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Gene	Condition Name	Inheritance Pattern	Risk Category	Example Report Language
ABCA4	ABCA4-related disorders	Autosomal recessive	Attenuated disease	Section: What does it mean to be a carrier? **Carriers of ABCA4 pathogenic variants may be at increased risk for age-related macular degeneration, but most carriers have no symptoms.** Clinical correlation and consultation with a healthcare provider is recommended. In both cases, the risk to have a child affected with an ABCA4-related disorder is increased. Testing of reproductive partners is recommended for carriers of an ABCA4-related disorder.
ABCB4	Progressive familial intrahepatic cholestasis 3	Autosomal recessive	Attenuated disease	Section: What does it mean to be a carrier? **Carriers of ABCB4 variants may develop intrahepatic cholestasis of pregnancy, but most carriers have no symptoms outside of pregnancy.** Clinical correlation and consultation with a healthcare provider is recommended. In both cases, the risk to have a child affected with PFIC3 is increased. Testing of reproductive partners is recommended for carriers of PFIC3.
ABCC8	Familial hyperinsulinism, ABCC8-related	Autosomal recessive	Other - alternate condition and male carrier risks	Section: What does it mean to be a carrier? **Carriers of familial hyperinsulinism, ABCC8-related may have neonatal diabetes mellitus, but most carriers have no symptoms.** Clinical correlation and consultation with a healthcare provider is recommended. In both cases, the risk to have a child affected with familial hyperinsulinism, ABCC8-related is increased. Testing of reproductive partners is recommended for carriers of familial hyperinsulinism, ABCC8-related.
ABCD1	Adrenoleukodystrophy, X-linked	X-linked	XL - symptomatic female	Section: What is... Adrenoleukodystrophy (ALD) is an inherited disorder affecting mostly males and involves the nervous system and adrenal glands. ALD is caused by pathogenic variants in the ABCD1 gene. In the most common type of ALD, the childhood cerebral form, symptoms usually appear in early childhood and rapidly worsen in the first two years of life. Males affected with ALD have progressive behavioral and learning problems; vision, hearing, and coordination can also be impaired. In the second most common type of ALD, called adrenomyeloneuropathy (AMN), symptoms usually appear in early adulthood. Symptoms are less severe, such as stiffness of the legs and bladder and bowel dysfunction, and progression is less rapid. Individuals affected with the third and rarest form of ALD usually only have issues with their adrenal glands and insufficient hormone production; some neurologic symptoms may appear later in life. Rarely, an individual may be asymptomatic. Different types of ALD may be seen even in family members. Life expectancy for ALD is variable depending on the severity of the condition. There is no cure for ALD; however, aggressive medical care and surveillance may help to improve some symptoms and overall quality of life. **Some females may exhibit symptoms of ALD, although onset of the condition is typically later than in males and does not progress as quickly; the symptoms most closely resemble the AMN type.**
AFF2	Fragile XE syndrome	X-linked	XL - symptomatic female	Section: What does it mean to be a carrier? A female who carries a full expansion (>200 CCG repeats), a premutation expansion (50-200 CCG repeats), or a different pathogenic variant in the AFF2 gene has a 50% chance of passing on the respective variant with each pregnancy. **Additionally, females with a full expansion may exhibit some features of fragile XE syndrome.**
AIPL1	Leber congenital amaurosis 4	Autosomal recessive	Attenuated disease	Section: What does it mean to be a carrier? **Carriers of AIPL1 variants may be at increased risk for retinal disorders, but most carriers have no symptoms.** For all carriers, the risk to have a child affected with LCA4 is increased. Testing of reproductive partners is recommended for carriers of LCA4.

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AIRE	Autoimmune polyglandular syndrome, type 1	Autosomal recessive	Attenuated disease	Section: What is... Autoimmune polyglandular syndrome, type 1 (APS1) is an inherited disorder that causes the immune system to attack the affected individual's own body. **APS1 is generally caused by biallelic pathogenic variants in the AIRE gene; more rarely, APS1 is caused by only one pathogenic variant.** Symptoms usually present in childhood starting with recurrent yeast infections in the skin and mucous membranes. Hypoparathyroidism and adrenal gland insufficiency often present later. Other symptoms may include liver disease, extreme fatigue, hair loss, and digestive and vision issues. There is no cure for APS1, however medical surveillance and care may help to improve some symptoms and overall condition of life.
ALG13	Developmental and epileptic encephalopathy 36	X-linked	XL - symptomatic female	Section: What does it mean to be a carrier? **Carriers of DEE36 may have a milder form of the disorder, but many carriers have no symptoms.** Clinical correlation and consultation with a healthcare provider is recommended. In both cases, the risk of having a child affected with DEE36 is increased.
ALPL	Hypophosphatasia	Autosomal recessive	Attenuated disease	Section: What does it mean to be a carrier? **Carriers of HPP may have a milder form of the disorder, but many carriers have no symptoms.** In both cases, the risk to have a child affected with HPP is increased. Testing of reproductive partners is recommended for carriers of HPP.
AQP2	Nephrogenic diabetes insipidus type 2	Autosomal recessive	Attenuated disease	Section: What does it mean to be a carrier? **Carriers of NDI type 2 may have a milder form of the disorder, but many carriers have no symptoms.** In both cases, the risk to have a child affected with NDI type 2 is increased. Testing of reproductive partners is recommended for carriers of NDI type 2.
AR	Androgen insensitivity syndrome	X-linked	XL - symptomatic female	Section: What does it mean to be a carrier? **Carriers of AIS may have a milder form of the disorder, but many carriers have no symptoms. It is estimated that 10% of carrier females with exhibit symptoms.** Clinical correlation and consultation with a healthcare provider is recommended. In both cases, the risk of having a child affected with AIS is increased.
ARX	X-linked developmental disorders, ARX-related	X-linked	XL - symptomatic female	Section: What does it mean to be a carrier? **Female carriers may have variable but milder symptoms such as intellectual disability, seizures, or agenesis of the corpus callosum, or may have no symptoms.** In both cases, the risk to have a child affected with an ARX-related disorder is increased.
ATM	Ataxia-telangiectasia	Autosomal recessive	Cancer Risk	Section: What does it mean to be a carrier? **Carriers of ataxia-telangiectasia do not have symptoms of the condition; however, they are at increased risk of developing certain types of cancer.** Clinical correlation and consultation with a healthcare provider is recommended. Additionally, the risk to have a child affected with ataxia-telangiectasia is increased. Testing of reproductive partners is recommended for carriers of ataxia-telangiectasia.
ATP7A	Menkes disease	X-linked	XL - symptomatic female	Section: What does it mean to be a carrier? **Some female carriers have pili torti, but typically do not exhibit other symptoms of Menkes disease.** However, the risk to have a child affected with Menkes disease is increased.

