CLINPATH PATHOLOGY

MQ190123

Copy to

Expanded reproductive carrier screen

P ease f nd attached the resu ts of the *Sonic Beacon Expanded Carrier Screen*, as reported by Fu gent Genet cs. Th s s a cover page on y and does not make up any part of the report.

Information sheets for patients

Informat on sheets to support the d scuss on of th s resu t w th nd v dua s and coup es can be found on the Son c Genet cs webs te, son cgenet cs.com.au/rcs/pat ent nformat on sheets.

Genetic Counselling

Genet c counse ng s ava ab e at no add t ona cost to e g b e coup es dent f ed as be ng at h gh reproduct ve r sk by th s test. P ease v s t son cgenet cs.com.au/rcs/gc for further nformat on about th s serv ce, nc ud ng a fu st of e g b ty cr ter a and the down oadab e referra form wh ch must be comp eted n order to access genet c counse ng.

Prenatal testing

If a fema e partner s found to be a carr er of an X nked cond t on, or a coup e s found to be carr ers for the same autosoma recess ve cond t on, prenata test ng for these spec f c var ants on a chor on c v ous or amn ocentes s samp e s ava ab e from th s aboratory.
Pr or to samp e co ect on or test request, p ease contact the aboratory on 1800 010 447 to d scuss w th a genet c patho og st. A ded cated request form must be used: son cgenet cs.com.au/bpns.

Panel gene content

P ease note that the expanded reproduct ve carr er screen pane gene content has been updated (Vers on 2.0 effect ve for samp es access oned at Fu gent Genet cs from 1 November 2022). P ease refer to the Supp ementa Tab e of the report for the gene st assessed n th s pat ent. For nd v dua partners tested separate y, p ease rev ew the assessed gene content when nterpret ng the reproduct ve r sk for the coup e.

Type of primary sample: SWAB

Cover

IS NOV22

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fulgent Sonic Genetics

Pa en nforma on:
MQ190123

Par ner nforma on:
Not Tested

Access on:
N/A

FINAL RESULTS

No carr er mutat ons dent f ed

TEST PERFORMED

Single Gene Carrier Screening: ACADM

(1 Gene Pane : *ACADM*; gene sequenc ng w th de et on and dup cat on ana ys s)

INTERPRETATION:

Notes and Recommendations:

- No carr er mutat ons were dent f ed n the subm tted spec men. A negat ve resu t does not ru e out the poss b ty of a genet c pred spos t on nor does t ru e out any pathogen c mutat ons n areas not assessed by th s test or n reg ons that were covered at a eve too ow to re ab y assess. A so, t does not ru e out mutat ons that are of the sort not quer ed by th s test; see Methods and L m tat ons for more nformat on.
- Th s carr er screen ng test does not screen for a poss b e genet c cond t ons, nor for a poss b e mutat ons n every gene tested. Ind v dua s w th negat ve test resu ts may st have up to a 3-4% r sk to have a ch d w th a b rth defect due to genet c and/or env ronmenta factors.
- Pat ents may w sh to d scuss any carr er resu ts w th b ood re at ves, as there s an ncreased chance that they are a so carr ers. These resu ts shou d be nterpreted n the context of th s nd v dua 's c n ca f nd ngs, b ochem ca prof e, and fam y h story.
- X- nked genes are not rout ne y ana yzed for ma e carr er screen ng tests. Gene spec f c notes and m tat ons may be present. See be ow.
- Th s report does not nc ude var ants of uncerta n s gn f cance.
- Genet c counse ng s recommended. Contact your phys c an about the ava ab e opt ons for genet c counse ng.

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GENES TESTED:

Custom Beacon Carrier Screening Panel - Gene

Th s ana ys s was run us ng the Custom Beacon Carr er Screen ng Pane gene st. 1 genes were tested w th 100.0% of targets sequenced at >20x coverage. For more gene spec f c nformat on and ass stance w th res dua r sk ca cu at on, see the SUPPLEMENTAL TABLE.

ACADM

METHODS:

Genom c DNA was so ated from the subm tted spec men nd cated above (f ce u ar mater a was subm tted). DNA was barcoded, and enr ched for the cod ng exons of targeted genes us ng hybr d capture techno ogy. Prepared DNA brar es were then sequenced us ng a Next Generat on Sequenc ng techno ogy. Fo ow ng a gnment to the human genome reference sequence (assemb y GRCh37), var ants were detected n reg ons of at east 10x coverage. For th s spec men, 100.00% and 100.00% of cod ng reg ons and sp c ng junct ons of genes sted had been sequenced w th coverage of at east 10x and 20x, respect ve y, by NGS or by Sanger sequenc ng. The rema n ng reg ons d d not have 10x coverage, and were not eva uated. Var ants were nterpreted manua y us ng ocus spec f c databases, terature searches, and other mo ecu ar b o og ca pr nc p es. To m n m ze fa se pos t ve resu ts, any var ants that do not meet nterna qua ty standards are conf rmed by Sanger sequenc ng. Var ants c ass f ed as pathogen c, ke y pathogen c, or r sk a e e wh ch are ocated n the cod ng reg ons and nearby ntron c reg ons (+/- 20bp) of the genes sted above are reported. Var ants outs de these nterva s may be reported but are typ ca y not guaranteed. When a s ng e pathogen c or ke y pathogen c var ant s dent f ed n a c n ca y re evant gene w th autosoma recess ve nher tance, the aboratory w attempt to ensure 100% coverage of cod ng sequences e ther through NGS or Sanger sequenc ng techno og es ("f - n"). A genes sted were eva uated for arge de et ons and/or dup cat ons. However, s ng e exon de et ons or dup cat ons w not be detected n th s assay, nor w copy number a terat ons n reg ons of genes w th s gn f cant pseudogenes. Putat ve de et ons or dup cat ons are ana yzed us ng Fu gent Germ ne propr etary p pe ne for th s spec men. B o nformat cs: The Fu gent Germ ne v2019.2 p pe ne was used to ana yze th s spec men.

LIMITATIONS:

General Limitations

These test resu ts and var ant nterpretat on are based on the proper dent f cat on of the subm tted spec men, accuracy of any stated fam a re at onsh ps, and use of the correct human reference sequences at the quer ed oc . In very rare nstances, errors may resu t due to m x-up or co-m ng ng of spec mens. Pos t ve resu ts do not mp y that there are no other contr butors, genet c or otherw se, to future pregnanc es, and negat ve resu ts do not ru e out the genet c r sk to a pregnancy. Off c a gene names change over t me. Fu gent uses the most up to date gene names based on HUGO Gene Nomenc ature Comm ttee (https://www.genenames.org) recommendat ons. If the gene name on report does not match that of ordered gene, p ease contact the aboratory and deta s can be prov ded. Resu t nterpretat on s based on the ava ab e c n ca and fam y h story nformat on for th s nd v dua , co ected pub shed nformat on, and A amut annotat on ava ab e at the t me of report ng. Th s assay s not des gned or va dated for the detect on of ow-eve mosa c sm or somat c mutat ons. Th s assay w not detect certa n types of genom c aberrat ons such as trans ocat ons, nvers ons, or repeat expans ons other than spec f ed genes. DNA a terat ons n regu atory reg ons or deep ntron c reg ons (greater than 20bp from an exon) may not be detected by th s test. Un ess otherw se nd cated, no add t ona assays have been performed to eva uate genet c changes n th s spec men. There are techn ca m tat ons on the ab ty of DNA sequenc ng to detect sma nsert ons and de et ons. Our aboratory uses a sens t ve detect on a gor thm, however these types of a terat ons are not detected as re ab y as s ng e nuc eot de var ants. Rare y, due to systemat c chem ca , computat ona , or human error, DNA var ants may be m ssed. A though next generat on sequenc ng techno og es and our b o nformat cs ana ys s s gn f cant y reduce the confound ng contr but on

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of pseudogene sequences or other h gh y-homo ogous sequences, somet mes these may st nterfere w th the techn ca ab ty of the assay to dent fy pathogen c a terat ons n both sequenc ng and de et on/dup cat on ana yses. De et on/dup cat on ana ys s can dent fy a terat ons of genom c reg ons wh ch nc ude one who e gene (bucca swab spec mens and who e b ood spec mens) and are two or more cont guous exons n s ze (who e b ood spec mens on y); s ng e exon de et ons or dup cat ons may occas ona y be dent f ed, but are not rout ne y detected by th s test. When nove DNA dup cat ons are dent f ed, t s not poss b e to d scern the genom c ocat on or or entat on of the dup cated segment, hence the effect of the dup cat on cannot be pred cted. Where de et ons are detected, t s not a ways poss b e to determ ne whether the pred cted product w rema n n-frame or not. Un ess otherw se nd cated, de et on/dup cat on ana ys s has not been performed n reg ons that have been sequenced by Sanger.

Gene Specific Notes and Limitations

No gene spec f c m tat ons app y to the genes on the tested pane .

SIGNATURE:

Dr. Harry Gao, DABMG, FACMG on **8/30/2023 3:15 PM PDT**
E ec ron ca y s gned

DISCLAIMER:

Th s test was deve oped and ts performance character st cs determ ned by **Fulgent Genetics**. It has not been c eared or approved by the FDA. The aboratory s regu ated under CLIA as qua f ed to perform h gh-comp ex ty test ng. Th s test s used for c n ca purposes. It shou d not be regarded as nvest gat ona or for research. S nce genet c var at on, as we as systemat c and techn ca factors, can affect the accuracy of test ng, the resu ts of test ng shou d a ways be nterpreted n the context of c n ca and fam a data. For ass stance w th nterpretat on of these resu ts, hea thcare profess ona s may contact us d rect y at **(626) 350-0537** or **info@fulgentgenetics.com**. It s recommended that pat ents rece ve appropr ate genet c counse ng to exp a n the mp cat ons of the test resu t, nc ud ng ts res dua r sks, uncerta nt es and reproduct ve or med ca opt ons.

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Supplemental Table

Gene	Condition	nheritance	Ethnicity	Carrier Rate	Detection Rate	Post test Carrier Probability*	Residual Risk*
ACADM	Medium chain acyl CoA dehydrogenase (MCAD) deficiency	AR	General Population	in 69	98%	in 3 40	in 938 676
			Caucasian / European Population	in 52	99%	in 5 0	in 06 008
			East Asian Population	in 98	99%	in 9 70	< in 0 million
			Native American Population	in 43	96%	in 05	in 80 772

* For genes that have tested negat ve
Abbrev at ons: AR, autosoma recess ve; XL, X- nked

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Pa en nforma on:
MQ190123

Par ner nforma on:
Not Tested

Access on:
N/A

FINAL RESULTS

No carr er mutat ons dent f ed

TEST PERFORMED

Sonic Beacon Expanded Carrier Screen v2.0 - Male
(361 Gene Pane ; gene sequenc ng w th de et on and dup cat on ana ys s)

INTERPRETATION:

Notes and Recommendations:

- No carr er mutat ons were dent f ed n the subm tted spec men. A negat ve resu t does not ru e out the poss b ty of a genet c pred spos t on nor does t ru e out any pathogen c mutat ons n areas not assessed by th s test or n reg ons that were covered at a eve too ow to re ab y assess. A so, t does not ru e out mutat ons that are of the sort not quer ed by th s test; see Methods and L m tat ons for more nformat on.
- Test ng for copy number changes n the SMN1 gene was performed to screen for the carr er status of Sp na Muscu ar Atrophy. The resu ts for th s nd v dua are w th n the norma range for non-carr ers. See L m tat ons sect on for more nformat on.
- Th s carr er screen ng test does not screen for a poss b e genet c cond t ons, nor for a poss b e mutat ons n every gene tested. Ind v dua s w th negat ve test resu ts may st have up to a 3-4% r sk to have a ch d w th a b rth defect due to genet c and/or env ronmenta factors.
- Pat ents may w sh to d scuss any carr er resu ts w th b ood re at ves, as there s an ncreased chance that they are a so carr ers. These resu ts shou d be nterpreted n the context of th s nd v dua 's c n ca f nd ngs, b ochem ca prof e, and fam y h story.
- X- nked genes are not rout ne y ana yzed for ma e carr er screen ng tests. Gene spec f c notes and m tat ons may be present. See be ow.
- Th s report does not nc ude var ants of uncerta n s gn f cance.
- Genet c counse ng s recommended. Contact your phys c an about the ava ab e opt ons for genet c counse ng.

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GENES TESTED:

Sonic Beacon Expanded Carrier Screen v2.0 - Male - 361 Genes

Th s ana ys s was run us ng the Son c Beacon Expanded Carr er Screen v2.0 - Ma e gene st cons st ng of 361 genes (v2, effect ve November 1st 2022). 361 genes were tested w th 99.51% of targets sequenced at >20x coverage. For more gene spec f c nformat on and ass stance w th res dua r sk ca cu at on, see the SUPPLEMENTAL TABLE.

