Personal Carrier Risk Genes: There may be detailed information on the report regarding additional health considerations when an individual is found to be a carrier of a variant in one of the following genes. All data and example report language are reported by Baylor Genetics®. 1/23/26

Quest Diagnostics®

Gene	Condition Name / Inheritance		Risk Category
ABCA4	ABCA4-related disorders	AR	Attenuated Dz
ABCB4	Progressive familial intrahepatic cholestasis 3	AR	Attenuated Dz
ABCC8	Familial hyperinsulinism, ABCC8-related	AR	Alt Dx and Male
ABCD1	Adrenoleukodystrophy, XL	XL	XL SF
AFF2	Fragile XE syndrome	XL	XL SF
AIPL1	Leber congenital amaurosis 4	AR	Attenuated Dz
AIRE	Autoimmune polyglandular syndrome, type 1	AR	Attenuated Dz
ALG13	Developmental and epileptic encephalopathy 36	XL	XL SF
ALPL	Hypophosphatasia	AR	Attenuated Dz
AQP2	Nephrogenic diabetes insipidus type 2	AR	Attenuated Dz
AR	Androgen insensitivity syndrome	XL	XL SF
ARX	XL developmental disorders, ARX-related	XL	XL SF
ATM	Ataxia-telangiectasia	AR	Onc
ATP7A	Menkes disease	XL	XL SF
ATP8B1	Progressive familial intrahepatic cholestasis 1 and benign recurrent intrahepatic cholestasis 1	AR	Attenuated Dz
ATRX	Alpha-thalassemia intellectual disability syndrome, XL	XL	XL SF
AVPR2	AVPR2-related disorders	XL	XL SF
BCHE	Pseudocholinesterase deficiency	AR	Attenuated Dz
BLM	Bloom syndrome	AR	Onc
BRIP1	Fanconi anemia, complementation group J	AR	Onc
BTK	XL agammaglobulinemia	XL	XL SF
CAPN3	Limb-girdle muscular dystrophy, type 2A	AR	Attenuated Dz
CASQ2	Catecholaminergic polymorphic ventricular tachycardia, type 2	AR	Attenuated Dz
CCDC88C	Congenital hydrocephalus 1	AR	Attenuated Dz
CD40LG	XL hyper IgM syndrome	XL	XL SF
CFTR	Cystic fibrosis	AR	Attenuated Dz
CHM	Choroideremia, XL	XL	XL SF
CHRNE	Congenital myasthenic syndrome, CHRNE-related	AR	Attenuated Dz
CLCN1	Myotonia congenita	AR	Alt Dx
CLCN5	Dent disease 1	XL	XL SF
COL11A2	COL11A2-related disorders	AR	Attenuated Dz
COL17A1	Junctional epidermolysis bullosa, COL17A1-related	AR	Attenuated Dz
COL4A3	Alport syndrome, COL4A3-related	AR	Attenuated Dz
COL4A4	Alport syndrome, COL4A4-related	AR	Attenuated Dz
COL4A5	Alport syndrome, COL4A5-related, XL	XL	XL SF

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COL7A1	Dystrophic epidermolysis bullosa, COL7A1-related	AR	Attenuated Dz
CPT1A	Carnitine palmitoyltransferase I deficiency	AR	PC
CPT2	Carnitine palmitoyltransferase II deficiency	AR	Attenuated Dz
CRB1	CRB1-related retinal dystrophies	AR	Attenuated Dz
CYBB	Chronic granulomatous disease, XL	XL	XL SF
CYP11A1	Congenital adrenal insufficiency, CYP11A1-related	AR	Attenuated Dz
DCX	DCX-related disorders	XL	XL SF
DDX11	Warsaw breakage syndrome	AR	Onc
DKC1	Dyskeratosis congenita, XL	XL	XL SF
DMD	Duchenne/Becker muscular dystrophy, XL	XL	XL SF and Alt Dx
DPYD	Dihydropyrimidine dehydrogenase deficiency	AR	PGx
DUOX2	Thyroid dyshormonogenesis 6	AR	Attenuated Dz
DYNC2H1	Short-rib thoracic dysplasia 3 with or without polydactyly	AR	Digenic
EDA	Hypohidrotic ectodermal dysplasia, XL	XL	XL SF
EMD	Emery-Dreifuss muscular dystrophy, XL	XL	XL SF
EPG5	EPG5-related disorder	AR	Attenuated Dz
ERCC2	ERCC2-related conditions	AR	PC
ERCC6	Cerebrooculofacioskeletal syndrome 1 / Cockayne syndrome, type B	AR	Alt Dx
F11	Factor XI deficiency / Hemophilia C	AR	Attenuated Dz
F2	Factor II deficiency / Prothrombin deficiency	AR	Attenuated Dz
F5	Factor V deficiency	AR	Alt Dx
F8	Factory VIII deficiency / Hemophilia A	XL	XL SF
FMR1	Fragile X syndrome	XL	XL SF and Alt Dx
FOXN1	Severe combined immunodeficiency, FOXN1-related	AR	Attenuated Dz
FRAS1	Fraser syndrome, type 1	AR	Attenuated Dz
FREM2	Fraser syndrome, type 2	AR	Attenuated Dz
G6PD	Glucose-6-phosphate dehydrogenase deficiency	XL	XL SF
GATM	Arginine:glycine amidinotransferase deficiency	AR	Alt Dx
GBA	Gaucher disease	AR	Alt Dx
GCH1	GCH1-related disorders	AR	Attenuated Dz
GDF5	GDF5-related disorders	AR	Attenuated Dz
GHR	Laron syndrome	AR	Attenuated Dz
GJB1	Charcot-Marie-Tooth disease, type 1X	XL	XL SF
GJB2	Nonsyndromic hearing loss and deafness (DFNB) 1	AR	Attenuated Dz
GLA	Fabry disease, XL	XL	Attenuated Dz