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Gene	Condition Name	Inheritance Pattern	Risk Category	Example Report Language
ATP8B1	Progressive familial intrahepatic cholestasis 1 and benign recurrent intrahepatic cholestasis 1	Autosomal recessive	Attenuated disease	Section: What does it mean to be a carrier? **Carriers may be at increased risk for intrahepatic cholestasis in pregnancy, which can cause fetal distress and prematurity; most individuals have no signs or symptoms associated with being a carrier for PFIC1 and BRIC1.** In both cases, the risk to have a child affected with PFIC1 and BRIC1 is increased. Testing of reproductive partners is recommended for carriers of PFIC1 and BRIC1.
ATRX	Alpha-thalassemia intellectual disability syndrome, X-linked	X-linked	XL - symptomatic female	Section: What does it mean to be a carrier? **Carriers of ATR-X may have a milder form of the disorder, but many carriers have no symptoms.** In both cases, the risk to have a child affected with ATR-X is increased.
AVPR2	AVPR2-related disorders	X-linked	XL - symptomatic female	Section: What does it mean to be a carrier? **Carriers of AVPR2 pathogenic variants may have a milder form of the disorder, but many carriers have no symptoms.** Clinical correlation and consultation with a healthcare provider is recommended. In both cases, the risk of having a child affected with AVPR2-related disorders is increased.
BCHE	Pseudocholinesterase deficiency	Autosomal recessive	Attenuated disease	Section: What does it mean to be a carrier? **There are generally no signs or symptoms associated with being a carrier for pseudocholinesterase deficiency. In rare cases, carriers of the disease may take a longer time to clear choline esterase from the body, which may result in mild symptoms of the disease.** The risk to have a child affected with the disease is increased. Testing of reproductive partners is recommended for carriers of pseudocholinesterase deficiency.
BLM	Bloom syndrome	Autosomal recessive	Cancer Risk	Section: What does it mean to be a carrier? There are generally no signs or symptoms associated with being a carrier of Bloom syndrome. Testing of reproductive partners, with consideration to their ethnicity, is recommended for carriers of Bloom syndrome. **Monoallelic germline alterations in BLM have been detected in individuals with breast cancer; however, current evidence is insufficient to determine if there is an increased risk of cancer over that of the general population.**
BRIP1	Fanconi anemia, complementation group J	Autosomal recessive	Cancer Risk	Section: What does it mean to be a carrier? **Carriers of Fanconi anemia, complementation group J have an increased risk to develop cancers associated with BRIP1**. Clinical correlation and consultation with a healthcare provider is recommended. The risk to have a child affected with Fanconi anemia, complementation group J or cancers associated with BRIP1 is increased. Testing of reproductive partners is recommended for carriers of an BRIP1 pathogenic variants.
BTK	X-linked agammaglobulinemia	X-linked	XL - symptomatic female	Section: What does it mean to be a carrier? **One female carrier of XLA has been reported to have symptoms of the disorder, but the majority of carriers have no symptoms.** In both cases, the risk to have a child affected with XLA is increased.
CAPN3	Limb-girdle muscular dystrophy, type 2A	Autosomal recessive	Attenuated disease	Section: What does it mean to be a carrier? **Carriers of CAPN3 may develop autosomal dominant limb-girdle muscular dystrophy 4, but most carriers have no symptoms.** Clinical correlation and consultation with a healthcare provider is recommended. In both cases, the risk to have a child affected with LGMD2A is increased. Testing of reproductive partners is recommended for carriers of LGMD2A.

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Gene	Condition Name	Inheritance Pattern	Risk Category	Example Report Language
CASQ2	Catecholaminergic polymorphic ventricular tachycardia, type 2	Autosomal recessive	Attenuated disease	Section: What does it mean to be a carrier? **Carriers of CPVT, type 2 may show mild symptoms of CPVT, but many carriers have no symptoms.** Clinical correlation and consultation with a healthcare provider is recommended. In both cases, the risk of having a child affected with CPVT, type 2 is increased.
CCDC88C	Congenital hydrocephalus 1	Autosomal recessive	Attenuated disease	Section: What does it mean to be a carrier? **Carriers of CCDC88C may develop SCA40, but most carriers have no symptoms.** Clinical correlation and consultation with a healthcare provider is recommended. In both cases, the risk to have a child affected with HYC1 is increased. Testing of reproductive partners is recommended for carriers of HYC1.
CD40LG	X-Linked hyper IgM syndrome	X-linked	XL - symptomatic female	Section: What does it mean to be a carrier? **Carriers of HIGM1 may have a milder form of the disorder, but many carriers have no symptoms.** In both cases, the risk to have a child affected with HIGM1 is increased.
CFTR	Cystic fibrosis	Autosomal recessive	Attenuated disease	Section: What is... Cystic fibrosis (CF) is an inherited disorder characterized by progressive respiratory damage due to a thick mucus buildup in the lungs and chronic digestive system problems. CF is caused by biallelic pathogenic variants in the CFTR gene and primarily affects the lungs and pancreas. Affected individuals may experience chronic lung infections with difficulty breathing, pancreatitis, poor growth, weight loss, greasy stools, and difficulty with bowel movements. Males with CF are typically infertile due to congenital absence of the vas deferens. There are over 1,700 variants in the CFTR gene and severity of CF symptoms is dependent on the pathogenic variants of affected individuals. Many individuals with CF now live well into adulthood secondary to improved treatments and symptom management. **Individuals who have only one pathogenic variant can be at increased risk to develop pancreatitis and other CF-related symptoms.**
CHM	Choroideremia, X-linked	X-linked	XL - symptomatic female	Section: What does it mean to be a carrier? **Carriers of choroideremia may develop night blindness or visual field loss, but many carriers have no symptoms.** In both cases, the risk to have a child affected with choroideremia is increased.
CHRNE	Congenital myasthenic syndrome, CHRNE-related	Autosomal recessive	Attenuated disease	Section: What is... Congenital myasthenic syndrome (CMS) is a group of inherited disorders characterized by skeletal muscle weakness and fatigue, including facial muscles and muscles controlling swallowing and sucking. **Pathogenic variants in the CHRNE gene are the most common cause of CMS cases. The majority are due to biallelic pathogenic variants; more rarely, CHRNE-related CMS is caused by only one pathogenic variant.** Onset and severity of the disorder are variable, but generally, the earlier the symptoms appear, the more pronounced the disease is likely to be. Affected infants may be delayed in learning to crawl or walk and later on display difficulty running or climbing stairs. Individuals may have joint deformities and breathing problems. There is no cure for CMS; however, there are medications available for some types of CMS that help to improve muscle strength and endurance.
CLCN1	Myotonia congenita	Autosomal recessive	Other - alternate condition	Section: What does it mean to be a carrier? **Carriers of CLCN1 may develop Thomsen disaese, but most carriers have no symptoms.** Clinical correlation and consultation with a healthcare provider is recommended. In both cases, the risk to have a child affected with myotonia congenita is increased. Testing of reproductive partners is recommended for carriers of myotonia congenita.