ABCA12	*ABCA3*	*ABCA4*	*ABCB11*	*ABCC8*	*ACAD9*
ACADVL	*ACAT1*	*ACOX1*	*ACSF3*	*ADA*	*ADAMTS2*
ADGRG1	*ADK*	*AGA*	*AGL*	*AGPS*	*AGXT*
AHI1	*AIPL1*	*ALDH3A2*	*ALDOB*	*ALG6*	*ALMS1*
ALPL	*AMT*	*AQP2*	*ARG1*	*ARL13B*	*ARSA*
ARSB	*ASL*	*ASNS*	*ASPA*	*ASS1*	*ATM*
ATP6V1B1	*ATP7B*	*BBS1*	*BBS10*	*BBS12*	*BBS2*
BCKDHA	*BCKDHB*	*BCS1L*	*BLM*	*BSND*	*CAPN3*
CASQ2	*CBS*	*CC2D2A*	*CCDC103*	*CCDC39*	*CCDC88C*
CDH23	*CEP290*	*CFTR*	*CHRNE*	*CHRNG*	*CHST6*
CIITA	*CLN3*	*CLN5*	*CLN6*	*CLN8*	*CLRN1*
CNGB3	*COL27A1*	*COL4A3*	*COL4A4*	*COL7A1*	*COX15*
CPS1	*CPT1A*	*CPT2*	*CRB1*	*CRYL1*	*CTNS*
CTSA	*CTSC*	*CTSD*	*CTSK*	*CYBA*	*CYP11A1*
CYP11B1	*CYP11B2*	*CYP17A1*	*CYP1B1*	*CYP21A2*	*CYP27A1*
DBT	*DCLRE1C*	*DDX11*	*DHCR7*	*DHDDS*	*DLD*
DNAH5	*DNAI1*	*DNAI2*	*DUOX2*	*DUOXA2*	*DYNC2H1*
DYSF	*EIF2AK3*	*EIF2B5*	*ELP1*	*ERCC2*	*ERCC5*
ERCC6	*ERCC8*	*ESCO2*	*ETFA*	*ETFB*	*ETFDH*
ETHE1	*EVC*	*EVC2*	*EXOSC3*	*F2*	*F5*
FAH	*FAM126A*	*FAM161A*	*FANCA*	*FANCC*	*FANCG*
FH	*FKRP*	*FKTN*	*FOXRED1*	*FTCD*	*FUCA1*
G6PC	*GAA*	*GALC*	*GALNS*	*GALT*	*GAMT*
GBA	*GBE1*	*GCDH*	*GDAP1*	*GDF5*	*GFM1*
GJB2	*GJB6*	*GLB1*	*GLDC*	*GLE1*	*GNE*
GNPTAB	*GNPTG*	*GNS*	*GSS*	*GUCY2D*	*GUSB*
HADHA	*HADHB*	*HAX1*	*HBA1*	*HBA2*	*HBB*
HEXA	*HEXB*	*HGSNAT*	*HJV*	*HLCS*	*HMGCL*
HOGA1	*HPS1*	*HPS3*	*HPS4*	*HSD17B4*	*HSD3B2*
HYLS1	*IDUA*	*IVD*	*IYD*	*JAK3*	*KCNJ11*
LAMA2	*LAMA3*	*LAMB3*	*LAMC2*	*LCA5*	*LDLRAP1*
LHX3	*LIFR*	*LIPA*	*LMBRD1*	*LOXHD1*	*LPL*
LRP2	*LRPPRC*	*LYST*	*MAN2B1*	*MANBA*	*MCOLN1*
MCPH1	*MED17*	*MESP2*	*MFSD8*	*MKS1*	*MLC1*
MLYCD	*MMAA*	*MMAB*	*MMACHC*	*MMADHC*	*MPI*
MPL	*MPV17*	*MTHFR*	*MTMR2*	*MTRR*	*MTTP*
MUT	*MVK*	*MYO7A*	*NAGA*	*NAGLU*	*NAGS*
NBN	*NDRG1*	*NDUFAF2*	*NDUFAF5*	*NDUFS4*	*NDUFS6*
NDUFS7	*NDUFV1*	*NEB*	*NEU1*	*NPC1*	*NPC2*
NPHP1	*NPHS1*	*NPHS2*	*NTRK1*	*OAT*	*OCA2*
OPA3	*OTOF*	*P3H1*	*PAH*	*PANK2*	*PC*
PCCA	*PCCB*	*PCDH15*	*PCNT*	*PDHB*	*PEX1*
PEX10	*PEX12*	*PEX2*	*PEX26*	*PEX6*	*PEX7*
PFKM	*PHGDH*	*PHYH*	*PKHD1*	*PLA2G6*	*PLOD1*
PMM2	*POLG*	*POLR1C*	*POMGNT1*	*POMT1*	*POMT2*
POR	*PPT1*	*PRF1*	*PROP1*	*PSAP*	*PTS*
PUS1	*QDPR*	*RAB23*	*RAG1*	*RAG2*	*RAPSN*

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RARS2	*RAX*	*RDH12*	*RMRP*	*RNASEH2B*	*RPE65*
RPGRIP1L	*RTEL1*	*SACS*	*SAMD9*	*SAMHD1*	*SCO2*
SEPSECS	*SERPINA1*	*SGCA*	*SGCB*	*SGCD*	*SGCG*
SGSH	*SH3TC2*	*SLC12A6*	*SLC17A5*	*SLC19A3*	*SLC1A4*
SLC22A5	*SLC25A13*	*SLC25A15*	*SLC26A2*	*SLC26A3*	*SLC35A3*
SLC37A4	*SLC39A4*	*SLC45A2*	*SLC46A1*	*SLC5A5*	*SLC7A7*
SMARCAL1	*SMN1*	*SMPD1*	*SPG11*	*SPINK5*	*STAR*
SUMF1	*SURF1*	*TCIRG1*	*TCTN2*	*TECPR2*	*TF*
TG	*TGM1*	*TH*	*TMEM216*	*TPO*	*TPP1*
TRDN	*TRIM32*	*TRMU*	*TSEN54*	*TSFM*	*TSHB*
TTC37	*TTPA*	*TYMP*	*TYR*	*TYRP1*	*UGT1A1*
USH1C	*USH1G*	*USH2A*	*VPS13A*	*VPS13B*	*VPS45*
VPS53	*VRK1*	*VSX2*	*WHRN*	*WRN*	*XPA*
XPC	*ZFYVE26*				

METHODS:

Genom c DNA was so ated from the subm tted spec men nd cated above (f ce u ar mater a was subm tted). DNA was barcoded, and enr ched for the cod ng exons of targeted genes us ng hybr d capture techno ogy. Prepared DNA brar es were then sequenced us ng a Next Generat on Sequenc ng techno ogy. Fo ow ng a gnment to the human genome reference sequence (assemb y GRCh37), var ants were detected n reg ons of at east 10x coverage. For th s spec men, 99.57% and 99.51% of cod ng reg ons and sp c ng junct ons of genes sted had been sequenced w th coverage of at east 10x and 20x, respect ve y, by NGS or by Sanger sequenc ng. The rema n ng reg ons d d not have 10x coverage, and were not eva uated. Var ants were nterpreted manua y us ng ocus spec f c databases, terature searches, and other mo ecu ar b o og ca pr nc p es. To m n m ze fa se pos t ve resu ts, any var ants that do not meet nterna qua ty standards are conf rmed by Sanger sequenc ng. Var ants c ass f ed as pathogen c, ke y pathogen c, or r sk a e e wh ch are ocated n the cod ng reg ons and nearby ntron c reg ons (+/- 20bp) of the genes sted above are reported. Var ants outs de these nterva s may be reported but are typ ca y not guaranteed. When a s ng e pathogen c or ke y pathogen c var ant s dent f ed n a c n ca y re evant gene w th autosoma recess ve nher tance, the aboratory w attempt to ensure 100% coverage of cod ng sequences e ther through NGS or Sanger sequenc ng techno og es ("f - n"). A genes sted were eva uated for arge de et ons and/or dup cat ons. However, s ng e exon de et ons or dup cat ons w not be detected n th s assay, nor w copy number a terat ons n reg ons of genes w th s gn f cant pseudogenes. Putat ve de et ons or dup cat ons are ana yzed us ng Fu gent Germ ne propr etary p pe ne for th s spec men. B o nformat cs: The Fu gent Germ ne v2019.2 p pe ne was used to ana yze th s spec men.

LIMITATIONS:

General Limitations

These test resu ts and var ant nterpretat on are based on the proper dent f cat on of the subm tted spec men, accuracy of any stated fam a re at onsh ps, and use of the correct human reference sequences at the quer ed oc . In very rare nstances, errors may resu t due to m x-up or co-m ng ng of spec mens. Pos t ve resu ts do not mp y that there are no other contr butors, genet c or otherw se, to future pregnanc es, and negat ve resu ts do not ru e out the genet c r sk to a pregnancy. Off c a gene names change over t me. Fu gent uses the most up to date gene names based on HUGO Gene Nomenc ature Comm ttee (https://www.genenames.org) recommendat ons. If the gene name on report does not match that of ordered gene, p ease contact the aboratory and deta s can be prov ded. Resu t nterpretat on s based on the ava ab e c n ca and fam y h story nformat on for th s nd v dua , co ected pub shed nformat on, and A amut annotat on ava ab e at the t me of report ng. Th s assay s not des gned or va dated for the detect on of ow- eve mosa c sm or somat c mutat ons. Th s assay w not detect certa n types of genom c aberrat ons such as trans ocat ons, nvers ons, or repeat expans ons other than spec f ed genes. DNA a terat ons n regu atory reg ons or deep ntron c reg ons (greater than 20bp from an exon) may not be detected by th s test. Un ess otherw se nd cated, no add t ona assays have been performed to eva uate genet c changes n th s spec men. There are techn ca m tat ons on the ab ty of DNA sequenc ng to detect sma nsert ons and de et ons. Our aboratory uses a sens t ve detect on a gor thm, however these types of a terat ons are not detected as re ab y as s ng e nuc eot de var ants. Rare y, due to systemat c chem ca , computat ona , or human error, DNA var ants may be m ssed. A though next generat on sequenc ng techno og es and our b o nformat cs ana ys s s gn f cant y reduce the confound ng contr but on of pseudogene sequences or other h gh y-homo ogous sequences, somet mes these may st nterfere w th the techn ca ab ty of the assay to dent fy pathogen c a terat ons n both sequenc ng and de et on/dup cat on ana yses. De et on/dup cat on ana ys s can dent fy a terat ons of genom c reg ons wh ch nc ude one who e

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gene (bucca swab spec mens and who e b ood spec mens) and are two or more cont guous exons n s ze (who e b ood spec mens on y); s ng e exon de et ons or dup cat ons may occas ona y be dent f ed, but are not rout ne y detected by th s test. When nove DNA dup cat ons are dent f ed, t s not poss b e to d scern the genom c ocat on or or entat on of the dup cated segment, hence the effect of the dup cat on cannot be pred cted. Where de et ons are detected, t s not a ways poss b e to determ ne whether the pred cted product w rema n n-frame or not. Un ess otherw se nd cated, de et on/dup cat on ana ys s has not been performed n reg ons that have been sequenced by Sanger.

