 GSD IIIb** **< 1 in 3500**
AGXT	**Hyperoxaluria, primary, type 1**	**General Population**	**>33%**	**< 1 in 159**	**< 1 in 236**
AIRE	**Autoimmune polyendocrinopathy syndrome , type I, with or without reversible metaphyseal dysplasia**	**Finnish** **Iranian Jewish**	**89%** **>99%**	**1 in 80** **~ 1 in 48**	**1 in 715** **< 1 in 4500**
ALPL	**Hypophosphatasia**	**Japanese** **Manitoba Mennonite**	**52%** **>90%**	**< 1 in 159** **1 in 25**	**< 1 in 300** **< 1 in 246**
ARSA		**Austrian**	**>70%**	**1 in 100**	**<1 in 333**

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Disease Information & Residual Risk					
Gene	**Disease**	**Ethnicity**	**Detection Rate**	**Carrier Frequency**	**Residual Risk**
	Metachromatic leukodystrophy	**European Caucasian**	**44%**	**1 in 100**	**1 in 179**
		Habbanite Jewish	**>50%**	**1 in 5**	**< 1 in 9**
ASL	**Argininosuccinicaciduria**	**Dutch**	**56%**	**1 in 133**	**1 in 300**
		Saudi Arabian	**52%**	**1 in 80**	**1 in 165**
ASPA	**Canavan disease**	**Ashkenazi Jewish**	**98%**	**1 in 55**	**1 in 2715**
		General Population	**50%**	**< 1 in 100**	**< 1 in 200**
ATM	**Ataxia-telangiectasia**	**Amish**	**99%**	**Unknown**	**< 1 in 500**
		Costa Rican	**56%**	**1 in 100**	**1 in 227**
		North African Jewish	**97%**	**1 in 82**	**1 in 2700**
		Norwegian	**57%**	**1 in 100**	**1 in 232**
Gene	**Disease**	**Ethnicity**	**Detection Rate**	**Carrier Frequency**	**Residual Risk**
ATP7B	**Wilson disease**	**Ashkenazi Jewish**	**67%**	**1 in 100**	**1 in 300**
		European Caucasian	**40%**	**1 in 87**	**1 in 145**

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Disease Information & Residual Risk					
Gene	**Disease**	**Ethnicity**	**Detection Rate**	**Carrier Frequency**	**Residual Risk**
BBS1	**Bardet-Biedl syndrome 1**	**General Population**	**65%**	**< 1 in 250 (BBS1 only)**	**< 1 in 700**
BBS10	**Bardet-Biedl syndrome 10**	**General Population**	**48%**	**< 1 in 250 (BBS10 only)**	**< 1 in 500**
BBS12	**Bardet-Biedl syndrome 12**	**Caucasian** **General Population**	**27%** **19%**	**< 1 in 500 (BBS12 only)** **< 1 in 500 (BBS12 only)**	**< 1 in 680** **< 1 in 600**
BCKDHA	**Maple syrup urine disease, type Ia**	**Mennonite**	**99%**	**< 1 in 7**	**< 1 in 568**
BCKDHB	**Maple syrup urine disease, type Ib**	**Ashkenazi Jewish**	**99%**	**1 in 80**	**1 in 7900**
BCS1L	**Bjornstad syndrome**	**Finnish**	**>99%**	**1 in 109**	**< 1 in 10,000**
BLM	**Bloom syndrome**	**Ashkenazi Jewish** **European** **Japanese**	**97%** **40%** **44%**	**1 in 107** **Unknown** **Unknown**	**1 in 3520** **< 1 in 250** **< 1 in 250**