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Gene	Condition Name	Inheritance Pattern	Risk Category	Example Report Language
CLCN5	Dent disease 1	X-linked	XL - symptomatic female	Section: What does it mean to be a carrier? **Carriers of Dent disease 1 may have kidney stones and protein in the urine, but many carriers have no symptoms.** Clinical correlation and consultation with a healthcare provider are recommended. In both cases, the risk to have a child affected with Dent disease is increased.
COL11A2	COL11A2-related disorders	Autosomal recessive	Attenuated disease	Section: What does it mean to be a carrier? **Carriers of COL11A2-related disorders may develop Deafness, autosomal dominant 13, Fibrochondrogenesis 2, or Otospondylomegaepiphyseal dysplasia.** Clinical correlation and consultation with a healthcare provider is recommended. In both cases, the risk to have a child affected with other COL11A2-related disorders is increased. Testing of reproductive partners is recommended for carriers of COL11A2-related disorders.
COL17A1	Junctional epidermolysis bullosa, COL17A1-related	Autosomal recessive	Attenuated disease	Section: What does it mean to be a carrier? **Carriers of JEB may have thinner tooth enamel, but many carriers have no symptoms.** Clinical correlation and consultation with a healthcare provider is recommended. In both cases, the risk to have a child affected with JEB is increased. Testing of reproductive partners is recommended for carriers of JEB.
COL4A3	Alport syndrome, COL4A3-related	Autosomal recessive	Attenuated disease	Section: What does it mean to be a carrier? **About 50% of carriers of autosomal recessive Alport syndrome will have intermittent or permanent blood present in the urine that is only detectable under a microscope (microhematuria).** The risk to have a child affected with autosomal recessive Alport syndrome is increased. Testing of reproductive partners is recommended for carriers of COL4A3 pathogenic variants.
COL4A4	Alport syndrome, COL4A4-related	Autosomal recessive	Attenuated disease	Section: What does it mean to be a carrier? **About 50% of carriers of autosomal recessive Alport syndrome will have intermittent or permanent blood present in the urine that is only detectable under a microscope (microhematuria).** The risk to have a child affected with autosomal recessive Alport syndrome is increased. Testing of reproductive partners is recommended for carriers of COL4A4 pathogenic variants.
COL4A5	Alport syndrome, COL4A5-related, X-linked	X-linked	XL - symptomatic female	Section: What does it mean to be a carrier? **Most female carriers of X-linked Alport syndrome will have intermittent or permanent blood present in the urine that is only detectable under a microscope (microhematuria) and are at risk to develop kidney problems later in life.** Clinical correlation is recommended.
COL7A1	Dystrophic epidermolysis bullosa, COL7A1-related	Autosomal recessive	Attenuated disease	Section: What does it mean to be a carrier? **Carriers of pathogenic COL7A1 variants may have DDEB and experience mild blistering, but many carriers have no symptoms.** Clinical correlation and consultation with a healthcare provider is recommended. In both cases, the risk to have a child affected with RDEB is increased. Testing of reproductive partners is recommended for carriers of RDEB.
CPT1A	Carnitine palmitoyltransferase I deficiency	Autosomal recessive	Other - pregnancy complications	Section: What does it mean to be a carrier? There are generally no signs or symptoms associated with being a carrier for CPT IA deficiency. **However, female carriers should be monitored for acute fatty liver during pregnancy.** Carriers have an increased risk to have children affected with CPT IA deficiency. Testing of reproductive partners is recommended for carriers of CPT IA deficiency.

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Gene	Condition Name	Inheritance Pattern	Risk Category	Example Report Language
CPT2	Carnitine palmitoyltransferase II deficiency	Autosomal recessive	Attenuated disease	Section: What does it mean to be a carrier? **Carriers of CPT II deficiency often have reduced carnitine palmitoyltransferase II enzyme levels when measured by biochemical analysis, however rarely do they exhibit symptoms of the disease.** Carriers do have an increased risk to have children affected with CPT II deficiency. Testing of reproductive partners is recommended for carriers of CPT II deficiency.
CRB1	CRB1-related retinal dystrophies	Autosomal recessive	Attenuated disease	Section: What does it mean to be a carrier? **Carriers of CRB1-related retinal dystrophies may have physical changes to the fundus of the eye, but this often does not cause any symptoms.** Clinical correlation and consultation with a healthcare provider is recommended. However, the risk to have a child affected with a CRB1-related retinal dystrophy is increased. Testing of reproductive partners is recommended for carriers of CRB1-related retinal dystrophies.
CYBB	Chronic granulomatous disease, X-linked	X-linked	XL - symptomatic female	Section: What does it mean to be a carrier? **Carriers of CGDX may be at increased risk for inflammatory disorders, but many carriers have no symptoms.** Clinical correlation and consultation with a healthcare provider are recommended. In both cases, the risk to have a child affected with CGDX is increased.
CYP11A1	Congenital adrenal insufficiency, CYP11A1-related	Autosomal recessive	Attenuated disease	Section: What does it mean to be a carrier? **Carriers of CYP11A1 may develop symptoms associated with the disorder, but most carriers have no symptoms.** Clinical correlation and consultation with a healthcare provider is recommended. In both cases, the risk to have a child affected with congenital adrenal insufficiency, CYP11A1-related is increased. Testing of reproductive partners is recommended for carriers of congenital adrenal insufficiency, CYP11A1-related.
DCX	DCX-related disorders	X-linked	XL - symptomatic female	Section: What does it mean to be a carrier? **50% of carriers of DCX-related disorders exhibit variable symptoms of SBH, while the other 50% are asymptomatic.** Clinical correlation and consultation with a healthcare provider are recommended. In both cases, the risk to have a child affected with DCX-related disorders is increased.
DDX11	Warsaw breakage syndrome	Autosomal recessive	Cancer Risk	Section: What does it mean to be a carrier? There are generally no signs or symptoms associated with being a carrier of Warsaw breakage syndrome. **Carriers may have an elevated risk of developing certain cancers, however, the data on this is limited and unclear.** For all carriers, the risk of having a child affected with Warsaw breakage syndrome is increased. Testing of reproductive partners is recommended for carriers of Warsaw breakage syndrome.
DKC1	Dyskeratosis congenita, X-linked	X-linked	XL - symptomatic female	Section: What does it mean to be a carrier? **Carriers of X-linked DC may have nail and skin issues and be at increased risk for bone marrow failure, but many carriers have no symptoms.** Clinical correlation and consultation with a healthcare provider are recommended. In both cases, the risk to have a child affected with X-linked DC is increased.
DMD	Duchenne/Becker muscular dystrophy, X-linked	X-linked	XL - symptomatic female and Other - alternate condition	Section: What does it mean to be a carrier? **Carrier females may be at an increased risk for cardiomyopathy or other heart problems in adulthood and may exhibit varying symptoms such as muscle weakness and cramping.** Genetic counseling and cardiology evaluation are recommended.