Gene Specific Notes and Limitations

CFTR: Ana ys s of he n ron 8 po ymorph c reg on (e.g. IVS8-5T a e e) s on y performed f he p.Arg117 s (R117) mu a on s de ec ed. S ng e exon de e on/dup ca on ana ys s s m ed o de e ons of prev ous y repor ed exons: 1, 2, 3, 11, 19, 20, 21. *CRYL1:* As mu a ons n he CRYL1 gene are no known o be assoc a ed w h any c n ca cond on, sequence var an s n h s gene are no ana yzed. owever, o ncrease copy number de ec on sens v y for arge de e ons nc ud ng h s gene and a ne ghbor ng on gene on he pane (GJB6, a so known as connex n 30), h s gene was eva ua ed for copy number var a on. *CYP11B1:* The curren es ng me hod s no ab e o re ab y de ec cer a n pa hogen c var an s n h s gene due o he n erference by h gh y homo ogous reg ons. Th s ana ys s s no des gned o de ec or ru e-ou copy-neu ra ch mer c CYP11B1/CYP11B2 gene. *CYP11B2:* The curren es ng me hod s no ab e o re ab y de ec cer a n pa hogen c var an s n h s gene due o he n erference by h gh y homo ogous reg ons. Th s ana ys s s no des gned o de ec or ru e-ou copy-neu ra ch mer c CYP11B1/CYP11B2 gene. *CYP21A2:* S gn f can pseudogene n erference and/or rec proca exchanges be ween he CYP21A2 gene and s pseudogene, CYP21A1P, have been known o occur and may mpac resu s. As such, he re evance of var an s repor ed n h s gene mus be n erpre ed c n ca y n he con ex of he c n ca f nd ngs, b ochem ca prof e, and fam y h s ory of each pa en . CYP21A2 var an s pr mar y assoc a ed w h non-c ass c congen a adrena hyperp as a (CA) are no nc uded n h s ana ys s (PubMed: 23359698). The var an s assoc a ed w h non-c ass c d sease, nc ud ng bu no m ed o c.188A>T (p. s63Leu), c.844G>T (p.Va 282Leu), c.1174G>A (p.A a392Thr), and c.1360C>T (p.Pro454Ser) w no be repor ed. LR-PCR s no rou ne y ordered for NM 000500.9:c.955C>T (p.G n319Ter). Ind v dua s w h c.955C>T (p.G n319Ter) w be repor ed as a Poss b e Carr er nd ca ng ha he prec se na ure of he var an has no been de erm ned by LR-PCR and ha he var an may occur n he CYP21A2 w d- ype gene or n he CYP21A1P pseudogene. The conf rma on es s recommended f he second reproduc ve par ner s es ed pos ve for var an s assoc a ed w h c ass c CA . *DUOX2:* The curren es ng me hod s no ab e o re ab y de ec var an s n exons 6-8 of he DUOX2 gene (NM 014080.5) due o s gn f can n erference by he h gh y homo ogous gene, DUOX1. *F2:* The common r sk a e e NM 000506.5:c.*97G>A s no nc uded n h s ana ys s. *F5:* The common Fac or 5 "Le den" a e e s no yp ca y repor ed; h s m d r sk a e e may be repor ed upon reques . *GALT:* In genera , he D2 "Duar e" a e e s no repor ed f de ec ed, bu can be repor ed upon reques . Wh e h s a e e can cause pos ve newborn screen ng resu s, s no known o cause c n ca symp oms n any s a e (PubMed: 25473725, 30593450). *GBA:* The curren es ng me hod may no be ab e o re ab y de ec cer a n pa hogen c var an s n he GBA gene due o homo ogous recomb na on be ween he pseudogene and he func ona gene. *HBA1:* The phase of he erozygous a era ons n he *HBA1* gene canno be de erm ned, bu can be conf rmed hrough paren a es ng. *HBA2:* The phase of he erozygous a era ons n he *HBA2* gene canno be de erm ned, bu can be conf rmed hrough paren a es ng. *MTHFR:* As recommended by ACMG, he wo common po ymorph sms n he MT FR gene - c.1286A>C (p.G u429A a, a so known as c.1298A>C) and c.665C>T (p.A a222Va , a so known as c.677C>T) - are no repor ed n h s es due o ack of suff c en c n ca u y o mer es ng (PubMed: 23288205). *NEB:* Th s gene con a ns a 32-kb r p ca e reg on (exons 82-105) wh ch s no amenab e o sequenc ng and de e on/dup ca on ana ys s. *NPHS2:* If de ec ed, he var an NM 014625.3:c.686G>A (p.Arg229G n) w no be repor ed as h s var an s no s gn f can y assoc a ed w h d sease when homozygous or n he compound he erozygous s a e w h var an s n exons 1-6 of NP S2. *SERPINA1:* If de ec ed he var an NM 000295.5:c.863A>T (p.G u288Va) w no be repor ed as h s var an s assoc a ed w h ow d sease pene rance and s no assoc a ed w h severe ear y onse d sease. *SMN1:* The curren es ng me hod de ec s sequenc ng var an s n exon 7 and copy number var a ons n exons 7-8 of he SMN1 gene (NM 022874.2). Sequenc ng and de e on/dup ca on ana ys s are no performed on any o her reg on n h s gene. Abou 5%-8% of he popu a on have wo cop es of SMN1 on a s ng e chromosome and a de e on on he o her chromosome, known as a [2+0] conf gura on (PubMed: 20301526). The curren es ng me hod canno d rec y de ec carr ers w h a [2+0] SMN1 conf gura on, bu can de ec nkage be ween he s en carr er a e e and cer a n popu a on-spec f c s ng e nuc eo de changes. As a resu , a nega ve resu for carr er es ng grea y reduces bu does no e m na e he chance ha a person s a carr er. On y abnorma resu s w be repor ed. *TRDN:* Due o h gh GC con en of cer a n exons, copy number ana ys s may have reduced sens v y for par a gene de e ons/dup ca ons of *TRDN*. Conf rma on of par a gene de e ons/dup ca ons are m ed o nd v dua s w h a pos ve persona h s ory of card ac arrhy hm a and/or nd v dua s carry ng a pa hogen c/ ke y pa hogen c sequence var an . *UGT1A1:* Common var an s n he *UGT1A1 gene* (popu a on a e e frequency >5%) are yp ca y no repor ed as hey do no cause a Mende an cond on. *WRN:* Due o he n erference by h gh y homo ogous reg ons w h n he WRN gene, our curren es ng me hod has ess sens v y o de ec var an s n exons 10-11 of WRN (NM 000553.6).

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Jianbo Song, Ph.D., ABMGG, CGMB, CCS, FACMG on **2/28/2023 06:06 PM PST**
E ec ron ca y s gned

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Supplemental Table

Gene	Condition	nheritance	Ethnicity	Carrier Rate	Detection Rate	Post test Carrier Probability*	Residual Risk*
ABCA12	Congenital ichthyosis ABCA 2 related	AR	General Population	< in 500	98%	in 24 95	< in 0 million
ABCA3	Surfactant metabolism dysfunction pulmonary 3	AR	General Population	in 6	99%	in 50	in 5 336 464
ABCA4	Stargardt disease	AR	General Population	in 5	98%	in 2 50	in 5 0 204
ABCB11	Progressive familial intrahepatic cholestasis	AR	General Population	in 2	98%	in 5 55	in 2 486 848
ABCC8	Familial hyperinsulinism	AR	General Population	in 2	98%	in 5 55	in 2 486 848
			Ashkenazi Jewish Population	in 44	98%	in 2 5	in 378 576
			Finnish Population	in 25	98%	in 20	in 20 00
			Middle Eastern Population	in 25	98%	in 20	in 20 00
ACAD9	Acyl CoA dehydrogenase 9 (ACAD9) deficiency	AR	General Population	< in 500	98%	in 24 95	< in 0 million
ACADVL	Very long chain acyl CoA dehydrogenase (VLCAD) deficiency	AR	General Population	in 8	93%	in 672	in 789 84
			Middle Eastern Population	in 74	93%	in 044	in 309 024
			Native American Population	in 6	93%	in 858	in 209 352
			South Asian/ ndian Population	in 73	93%	in 030	in 300 760
ACAT1	3 ketothiolase deficiency	AR	General Population	< in 500	98%	in 24 95	< in 0 million
ACOX1	Peroxisomal acyl CoA oxidase deficiency	AR	General Population	< in 500	98%	in 24 95	< in 0 million
ACSF3	Combined malonic and methylmalonic aciduria	AR	General Population	< in 500	98%	in 24 95	< in 0 million
ADA	Adenosine deaminase deficiency	AR	General Population	in 224	93%	in 3 87	in 2 855 552
ADAMTS2	Ehlers Danlos syndrome dermatosparaxis type	AR	General Population	< in 500	98%	in 24 95	< in 0 million
			Ashkenazi Jewish Population	in 248	98%	in 2 35	< in 0 million
ADGRG1	Bilateral frontoparietal polymicrogyria	AR	General Population	< in 500	98%	in 24 95	< in 0 million
ADK	Hypermethioninemia due to adenosine kinase deficiency	AR	General Population	< in 500	99%	in 49 90	< in 0 million
AGA	Aspartylglucosaminuria	AR	General Population	< in 500	98%	in 24 95	< in 0 million
			Finnish Population	in 7	98%	in 3 50	in 994 284
AGL	Glycogen storage disease type	AR	General Population	in 58	95%	in 3 4	in 985 2
			Faroese Population	in 28	95%	in 54	in 60 592
			nuit Population	in 25	95%	in 48	in 48 00
			North African Jewish Population	in 37	95%	in 72	in 06 708
AGPS	Rhizomelic chondrodysplasia punctata type 3	AR	General Population	< in 500	98%	in 24 95	< in 0 million
AGXT	Primary hyperoxaluria type	AR	General Population	in 20	99%	in 90	in 5 7 2 480
			Caucasian / European Population	in 73	99%	in 7 20	< in 0 million
AHI1	Joubert syndrome AH related	AR	General Population	in 448	99%	in 44 70	< in 0 million
AIPL1	Childhood onset severe retinal dystrophy A PL related	AR	General Population	in 409	99%	in 40 80	< in 0 million
ALDH3A2	Sjögren Larsson syndrome	AR	General Population	in 250	98%	in 2 45	< in 0 million
ALDOB	Hereditary fructose intolerance	AR	General Population	in 22	99%	in 2 0	in 5 905 288
			African/African American Population	in 250	99%	in 24 90	< in 0 million
			Caucasian / European Population	in 67	99%	in 6 60	in 769 068
			Middle Eastern Population	in 97	99%	in 9 60	in 3 725 88
ALG6	Congenital disorder of glycosylation type c	AR	General Population	< in 500	98%	in 24 95	< in 0 million
ALMS1	Alstrom syndrome	AR	General Population	in 500	98%	in 24 95	< in 0 million
ALPL	Hypophosphatasia	AR	General Population	in 58	95%	in 3 4	in 985 2
			Caucasian / European Population	in 274	95%	in 5 46	in 5 985 256
			Mennonite Population	in 25	95%	in 48	in 48 00
AMT	Glycine encephalopathy	AR	General Population	in 373	98%	in 8 60	< in 0 million
			Finnish Population	in 7	98%	in 5 80	in 2 7 4 868
AQP2	Nephrogenic diabetes insipidus	AR	General Population	< in 500	95%	in 9 98	< in 0 million
			Finnish Population	in 69	95%	in 3 36	in 2 272 036
ARG1	Arginase deficiency	AR	General Population	in 296	98%	in 4 75	< in 0 million
ARL13B	Joubert syndrome ARL 3B related	AR	General Population	< in 500	99%	in 49 90	< in 0 million
ARSA	Metachromatic leukodystrophy	AR	General Population	in 00	99%	in 9 90	in 3 960 400
			Caucasian / European Population	in 78	99%	in 7 70	in 2 402 7 2
			Yemenite Jewish Population	in 75	99%	in 7 40	in 2 220 300
ARSB	Mucopolysaccharidosis type V (Maroteaux Lamy syndrome)	AR	General Population	in 250	98%	in 2 45	< in 0 million
			Western Australian Population	in 283	98%	in 4 0	< in 0 million
ASL	Argininosuccinate lyase deficiency	AR	General Population	in 32	90%	in 3	in 692 208
ASNS	Asparagine synthetase deficiency	AR	General Population	< in 500	99%	in 49 90	< in 0 million
			ranian Jewish Population	in 80	99%	in 7 90	in 2 528 320
ASPA	Canavan disease	AR	General Population	in 300	97%	in 9 968	< in 0 million
			Ashkenazi Jewish Population	in 55	96%	in 35	in 297 220
ASS1	Citrullinemia	AR	General Population	in 9	96%	in 2 95	in 404 676
			East Asian Population	in 32	96%	in 3 276	in 729 728