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Disease Information & Residual Risk					
Gene	**Disease**	**Ethnicity**	**Detection Rate**	**Carrier Frequency**	**Residual Risk**
CAPN3	**Muscular dystrophy, limb-girdle, type 2A**	**Bulgarian**	**58%**	**1 in 100**	**1 in 246**
		Croatian	**76%**	**1 in 133**	**1 in 550**
		Italian (Northeastern)	**38%**	**1 in 163**	**1 in 263**
		Russian	**45%**	**< 1 in 100**	**< 1 in 180**
		Turkish	**35%**	**1 in 100**	**1 in 160**
CBS	**Homocystinuria, B6-responsive and nonresponsive types**	**Irish**	**70%**	**1 in 128**	**< 1 in 400**
		Norwegian	**75%**	**1 in 41**	**< 1 in 150**
		Qatari	**>92%**	**< 1 in 22**	**< 1 in 260**
CDH23	**Deafness**	**General Population**	**9%**	**~1 in 134**	**< 1 in 147**
CEP290	**Bardet-Biedl syndrome 14**	**Northern European**	**48%**	**~ 1 in 224**	**1 in 430**
CFTR	**Cystic Fibrosis**	**African American**	**77%**	**1 in 61**	**1 in 262**
		Ashkenazi Jewish	**99%**	**1 in 24**	**1 in 2301**
		Asian	**55%**	**1 in 94**	**1 in 205**
		Caucasian	**92%**	**1 in 25**	**1 in 301**

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Disease Information & Residual Risk					
Gene	**Disease**	**Ethnicity**	**Detection Rate**	**Carrier Frequency**	**Residual Risk**
		Hispanic	**83%**	**1 in 58**	**1 in 336**
CHM	**Choroideremia**	**Finnish**	**90%**	**< 1 in 5000**	**< 1 in 57000**
CLN5	**Ceroid lipofuscinosis, neuronal, 5**	**Finnish**	**94%**	**1 in 100**	**< 1 in 1700**
CLN6	**Ceroid lipofuscinosis, neuronal, 6**	**Portuguese**	**80%**	**1 in 139**	**< 1 in 600**
CLN8	**Ceroid lipofuscinosis, neuronal, 8**	**Finnish**	**99%**	**1 in 135**	**< 1 in 13,000**
CLRN1	**Retinitis pigmentosa 61**	**Ashkenazi Jewish** **Finnish**	**92%** **95%**	**1 in 140** **1 in 100**	**< 1 in 13000** **1 in 1981**
CNGB3	**Achromatopsia 3**	**European** **Pingelapese** **(Micronesian)**	**83%** **99%**	**1 in 123** **1 in 3**	**< 1 in 700** **< 1 in 189**
CPT1A	**CPT deficiency, hepatic, type IA**	**Hutterite**	**95%**	**1 in 16**	**< 1 in 300**
CPT2	**Carnitinepalmitoyltransferase II deficiency**	**General Population**	**>50%**	**Unknown**	**< 1 in 500**
CTNS	**Cystinosis, nephropathic**	**French Canadian** **General Population (US)**	**54%** **62%**	**1 in 39** **1 in 159**	**1 in 84** **1 in 416**

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Disease Information & Residual Risk					
Gene	**Disease**	**Ethnicity**	**Detection Rate**	**Carrier Frequency**	**Residual Risk**
		Italian	**17%**	**1 in 159**	**1 in 191**
CTSK	**Pycnodysostosis**	**General Population**	**Unknown**	**Rare**	**< 1 in 380**
CYP17A1	**17,20-lyase deficiency**	**Brazilian** **Canadian Mennonite** **and Dutch Freislander** **Chinese**	**87%** **92%** **32%**	**< 1 in 112** **< 1 in 112** **< 1 in 112**	**< 1 in 850** **< 1 in 1300** **< 1 in 165**
CYP27A1	**Cerebrotendinousxanthomatosis**	**Caucasian**	**9%**	**1 in 115**	**1 in 127**
DCLRE1C	**Omenn syndrome**	**Navajo and Apache** **(Athabascan-speaking)**	**98%**	**1 in 23**	**< 1 in 1000**
DLD	**Dihydrolipoamide dehydrogenase deficiency**	**Ashkenazi Jewish**	**95%**	**< 1 in 80**	**< 1 in 1500**
DPYD	**Dihydropyrimidine dehydrogenase deficiency**	**General Population**	**52%**	**~ 1 in 51**	**~1 in 104**
ETFA	**Glutaricaciduria, type IA**	**European Caucasian**	**25%**	**Very rare**	**< 1 in 500**
Disease information & Residual Risk (cont)					