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DPYD	Dihydropyrimidine dehydrogenase deficiency	Autosomal recessive	Other - pharmacogenomic risk	Section: What does it mean to be a carrier? **Carriers of DPD deficiency have no symptoms of the condition but may be at risk for fluoropyrimidine toxicity.** Clinical correlation and consultation with a healthcare provider is recommended. In both cases, the risk to have a child affected with DPD deficiency is increased. Testing of reproductive partners is recommended for carriers of DPD deficiency.
DUOX2	Thyroid dyshormonogenesis 6	Autosomal recessive	Attenuated disease	Section: What is... Thyroid dyshormonogenesis is a type of congenital hypothyroidism that affects the production of thyroid hormones. Thyroid dyshormonogenesis 6 is caused by biallelic pathogenic variants in DUOX2. Affected infants are typically born without symptoms but present in the first few weeks of life with failure to thrive, constipation, and lethargy. If untreated, these individuals experience goiters, intellectual disability, developmental delays, and poor growth. Most infants develop normally after treatment is initiated. **Some carriers of pathogenic DUOX2 variants have been reported to have mild transient congenital hypothyroidism that resolves, often spontaneously, in the first few months of life.**
DYNC2H1	Short-rib thoracic dysplasia 3 with or without polydactyly	Autosomal recessive	Other - potential digenic inheritance	Section: How is...inherited SRTD3 is inherited in an autosomal recessive manner. Both parents must be carriers of a pathogenic variant in the gene in order to be at risk to have an affected child. This type of inheritance requires the presence of two copies of a pathogenic variant in the gene, one inherited from each carrier parent, for the child to have the genetic disease. There is a 1 in 4 chance that a child will inherit two pathogenic variants, one in each copy of the gene, and be affected when both parents are carriers. **There is some evidence that this condition can be caused by digenic biallelic mutation in the DYNC2H1 and NEK1 genes.**
EDA	Hypohidrotic ectodermal dysplasia, X-linked	X-linked	XL - symptomatic female	Section: What does it mean to be a carrier? **There are generally no signs or symptoms associated with being a carrier for hypohidrotic ectodermal dysplasia, though female carriers may occasionally exhibit mild symptoms of ectodermal dysplasia.** Genetic counseling and clinical correlation are recommended.
EMD	Emery-Dreifuss muscular dystrophy, X-linked	X-linked	XL - symptomatic female	Section: What does it mean to be a carrier? **Generally, there are no signs or symptoms associated with being a carrier for Emery-Dreifuss muscular dystrophy, however, female carriers may be mildly affected with milder progressive muscle weakness or heart problems.** The risk to have a child affected with Emery-Dreifuss muscular dystrophy is increased. Genetic counseling and clinical correlation are recommended.
EPG5	EPG5-related disorder	Autosomal recessive	Attenuated disease	Section: What does it mean to be a carrier? There are generally no signs or symptoms associated with being a carrier for EPG5-related disorder. **Some carriers may develop subtle signs of the syndrome, such as early-onset cataracts, vitiligo, and higher frequency of certain tumor disorders, however, the data on this is limited and unclear.** In general, the risk to have a child affected with EPG5-related disorder is increased. Testing of reproductive partners is recommended for carriers of EPG5-related disorder.

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ERCC2	ERCC2-related conditions	Autosomal recessive	Other - pregnancy complications	Section: What does it mean to be a carrier? There are typically no signs or symptoms associated with being a carrier for an ERCC2-related pathogenic variant. However, the risk to have a child affected with an ERCC2-related disorder is increased. The testing of reproductive partners is recommended for carriers of an ERCC2 pathogenic variant. **Of note, pregnant women carrying a fetus affected with trichothiodystrophy 1, photosensitive type may experience problems during pregnancy including pregnancy-induced high blood pressure and a related condition called HELLP syndrome that can damage the liver.**
ERCC6	Cerebrooculofacioskeletal syndrome 1 / Cockayne syndrome, type B	Autosomal recessive	Other - alternate condition	Section: What does it mean to be a carrier? **Some carriers of ERCC6-related CS have evidence of premature ovarian failure, but most carriers have no symptoms.** Clinical correlation and consultation with a healthcare provider are recommended. In both cases, the risk to have a child affected with ERCC6-related CS is increased. Testing of reproductive partners is recommended for carriers of ERCC6-related CS.
F11	Factor XI deficiency / Hemophilia C	Autosomal recessive	Attenuated disease	Section: What does it mean to be a carrier? There are generally no signs or symptoms associated with being a carrier for factor XI deficiency. **However, the risk to have a child affected with factor XI deficiency is increased as high as 50% if it is autosomal dominant in the family.** Testing of reproductive partners is recommended for carriers of factor XI deficiency.
F2	Factor II deficiency / Prothrombin deficiency	Autosomal recessive	Attenuated disease	Section: What does it mean to be a carrier? There are generally no signs or symptoms associated with being a carrier for factor II deficiency. However, the risk to have a child affected with factor II deficiency is increased. Testing of reproductive partners is recommended for carriers of factor II deficiency. **Individuals with prothrombin thrombophilia are at increased risk to develop blood clots and to have children with prothrombin thrombophilia.**
F5	Factor V deficiency	Autosomal recessive	Other - alternate condition	Section: What does it mean to be a carrier? There are generally no signs or symptoms associated with being a carrier for factor V deficiency. However, the risk to have a child affected with factor V deficiency is increased. Testing of reproductive partners is recommended for carriers of factor V deficiency. **Individuals with factor V Leiden thrombophilia are at increased risk to develop blood clots and to have children with factor V Leiden thrombophilia.**
F8	Factory VIII deficiency / Hemophilia A	X-linked	XL - symptomatic female	Section: What does it mean to be a carrier? **Carriers of hemophilia A may have a milder form of the disorder, but many carriers have no symptoms.** Clinical correlation and consultation with a healthcare provider is recommended, particularly during pregnancy. In both cases, the risk to have a child affected with hemophilia A is increased.
F9	Factor IX deficiency / Hemophilia B	X-linked	XL - symptomatic female	Section: What does it mean to be a carrier? **Females who are carriers may also be affected but show milder symptoms, and may need specific treatment during pregnancy or postpartum care.** Testing is recommended for women who are at risk for being carriers of factor IX deficiency. Clinical correlation and consultation with a healthcare provider are recommended.