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Supplemental Table

Gene	Condition	nheritance	Ethnicity	Carrier Rate	Detection Rate	Post test Carrier Probability*	Residual Risk*
ATM	Ataxia telangiectasia	AR	General Population	in 00	92%	in 239	in 495 600
ATP6V1B1	Renal tubular acidosis with deafness	AR	General Population	< in 500	98%	in 24 95	< in 0 million
ATP7B	Wilson disease	AR	General Population	in 87	98%	in 4 30	in 496 748
			Caucasian / European Population	in 42	98%	in 2 05	in 344 568
			Ashkenazi Jewish Population	in 70	98%	in 3 45	in 966 280
BBS1	Bardet Biedl syndrome type	AR	General Population	in 367	99%	in 36 60	< in 0 million
BBS10	Bardet Biedl syndrome type 0	AR	General Population	in 395	99%	in 39 40	< in 0 million
BBS12	Bardet Biedl syndrome type 2	AR	General Population	in 79	99%	in 79 00	< in 0 million
BBS2	Bardet Biedl syndrome 2	AR	General Population	in 62	99%	in 62 00	< in 0 million
			Ashkenazi Jewish Population	in 07	99%	in 0 60	in 4 537 228
BBS2	Retinitis Pigmentosa 74	AR	General Population	in 62	99%	in 62 00	< in 0 million
			Ashkenazi Jewish Population	in 07	99%	in 0 60	in 4 537 228
BCKDHA	Maple syrup urine disease type a	AR	General Population	in 32	98%	in 6 00	< in 0 million
			Mennonite Population	in 0	98%	in 45	in 8 040
BCKDHB	Maple syrup urine disease type b	AR	General Population	in 364	98%	in 8 5	< in 0 million
			Ashkenazi Jewish Population	in 97	98%	in 4 80	in 862 788
BCS1L	Björnstad syndrome	AR	General Population	< in 500	98%	in 24 95	< in 0 million
BCS1L	GRAC LE syndrome	AR	General Population	< in 500	98%	in 24 95	< in 0 million
BCS1L	Mitochondrial complex deficiency	AR	General Population	< in 500	98%	in 24 95	< in 0 million
BLM	Bloom syndrome	AR	General Population	in 800	87%	in 6 47	< in 0 million
			Ashkenazi Jewish Population	in 34	99%	in 3 30	in 7 29 336
BSND	Bartter syndrome	AR	General Population	in 500	98%	in 24 95	< in 0 million
CAPN3	Limb girdle muscular dystrophy type 2A	AR	General Population	< in 500	98%	in 24 95	< in 0 million
			Caucasian / European Population	in 03	98%	in 5 0	in 2 0 6 2
CASQ2	Catecholaminergic polymorphic ventricular tachycardia	AR	General Population	in 224	99%	in 22 30	< in 0 million
CBS	Homocystinuria due to cystathionine beta synthase deficiency	AR	General Population	in 224	99%	in 22 30	< in 0 million
			Caucasian / European Population	in 86	99%	in 8 50	in 2 924 344
			Middle Eastern Population	in 2	99%	in 2 00	in 68 084
CC2D2A	Joubert syndrome 9	AR	General Population	in 20	99%	in 20 00	in 6 080 804
CCDC103	Primary ciliary dyskinesia type 7	AR	General Population	in 3 6	98%	in 5 75	< in 0 million
CCDC39	Primary ciliary dyskinesia type 4	AR	General Population	in 2	98%	in 0 50	in 8 862 844
CCDC88C	Congenital hydrocephalus	AR	General Population	in 37	99%	in 3 60	in 7 453 348
CDH23	Usher syndrome type D	AR	General Population	in 285	90%	in 2 84	in 364
CEP290	Joubert syndrome 5	AR	General Population	in 90	98%	in 9 45	in 7 82 760
CEP290	Leber congenital amaurosis 0	AR	General Population	in 90	98%	in 9 45	in 7 82 760
CEP290	Bardet Biedl syndrome 4	AR	General Population	in 90	98%	in 9 45	in 7 82 760
CEP290	CEP290 related disorders	AR	General Population	in 90	98%	in 9 45	in 7 82 760
CEP290	Senior Løken syndrome 6	AR	General Population	in 90	98%	in 9 45	in 7 82 760
CEP290	Meckel syndrome 4	AR	General Population	in 90	98%	in 9 45	in 7 82 760
CFTR	Cystic Fibrosis	AR	General Population	in 32	99%	in 3 0	in 396 928
			African/African American Population	in 6	99%	in 6 00	in 464 244
			Ashkenazi Jewish Population	in 24	99%	in 2 30	in 220 896
			Caucasian / European Population	in 25	99%	in 2 40	in 240 00
			East Asian Population	in 94	99%	in 9 30	in 3 497 76
			Latino Population	in 58	99%	in 5 70	in 322 632
CHRNE	Congenital myasthenic syndrome	AR	General Population	in 408	99%	in 40 70	< in 0 million
CHRNG	Multiple pterygium syndrome	AR	General Population	< in 500	98%	in 24 95	< in 0 million
CHST6	Macular corneal dystrophy CHST6 related	AR	General Population	in 79	99%	in 7 80	in 2 465 6
CIITA	Bare lymphocyte syndrome type	AR	General Population	< in 500	98%	in 24 95	< in 0 million
CLN3	Neuronal ceroid lipofuscinosis	AR	General Population	in 230	98%	in 45	< in 0 million
			Finnish Population	in 72	98%	in 3 55	in 022 688
CLN5	Neuronal ceroid lipofuscinosis 5	AR	General Population	< in 500	95%	in 9 98	< in 0 million
			Finnish Population	in 5	95%	in 2 28	in 049 260
CLN6	Neuronal ceroid lipofuscinosis CLN6 related	AR	General Population	< in 500	92%	in 6 239	< in 0 million
CLN8	Neuronal ceroid lipofuscinosis CLN8 related	AR	General Population	< in 500	95%	in 9 98	< in 0 million
			Finnish Population	in 35	95%	in 2 68	in 447 740
CLRN1	Usher syndrome type 3A	AR	General Population	in 500	98%	in 24 95	< in 0 million
			Ashkenazi Jewish Population	in 20	98%	in 5 95	in 2 856 480
			Finnish Population	in 70	98%	in 3 45	in 966 280
CNGB3	Achromatopsia	AR	General Population	in 87	99%	in 8 60	in 2 993 48
			Micronesian Population	in 2	99%	in 0	in 808

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COL27A1	Steel syndrome	AR	General Population	< in 500	98%	in 24 95	< in 0 million
COL4A3	Alport syndrome COL4A3 related	AR	General Population	in 267	98%	in 3 30	< in 0 million
			Ashkenazi Jewish Population	in 88	98%	in 9 35	in 7 03 952
COL4A4	Alport syndrome COL4A4 related	AR	General Population	in 267	98%	in 3 30	< in 0 million
COL7A1	Dystrophic epidermolysis bullosa	AR	General Population	in 96	97%	in 6 50	in 5 096 784
COX15	Mitochondrial complex V deficiency	AR	General Population	< in 500	99%	in 49 90	< in 0 million
CPS1	Carbamoylphosphate synthetase deficiency	AR	General Population	in 570	98%	in 28 45	< in 0 million
CPT1A	Carnitine palmitoyltransferase A deficiency	AR	General Population	in 354	90%	in 3 53	in 4 999 896
			Hutterite Population	in 6	90%	in 5	in 9 664
CPT2	Carnitine palmitoyltransferase deficiency	AR	General Population	< in 500	95%	in 9 98	< in 0 million
			Ashkenazi Jewish Population	in 5	95%	in 00	in 204 204
CRB1	Leber congenital amaurosis 8	AR	General Population	in 04	98%	in 5 5	in 2 42 8 6
CRB1	Retinitis pigmentosa 2	AR	General Population	in 04	98%	in 5 5	in 2 42 8 6
CRYL1	GJB6 CRYL related nonsyndromic hearing loss	UK	General Population	in 423	99%	in 42 20	< in 0 million
CTNS	Cystinosis	AR	General Population	in 58	99%	in 5 70	in 9 923 032
			British Population	in 8	99%	in 8 00	in 2 592 324
			Moroccan Jewish Population	in 00	99%	in 9 90	in 3 960 400
CTSA	Galactosialidosis	AR	General Population	< in 500	99%	in 49 90	< in 0 million
CTSC	Papillon Lefevre syndrome	AR	General Population	< in 500	98%	in 24 95	< in 0 million
CTSD	Neuronal ceroid lipofuscinosis CTSD related	AR	General Population	< in 500	99%	in 49 90	< in 0 million
CTSK	Pycnodysostosis	AR	General Population	< in 500	98%	in 24 95	< in 0 million
CYBA	Chronic granulomatous disease	AR	General Population	in 224	99%	in 22 30	< in 0 million
CYP11A1	Congenital adrenal insufficiency	AR	General Population	in 4	99%	in 30	in 5 53 256
CYP11B1	Congenital adrenal hyperplasia due to beta hydroxylase deficiency	AR	General Population	in 58	98%	in 7 85	in 4 96 832
			Morrocan Jewish Population	in 35	98%	in 70	in 238 40
CYP11B2	Corticosterone methyloxidase deficiency	AR	General Population	< in 500	98%	in 24 95	< in 0 million
CYP17A1	Congenital adrenal hyperplasia due to 7 alpha hydroxylase deficiency	AR	General Population	in 500	98%	in 24 95	< in 0 million
CYP1B1	Primary congenital glaucoma	AR	General Population	in 50	99%	in 4 90	in 980 200
CYP21A2	Congenital adrenal hyperplasia due to 2 hydroxylase deficiency	AR	General Population	in 6	99%	in 6 00	in 464 244
			nuit Population	in 9	99%	in 80	in 28 836
			Middle Eastern Population	in 35	99%	in 3 40	in 476 40
CYP27A1	Cerebrotendinous xanthomatosis	AR	General Population	in 500	98%	in 24 95	< in 0 million
			Morrocan Jewish Population	in 5	98%	in 20	in 4 020
DBT	Maple syrup urine disease type	AR	General Population	in 48	98%	in 24 00	< in 0 million
DCLRE1C	Severe combined immunodeficiency with sensitivity to ionizing radiation	AR	General Population	< in 500	98%	in 24 95	< in 0 million
DDX11	Warsaw breakage syndrome	AR	General Population	< in 500	99%	in 49 90	< in 0 million
			Ashkenazi Jewish Population	in 68	99%	in 6 70	in 822 672
DHCR7	Smith Lemli Opitz syndrome	AR	General Population	in 30	96%	in 726	in 87 20
			African/African American Population	in 38	96%	in 3 426	in 89 52
			Ashkenazi Jewish Population	in 36	96%	in 876	in 26 44
DHDDS	Retinitis pigmentosa 59	AR	General Population	in 296	98%	in 4 75	< in 0 million
			Ashkenazi Jewish Population	in 8	98%	in 5 85	in 2 76 672
DLD	Dihydrolipoamide dehydrogenase deficiency	AR	General Population	in 500	98%	in 24 95	< in 0 million
			Ashkenazi Jewish Population	in 07	98%	in 5 30	in 2 268 828
DNAH5	Primary ciliary dyskinesia DNAH5 related	AR	General Population	in 42	98%	in 7 05	in 4 004 968
			Ashkenazi Jewish Population	in 3	99%	in 20	in 5 062 852
DNAI1	Primary ciliary dyskinesia DNA related	AR	General Population	in 230	98%	in 45	< in 0 million
DNAI2	Primary ciliary dyskinesia DNA 2 related	AR	General Population	in 447	98%	in 22 30	< in 0 million
DUOX2	Congenital hypothyroidism DUOX2 related	AR	General Population	in 366	9 %	in 4 057	in 5 938 797
DUOXA2	Congenital hypothyroidism DUOXA2 related	AR	General Population	< in 500	99%	in 49 90	< in 0 million
DYNC2H1	Short rib thoracic dysplasia 3 with or without polydactyly	AR	General Population	in 68	98%	in 3 35	in 924 876
DYSF	Limb girdle muscular dystrophy type 2B	AR	General Population	< in 500	95%	in 9 98	< in 0 million
			Japanese Population	in 332	95%	in 6 62	in 8 792 688
			Libyan Jewish Population	in 8	95%	in 34	in 24 552
EIF2AK3	Wolcott Rallison Syndrome	AR	General Population	< in 500	98%	in 24 95	< in 0 million
EIF2B5	Leukoencephalopathy with vanishing white matter	AR	General Population	< in 500	99%	in 49 90	< in 0 million
ELP1	Familial Dysautonomia	AR	General Population	in 300	99%	in 29 90	< in 0 million
			Ashkenazi Jewish Population	in 3	99%	in 3 00	in 372 24