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Disease Information & Residual Risk					
Gene	**Disease**	**Ethnicity**	**Detection Rate**	**Carrier Frequency**	**Residual Risk**
Gene	**Disease**	**Ethnicity**	**Detection Rate**	**Carrier Frequency**	**Residual Risk**
ETFDH	**Glutaricaciduria, type IC**	**European Caucasian**	**17%**	**Very rare**	**< 1 in 500**
ETHE1	**Ethylmalonic encephalopathy**	**General Population**	**11%**	**Very rare**	**< 1 in 500**
F11	**Factor XI deficiency**	**Ashkenazi Jewish**	**95%**	**1 in 11**	**< 1 in 200**
		General Population	**12%**	**1 in 500**	**1 in 569**
FANCC	**Fanconianemia, complementation group C**	**Ashkenazi Jewish**	**99%**	**1 in 89**	**1 in 8801**
FANCG	**Fanconianemia, complementation group G**	**Brazilian**	**99%**	**Very rare**	**< 1 in 1000**
		Japanese	**65%**	**Very rare**	**< 1 in 1000**
FKTN	**Cardiomyopathy, dilated, 1L**	**Ashkenazi Jewish**	**99%**	**1 in 144**	**1 in 14179**
G6PC	**Glycogen storage disease Ia**	**Ashkenazi Jewish**	**99%**	**1 in 71**	**1 in 7022**
		Caucasian	**60%**	**1 in 159**	**1 in 395**
		Chinese	**80%**	**1 in 159**	**1 in 789**
		Hispanic	**<54%**	**1 in 159**	**< 1 in 344**

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Disease Information & Residual Risk					
Gene	**Disease**	**Ethnicity**	**Detection Rate**	**Carrier Frequency**	**Residual Risk**
		Japanese	**90%**	**1 in 159**	**< 1 in 1577**
		Korean	**75%**	**1 in 159**	**1 in 631**
GAA	**Glycogen storage disease II**	**African American**	**43%**	**1 in 60**	**1 in 104**
		Dutch	**32%**	**1 in 100**	**1 in 147**
GALC	**Krabe disease**	**European Caucasian**	**22%**	**1 in 159**	**1 in 191**
		Japanese	**57%**	**Unknown**	**< 1 in 350**
GALT	**Galactosemia**	**Ashkenazi Jewish**	**87.50%**	**1 in 127**	**< 1 in 1000**
		General Population	**~84%**	**1 in 87**	**< 1 in 500**
GBA	**Gaucher disease**	**Ashkenazi Jewish**	**96%**	**1 in 15**	**1 in 354**
		General Population (non-Jewish)	**70%**	**< 1 in 100**	**< 1 in 331**
GCDH	**Glutaricaciduria, type I**	**Amish**	**99%**	**1 in 12**	**< 1 in 1000**
		Caucasian	**>40%**	**1 in 112**	**< 1 in 187**
GNE	**Nonaka myopathy**	**Iranian Jewish**	**99%**	**1 in 20**	**< 1 in 1800**

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Disease Information & Residual Risk					
Gene	**Disease**	**Ethnicity**	**Detection Rate**	**Carrier Frequency**	**Residual Risk**
		Japanese	**73%**	**Unknown**	**< 1 in 500**
		Korean	**80%**	**Unknown**	**< 1 in 500**
GRHPR	**Hyperoxaluria, primary, type II**	**European Caucasian**	**30%**	**1 in 500**	**< 1 in 715**
HADHA	**Trifunctional protein deficiency**	**Northern European**	**71%**	**1 in 177**	**1 in 602**
HBB	**Sickle cell anemia**	**African American**	**80%**	**< 1 in 8**	**1 in 38**
		Indian	**>45%**	**1 in 20**	**< 1 in 35**
		Mediterranean	**>75%**	**1 in 7**	**1 in 24**
		Northern Spain (Seville)	**80%**	**1 in 8**	**1 in 75**
HEXB	**Sandhoff disease, infantile, juvenile, and adult forms**	**Argentinian Creole**	**97%**	**1 in 183**	**< 1 in 6000**
HGSNAT	**Mucopolysaccharidosis type IIIC (Sanfilippo C)**	**European Caucasian**	**80%**	**< 1 in 300**	**< 1 in 1700**
HLCS	**Holocarboxylasesynthetase deficiency**	**Faroese**	**Unknown**	**1 in 51**	**< 1 in 51**
		General Population	**44%**	**1 in 148**	**1 in 263**
		Japanese	**42%**	**1 in 159**	**1 in 273**