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FH	Fumarase deficiency	Autosomal recessive	Cancer Risk	Section: What does it mean to be a carrier? **Carriers of FH deficiency are at increased risk for fibroids, particularly of the uterus, and kidney tumors.** Clinical correlation and consultation with a healthcare provider are recommended. In both cases, the risk to have a child affected with FH deficiency is increased. Testing of reproductive partners is recommended for carriers of FH deficiency.
FHL1	FHL1-related disorders	X-linked	XL - symptomatic female	Section: What does it mean to be a carrier? **Carriers of FHL1-related disorders may be at risk for muscle weakness and cardiomyopathy, but many carriers do not have symptoms.** Clinical correlation and consultation with a healthcare provider is recommended. In both cases, the risk to have a child affected with FHL1-related disorders is increased.
FMR1	Fragile X syndrome	X-linked	XL - symptomatic female and Other - alternate condition	Section: What does it mean to be a carrier? A female who carries a full expansion (>200 CGG repeats), a premutation expansion (55-200 CGG repeats), or a different pathogenic variant in the FMR1 gene has a 50% chance of passing on the respective variant with each pregnancy. **Additionally, females with a full expansion may exhibit some features of FXS. Female premutation carriers (55-200 CGG repeats) are generally unaffected but are at an increased risk for disorders called Fragile X-associated primary ovarian insufficiency (FXPOI) and Fragile X-associated tremor/ataxia syndrome (FXTAS). Males who carry the premutation variant are also at increased risk for FXTAS. Female carriers with an intermediate size (45-54 CGG repeats) are not at risk for FXPOI or FXTAS but have a small risk of repeat expansion with each pregnancy.**
FOXN1	Severe combined immunodeficiency, FOXN1-related	Autosomal recessive	Attenuated disease	Section: What does it mean to be a carrier? **Carriers of FOXN1 pathogenic variants may develop T-cell lymphopenia, infantile, with or without nail dystrophy, autosomal dominant.** Clinical correlation and consultation with a healthcare provider is recommended. In both cases, the risk to have a child affected with FOXN1-related disorders is increased. Testing of reproductive partners is recommended for carriers of FOXN1-related disorders.
FRAS1	Fraser syndrome, type 1	Autosomal recessive	Attenuated disease	Section: What does it mean to be a carrier? There are generally no signs or symptoms associated with being a carrier for FRASRS1. **Some carriers may have an increased risk of cataracts.** The risk to have a child affected with FRASRS1 is increased. Testing of reproductive partners is recommended for carriers of FRASRS1.
FREM2	Fraser syndrome, type 2	Autosomal recessive	Attenuated disease	Section: What does it mean to be a carrier? There are generally no signs or symptoms associated with being a carrier for FRASRS2. **Some carriers may have an increased risk of cataracts.** The risk to have a child affected with FRASRS2 is increased. Testing of reproductive partners is recommended for carriers of FRASRS2.
G6PD	Glucose-6-phosphate dehydrogenase deficiency	X-linked	XL - symptomatic female	Section: What does it mean to be a carrier? **Carriers of G6PD deficiency may have a milder form of the disorder, but many carriers have no symptoms.** Clinical correlation and consultation with a healthcare provider is recommended. In both cases, the risk to have a child affected with G6PD deficiency is increased.

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GATM	Arginine:glycine amidinotransferase deficiency	Autosomal recessive	Other - alternate condition	Section: What is... Arginine:glycine amidinotransferase (AGAT) deficiency, also known as cerebral creatine deficiency syndrome 3, is an inherited metabolic condition characterized by a decrease in creatine, which is an energy source for many organs including the brain. It is caused by biallelic pathogenic variants in the GATM gene. Onset of symptoms occurs in infancy. Affected individuals typically have mild to moderate intellectual disability as well as delays in speech and motor skills development. Children with AGAT deficiency may also have failure to thrive due to inability to gain weight or grow at the expected rate. Treatment typically involves oral administration of creatine. **Individuals with one pathogenic variant in the GATM gene may develop a condition called Fanconi renotubular syndrome 1 (FRTS1). This is characterized by excessive thirst (polydipsia), short stature, skeletal system abnormalities, muscle weakness, and kidney issues. Onset of symptoms typically occurs in the first decade of life and can be highly variable. Some individuals with FRTS1 may need kidney transplantation.**
GBA	Gaucher disease	Autosomal recessive	Other - alternate condition	Section: What does it mean to be a carrier? **Carriers of Gaucher disease do not have symptoms of the condition; however, the risk of developing Parkinson's disease is increased over the general population.** Additionally, the risk to have a child affected with the disease is increased. Testing of reproductive partners is recommended for carriers of Gaucher disease.
GCH1	GCH1-related disorders	Autosomal recessive	Attenuated disease	Section: What does it mean to be a carrier? **Carriers of GCH1-related disorders may develop DOPA-responsive dystonia.** For all carriers, the risk to have a child affected with GCH1-related disorders is increased. Testing of reproductive partners is recommended for carriers of GCH1-related disorders.
GDF5	GDF5-related disorders	Autosomal recessive	Attenuated disease	Section: What does it mean to be a carrier? **Carriers of GDF5-related disorders may develop bone abnormalities, but many carriers do not have symptoms.** Clinical correlation and consultation with a healthcare provider is recommended. In both cases, the risk to have a child affected with GDF5-related disorders is increased.
GHR	Laron syndrome	Autosomal recessive	Attenuated disease	Section: What does it mean to be a carrier? **Carriers of GHR variants may develop partial growth hormone insensitivity and increased responsiveness to growth hormone, but many carriers remain unaffected.** In both cases, the risk to have a child affected with Laron syndrome is increased. Testing of reproductive partners is recommended for carriers of Laron syndrome.
GJB1	Charcot-Marie-Tooth disease, type 1X	X-linked	XL - symptomatic female	Section: What does it mean to be a carrier? **Carriers of CMT1X may have a milder form of the disorder, but some carriers have no symptoms.** Clinical correlation and consultation with a healthcare provider is recommended. In both cases, the risk to have a child affected with CMT1X is increased.
GJB2	Nonsyndromic hearing loss and deafness (DFNB) 1	Autosomal recessive	Attenuated disease	Section: What does it mean to be a carrier? **There are generally no signs or symptoms associated with being a carrier for DFNB1, but clinical correlation and consultation with a healthcare provider are recommended due to the potential for autosomal dominant inheritance**. The risk to have a child affected with DFNB1 is increased. Testing of reproductive partners is recommended.

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Gene	Condition Name	Inheritance Pattern	Risk Category	Example Report Language
GLA	Fabry disease, X-linked	X-linked	Attenuated disease	Section: What does it mean to be a carrier? **Carriers of Fabry disease may develop symptoms such as pain, kidney issues, and neurological abnormalities, but some carriers have no symptoms.** Clinical correlation and consultation with a healthcare provider is recommended. In both cases, the risk to have a child affected with Fabry disease is increased.
GNE	GNE myopathy	Autosomal recessive	Attenuated disease	Section: What does it mean to be a carrier? **Carriers of GNE myopathy may have symptoms of sialuria, but most carriers have no symptoms.** Clinical correlation and consultation with a healthcare provider is recommended. In both cases, the risk to have a child affected with GNE myopathy is increased. Testing of reproductive partners is recommended for carriers of GNE myopathy.
GP1BA	Bernard-Soulier syndrome, type A	Autosomal recessive	Attenuated disease	Section: What does it mean to be a carrier? **Rarely, carriers of BSS type A may have symptoms of the disease, but many carriers have no symptoms.** Clinical correlation and consultation with a healthcare provider are recommended. In both cases, the risk to have a child affected with BSS is increased. Testing of reproductive partners is recommended for carriers of BSS.
GUCY2D	Leber congenital amaurosis 1	Autosomal recessive	Attenuated disease	Section: What does it mean to be a carrier? **Carriers of GUCY2D variants may develop a similar retinal disorder called cone-rod dystrophy 6, but most carriers have no symptoms.** For all carriers, the risk to have a child affected with LCA1 is increased. Testing of reproductive partners is recommended for carriers of LCA1.
HADH	3-hydroxyacyl-CoA dehydrogenase deficiency	Autosomal recessive	Attenuated disease	Section: What does it mean to be a carrier? **Carriers of HADH may develop familial hyperinsulinemic hypoglycemia-4, but most carriers have no symptoms.** Clinical correlation and consultation with a healthcare provider is recommended. In both cases, the risk to have a child affected with HADH-related disorders is increased. Testing of reproductive partners is recommended for carriers of HADH-related disorders.
HADHA	ain 3-hydroxyacyl-CoA dehydrogenase d	Autosomal recessive	Other - pregnancy complications	Section: What does it mean to be a carrier? There are typically no signs or symptoms associated with being a carrier for LCHAD deficiency or TFP deficiency. However, the risk to have a child affected with these conditions is increased. The testing of reproductive partners is recommended for carriers of LCHAD deficiency or TFP deficiency. **Pregnancy complications such as HELLP (hemolysis, elevated liver enzymes, and low platelet count) syndrome and acute fatty liver of pregnancy are seen in about 15%-25% of pregnancies in women carrying a fetus affected with LCHAD/TFP deficiency.**
HBA1/HBA2	Alpha-thalassemia	Autosomal recessive	Attenuated disease	Section: What does it mean to be a carrier? **Silent carriers of alpha thalassemia generally do not have any disease symptoms but may have smaller than normal red blood cells. Carriers of alpha thalassemia may have mild anemia or no medical issues but are at increased risk to have children with alpha thalassemia.** Testing of reproductive partners is recommended for carriers of alpha-thalassemia.