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Supplemental Table

Gene	Condition	nheritance	Ethnicity	Carrier Rate	Detection Rate	Post test Carrier Probability*	Residual Risk*
ERCC2	Xeroderma pigmentosum group D	AR	General Population	in 65	99%	in 6 40	in 664 260
ERCC2	Photosensitive trichothiodystrophy	AR	General Population	in 65	99%	in 6 40	in 664 260
ERCC2	Cerebrooculofacioskeletal syndrome 2	AR	General Population	in 65	99%	in 6 40	in 664 260
ERCC5	Xeroderma Pigmentosa group G	AR	General Population	< in 500	99%	in 49 90	< in 0 million
ERCC6	De Sanctis Cacchione syndrome	AR	General Population	in 500	99%	in 49 90	< in 0 million
			Japanese Population	in 74	99%	in 7 30	in 2 6 096
ERCC6	Cockayne syndrome type B	AR	General Population	in 500	99%	in 49 90	< in 0 million
			Japanese Population	in 74	99%	in 7 30	in 2 6 096
ERCC8	Cockayne syndrome type A	AR	General Population	in 822	98%	in 4 05	< in 0 million
ESCO2	Roberts syndrome	AR	General Population	< in 500	99%	in 49 90	< in 0 million
ETFA	Glutaric aciduria A	AR	General Population	in 500	98%	in 24 95	< in 0 million
ETFB	Glutaric aciduria B	AR	General Population	in 500	98%	in 24 95	< in 0 million
ETFDH	Glutaric aciduria C	AR	General Population	in 250	98%	in 2 45	< in 0 million
			East Asian Population	in 74	98%	in 3 65	in 080 696
ETHE1	Ethylmalonic encephalopathy	AR	General Population	< in 500	98%	in 24 95	< in 0 million
EVC	Weyers acrofacial dysostosis EVC related	AR	General Population	in 42	98%	in 7 05	in 4 004 968
			Amish Population	in 7	98%	in 30	in 8 428
EVC	Ellis van Creveld syndrome EVC related	AR	General Population	in 42	98%	in 7 05	in 4 004 968
			Amish Population	in 7	98%	in 30	in 8 428
EVC2	Weyers acrodental dysostosis EVC2 related	AR	General Population	in 240	98%	in 95	< in 0 million
			Amish Population	in 7	98%	in 30	in 8 428
EVC2	Ellis van Creveld syndrome EVC2 related	AR	General Population	in 240	98%	in 95	< in 0 million
			Amish Population	in 7	98%	in 30	in 8 428
EXOSC3	Pontocerebellar hypoplasia type B	AR	General Population	< in 500	98%	in 24 95	< in 0 million
F2	Prothrombin related conditions	AR	General Population	in 33	99%	in 3 20	in 422 532
			Caucasian / European Population	in 4	99%	in 30	in 4 8 6
F5	Factor V deficiency	AR	General Population	in 36	99%	in 3 50	in 504 44
			Caucasian / European Population	in 9	99%	in 80	in 36 876
			Latino Population	in 45	99%	in 4 40	in 792 80
			African/African American Population	in 83	99%	in 8 20	in 2 722 732
			East Asian Population	in 222	99%	in 22 0	< in 0 million
			Native American Population	in 80	99%	in 7 90	in 2 528 320
FAH	Tyrosinemia type	AR	General Population	in 99	95%	in 96	in 776 556
			Ashkenazi Jewish Population	in 50	95%	in 2 98	in 788 600
			Finnish Population	in 22	95%	in 2 42	in 8 448
			French Canadian Population	in 66	95%	in 30	in 343 464
			South Asian/ ndian Population	in 72	95%	in 3 42	in 2 353 648
FAM126A	Hypomyelinating leukodystropy type 5	AR	General Population	< in 500	99%	in 49 90	< in 0 million
FAM161A	Retinitis pigmentosa 28	AR	General Population	in 296	98%	in 4 75	< in 0 million
FANCA	Fanconi anemia group A	AR	General Population	in 239	99%	in 23 80	< in 0 million
			Moroccan Jewish	in 00	99%	in 9 90	in 3 960 400
			ndian Jewish Population	in 27	99%	in 2 60	in 280 908
FANCC	Fanconi anemia group C	AR	General Population	in 535	99%	in 53 40	< in 0 million
			Ashkenazi Jewish Population	in 99	99%	in 9 80	in 3 88 96
FANCG	Fanconi anemia group G	AR	General Population	in 632	90%	in 6 3	< in 0 million
FH	Fumarase deficiency	AR	General Population	< in 500	99%	in 49 90	< in 0 million
			Ashkenazi Jewish Population	in 99	99%	in 9 80	in 3 88 96
FKRP	Muscular dystrophy dystroglycanopathy FKRP related	AR	General Population	in 58	98%	in 7 85	in 4 96 832
FKTN	Muscular dystrophy dystroglycanopathy FKTN related	AR	General Population	< in 500	99%	in 49 90	< in 0 million
			Ashkenazi Jewish Population	in 50	99%	in 4 90	in 8 940 600
			Japanese Population	in 82	99%	in 8 0	in 2 657 28
FKTN	Fukuyama congenital muscular dystrophy	AR	General Population	< in 500	99%	in 49 90	< in 0 million
			Ashkenazi Jewish Population	in 50	99%	in 4 90	in 8 940 600
			Japanese Population	in 82	99%	in 8 0	in 2 657 28
FOXRED1	Mitochondrial complex deficiency	AR	General Population	< in 500	99%	in 49 90	< in 0 million
FTCD	Glutamate formiminotransferase deficiency	AR	General Population	< in 500	99%	in 49 90	< in 0 million
FUCA1	Fucosidosis	AR	General Population	< in 500	99%	in 49 90	< in 0 million
G6PC	Glycogen storage disease type a	AR	General Population	in 77	95%	in 3 52	in 2 492 868
			Ashkenazi Jewish Population	in 64	95%	in 26	in 322 8 6
GAA	Pompe disease	AR	General Population	in 00	98%	in 4 95	in 980 400
			African/African American Population	in 60	98%	in 2 95	in 708 240
			East Asian Population	in 2	98%	in 5 55	in 2 486 848
			Ashkenazi Jewish Population	in 76	99%	in 7 50	in 2 280 304

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GALC	Krabbe disease	AR	General Population	in 58	99%	in 5 70	in 9 923 032
			sraeli Druze Population	in 6	99%	in 50	in 2 024
GALNS	Mucopolysaccharidosis VA (Morquio syndrome A)	AR	General Population	in 224	97%	in 7 434	in 6 660 864
GALT	Galactosemia	AR	General Population	in 0	99%	in 0 90	in 4 796 440
			African/African American Population	in 94	99%	in 9 30	in 3 497 76
			Ashkenazi Jewish Population	in 27	99%	in 2 60	in 6 40 308
GAMT	Guanidinoacetate methyltransferase deficiency	AR	General Population	in 37	99%	in 37 00	< in 0 million
GBA	Gaucher disease	AR	General Population	in 77	99%	in 7 60	in 2 34 08
			African/African American Population	in 35	99%	in 3 40	in 476 40
			Ashkenazi Jewish Population	in 5	99%	in 40	in 84 060
GBE1	Glycogen storage disease V	AR	General Population	in 387	99%	in 38 60	< in 0 million
GCDH	Glutaric aciduria type	AR	General Population	in 87	98%	in 4 30	in 496 748
			Amish Population	in 9	98%	in 40	in 4 436
GDAP1	Charcot Marie Tooth disease GDAP related	AR	General Population	in 52	99%	in 5 0	in 9 8 408
GDF5	Du Pan Syndrome	AR	General Population	< in 500	98%	in 24 95	< in 0 million
GFM1	Combined oxidative phosphorylation deficiency GFM related	AR	General Population	< in 500	98%	in 24 95	< in 0 million
GJB2	Nonsyndromic hearing loss A	AR	General Population	in 42	99%	in 4 0	in 688 968
			African/African American Population	in 25	99%	in 2 40	in 240 00
			Ashkenazi Jewish Population	in 2	99%	in 2 00	in 68 084
			Caucasian / European Population	in 33	99%	in 3 20	in 422 532
			Latino Population	in 00	99%	in 9 90	in 3 960 400
			Middle Eastern Population	in 83	99%	in 8 20	in 2 722 732
			South Asian/ ndian Population	in 48	99%	in 4 70	in 8 702 992
GJB6	GJB6 CRYL related nonsyndromic hearing loss	AR	General Population	in 423	99%	in 42 20	< in 0 million
GLB1	GM gangliosidosis	AR	General Population	in 34	99%	in 3 30	in 7 29 336
			Maltese Population	in 30	99%	in 2 90	in 348 20
			Roma Population	in 50	99%	in 4 90	in 980 200
GLB1	Mucopolysaccharidosis type VB (Morquio syndrome B)	AR	General Population	in 34	99%	in 3 30	in 7 29 336
			Maltese Population	in 30	99%	in 2 90	in 348 20
			Roma Population	in 50	99%	in 4 90	in 980 200
GLDC	Glycine encephalopathy GLDC related	AR	General Population	in 93	98%	in 9 60	in 7 4 972
			British Columbia Canadian Population	in 25	99%	in 2 40	in 6 200 500
			Finnish Population	in 7	99%	in 60	in 5 429 268
GLE1	Lethal congenital contracture syndrome	AR	General Population	< in 500	98%	in 24 95	< in 0 million
			Finnish Population	in 80	98%	in 3 95	in 264 320
GNE	nclusion body myopathy type 2 (Nonaka myopathy)	AR	General Population	< in 500	99%	in 49 90	in 99 802 000
			ranian Jewish Population	in	99%	in 00	in 44 044
GNPTAB	Mucolipidosis alpha/beta	AR	General Population	< in 500	95%	in 9 98	< in 0 million
GNPTAB	Mucolipidosis alpha/beta	AR	General Population	< in 500	95%	in 9 98	< in 0 million
GNPTG	Mucolipidosis gamma	AR	General Population	< in 500	95%	in 9 98	< in 0 million
GNS	Mucopolysaccharidosis D (Sanfilippo syndrome D)	AR	General Population	in 500	98%	in 24 95	< in 0 million
GSS	Glutathione synthetase deficiency	AR	General Population	< in 500	99%	in 49 90	< in 0 million
GUCY2D	Leber congenital amaurosis	AR	General Population	< in 500	98%	in 24 95	< in 0 million
GUSB	Mucopolysaccharidosis type V	AR	General Population	in 250	98%	in 2 45	< in 0 million
HADHA	Trifunctional protein deficiency	AR	General Population	< in 500	98%	in 24 95	< in 0 million
			Finnish Population	in 24	98%	in 6 5	in 3 050 896
HADHA	Long chain 3 hydroxyacyl CoA dehydrogenase (LCHAD) deficiency	AR	General Population	< in 500	98%	in 24 95	< in 0 million
			Finnish Population	in 24	98%	in 6 5	in 3 050 896
HADHB	Trifunctional protein deficiency	AR	General Population	< in 500	98%	in 24 95	< in 0 million
			Finnish Population	in 24	98%	in 6 5	in 3 050 896
HAX1	Severe congenital neutropenia HAX related	AR	General Population	in 224	98%	in 5	in 9 99 296
HBA1	Alpha thalassemia	AR	General Population	in 000	98%	in 860	in 3 440 364
			General Population	in 8	98%	in 860	in 3 440 364
			Southeast Asian Population	≤ in 7	98%	≤ in 305	≤ in 7 228
			Southeast Asian Population	≤ in 4	98%	≤ in 305	≤ in 7 228
			Mediterranean Population	≤ in 6	98%	≤ in 229	≤ in 457 556
			Mediterranean Population	in 500	98%	≤ in 229	≤ in 457 556
			African/African American Population	in 30	98%	in 45	in 5 804 000