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Disease Information & Residual Risk					
Gene	**Disease**	**Ethnicity**	**Detection Rate**	**Carrier Frequency**	**Residual Risk**
HMGCL	**HMG-CoA lyase deficiency**	**Iberian Peninsula**	**84%**	**Unknown**	**< 1 in 500**
		Saudi Arabian	**94%**	**< 1 in 50**	**< 1 in 800**
HPS3	**Hermansky-Pudlak syndrome 3**	**Ashkenazi Jewish**	**89%**	**1 in 235**	**< 1 in 2000**
IDUA	**MucopolysaccharidosisIh/s**	**European Caucasian**	**35%**	**1 in 159**	**1 in 243**
		General Population	**21%**	**1 in 159**	**1 in 200**
		Italian	**39%**	**1 in 159**	**1 in 259**
		Moroccan	**92%**	**1 in 159**	**< 1 in 2000**
		Scandinavian	**62%**	**1 in 159**	**1 in 416**
IKBKAP	**Dysautonomia, familial**	**Ashkenazi Jewish**	**>99%**	**1 in 30**	**1 in 3000**
IL2RG	**Combined immunodeficiency, X-linked, moderate**	**General Population**	**19%**	**1 in 25,000**	**1 in 30,000**
LAMA3	**Epidermolysis bullosa, generalized atrophic benign**	**Pakistani**	**99%**	**Unknown**	**< 1 in 500**
LAMC2	**Epidermolysisbullosa, junctional, Herlitz type**	**Italian**	**33%**	**Unknown**	**< 1 in 500**
LRPPRC	**Leigh syndrome, French-Canadian type**	**French Canadian**	**95%**	**1 in 23**	**< 1 in 400**

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Disease Information & Residual Risk					
Gene	**Disease**	**Ethnicity**	**Detection Rate**	**Carrier Frequency**	**Residual Risk**
MCOLN1	**Mucolipidosis IV**	**Ashkenazi Jewish**	**95%**	**1 in 96**	**<1 in 1900**
MEFV	**Familial Mediterranean fever**	**Armenian**	**69%**	**< 1 in 5**	**< 1 in 14**
		Ashkenazi Jewish			
		Mediterranean			
		North African Jewish Turkish			
MKS1	**Bardet-Biedl syndrome 13**	**European**	**12%**	**1 in 188**	**1 in 212**
		Finnish	**55%**	**1 in 48**	**1 in 106**
		German	**47%**	**1 in 184**	**1 in 344**
MLC1	**Megalencephalic leukoencephalopathy with subcortical cysts**	**Libyan Jewish**	**>99%**	**1 in 40**	**< 1 in 4000**
MMAA	**Methylmalonic aciduria, vitamin B12-responsive**	**Caucasian**	**45%**	**Unknown**	**< 1 in 400**
MMACHC	**Methylmalonicaciduria and homocystinuria, cblC type**	**Chinese**	**54%**	**Very rare**	**< 1 in 500**
		General Population	**65%**	**Very rare**	**< 1 in 500**
		Italian	**75%**	**Very rare**	**< 1 in 500**
		Portuguese	**91%**	**Very rare**	**< 1 in 500**

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Disease Information & Residual Risk					
Gene	**Disease**	**Ethnicity**	**Detection Rate**	**Carrier Frequency**	**Residual Risk**
MPI	**Congenital disorder of glycosylation, type Ib**	**General Population**	**Unknown**	**Very rare**	**< 1 in 400**
MPL	**Thrombocytopenia, congenital amegakaryocytic**	**European Caucasian**	**~30%**	**Unknown**	**< 1 in 500**
MPV17	**Mitochondrial DNA depletion syndrome 6 (hepatocerebral type)**	**Navajo**	**99%**	**1 in 20**	**1 in 1950**
MTTP	**Abetalipoproteinemia**	**Ashkenazi Jewish**	**75%**	**1 in 131**	**< 1 in 500**
MUT	**Methylmalonic aciduria**	**African American**	**34%**	**Unknown**	**Unknown**
		European Caucasian	**20%**	**Unknown**	**Unknown**
		Hispanic	**55%**	**Unknown**	**Unknown**
		Japanese	**26%**	**Unknown**	**Unknown**
MYO7A	**Deafness**	**General Population**	**Unknown**	**Unknown**	**Unknown**
		Moroccan	**85%**	**Unknown**	**Unknown**
NAGLU	**Mucopolysaccharidosis type IIIB (Sanfilippo B)**	**Japanese**	**42%**	**1 in 200**	**1 in 345**
		Spanish Portuguese	**38%**	**1 in 187**	**1 in 300**