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Gene	Condition Name	Inheritance Pattern	Risk Category	Example Report Language
HBB	Beta hemoglobinopathies	Autosomal recessive	Attenuated disease	Section: What does it mean to be a carrier? Individuals who are carriers of sickle cell disease or Hemoglobin SC disease generally do not have symptoms of the condition. **Individuals who are carriers of beta-thalassemia may be referred to as having beta-thalassemia minor and may have mild anemia.** Carriers of sickle cell disease, Hemoglobin SC disease, beta-thalassemia or other beta hemoglobinopathies are at higher risk of having children affected with each condition. Testing of reproductive partners is recommended for carriers of beta hemoglobinopathies.
HCFC1	Methylmalonic aciduria and homocystinuria, cblX type	X-linked	XL - symptomatic female	Section: What does it mean to be a carrier? **Carriers of MAHCX may have mild symptoms of the disorder, but most carriers do not have symptoms.** Clinical correlation and consultation with a healthcare provider is recommended. In both cases, the risk to have a child affected with MAHCX is increased.
HPD	Tyrosinemia, type III	Autosomal recessive	Attenuated disease	Section: What does it mean to be a carrier? **Some carriers of tyrosinemia, type III have hawkinsinuria, but most carriers have no symptoms.** In both cases, the risk to have a child affected with tyrosinemia, type III is increased. Testing of reproductive partners is recommended for carriers of tyrosinemia, type III.
HPRT1	HPRT1-related disorders	X-linked	XL - symptomatic female	Section: What does it mean to be a carrier? **Carriers of HPRT1-related disorders may have biochemical features, but most carriers have no symptoms.** Clinical correlation and consultation with a healthcare provider is recommended. In both cases, the risk to have a child affected with HPRT1-related disorders is increased.
HSD17B10	HSD10 disease	X-linked	XL - symptomatic female	Section: What does it mean to be a carrier? **Carriers of HSD10 disease may developmental and intellectual differences, seizures, or movement issues, but many carriers have no symptoms.** Clinical correlation and consultation with a healthcare provider is recommended. In both cases, the risk to have a child affected with HSD10 disease is increased.
IDS	Mucopolysaccharidosis, type II / Hunter syndrome	X-linked	XL - symptomatic female	Section: What does it mean to be a carrier? **Carriers of MPS II may have a milder form of the disorder or suggestive findings such as short stature, joint contractures, or mild characteristic facies of the condition; however, many carriers have no symptoms.** Clinical correlation and consultation with a healthcare provider are recommended. In both cases, the risk to have a child affected with MPS II is increased.
IKBKB	Severe combined immunodeficiency, IKBKB-related	Autosomal recessive	Attenuated disease	Section: What does it mean to be a carrier? **Carriers of IKBKB pathogenic variants may develop Immunodeficiency 15A.** Clinical correlation and consultation with a healthcare provider is recommended. In both cases, the risk to have a child affected with IKBKB-related disorders is increased. Testing of reproductive partners is recommended for carriers of IKBKB-related disorders.
ITGB3	ITGB3-related disorders	Autosomal recessive	Attenuated disease	Section: What does it mean to be a carrier? **Carriers of ITGB3 pathogenic variants may have abnormal platelet production and bleeding tendency, although it tends to be mild in severity; many carriers have no symptoms.** Clinical correlation and consultation with a healthcare provider is recommended. In both cases, the risk to have a child affected with GT2 is increased. Testing of reproductive partners is recommended for carriers of GT2.

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Gene	Condition Name	Inheritance Pattern	Risk Category	Example Report Language
KCNJ11	Familial hyperinsulinism, KCNJ11-related	Autosomal recessive	Other - alternate condition and male carrier risks	Section: What does it mean to be a carrier? **Carriers of FHI may be at risk for gestational diabetes, diabetes mellitus type 2 and MODY13, but many carriers have no symptoms.** Clinical correlation and consultation with a healthcare provider is recommended. In both cases, the risk to have a child affected with FHI is increased. Testing of reproductive partners is recommended for carriers of FHI.
LAMA3	Junctional epidermolysis bullosa, LAMA3-related	Autosomal recessive	Attenuated disease	Section: What does it mean to be a carrier? **Carriers of JEB may have thinner tooth enamel, but many carriers have no symptoms.** Clinical correlation and consultation with a healthcare provider is recommended. In both cases, the risk to have a child affected with JEB is increased. Testing of reproductive partners is recommended for carriers of JEB.
LAMB3	Junctional epidermolysis bullosa, LAMB3-related	Autosomal recessive	Attenuated disease	Section: What does it mean to be a carrier? **Carriers of JEB may have thinner tooth enamel, but many carriers have no symptoms.** Clinical correlation and consultation with a healthcare provider is recommended. In both cases, the risk to have a child affected with JEB is increased. Testing of reproductive partners is recommended for carriers of JEB.
LDLR	Familial hypercholesterolemia, LDLR-related	Autosomal recessive	Attenuated disease	Section: What does it mean to be a carrier? **Individuals with one pathogenic variant for LDLR-related familial hypercholesterolemia often experience some symptoms, but with variable severity and later age of onset. Individuals with two pathogenic variants for LDLR-related familial hypercholesterolemia typically have more severe disease.** Clinical correlation and consultation with a healthcare provider are recommended to review recommended screening. The risk to have a child affected with familial hypercholesterolemia is up to 50%, but severity depends on whether both the patient and partner carry a variant. Testing of reproductive partners is recommended for individuals with pathogenic variants for familial hypercholesterolemia.
LIG4	LIG4 syndrome	Autosomal recessive	Attenuated disease	Section: What does it mean to be a carrier? **Carriers of LIG4 pathogenic variants may have a degree of immune dysregulation, but most carriers have no symptoms.** Clinical correlation and consultation with a healthcare provider is recommended. In both cases, carriers are at an increased risk of having a child affected with LIG4 syndrome. Testing of reproductive partners is recommended for carriers of LIG4 syndrome.
LPL	Lipoprotein lipase deficiency	Autosomal recessive	Attenuated disease	Section: What does it mean to be a carrier? **Carriers of LPL deficiency may have increased triglycerides in the blood and be at increased risk for cardiovascular disease, but many carriers have no symptoms.** Clinical correlation and consultation with a healthcare provider are recommended. In both cases, the risk to have a child affected with LPL deficiency is increased. Testing of reproductive partners is recommended for carriers of LPL deficiency.
MEFV	Familial Mediterranean fever	Autosomal recessive	Attenuated disease	Section: What does it mean to be a carrier? **Carriers of FMF may have a milder form of the disorder that presents later in life, but many carriers have no symptoms.** Clinical correlation and consultation with a healthcare provider is recommended. In both cases, the risk to have a child affected with FMF is increased. Testing of reproductive partners is recommended for carriers of FMF.