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HBA2	Alpha thalassemia	AR	General Population	in 000	98%	in 860	in 3 440 364
			General Population	in 8	98%	in 860	in 3 440 364
			Southeast Asian Population	≤ in 7	98%	≤ in 305	≤ in 7 228
			Southeast Asian Population	≤ in 4	98%	≤ in 305	≤ in 7 228
			Mediterranean Population	≤ in 6	98%	≤ in 229	≤ in 457 556
			Mediterranean Population	in 500	98%	≤ in 229	≤ in 457 556
			African/African American Population	in 30	98%	in 45	in 5 804 000
HBB	Sickle cell disease	AR	General Population	in 58	95%	in 3 4	in 985 2
			African/African American Population	in 0	95%	in 8	in 7 240
			East Asian Population	in 50	95%	in 98	in 96 200
			Latino Population	in 28	95%	in 2 54	in 300 992
			Mediterranean Population	in 3	95%	in 4	in 492
			South Asian/ ndian Population	in 25	95%	in 48	in 48 00
HBB	Hemoglobin C disease	AR	General Population	in 58	95%	in 3 4	in 985 2
			African/African American Population	in 0	95%	in 8	in 7 240
			East Asian Population	in 50	95%	in 98	in 96 200
			Latino Population	in 28	95%	in 2 54	in 300 992
			Mediterranean Population	in 3	95%	in 4	in 492
			South Asian/ ndian Population	in 25	95%	in 48	in 48 00
HBB	Beta thalassemia	AR	General Population	in 58	95%	in 3 4	in 985 2
			African/African American Population	in 0	95%	in 8	in 7 240
			East Asian Population	in 50	95%	in 98	in 96 200
			Latino Population	in 28	95%	in 2 54	in 300 992
			Mediterranean Population	in 3	95%	in 4	in 492
			South Asian/ ndian Population	in 25	95%	in 48	in 48 00
HEXA	Tay Sachs disease	AR	General Population	in 300	99%	in 29 90	< in 0 million
			Ashkenazi Jewish Population	in 27	99%	in 2 60	in 280 908
			Moroccan Jewish Population	in 0	99%	in 0 90	in 4 796 440
HEXB	Sandhoff disease	AR	General Population	in 600	98%	in 29 95	< in 0 million
HGSNAT	Mucopolysaccharidosis type C (Sanfilippo syndrome C)	AR	General Population	in 434	98%	in 2 65	< in 0 million
			Caucasian / European Population	in 345	98%	in 7 20	< in 0 million
HJV	Hemochromatosis type 2A	AR	General Population	in 500	99%	in 49 90	< in 0 million
HLCS	Holocarboxylase synthetase deficiency	AR	General Population	in 500	98%	in 24 95	< in 0 million
HMGCL	3 hydroxy 3 methylglutaryl CoA lyase deficiency	AR	General Population	< in 500	98%	in 24 95	< in 0 million
HOGA1	Primary hyperoxaluria type	AR	General Population	in 84	99%	in 8 30	< in 0 million
HPS1	Hermansky Pudlak syndrome	AR	General Population	in 354	98%	in 7 65	< in 0 million
			Puerto Rican Population	in 2	98%	in 00	in 84 084
HPS3	Hermansky Pudlak syndrome 3	AR	General Population	in 354	98%	in 7 65	< in 0 million
HPS4	Hermansky Pudlak syndrome 4	AR	General Population	< in 500	98%	in 24 95	< in 0 million
HSD17B4	D bifunctional protein deficiency	AR	General Population	in 58	98%	in 7 85	in 4 96 832
HSD3B2	Congenital adrenal hyperplasia due to 3 beta hydroxysteroid dehydrogenase 2 deficiency	AR	General Population	< in 500	98%	in 24 95	< in 0 million
HYLS1	Hydrolethalus syndrome	AR	General Population	< in 500	98%	in 24 95	< in 0 million
			Finnish Population	in 50	98%	in 2 45	in 490 200
IDUA	Mucopolysaccharidosis type (Hurler syndrome)	AR	General Population	< in 500	95%	in 9 98	< in 0 million
			Caucasian / European Population	in 53	95%	in 3 04	in 86 092
IVD	sovaleric Acidemia	AR	General Population	in 67	90%	in 66	in 09 548
			African/African American Population	in 00	90%	in 99	in 396 400
			Caucasian / European Population	in 5	90%	in 4	in 524 860
			East Asian Population	in 407	90%	in 4 06	in 6 6 308
IYD	Thyroid dyshormonogenesis YD related	AR	General Population	< in 500	99%	in 49 90	< in 0 million
JAK3	Severe combined immunodeficiency JAK3 related	AR	General Population	in 299	99%	in 29 80	< in 0 million
KCNJ11	Congenital hyperinsulinism	AR	General Population	in 423	99%	in 42 20	< in 0 million
			Caucasian / European Population	in 232	99%	in 23 0	< in 0 million
KCNJ11	Permanent neonatal diabetes mellitus	AR	General Population	in 423	99%	in 42 20	< in 0 million
			Caucasian / European Population	in 232	99%	in 23 0	< in 0 million
LAMA2	Muscular dystrophy LAMA2 related	AR	General Population	< in 500	99%	in 49 90	< in 0 million
			Caucasian / European Population	in 25	99%	in 2 40	in 6 200 500
LAMA3	Junctional epidermolysis bullosa LAMA3 related	AR	General Population	in 78	98%	in 39 00	< in 0 million
LAMA3	Laryngo onycho cutaneous syndrome	AR	General Population	in 78	98%	in 39 00	< in 0 million
LAMB3	Junctional epidermolysis bullosa LAMB3 related	AR	General Population	in 78	98%	in 39 00	< in 0 million
LAMC2	Junctional epidermolysis bullosa LAMC2 related	AR	General Population	in 78	98%	in 39 00	< in 0 million
LCA5	Leber congenital amaurosis 5	AR	General Population	in 500	98%	in 24 95	< in 0 million

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LDLRAP1	Familial Hypercholesterolemia	AR	General Population	in 8	99%	in 70	in 22 432
			Amish Population	in 2	99%	in 0	in 808
			Caucasian / European Population	in 7	99%	in 60	in 6 828
			French Canadian Population	in 8	99%	in 70	in 22 432
LHX3	Combined pituitary hormone deficiency 3	AR	General Population	in 45	98%	in 2 20	in 396 80
LIFR	Stuve Wiedemann syndrome	AR	General Population	< in 500	98%	in 24 95	< in 0 million
LIPA	Lysosomal acid lipase deficiency	AR	General Population	< in 500	99%	in 49 90	< in 0 million
			Caucasian / European Population	in 2	99%	in 0	in 4 973 248
			ranian Jewish Population	in 26	99%	in 2 50	in 260 04
LMBRD1	Methylmalonic aciduria and homocystinuria cblF type	AR	General Population	< in 500	99%	in 49 90	< in 0 million
LOXHD1	Nonsyndromic hearing loss 77	AR	General Population	in 500	98%	in 24 95	< in 0 million
			Ashkenazi Jewish Population	in 80	98%	in 8 95	in 6 444 720
LPL	Familial lipoprotein lipase deficiency	AR	General Population	in 500	99%	in 49 90	< in 0 million
			French Canadian Population	in 46	99%	in 4 50	in 828 84
LRP2	Donnai Barrow syndrome	AR	General Population	in 2 4	99%	in 0 65	in 9 7 256
LRPPRC	Leigh syndrome with Complex V deficiency	AR	General Population	in 447	98%	in 22 30	< in 0 million
			Faroese Population	in 2	98%	in 00	in 84 084
			French Canadian Population	in 22	98%	in 05	in 92 488
LYST	Chediak Higashi syndrome	AR	General Population	< in 500	90%	in 4 99	in 9 982 000
MAN2B1	Alpha Mannosidosis	AR	General Population	in 354	99%	in 35 30	< in 0 million
			Caucasian / European Population	in 274	99%	in 27 30	< in 0 million
MANBA	Beta Mannosidosis	AR	General Population	< in 500	99%	in 49 90	< in 0 million
MCOLN1	Mucolipidosis V	AR	General Population	in 300	99%	in 29 90	< in 0 million
			Ashkenazi Jewish Population	in 00	99%	in 9 90	in 3 960 400
MCPH1	Primary microcephaly recessive	AR	General Population	in 47	99%	in 4 60	in 8 585 388
MED17	Postnatal Progressive Microcephaly with Seizures and Brain Atrophy	AR	General Population	< in 500	99%	in 49 90	< in 0 million
			Bukharan/Kurdish Jewish Population	in 20	99%	in 90	in 52 080
MESP2	Spondylocostal dysostosis	AR	General Population	< in 500	98%	in 24 95	< in 0 million
MFSD8	Neuronal ceroid lipofuscinosis MFSD8 related	AR	General Population	< in 500	95%	in 9 98	< in 0 million
MKS1	Bardet Biedl syndrome 3	AR	General Population	in 260	98%	in 2 95	< in 0 million
			Finnish Population	in 47	98%	in 2 30	in 432 588
MKS1	Joubert syndrome 28	AR	General Population	in 260	98%	in 2 95	< in 0 million
			Finnish Population	in 47	98%	in 2 30	in 432 588
MKS1	Meckel syndrome	AR	General Population	in 260	98%	in 2 95	< in 0 million
			Finnish Population	in 47	98%	in 2 30	in 432 588
MLC1	Megalencephalic leukoencephalopathy with subcortical cysts	AR	General Population	< in 500	99%	in 49 90	< in 0 million
			Libyan Jewish Population	in 40	99%	in 3 90	in 624 60
MLYCD	Malonyl CoA decarboxylase deficiency	AR	General Population	< in 500	98%	in 24 95	< in 0 million
MMAA	Methylmalonic aciduria cblA type	AR	General Population	in 30	97%	in 0 00	< in 0 million
MMAB	Methylmalonic aciduria cblB type	AR	General Population	in 435	98%	in 2 70	< in 0 million
MMACHC	Methylmalonic aciduria and homocystinuria cblC type	AR	General Population	in 34	90%	in 33	in 7 3 4 6
MMADHC	Methylmalonic aciduria and homocystinuria cblD type	AR	General Population	< in 500	98%	in 24 95	< in 0 million
MPI	Congenital disorder of glycosylation type b	AR	General Population	< in 500	98%	in 24 95	< in 0 million
MPL	Congenital amegakaryocytic thrombocytopenia	AR	General Population	in 02	98%	in 5 05	in 2 060 808
			Ashkenazi Jewish Population	in 55	98%	in 2 70	in 594 220
MPV17	Hepatocerebral mitochondrial DNA depletion syndrome MPV 7 related	AR	General Population	< in 500	96%	in 2 476	< in 0 million
			Native American Population	in 20	96%	in 476	in 38 080
MTHFR	Homocystinuria MTHFR related	AR	General Population	in 224	98%	in 5	in 9 99 296
MTMR2	Charcot Marie Tooth disease type 4B	AR	General Population	< in 500	99%	in 49 90	< in 0 million
MTRR	Homocystinuria megaloblastic anemia cobalamin E type	AR	General Population	< in 500	98%	in 24 95	< in 0 million
MTTP	Abetalipoproteinemia	AR	General Population	< in 500	98%	in 24 95	< in 0 million
			Ashkenazi Jewish Population	in 80	98%	in 8 95	in 6 444 720
MUT	Methylmalonic acidemia MUT related	AR	General Population	in 95	96%	in 4 85	in 3 783 780
			East Asian Population	in 53	96%	in 30	in 275 8 2
			Middle Eastern Population	in 52	96%	in 276	in 265 408
MUT	Methylmalonic aciduria methylmalonyl CoA mutase deficiency	AR	General Population	in 00	99%	in 9 90	in 3 960 400
MVK	Hyperimmunoglobulinemia D syndrome	AR	General Population	< in 500	99%	in 49 90	< in 0 million
MVK	Mevalonate kinase deficiency	AR	General Population	< in 500	99%	in 49 90	< in 0 million
MYO7A	Usher syndrome type B	AR	General Population	in 206	98%	in 0 25	in 8 446 824
			East Asian Population	in 62	98%	in 3 05	in 756 648

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MYO7A	Non syndromic hearing loss MYO7A related	AR	General Population	in 206	98%	in 0 25	in 8 446 824
			East Asian Population	in 62	98%	in 3 05	in 756 648
NAGA	Schindler disease types and 3	AR	General Population	in 94	99%	in 9 30	in 3 497 76
NAGLU	Mucopolysaccharidosis type B (Sanfilippo syndrome B)	AR	General Population	< in 500	99%	in 49 90	< in 0 million
			Caucasian / European Population	in 346	99%	in 34 50	< in 0 million
			East Asian Population	in 298	99%	in 29 70	< in 0 million
NAGS	N acetylglutamate synthase deficiency	AR	General Population	< in 500	98%	in 24 95	< in 0 million
NBN	Nijmegen breakage syndrome	AR	General Population	in 58	99%	in 5 70	in 9 923 032
NDRG1	Charcot Marie Tooth disease type 4D	AR	General Population	in 22	98%	in 05	in 92 488
NDUFAF2	Mitochondrial complex deficiency	AR	General Population	< in 500	99%	in 49 90	< in 0 million
NDUFAF5	Mitochondrial complex deficiency (Leigh syndrome)	AR	General Population	in 447	98%	in 22 30	< in 0 million
			Ashkenazi Jewish Population	in 290	98%	in 4 45	< in 0 million
NDUFS4	Mitochondrial complex deficiency	AR	General Population	< in 500	99%	in 49 90	< in 0 million
NDUFS4	Mitochondrial complex deficiency	AR	General Population	< in 500	99%	in 49 90	< in 0 million
			Hutterite Population	in 27	99%	in 2 60	in 280 908
NDUFS6	Mitochondrial complex deficiency (Leigh syndrome)	AR	General Population	< in 500	99%	in 49 90	< in 0 million
			Bukharan/Kurdish Jewish Population	in 24	99%	in 2 30	in 220 896
NDUFS7	Mitochondrial complex deficiency	AR	General Population	< in 500	99%	in 49 90	< in 0 million
NDUFV1	Mitochondrial complex deficiency nuclear type 4	AR	General Population	< in 500	99%	in 49 90	< in 0 million
NEB	Nemaline myopathy	AR	General Population	in 2	98%	in 5 55	in 2 486 848
			Amish Population	in	98%	in 50	in 22 044
			Ashkenazi Jewish Population	in 08	98%	in 5 35	in 2 3 632
			Finnish Population	in 2	98%	in 5 55	in 2 486 848
NEU1	Sialidosis type and	AR	General Population	< in 500	99%	in 49 90	< in 0 million
NPC1	Niemann Pick disease type C	AR	General Population	in 94	90%	in 93	in 498 456
NPC2	Niemann Pick disease type C2	AR	General Population	in 94	99%	in 9 30	< in 0 million
NPHP1	Joubert syndrome 4	AR	General Population	in 480	98%	in 23 95	< in 0 million
			Finnish Population	in 24	98%	in 6 5	in 3 050 896
NPHP1	Nephronophthisis	AR	General Population	in 480	98%	in 23 95	< in 0 million
			Finnish Population	in 24	98%	in 6 5	in 3 050 896
NPHP1	NPHP related disorders	AR	General Population	in 480	98%	in 23 95	< in 0 million
			Finnish Population	in 24	98%	in 6 5	in 3 050 896
NPHP1	Senior Løken syndrome	AR	General Population	in 480	98%	in 23 95	< in 0 million
			Finnish Population	in 24	98%	in 6 5	in 3 050 896
NPHS1	Congenital nephrotic syndrome type	AR	General Population	in 289	98%	in 4 40	< in 0 million
			Finnish Population	in 50	98%	in 2 45	in 490 200
NPHS2	Congenital nephrotic syndrome type 2	AR	General Population	in 289	98%	in 4 40	< in 0 million
			Finnish Population	in 50	98%	in 2 45	in 490 200
NTRK1	Congenital insensitivity to pain with anhidrosis	AR	General Population	< in 500	99%	in 49 90	< in 0 million
OAT	Gyrate atrophy of choroid and retina	AR	General Population	< in 500	98%	in 24 95	< in 0 million
OCA2	Oculocutaneous albinism type	AR	General Population	in 76	99%	in 7 50	in 2 280 304
OPA3	Costeff syndrome	AR	General Population	< in 500	98%	in 24 95	< in 0 million
			raqi Jewish Population	in 50	98%	in 2 45	in 490 200
OTOF	Nonsyndromic hearing loss OTOF related	AR	General Population	< in 500	99%	in 49 90	< in 0 million
			Spanish Population	in 06	99%	in 0 50	in 4 452 424
P3H1	Osteogenesis imperfecta type V	AR	General Population	< in 500	99%	in 49 90	< in 0 million
			West African Population	in 67	99%	in 6 60	in 769 068
			African American Population	in 250	99%	in 24 90	< in 0 000 000
PAH	Phenylalanine Hydroxylase deficiency (Phenylketonuria)	AR	General Population	in 93	99%	in 9 20	in 3 422 772
			Caucasian / European Population	in 63	99%	in 6 20	in 562 652
			Middle Eastern Population	in 74	99%	in 7 30	in 2 6 096
			South East Asian	in 59	99%	in 5 80	in 369 036
PANK2	Pantothenate kinase associated neurodegeneration	AR	General Population	in 289	99%	in 28 80	< in 0 million
PC	Pyruvate carboxylase deficiency	AR	General Population	in 250	95%	in 4 98	in 4 98 000
PCCA	Propionic acidemia PCCA related	AR	General Population	in 224	96%	in 5 576	in 4 996 096
			Native American Population	in 85	96%	in 2 0	in 7 4 340
PCCB	Propionic acidemia PCCB related	AR	General Population	in 224	99%	in 22 30	< in 0 million
			Native American Population	in 85	99%	in 8 40	in 2 856 340
PCDH15	Non syndromic hearing loss PCDH 5 related	AR	General Population	in 395	98%	in 9 70	in 78 804
			Ashkenazi Jewish Population	in 72	98%	in 3 55	in 4 204
PCDH15	Usher syndrome type F	AR	General Population	in 395	98%	in 9 70	in 78 804
			Ashkenazi Jewish Population	in 72	98%	in 3 55	in 4 204

