Donor MQ191022

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.
- 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:

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, 99.55% and 99.50% 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. New York pat ents: d agnost c f nd ngs are conf rmed by Sanger, MLPA, or qPCR; except on SNV var ants n genes for wh ch conf rmat on of NGS resu ts has been performed >=10 t mes may not be conf rmed f dent f ed w th h gh qua ty by NGS. 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 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 . The var an s c.188A>T (p. s63Leu), c.844G>T (p.Va 282Leu), c.1174G>A (p.A a392Thr), and c.1360C>T (p.Pro454Ser) n CYP21A2 w no be rou ne y repor ed as hese var an s are pr mar y assoc a ed w h non-c ass c congen a adrena hyperp as a and ow d sease pene rance. Add ona y, he var an c.955C>T (p.G n319Ter) s n he reg on w h pseudogene n erference, and he probab y of h s var an occurr ng n he rea gene s grea er han 50%. When observed, h s var an w be repor ed as a poss b e carr er w hou LR-PCR. The conf rma on es s recommended f he second reproduc ve par ner s es s pos ve for var an s n CYP21A2. *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. *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).

SIGNATURE:

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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	Inheritance	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

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 CLN5 related	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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Supplemental Table

Gene	Condition	nheritance	Ethnicity	Carrier Rate	Detection Rate	Post test Carrier Probability*	Residual Risk*
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	98%	in 24 95	< 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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Supplemental Table

Gene	Condition	nheritance	Ethnicity	Carrier Rate	Detection Rate	Post test Carrier Probability*	Residual Risk*
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 GJB2 related	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 8	98%	in 860	in 3 440 364
			General Population	in 000	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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Supplemental Table

Gene	Condition	nheritance	Ethnicity	Carrier Rate	Detection Rate	Post test Carrier Probability*	Residual Risk*
HBA2	Alpha thalassemia	AR	General Population	in 8	98%	in 860	in 3 440 364
			General Population	in 000	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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Supplemental Table

Gene	Condition	nheritance	Ethnicity	Carrier Rate	Detection Rate	Post test Carrier Probability*	Residual Risk*
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 LOXHD related	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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Supplemental Table

Gene	Condition	Inheritance	Ethnicity	Carrier Rate	Detection Rate	Post test Carrier Probability*	Residual Risk*
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 00	98%	in 4 95	in 980 400
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

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Supplemental Table

Gene	Condition	nheritance	Ethnicity	Carrier Rate	Detection Rate	Post test Carrier Probability*	Residual Risk*
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
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	in 500	95%	in 9 98	< in 0 million
			Japanese Population	in 354	95%	in 7 06	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	in 500	95%	in 9 98	< in 0 million
			Ashkenazi Jewish Population	in 23	95%	in 2 44	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	in 280	99%	in 27 90	< in 0 million
			Yemenite Jewish Population	in 8	99%	in 70	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	< in 500	99%	in 49 90	< in 0 million
			Ashkenazi Jewish Population	in 20	99%	in 90	in 5 7 2 480
PHGDH	Phosphoglycerate dehydrogenase deficiency	AR	General Population	< in 500	98%	in 24 95	< in 0 million
			Ashkenazi Jewish Population	in 280	98%	in 3 95	< 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	in 70	98%	in 3 45	in 966 280
			Ashkenazi Jewish Population	in 07	98%	in 5 30	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	< in 500	99%	in 49 90	< in 0 million
			Ashkenazi Jewish Population	in 57	99%	in 5 60	in 277 028
			Caucasian / European Population	in 7	99%	in 7 00	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	in 462	98%	in 23 05	< in 0 million
			Finnish Population	in	98%	in 5 50	in 2 442 444
POMGNT1	Retinitis pigmentosa 76	AR	General Population	in 462	98%	in 23 05	< in 0 million
			Finnish Population	in	98%	in 5 50	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	in 368	98%	in 8 35	< in 0 million
			Caucasian / European Population	in 488	98%	in 24 35	< in 0 million
			Finnish Population	in 75	98%	in 3 70	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

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Supplemental Table

Gene	Condition	nheritance	Ethnicity	Carrier Rate	Detection Rate	Post test Carrier Probability*	Residual Risk*
RDH12	Leber congenital amaurosis type 3	AR	General Population	< in 500	98%	in 24 95	< in 0 million
			Caucasian / European Population	in 456	98%	in 22 75	< in 0 million
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

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Supplemental Table

Gene	Condition	nheritance	Ethnicity	Carrier Rate	Detection Rate	Post test Carrier Probability*	Residual Risk*
SLC26A2	Diastrophic dysplasia	AR	General Population	in 58	90%	in 57	in 992 872
			Finnish Population	in 50	90%	in 49	in 98 200
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
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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Supplemental Table

Gene	Condition	nheritance	Ethnicity	Carrier Rate	Detection Rate	Post test Carrier Probability*	Residual Risk*
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 a pha tha assem a tra t c s s descr bed n rows marked w th a dagger symbo .
Abbrev at ons: AR, autosoma recess ve; XL, X- nked