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Gene	Condition Name	Inheritance Pattern	Risk Category	Example Report Language
MID1	X-linked Opitz G/BBB syndrome	X-linked	XL - symptomatic female	Section: How is...inherited GBBB is inherited in an X-linked recessive manner. This type of inheritance requires the presence of one copy of a pathogenic variant in the gene located on the X-chromosome for males to be affected. When a female carries this variant on one of her X chromosomes, male children have a 50% risk to be affected, and female children have a 50% risk to be carriers. **Females who carry one copy of this variant may also be affected but show milder symptoms.**
MPL	Congenital amegakaryocytic thrombocytopenia	Autosomal recessive	Other - alternate condition	Section: What does it mean to be a carrier? **There are generally no signs or symptoms associated with being a carrier for CAMT.** However, the risk to have a child affected with CAMT is increased. Testing of reproductive partners is recommended for carriers of CAMT. **Several pathogenic variants in MPL have been linked to autosomal dominant familial essential thrombocythemia, a condition characterized by increased platelet production; carriers may be at risk for blood clots and abnormal bleeding**. Clinical correlation and consultation with a healthcare provider is recommended.
MTM1	X-linked myotubular myopathy	X-linked	XL - symptomatic female	Section: What does it mean to be a carrier? **Carriers of X-MTM may have mild to moderate muscle weakness, but many carriers have no symptoms.** Clinical correlation and consultation with a healthcare provider is recommended. In both cases, the risk to have a child affected with X-MTM is increased.
MVK	Mevalonic aciduria / Hyper-IgD syndrome	Autosomal recessive	Attenuated disease	Section: What does it mean to be a carrier? **Carriers of MVK may develop porokeratosis, but most carriers have no symptoms.** Clinical correlation and consultation with a healthcare provider is recommended. In both cases, the risk to have a child affected with mevalonic aciduria or hyper-IgD syndrome is increased. Testing of reproductive partners is recommended for carriers of mevalonic aciduria or hyper-IgD syndrome.
MYO7A	Usher syndrome, type 1B	Autosomal recessive	Other - alternate condition	Section: What does it mean to be a carrier? There are generally no signs or symptoms associated with being a carrier for variants associated with USH1B. However, the risk to have a child affected with USH1B is increased. Testing of reproductive partners is recommended for carriers of USH1B. **Individuals with specific variants have been reported to be at risk for autosomal dominant deafness-11.**
NBN	Nijmegen breakage syndrome	Autosomal recessive	Cancer Risk	Section: What does it mean to be a carrier? There are generally no signs or symptoms associated with being a carrier of Nijmegen breakage syndrome. However, the risk to have a child affected with NBS is increased. Testing of reproductive partners is recommended for carriers of NBS. **Monoallelic germline alterations in NBN have been detected in individuals with cancer, including individuals with breast cancer, prostate cancer, and melanoma; however, current evidence is insufficient to determine if there is an increased risk of cancer over that of the general population.**
NPC1	Niemann-Pick disease, type C1	Autosomal recessive	Attenuated disease	Section: What does it mean to be a carrier? **Carriers of Niemann-Pick disease, type C may have mild clinical and biochemical features of the condition, but many carriers are asymptomatic.** Clinical correlation and consultation with a healthcare provider are recommended. In both cases, the risk to have a child affected with Niemann-Pick disease, type C is increased. Testing of reproductive partners is recommended for carriers of Niemann-Pick disease, type C.

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Gene	Condition Name	Inheritance Pattern	Risk Category	Example Report Language
NPC2	Niemann-Pick disease, type C2	Autosomal recessive	Attenuated disease	Section: What does it mean to be a carrier? **Carriers of Niemann-Pick disease, type C may have mild clinical and biochemical features of the condition, but many carriers are asymptomatic.** Clinical correlation and consultation with a healthcare provider are recommended. In both cases, the risk to have a child affected with Niemann-Pick disease, type C is increased. Testing of reproductive partners is recommended for carriers of Niemann-Pick disease, type C.
NR2E3	Enhanced S-cone syndrome	Autosomal recessive	Other - alternate condition	Section: What does it mean to be a carrier? **Carriers of ESCS may be at risk for retinitis pigmentosa, a different eye disorder, but many carriers have no symptoms.** Clinical correlation and consultation with a healthcare provider are recommended. In both cases, the risk to have a child affected with ESCS is increased. Testing of reproductive partners is recommended for carriers of ESCS.
OCRL	Lowe syndrome, X-linked	X-linked	XL - symptomatic female	Section: What does it mean to be a carrier? **95% of carrier females have lens opacities; 10% have cataracts that may be visually-significant and require treatment. Carrier females with other symptoms of Lowe syndrome are rare.** Clinical correlation and consultation with a healthcare provider is recommended. In both cases, the risk to have a child affected with Lowe syndrome is increased.
OPA3	3-methylglutaconic aciduria, type III / Costeff syndrome	Autosomal recessive	Other - alternate condition	Section: What does it mean to be a carrier? **Carriers of OPA3 may develop autosomal dominant optic atrophy and cataract, but most carriers have no symptoms.** Clinical correlation and consultation with a healthcare provider is recommended. In both cases, the risk to have a child affected with Costeff syndrome is increased. Testing of reproductive partners is recommended for carriers of Costeff syndrome.
OTC	Ornithine transcarbamylase deficiency, X-linked	X-linked	XL - symptomatic female	Section: What does it mean to be a carrier? **Females who carry one pathogenic variant of OTC deficiency may be affected, with symptoms ranging from asymptomatic to severely affected due to X-chromosome inactivation. Females affected with OTC deficiency need to be closely monitored during pregnancy, especially in the postpartum period.** The risk to have a child affected with OTC deficiency is increased.
PDHA1	Pyruvate dehydrogenase E1-alpha deficiency	X-linked	XL - symptomatic female	Section: What does it mean to be a carrier? **Most female carriers of pyruvate dehydrogenase E1-alpha deficiency have symptoms of the condition but are often more mildly affected than males.** Clinical correlation and consultation with a healthcare provider are recommended. The risk to have a child affected with pyruvate dehydrogenase E1-alpha deficiency is increased.
PEX6	Zellweger spectrum disorders, PEX6-related	Autosomal recessive	Other - one variant issue	Section: What does it mean to be a carrier? **Carriers of the p.Arg860Trp variant in PEX6 may have ZSD in the heterozygous state, but most carriers of PEX6 have no symptoms.** Clinical correlation and consultation with a healthcare provider is recommended. In both cases, the risk to have a child affected with PEX6-related Zellweger spectrum disorder is increased. Testing of reproductive partners is recommended for carriers of PEX6-related Zellweger spectrum disorder.