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Disease Information & Residual Risk					
Gene	**Disease**	**Ethnicity**	**Detection Rate**	**Carrier Frequency**	**Residual Risk**
NBN	**Nijmegen breakage syndrome**	**Eastern European**	**85%**	**1 in 155**	**< 1 in 1000**
NEB	**Nemaline myopathy 2, autosomal recessive**	**Ashkenazi Jewish**	**99%**	**< 1 in 108**	**< 1 in 10000**
NPC1	**Niemann-Pick disease, type C1,D**	**General Population**	**>15%**	**>1 in 174**	**<1 in 200**
NPHS1	**Nephrotic syndrome, type 1**	**Finnish**	**16%**	**1 in 46**	**1 in 54**
NPHS2	**Nephrotic syndrome, type 2**	**European** **Israeli-Arab**	**< 20%** **55%**	**Unknown** **Unknown**	**< 1 in 300** **< 1 in 500**
PAH	**Phenylketonuria**	**Caucasian** **Irish**	**47%** **68%**	**1 in 50** **1 in 34**	**1 in 94** **1 in 104**
PCCA	**Propionicacidemia**	**Japanese**	**15%**	**1 in 66**	**1 in 78**
PCCB	**Propionicacidemia**	**Japanese** **Spanish/Latin American**	**32%** **50%**	**< 1 in 66** **< 1 in 159**	**< 1 in 97** **< 1 in 316**
PEX1	**Heimler syndrome 1**	**General Population**	**>80%**	**1 in 140**	**< 1 in 700**
PEX7	**Peroxisome biogenesis disorder 9B**	**European Caucasian**	**72%**	**< 1 in 159**	**< 1 in 550**

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Disease Information & Residual Risk					
Gene	**Disease**	**Ethnicity**	**Detection Rate**	**Carrier Frequency**	**Residual Risk**
PKHD1	**Polycystic kidney disease 4, with or without hepatic disease**	**Caucasian**	**>20%**	**1 in 71**	**< 1 in 89**
		Finnish	**75%**	**1 in 71**	**1 in 282**
PMM2	**Congenital disorder of glycosylation, type Ia**	**European Caucasian**	**53%**	**1 in 71**	**1 in 150**
POLG	**Mitochondrial recessive ataxia syndrome**	**Scandinavian**	**59%**	**1 in 100**	**1 in 244**
POR	**Antley-Bixler syndrome**	**European Caucasian**	**40%**	**Unknown**	**< 1 in 500**
		General	**50%**	**Unknown**	**< 1 in 500**
			60%	**Unknown**	**< 1 in 500**
PPT1	**Ceroid lipofuscinosis, neuronal, 1**	**Finnish**	**98%**	**1 in 70**	**< 1 in 3000**
		General Population (US)	**59%**	**< 1 in 139**	**< 1 in 300**
PTS	**Hyperphenylalaninemia, BH4-deficient, A**	**Chinese**	**70%**	**1 in 180**	**< 1 in 600**
PYGM	**McArdle disease**	**Caucasian**	**>62%**	**1 in 159**	**< 1 in 400**
		Japanese	**71%**	**Unknown**	**Unknown**