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Supplemental Table

Gene	Condition	nheritance	Ethnicity	Carrier Rate	Detection Rate	Post test Carrier Probability*	Residual Risk*
PCNT	Microcephalic osteodysplastic primordial dwarfism type	AR	General Population	< in 500	98%	in 24 95	< in 0 million
PDHB	Pyruvate dehydrogenase E beta deficiency	AR	General Population	< in 500	98%	in 24 95	< in 0 million
PEX1	Zellweger syndrome PEX related	AR	General Population	in 47	95%	in 2 92	in 7 7 548
PEX10	Zellweger syndrome PEX 0 related	AR	General Population Japanese Population	in 500 in 354	95% 95%	in 9 98 in 7 06	< in 0 million in 9 998 376
PEX12	Zellweger syndrome PEX 2 related	AR	General Population	in 373	95%	in 7 44	< in 0 million
PEX2	Zellweger syndrome PEX2 related	AR	General Population Ashkenazi Jewish Population	in 500 in 23	95% 95%	in 9 98 in 2 44	< in 0 million in 200 972
PEX26	Zellweger syndrome	AR	General Population	< in 500	99%	in 49 90	< in 0 million
PEX6	Zellweger syndrome PEX6 related	AR	General Population Yemenite Jewish Population	in 280 in 8	99% 99%	in 27 90 in 70	< in 0 million in 22 472
PEX7	Rhizomelic chondrodysplasia punctata type	AR	General Population	in 58	99%	in 5 70	in 9 923 032
PFKM	Glycogen storage disease V	AR	General Population Ashkenazi Jewish Population	< in 500 in 20	99% 99%	in 49 90 in 90	< in 0 million in 5 7 2 480
PHGDH	Phosphoglycerate dehydrogenase deficiency	AR	General Population Ashkenazi Jewish Population	< in 500 in 280	98% 98%	in 24 95 in 3 95	< in 0 million < in 0 million
PHYH	Refsum disease	AR	General Population	< in 500	99%	in 49 90	< in 0 million
PKHD1	Polycystic kidney disease PKHD related	AR	General Population Ashkenazi Jewish Population	in 70 in 07	98% 98%	in 3 45 in 5 30	in 966 280 in 2 268 828
PLA2G6	nfantile neuroaxonal dystrophy	AR	General Population	in 500	97%	in 6 634	< in 0 million
PLOD1	Ehlers Danlos syndrome with kyphoscoliosis PLOD related	AR	General Population	in 59	99%	in 5 80	< in 0 million
PMM2	Congenital disorder of glycosylation type a	AR	General Population Ashkenazi Jewish Population Caucasian / European Population	in 63 in 57 in 7	99% 99% 99%	in 6 20 in 5 60 in 7 00	in 562 652 in 277 028 in 988 284
POLG	Ataxia neuropathy spectrum	AR	General Population	in 3	95%	in 2 24	in 0 2 932
POLG	Progressive external ophthalmoplegia	AR	General Population	in 3	95%	in 2 24	in 0 2 932
POLG	Myocerebrohepatopathy syndrome	AR	General Population	in 3	95%	in 2 24	in 0 2 932
POLG	POLG related disorders	AR	General Population	in 3	95%	in 2 24	in 0 2 932
POLG	Alpers Huttenlocher syndrome	AR	General Population	in 3	95%	in 2 24	in 0 2 932
POLR1C	Hypomyelinating Leukodystrophy POLR C related	AR	General Population	< in 500	99%	in 49 90	< in 0 million
POLR1C	Treacher Collins syndrome POLR C related	AR	General Population	< in 500	99%	in 49 90	< in 0 million
POMGNT1	Muscular dystrophy dystroglycanopathy	AR	General Population Finnish Population	in 462 in	98% 98%	in 23 05 in 5 50	< in 0 million in 2 442 444
POMGNT1	Retinitis pigmentosa 76	AR	General Population Finnish Population	in 462 in	98% 98%	in 23 05 in 5 50	< in 0 million in 2 442 444
POMT1	Muscular dystrophy dystroglycanopathy POMT related	AR	General Population	in 290	99%	in 28 90	< in 0 million
POMT2	Muscular dystrophy dystroglycanopathy POMT2 related	AR	General Population	in 37	99%	in 37 00	< in 0 million
POR	Antley Bixler syndrome	AR	General Population	in 59	98%	in 7 90	in 5 025 036
PPT1	Neuronal ceroid lipofuscinosis PPT related	AR	General Population Caucasian / European Population Finnish Population	in 368 in 488 in 75	98% 98% 98%	in 8 35 in 24 35 in 3 70	< in 0 million < in 0 million in 0 300
PRF1	Hemophagocytic lymphohistiocytosis familial 2	AR	General Population	in 49	99%	in 4 80	in 8 82 396
PROP1	Combined pituitary hormone deficiency 2	AR	General Population	in 45	98%	in 2 20	in 396 80
PSAP	Metachromatic leukodystrophy due to saposin b deficiency	AR	General Population	< in 500	98%	in 24 95	< in 0 million
PTS	Tetrahydrobiopterin deficiency	AR	General Population	in 354	96%	in 8 826	< in 0 million
PUS1	Mitochondrial myopathy and sideroblastic anemia	AR	General Population	< in 500	98%	in 24 95	< in 0 million
QDPR	Tetrahydrobiopterin deficiency QDPR related	AR	General Population	< in 500	99%	in 49 90	< in 0 million
RAB23	Carpenter syndrome	AR	General Population	< in 500	98%	in 24 95	< in 0 million
RAG1	Omenn syndrome RAG related	AR	General Population	in 290	98%	in 4 45	in 6 763 60
RAG2	Omenn syndrome RAG2 related	AR	General Population	in 37	98%	in 6 80	in 3 726 948
RAPSN	Congenital myasthenic syndrome RAPSN related	AR	General Population	< in 500	99%	in 49 90	< in 0 million
RAPSN	Fetal akinesia deformation sequence	AR	General Population	< in 500	99%	in 49 90	< in 0 million
RARS2	Pontocerebellar hypoplasia type 6	AR	General Population	< in 500	98%	in 24 95	< in 0 million
RAX	Microphthalmia isolated 3	AR	General Population	in 289	99%	in 28 80	< in 0 million
RDH12	Leber congenital amaurosis type 3	AR	General Population Caucasian / European Population	< in 500 in 456	98% 98%	in 24 95 in 22 75	< in 0 million < in 0 million

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RMRP	Metaphyseal dysplasia without hypotrichosis	AR	General Population	< in 500	99%	in 49 90	< in 0 million
			Amish Population	in 6	99%	in 50	in 96 064
			Finnish Population	in 76	99%	in 7 50	in 2 280 304
RMRP	Cartilage Hair Hypoplasia Anauxetic Dysplasia Spectrum Disorder	AR	General Population	< in 500	99%	in 49 90	< in 0 million
			Amish Population	< in 500	99%	in 49 90	< in 0 million
			Finnish Population	< in 500	99%	in 49 90	< in 0 million
RMRP	Anauxetic dysplasia	AR	General Population	< in 500	99%	in 49 90	< in 0 million
			Amish Population	in 6	99%	in 50	in 96 064
			Finnish Population	in 76	99%	in 7 50	in 2 280 304
RMRP	Cartilage hair hypoplasia	AR	General Population	< in 500	99%	in 49 90	< in 0 million
			Amish Population	in 6	99%	in 50	in 96 064
			Finnish Population	in 76	99%	in 7 50	in 2 280 304
RNASEH2B	Aicardi Goutieres syndrome 2	AR	General Population	in 2 7	99%	in 0 80	in 9 375 268
RPE65	Retinitis pigmentosa 20	AR	General Population	in 228	98%	in 35	< in 0 million
RPE65	Leber congenital amaurosis 2	AR	General Population	in 228	98%	in 35	< in 0 million
RPGRIP1L	COACH syndrome	AR	General Population	in 259	98%	in 2 90	< in 0 million
RPGRIP1L	Joubert syndrome 7	AR	General Population	in 259	98%	in 2 90	< in 0 million
RPGRIP1L	Meckel syndrome 5	AR	General Population	in 259	98%	in 2 90	< in 0 million
RTEL1	Dyskeratosis congenita type 5	AR	General Population	in 500	99%	in 49 90	< in 0 million
			Ashkenazi Jewish Population	in 203	99%	in 20 20	< in 0 million
SACS	Autosomal recessive spastic ataxia of Charlevoix Saguenay	AR	General Population	< in 500	95%	in 9 98	< in 0 million
			French Canadian Population	in 9	95%	in 36	in 27 436
SAMD9	Normophosphatemic Familial Tumoral Calcinosis	AR	General Population	< in 500	99%	in 49 90	< in 0 million
			Yemeni Jewish Population	in 25	99%	in 2 40	in 240 00
SAMHD1	Aicardi Goutieres syndrome	AR	General Population	< in 500	95%	in 9 98	< in 0 million
SCO2	Mitochondrial complex V deficiency	AR	General Population	in 50	99%	in 4 90	in 8 940 600
SEPSECS	Pontocerebellar hypoplasia type 2D	AR	General Population	< in 500	99%	in 49 90	< in 0 million
			Moroccan/ raqi Jewish Population	in 44	99%	in 4 30	in 756 976
SERPINA1	Alpha antitrypsin deficiency	AR	General Population	in 33	95%	in 64	in 84 6 2
			Caucasian / European Population	in 9	95%	in 36	in 27 436
SGCA	Limb girdle muscular dystrophy type 2D	AR	General Population	< in 500	98%	in 24 95	< in 0 million
			Caucasian / European Population	in 288	98%	in 4 35	< in 0 million
			Finnish Population	in 50	98%	in 7 45	in 4 470 600
SGCB	Limb girdle muscular dystrophy type 2E	AR	General Population	in 500	98%	in 24 95	< in 0 million
			Caucasian / European Population	in 406	98%	in 20 25	< in 0 million
SGCD	Limb girdle muscular dystrophy type 2F	AR	General Population	< in 500	98%	in 24 95	< in 0 million
SGCG	Limb girdle muscular dystrophy type 2C	AR	General Population	in 38	98%	in 9 00	< in 0 million
			Moroccan Population	in 250	98%	in 2 45	< in 0 million
			Roma / Gypsy Population	in 96	98%	in 4 75	in 824 384
SGSH	Mucopolysaccharidosis A (Sanfilippo syndrome A)	AR	General Population	in 454	98%	in 22 65	< in 0 million
			Caucasian / European Population	in 253	98%	in 2 60	< in 0 million
SH3TC2	Charcot Marie Tooth disease SH3TC2 related	AR	General Population	in 69	99%	in 6 80	in 877 076
SLC12A6	Andermann syndrome	AR	General Population	< in 500	98%	in 24 95	< in 0 million
			French Canadian Population	in 23	99%	in 2 20	in 202 492
SLC17A5	Sialic acid storage disorder	AR	General Population	< in 500	9 %	in 5 545	< in 0 million
			Finnish Population	in 00	9 %	in 0	in 440 400
SLC19A3	Refsum disease	AR	General Population	< in 500	99%	in 49 90	< in 0 million
SLC19A3	Biotin responsive basal ganglia disease	AR	General Population	in 09	99%	in 5 40	in 2 354 836
SLC1A4	Spastic tetraplegia thin corpus callosum and progressive microcephaly syndrome	AR	General Population	< in 500	99%	in 49 90	< in 0 million
			Ashkenazi Jewish Population	in 06	99%	in 0 50	in 4 452 424
SLC22A5	Systemic primary carnitine deficiency	AR	General Population	in 29	99%	in 2 80	in 6 605 3 6
			African/African American Population	in 86	99%	in 8 50	in 2 924 344
			East Asian Population	in 77	99%	in 7 60	in 2 34 08
			Faroese Population	in 9	99%	in 80	in 28 836
			Pacific slander Population	in 37	99%	in 3 60	in 532 948
			South Asian/ ndian Population	in 5	99%	in 5 00	in 020 204
SLC25A13	Citrin deficiency	AR	General Population	< in 500	95%	in 9 98	< in 0 million
			East Asian Population	in 65	95%	in 28	in 333 060
SLC25A15	Hyperornithinemia hyperammonemia homocitrullinemia syndrome (Triple H syndrome)	AR	General Population	< in 500	99%	in 49 90	< in 0 million
			French Canadian Population	in 37	99%	in 3 60	in 532 948
SLC26A2	Diastrophic dysplasia	AR	General Population	in 58	90%	in 57	in 992 872
			Finnish Population	in 50	90%	in 49	in 98 200