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Gene	Condition Name / Inheritance		Risk Category
GNE	GNE myopathy	AR	Attenuated Dz
GP1BA	Bernard-Soulier syndrome, type A	AR	Attenuated Dz
GUCY2D	Leber congenital amaurosis 1	AR	Attenuated Dz
HADH	3-hydroxyacyl-CoA dehydrogenase deficiency	AR	Attenuated Dz
HADHA	Long chain 3-hydroxyacyl-CoA dehydrogenase deficiency	AR	PC
HBA1/HBA2	Alpha-thalassemia	AR	Attenuated Dz
HBB	Beta hemoglobinopathies	AR	Attenuated Dz
HCFC1	Methylmalonic aciduria and homocystinuria, cblX type	XL	XL SF
HPD	Tyrosinemia, type III	AR	Attenuated Dz
HPRT1	HPRT1-related disorders	XL	XL SF
HSD17B10	HSD10 disease	XL	XL SF
IDS	Mucopolysaccharidosis, type II / Hunter syndrome	XL	XL SF
IKBKB	Severe combined immunodeficiency, IKBKB-related	AR	Attenuated Dz
ITGB3	ITGB3-related disorders	AR	Attenuated Dz
KCNJ11	Familial hyperinsulinism, KCNJ11-related	AR	Alt Dx and Male
LAMA3	Junctional epidermolysis bullosa, LAMA3-related	AR	Attenuated Dz
LAMB3	Junctional epidermolysis bullosa, LAMB3-related	AR	Attenuated Dz
LDLR	Familial hypercholesterolemia, LDLR-related	AR	Attenuated Dz
LIG4	LIG4 syndrome	AR	Attenuated Dz
LPL	Lipoprotein lipase deficiency	AR	Attenuated Dz
MEFV	Familial Mediterranean fever	AR	Attenuated Dz
MID1	XL Opitz G/BBB syndrome	XL	XL SF
MPL	Congenital amegakaryocytic thrombocytopenia	AR	Alt Dx
MTM1	XL myotubular myopathy	XL	XL SF
MVK	Mevalonic aciduria / Hyper-IgD syndrome	AR	Attenuated Dz
MYO7A	Usher syndrome, type 1B	AR	Alt Dx
NBN	Nijmegen breakage syndrome	AR	Onc
NPC1	Niemann-Pick disease, type C1	AR	Attenuated Dz
NPC2	Niemann-Pick disease, type C2	AR	Attenuated Dz
NR2E3	Enhanced S-cone syndrome	AR	Alt Dx
OCRL	Lowe syndrome, XL	XL	XL SF
OPA3	3-methylglutaconic aciduria, type III / Costeff syndrome	AR	Alt Dx
OTC	Ornithine transcarbamylase deficiency, XL	XL	XL SF
PDHA1	Pyruvate dehydrogenase E1-alpha deficiency	XL	XL SF
PEX6	Zellweger spectrum disorders, PEX6-related	AR	Variant
PLP1	PLP1-related disorders	XL	XL SF

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Gene	Condition Name / Inheritance		Risk Category
POLG	POLG-related disorders	AR	Alt Dx
POU1F1	Combined or isolated pituitary hormone deficiency, type 1	AR	Attenuated Dz
PRPS1	PRPS1-related disorders	XL	XL SF
RDH12	Leber congenital amaurosis 13	AR	Alt Dx
RLBP1	RLBP1-related retinopathies	AR	Attenuated Dz
RP2	Retinitis pigmentosa 2	XL	XL SF
RPE65	Leber congenital amaurosis 2	AR	Alt Dx
RPGR	Retinitis pigmentosa 3	XL	XL SF
RS1	Juvenile retinoschisis, XL	XL	XL SF
SEC23B	Congenital dyserythropoietic anemia, type II	AR	Attenuated Dz
SERPINA1	Alpha-1 antitrypsin deficiency	AR	Attenuated Dz
SLC12A6	Agenesis of the corpus callosum with peripheral neuropathy	AR	Alt Dx
SLC37A4	Glycogen storage disease, type Ib / IIw	AR	Alt Dx
SLC4A11	Corneal dystrophy and perceptive deafness syndrome	AR	Alt Dx
SLC6A8	Creatine transporter defect, SLC6A8-related, XL / Cerebral creatine deficiency syndrome	XL	XL SF
SPR	Sepiapterin reductase deficiency	AR	Attenuated Dz
TAFAZZIN	Barth syndrome	XL	XL SF
TERT	Dyskeratosis congenita spectrum disorders	AR	Attenuated Dz
TMC1	Nonsyndromic hearing loss and deafness (DFNB) 7	AR	Attenuated Dz
TREX1	TREX1-related disorders	AR	Attenuated Dz
TSHR	Congenital hypothyroidism, TSHR-related	AR	Attenuated Dz
USH1C	Usher syndrome, type 1C	AR	Variant
WAS	Wiskott-Aldrich syndrome, XL	XL	XL SF
WNT10A	Odonto-onycho-dermal dysplasia / Schopf-Schulz-Passarge syndrome	AR	Attenuated Dz
ZIC3	XL heterotaxy-1	XL	XL SF

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