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Disease Information & Residual Risk					
Gene	**Disease**	**Ethnicity**	**Detection Rate**	**Carrier Frequency**	**Residual Risk**
RAB23	**Carpenter syndrome**	**General Population**	**67%**	**< 1 in 500**	**< 1 in 1500**
		Northern European	**75%**	**< 1 in 500**	**< 1 in 2000**
RDH12	**Leber congenital amaurosis 13**	**General Population**	**40%**	**1 in 500**	**< 1 in 800**
RLBP1	**Bothnia retinal dystrophy, Retinitis punctataalbescens**	**Newfoundland, Northern Swedish**	**99%**	**Unknown**	**< 1 in 500**
			94%	**1 in 60 (Bothnia dystrophy)**	**< 1 in 900**
RS1	**Retinoschisis**	**European Caucasian**	**35%**	**< 1 in 2500**	**< 1 in 3800**
		Finnish	**95%**	**< 1 in 7500**	**< 1 in 150,000**
SGCA	**Muscular dystrophy, limb-girdle, type 2D**	**Brazilian**	**64%**	**1 in 250**	**1 in 694**
		European Caucasian	**23%**	**1 in 250**	**1 in 325**
SGCB	**Muscular dystrophy, limb-girdle, type 2E**	**Amish**	**99% (Indiana)**	**Unknown**	**< 1 in 500**
		General Population	**Unknown**	**Unknown**	**< 1 in 500**

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Disease Information & Residual Risk					
Gene	**Disease**	**Ethnicity**	**Detection Rate**	**Carrier Frequency**	**Residual Risk**
SGCG	**Muscular dystrophy, limb-girdle, type 2C**	**General Population**	**Unknown**	**~ 1 in 350**	**1 in 350**
		Gypsy/Romani	**99%**	**< 1 in 50**	**< 1 in 5000**
SGSH	**Mucopolysaccharidosis type IIIA (Sanfilippo A)**	**Italian**	**29%**	**1 in 126**	**1 in 176**
SLC12A6	**Agenesis of the corpus callosum with peripheral Neuropathy**	**French Canadian**	**99%**	**1 in 23**	**1 in 2200**
SLC17A5	**Sialic acid storage disorder, infantile**	**Finnish**	**97%**	**1 in 100 to 1 in 200**	**< 1 in 3000**
SLC25A15	**Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome**	**French Canadian**	**96%**	**1 in 20**	**1 in 472**
SLC26A4	**Deafness**	**European Caucasian**	**~20%**	**~1 in 58**	**~1 in 73**
SLC37A4	**Glycogen storage disease Ib**	**Caucasian**	**46%**	**1 in 350**	**< 1 in 650**
SLC45A2	**Albinism, oculocutaneous, type IV**	**Japanese**	**39%**	**1 in 146**	**1 in 239**
SLC7A7	**Lysinuric protein intolerance**	**Finnish**	**99%**	**1 in 138**	**< 1 in 10,000**
		Italian	**44%**	**< 1 in 120**	**< 1 in 200**
		Japanese	**64%**	**1 in 120**	**1 in 330**

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Disease Information & Residual Risk					
Gene	**Disease**	**Ethnicity**	**Detection Rate**	**Carrier Frequency**	**Residual Risk**
SMPD1	**Niemann-Pick disease, type A**	**Ashkenazi Jewish**	**95%**	**1 in 90 (Type A)**	**1 in 1780**
		General Population	**20%**	**1 in 159 (Type B)**	**1 in 200**
		North African	**87%**	**Unknown (Type B)**	**< 1 in 500**
		Saudi Arabian	**85%**	**1 in 100 (Type B)**	**< 1 in 650**
TGM1	**Ichthyosis, congenital**	**General Population**	**28%**	**1 in 224**	**< 1 in 300**
		Norwegian	**80%**	**1 in 151**	**< 1 in 750**
TMEM216	**Joubert syndrome 2**	**Ashkenazi Jewish**	**99%**	**1 in 92**	**1 in 9122**
TPP1	**Ceroid lipofuscinosis, neuronal, 2**	**European Caucasian**	**63%**	**1 in 139**	**< 1 in 350**
		Newfoundland	**67%**	**1 in 53**	**1 in 159**
TTPA	**Ataxia with isolated vitamin E deficiency**	**Italian**	**>50%**	**1 in 268**	**< 1 in 535**
		North African	**>80%**	**1 in 159**	**<1 in 789**
TYR	**Albinism, oculocutaneous, type IA**	**Chinese**	**11%**	**1 in 100**	**1 in 113**
UGT1A1	**Crigler-Najjar syndrome, type I**	**Dutch**	**34%**	**1 in 500**	**1 in 750**

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Genetic Carrier Screening Report