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SLC26A2	Achondrogenesis type B	AR	General Population	in 58	90%	in 57	in 992 872
			Finnish Population	in 50	90%	in 49	in 98 200
SLC26A2	Multiple epiphyseal dysplasia	AR	General Population	in 58	90%	in 57	in 992 872
			Finnish Population	in 50	90%	in 49	in 98 200
SLC26A2	Atelosteogenesis	AR	General Population	in 58	90%	in 57	in 992 872
			Finnish Population	in 50	90%	in 49	in 98 200
SLC26A3	Congenital secretory chloride diarrhea	AR	General Population	< in 500	98%	in 24 95	< in 0 million
			Middle Eastern Population	in 57	98%	in 2 80	in 638 628
SLC35A3	Arthrogryposis intellectual disability and seizures	AR	General Population	< in 500	98%	in 24 95	< in 0 million
			Ashkenazi Jewish Population	in 453	98%	in 22 60	< in 0 million
SLC37A4	Glycogen storage disease type b	AR	General Population	in 58	95%	in 3 4	in 985 2
			Ashkenazi Jewish Population	in 7	95%	in 40	in 397 884
SLC39A4	Acrodermatitis enteropathica	AR	General Population	< in 500	98%	in 24 95	< in 0 million
SLC45A2	Oculocutaneous albinism type V	AR	General Population	in 59	98%	in 7 90	in 5 025 036
			Japanese Population	in 46	98%	in 7 25	in 4 234 584
SLC46A1	Hereditary folate malabsorption	AR	General Population	< in 500	99%	in 49 90	< in 0 million
			Puerto Rican Population	in 500	99%	in 49 90	< in 0 million
SLC5A5	Thyroid dyshormonogenesis SLC5A5 related	AR	General Population	< in 500	99%	in 49 90	< in 0 million
SLC7A7	Lysinuric protein intolerance	AR	General Population	< in 500	95%	in 9 98	< in 0 million
			Finnish Population	in 22	95%	in 2 42	in 8 448
			Japanese Population	in 9	95%	in 2 36	in 23 836
SMARCAL1	Schimke immunoosseous dysplasia	AR	General Population	in 500	90%	in 4 99	in 9 982 000
SMN1	Spinal muscular atrophy	AR	General Population	in 54	9 %	in 590	in 27 440
			African/African American Population	in 72	7 %	in 246	in 70 848
			Ashkenazi Jewish Population	in 67	9 %	in 734	in 96 7 2
			Caucasian / European Population	in 47	95%	in 92	in 73 48
			East Asian Population	in 59	93%	in 830	in 95 880
			Latino Population	in 68	90%	in 67	in 82 5 2
			Sephardic Jewish Population	in 34	96%	in 826	in 2 336
SMN1	Spinal muscular atrophy silent carrier	AR	General Population	in 54	9 %	in 590	in 27 440
SMPD1	Niemann Pick disease type A/B	AR	General Population	in 250	95%	in 4 98	in 4 98 000
			Ashkenazi Jewish Population	in 5	95%	in 2 28	in 049 260
			Latino Population	in 06	95%	in 2 0	in 890 824
SPG11	SPG related Neuromuscular Disorders	AR	General Population	in 59	99%	in 5 80	< in 0 million
SPINK5	Netherton syndrome	AR	General Population	in 224	99%	in 23 30	< in 0 million
			Ashkenazi Jewish Population	in 7	99%	in 60	in 08 868
STAR	Lipoid congenital adrenal hyperplasia	AR	General Population	< in 500	98%	in 24 95	< in 0 million
SUMF1	Multiple sulfatase deficiency	AR	General Population	in 500	98%	in 24 95	< in 0 million
			Ashkenazi Jewish Population	in 320	98%	in 5 95	< in 0 million
SURF1	Charcot Marie Tooth disease SURF related	AR	General Population	< in 500	99%	in 49 90	< in 0 million
SURF1	Leigh syndrome SURF related	AR	General Population	< in 500	99%	in 49 90	< in 0 million
TCIRG1	Osteopetrosis TC RG related	AR	General Population	in 250	98%	in 2 45	< in 0 million
TCTN2	Meckel syndrome 8	AR	General Population	< in 500	99%	in 49 90	< in 0 million
			Ethiopian Jewish Population	in 42	99%	in 4 0	in 688 968
			Yemenite Jewish Population	in 78	99%	in 7 70	in 2 402 7 2
TCTN2	Joubert syndrome 24	AR	General Population	< in 500	99%	in 49 90	< in 0 million
TECPR2	Spastic paraplegia 49	AR	General Population	< in 500	98%	in 24 95	< in 0 million
TF	Atransferrinemia	AR	General Population	in 6	99%	in 50	in 5 336 464
TG	Thyroid dyshormonogenesis TG related	AR	General Population	in 24	99%	in 24 00	< in 0 million
TGM1	Congenital ichthyosis	AR	General Population	in 224	95%	in 4 46	in 3 997 056
TH	Segawa syndrome	AR	General Population	in 224	98%	in 5	in 9 99 296
TMEM216	Joubert syndrome 2	AR	General Population	in 4	98%	in 7 00	in 3 948 564
			Ashkenazi Jewish Population	in 92	98%	in 4 55	in 674 768
TMEM216	Meckel syndrome 2	AR	General Population	in 4	98%	in 7 00	in 3 948 564
			Ashkenazi Jewish Population	in 92	98%	in 4 55	in 674 768
TPO	Thyroid dyshormonogenesis TPO related	AR	General Population	in 373	99%	in 37 20	< in 0 million
TPP1	Neuronal ceroid lipofuscinosis TPP related	AR	General Population	in 252	97%	in 8 368	in 8 434 944
			French Canadian Population	in 53	97%	in 734	in 367 608
TRDN	Catecholaminergic polymorphic ventricular tachycardia	AR	General Population	in 354	98%	in 7 65	< in 0 million
TRIM32	Limb girdle muscular dystrophy type 2H	AR	General Population	< in 500	98%	in 24 95	< in 0 million
			Hutterite Population	in 2	98%	in 55	in 26 448

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TRIM32	Bardet Biedl syndrome	AR	General Population	< in 500	98%	in 24 95	< in 0 million
			Hutterite Population	in 2	98%	in 55	in 26 448
TRMU	Liver failure acute infantile	AR	General Population	< in 500	98%	in 24 95	< in 0 million
			Yemeni Jewish Population	in 34	98%	in 65	in 224 536
TSEN54	Pontocerebellar hypoplasia TSEN54 related	AR	General Population	in 250	98%	in 2 45	< in 0 million
TSFM	Combined oxidative phosphorylation deficiency TSFM related	AR	General Population	< in 500	98%	in 24 95	< in 0 million
			Finnish Population	in 80	98%	in 3 95	in 264 320
TSHB	Congenital hypothyroidism TSHB related	AR	General Population	in 500	99%	in 49 90	< in 0 million
TTC37	Trichohepatoenteric syndrome	AR	General Population	in 500	98%	in 24 95	< in 0 million
TTPA	Ataxia with isolated vitamin E deficiency	AR	General Population	< in 500	98%	in 24 95	< in 0 million
			Caucasian / European Population	in 267	90%	in 2 66	in 2 84 948
TYMP	Mitochondrial neurogastrointestinal encephalopathy (MNG E) disease	AR	General Population	< in 500	98%	in 24 95	< in 0 million
TYR	Oculocutaneous albinism types A and B	AR	General Population	in 20	99%	in 90	in 52 080
TYRP1	Oculocutaneous albinism type	AR	General Population	< in 500	98%	in 24 95	< in 0 million
			African Population	in 47	98%	in 2 30	in 432 588
UGT1A1	Crigler Najjar syndrome	AR	General Population	< in 500	98%	in 24 95	< in 0 million
USH1C	Usher syndrome type C	AR	General Population	in 353	90%	in 3 52	in 4 97 652
			French Canadian Population	in 227	90%	in 2 26	in 2 052 988
USH1C	Non syndromic hearing loss USH C related	AR	General Population	in 353	90%	in 3 52	in 4 97 652
			French Canadian Population	in 227	90%	in 2 26	in 2 052 988
USH1G	Usher syndrome type G	AR	General Population	in 434	99%	in 43 30	< in 0 million
USH2A	Usher syndrome type 2A	AR	General Population	in 26	96%	in 3 26	in 575 504
			Caucasian / European Population	in 73	96%	in 80	in 525 892
			Ashkenazi Jewish Population	in 35	99%	in 3 40	in 476 40
			ranian Jewish Population	in 60	99%	in 5 90	in 4 6 240
VPS13A	Choreoacanthocytosis	AR	General Population	< in 500	98%	in 24 95	< in 0 million
VPS13B	Cohen syndrome	AR	General Population	< in 500	98%	in 24 95	< in 0 million
VPS45	Severe congenital neutropenia VPS45 related	AR	General Population	in 224	98%	in 5	in 9 99 296
VPS53	Pontocerebellar hypoplasia VPS53 related	AR	General Population	< in 500	98%	in 24 95	< in 0 million
			Moroccan Jewish Population	in 37	98%	in 80	in 266 548
VRK1	Pontocerebellar hypoplasia type A	AR	General Population	< in 500	98%	in 24 95	< in 0 million
VSX2	Microphthalmia with or without coloboma	AR	General Population	in 9	98%	in 4 50	in 638 364
WHRN	Usher syndrome type 2D	AR	General Population	in 282	99%	in 28 0	< in 0 million
WRN	Werner syndrome	AR	General Population	in 308	98%	in 5 35	< in 0 million
			Caucasian / European Population	in 2	98%	in 5 55	in 2 486 848
			Japanese Population	in 7	98%	in 3 50	in 994 284
XPA	Xeroderma pigmentosum group A	AR	General Population	in 500	99%	in 49 90	< in 0 million
			Japanese Population	in 74	99%	in 7 30	in 2 6 096
XPC	Xeroderma pigmentosum group C	AR	General Population	in 500	99%	in 49 90	< in 0 million
ZFYVE26	Spastic paraplegia 5	AR	General Population	< in 500	98%	in 24 95	< in 0 million

* For genes that have tested negat ve
† The carr er frequency for heterozygous a pha tha assem a carr ers (αα/α-) s descr bed n rows marked w th a dagger symbo . The carr er frequency for a pha tha assem a tra t c s (αα/- -) s 1 n 1000.
Abbrev at ons: AR, autosoma recess ve; XL, X- nked