Disease Information & Residual Risk					
Gene	**Disease**	**Ethnicity**	**Detection Rate**	**Carrier Frequency**	**Residual Risk**
		Tunisian	**84%**	**1 in 500**	**< 1 in 3000**
USH1C	**Deafness 18A**	**Acadian**	**99%**	**Unknown**	**< 1 in 500**
		French Canadian	**40%**	**< 1 in 100**	**< 1 in 280**
USH2A	**Usher syndrome, type 2A**	**French Canadian**	**>55%**	**~ 1 in 125**	**< 1 in 275**
		General Population	**>20%**	**~ 1 in 125**	**< 1 in 150**
VPS13B	**Cohen syndrome**	**Amish (Ohio)**	**> 99%**	**1 in 12**	**< 1 in 1000**
		Finnish	**75%**	**1 in 120 - 1 in 160**	**< 1 in 480**
WRN	**Werner syndrome**	**Caucasian**	**29%**	**1 in 224**	**< 1 in 315**
		Japanese	**78%**	**< 1 in 71**	**< 1 in 315**
ABCC6	**Pseudoxanthoma Elasticum**	**European**	**28%**	**1/80 to 1/160**	**< 1 in 110**
ALDH7A1	**Pyridoxine-Dependent Epilepsy**	**Dutch**	**64%**	**< 1 in 260**	**< 1 in 725**
		European Caucasian	**33%**	**< 1 in 260**	**< 1 in 390**
CHRNE	**Congenital Myasthenic Syndrome, CHRNE-associated**	**European/Gypsy**	**>50%**	**< 1 in 20**	**< 1 in 39**

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Disease Information & Residual Risk					
Gene	**Disease**	**Ethnicity**	**Detection Rate**	**Carrier Frequency**	**Residual Risk**
		North African	**>44%**	**Unknown**	**< 1 in 400**
CRB1	**CRB1-associated Retinal Dystrophies**	**European Caucasian**	**~20%**	**~1 in 175**	**~ 1 in 220**
CYP1B1	**Primary Congenital Glaucoma**	**Caucasian**	**19%**	**1 in 51**	**1 in 62**
		Indian	**8% North, 17% South**	**< 1 in 29**	**< 1 in 32**
		Saudi Arabian	**10%**	**1 in 26**	**1 in 28**
		Slovakian Gypsy (Rom)	**99%**	**< 1 in 9**	**< 1 in 800**
CYP27B1	**Vitamin D-dependent Rickets, Type I**	**French Canadian**	**>89%**	**1 in 26**	**< 1 in 228**
DNAH5	**Primary Ciliary Dyskinesia, DNAH5-associated**	**Caucasian**	**15%**	**~1 in 120**	**~1 in 141**
DNAI1	**Primary Ciliary Dyskinesia, DNAI1-associated**	**Caucasian**	**17%**	**~ 1 in 200**	**~ 1 in 240**
		Polish	**33%**	**~ 1 in 200**	**~1 in 300**
EIF2B5	**Leukoencephalopathy with Vanishing White Matter**	**General Population**	**34%**	**Unknown**	**< 1 in 500**

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Disease Information & Residual Risk					
Gene	**Disease**	**Ethnicity**	**Detection Rate**	**Carrier Frequency**	**Residual Risk**
EYS	**Retinitis Pigmentosa, EYS-associated**	**Moroccan Jewish**	**Unknown**	**Unknown**	**< 1 in 34**
GP1BA	**Bernard-Soulier Syndrome, Type A1**	**General Population**	**Unknown**	**Very rare**	**< 1 in 500**
GP9	**Bernard-Soulier Syndrome, Type C**	**General Population**	**Unknown**	**Very rare**	**< 1 in 500**
GPR56	**Bilateral Frontoparietal Polymicrogyria (BFPP)**	**General Population**	**Unknown**	**Unknown**	**< 1 in 500**
LDLRAP1	**Familial Hypercholesterolemia, LDLRAP1 associated**	**Sardinian**	**54%**	**< 1 in 100**	**< 1 in 200**
MTRR	**Homocystinuria, cblE type**	**European**	**60%**	**Very rare**	**< 1 in 500**
NDRG1	**Charcot-Marie-Tooth Disease, Type 4D (CMT4D)**	**Gypsy/Romani**	**>99%**	**1 in 11**	**< 1 in 989**
PC	**Pyruvate Carboxylase Deficiency**	**Canadian Indian** **General Population**	**> 99%** **13%**	**1 in 10** **1 in 250**	**< 1 in 850** **1 in 288**
PEPD	**Prolidase Deficiency**	**Druze**	**67%**	**1 in 21**	**1 in 62**
RAPSN	**Congenital Myasthenic Syndrome, RAPSN-associated**	**General Population**	**70%**	**Unknown**	**< 1 in 500**

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Genetic Carrier Screening Report

Disease Information & Residual Risk					
Gene	**Disease**	**Ethnicity**	**Detection Rate**	**Carrier Frequency**	**Residual Risk**
SACS	**Autosomal Recessive Spastic Ataxia**	**Northeastern Quebec**	**95%**	**1 in 22**	**1 in 431**
SLC25A13	**Citrin Deficiency**	**Japanese**	**>30%**	**1 in 70**	**< 1 in 100**
WISP3	**Progressive Pseudorheumatoid Dysplasia (PPD)**	**Middle Eastern**	**~57%**	**Unknown**	**< 1 in 500**
WNT10A	**Odonto-onycho-dermal dysplasia/Schopf-Schulz-Passarge Syndrome**	**General Population**	**>36%**	**Unknown**	**< 1 in 500**

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Govind M Suresh
Senior Genome Analyst

Dr. Aruna Priya
Lead – Genetic Counselor & Reporting

Dr. Himani Pandey
Lab Head - Clinical Genomics
Post-Doc. Fellowship
(Medical Genetics) SGPGIMS

Dr. Shivani Mishra
DM (Medical Genetics, SGPGIMS)
MD(Obgyn), MBBS
Former Associate Professor,
KMC Hospital Manipal (MAHE)
Consultant Clinical Geneticist

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Genetic Carrier Screening Report

Conditions for Reporting

1. It is presumed that specimen belongs to patient named or identified, such verification being carried out at the point of generation of said specimen.
2. A test might not be performed due to following reasons:

a. Specimen Quantity not sufficient (Inadequate collection/spillage during transit).

b. Specimen Quality not acceptable (Hemolysis/clotted/lipemic.).

c. Incorrect sample type.

d. Test cancelled either on request of patient or doctor.

3. In any of the above case a fresh specimen will be required for testing and reporting.
4. The results of the tests may vary from lab to lab, time to time for the same patient.
5. The reported results are dependent on individual assay methods, equipment, method sensitivity, specificity and quality of the specimen received.
6. Partial representation of report is not allowed.
7. The reported tests are for the notification of the referring doctor, only to assist him/her in the diagnosis and management of the patient.
8. Report with status "Preliminary" means one or more tests are yet to be reported.
9. This report is not valid for Medico Legal Purpose.
10. Applicable Jurisdiction will be of "Delhi" for any dispute/claim concerning the test(s) & results of the test(s).

Disclaimer: Method given in report are only indicative and can be changed depending upon type of machine and kit available at time of testing.

Not all tests at all locations are under NABL scope. Availability of tests under NABL scope varies from lab to lab.

Terms and Conditions of Reporting

1. The presented findings in the Reports are intended solely for informational and interpretational purposes by the referring physician or other qualified medical professionals possessing a comprehensive understanding of reporting units, reference ranges, and technological limitations. The laboratory shall not be held liable for any interpretation or misinterpretation of the results, nor for any consequential or incidental damages arising from such interpretation.

2. It is to be presumed that the tests performed pertain to the specimen/sample attributed to the Customer's name or identification. It is presumed that the verification particulars have been cleared out by the customer or his/her representation at the point of generation of said specimen / sample. It is hereby clarified that the reports furnished are restricted solely to the given specimen only.

3. It is to be noted that variations in results may occur between different laboratories and over time, even for the same parameter for the same Customer. The assays are performed and conducted in accordance with standard procedures, and the reported outcomes are contingent on the specific individual assay methods and equipment(s) used, as well as the quality of the received specimen.

4. This report shall not be deemed valid or admissible for any medico-legal purposes.

5. The Customers assume full responsibility for apprising the Company of any factors that may impact the test finding. These factors, among others, includes dietary intake, alcohol, or medication / drug(s) consumption, or fasting. This list of factors is only representative and not exhaustive.