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Gene	Condition Name	Inheritance Pattern	Risk Category	Example Report Language
PLP1	PLP1-related disorders	X-linked	XL - symptomatic female	Section: What does it mean to be a carrier? **Carriers of PLP1-related disorders may have a milder form of the disorder, but many carriers have no symptoms.** Clinical correlation and consultation with a healthcare provider is recommended. In both cases, the risk to have a child affected with PLP1-related disorders is increased.
POLG	POLG-related disorders	Autosomal recessive	Other - alternate condition	Section: What is... POLG-related disorders are a group of inherited disorders that affect the brain, nerves, muscle, and liver, and include Alpers-Huttenlocher syndrome (AHS), Childhood myocerebrohepatopathy spectrum (MCHS), Myoclonic epilepsy myopathy sensory ataxia (MEMSA), ataxia neuropathy spectrum (ANS), Autosomal recessive progressive external ophthalmoplegia (arPEO), and **Autosomal dominant progressive external ophthalmoplegia (adPEO)**. These disorders are caused by pathogenic variants in the POLG gene which causes a deficiency of DNA polymerase gamma, the enzyme required for mitochondrial DNA synthesis. Ages of onset range from early childhood to adulthood and many individuals have some, but not all, of the symptoms of one or more of the individual disorders. Symptoms and severity are not predictable based on the pathogenic variant(s) identified. AHS, MCHS, MEMSA, ANS, and arPEO are typically inherited in an autosomal recessive manner. **adPEO is typically inherited in an autosomal dominant manner.**
POU1F1	Combined or isolated pituitary hormone deficiency, type 1	Autosomal recessive	Attenuated disease	Section: What does it mean to be a carrier? **Carriers of CPHD1 may develop symptoms of the disorder, but many carriers have no symptoms.** Clinical correlation and consultation with a healthcare provider is recommended. In both cases, the risk to have a child affected with a CPHD1 is increased. Testing of reproductive partners is recommended for carriers of CPHD1.
PRPS1	PRPS1-related disorders	X-linked	XL - symptomatic female	Section: What does it mean to be a carrier? **Carriers of PRPS1-related disorders may have a milder form of the disorders, but many carriers have no symptoms.** Clinical correlation and consultation with a healthcare provider is recommended. In both cases, the risk to have a child affected with PRPS1-related disorders is increased.
RDH12	Leber congenital amaurosis 13	Autosomal recessive	Other - alternate condition	Section: What does it mean to be a carrier? **Carriers of LCA13 are at increased risk for autosomal dominant retinitis pigmentosa, but many carriers have no symptoms.** Clinical correlation and consultation with a healthcare provider is recommended. In both cases, the risk to have a child affected with LCA13 is increased. Testing of reproductive partners is recommended for carriers of LCA13.
RLBP1	RLBP1-related retinopathies	Autosomal recessive	Attenuated disease	Section: What does it mean to be a carrier? **Carriers of RLBP1 may develop retinal disease, but most carriers have no symptoms.** Clinical correlation and consultation with a healthcare provider is recommended. In both cases, the risk to have a child affected with a retinopathy is increased. Testing of reproductive partners is recommended for carriers of RLBP1-related retinopathies.
RP2	Retinitis pigmentosa 2	X-linked	XL - symptomatic female	Section: What does it mean to be a carrier? **Carriers of RP2 may have a milder form of the disorder, but many carriers have no symptoms.** Clinical correlation and consultation with a healthcare provider is recommended. In both cases, the risk to have a child affected with RP2 is increased.

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Gene	Condition Name	Inheritance Pattern	Risk Category	Example Report Language
RPE65	Leber congenital amaurosis 2	Autosomal recessive	Other - alternate condition	Section: What does it mean to be a carrier? **Carriers of LCA2 are at increased risk for autosomal dominant retinitis pigmentosa, but many carriers have no symptoms.** Clinical correlation and consultation with a healthcare provider is recommended. In both cases, the risk to have a child affected with LCA2 is increased. Testing of reproductive partners is recommended for carriers of LCA2.
RPGR	Retinitis pigmentosa 3	X-linked	XL - symptomatic female	Section: What is... Retinitis pigmentosa is an inherited disorder affecting the retina, the light-sensitive tissue of the eyes. There are multiple genetic causes of nonsyndromic, or isolated, retinitis pigmentosa; retinitis pigmentosa 3 is caused by pathogenic variants in RPGR and is inherited in an X-linked manner. In most affected individuals, symptoms begin in early adulthood with decreased vision at night and the development of tunnel vision; central vision, causing issues with reading and driving, deteriorates over time. The condition often progresses over years to legal blindness in adulthood. **Females may be unaffected or have milder symptoms of the condition.** Pathogenic variants in RPGR can also cause cone-rod dystrophy, a different progressive visual condition resulting in increased light sensitivity and decreased sharpness of vision.
RS1	Juvenile retinoschisis, X-linked	X-linked	XL - symptomatic female	Section: What does it mean to be a carrier? **Carriers of XLRS may have eye changes and vision issues, but most carriers have no symptoms.** Clinical correlation and consultation with a healthcare provider is recommended. In both cases, the risk to have a child affected with XLRS is increased.
RTEL1	Dyskeratosis congenita, RTEL1-related	Autosomal recessive	Attenuated disease	Section: What does it mean to be a carrier? **Some carriers may have symptoms of the condition, as specific variants in RTEL1 are associated with autosomal dominant DC.** The risk to have a child affected with DC is increased. Testing of reproductive partners is recommended for carriers of DC.
RYR1	RYR1-related disorders	Autosomal recessive	Other - alternate condition	Section: What does it mean to be a carrier? **Carriers of RYR1-related conditions may develop the signs and symptoms of autosomnal dominant conditions associated with RYR1, such as malignant hyperthermia, congenital myopathy 1A with susceptibility to malignant hyperthermia, and King-Denborough syndrome.** Clinical correlation and consultation with a healthcare provider is recommended. In general, the risk to have a child affected with an RYR1-related condition is increased. Testing of reproductive partners is recommended for carriers of RYR1-related conditions.
SAMD9	Normophosphatemic familial tumoral calcinosis	Autosomal recessive	Other - alternate condition	Section: What is... Familial tumoral calcinosis (FTC) is an inherited disorder causing the abnormal development of calcified masses throughout the body. There are two forms of the disease that differ by the amount of phosphate in the blood; normophosphatemic familial tumoral calcinosis (NFTC) is caused by biallelic pathogenic variants in SAMD9. Affected individuals develop the first calcified masses in childhood; they can appear around but not in the joints (periarticular) and under the skin (subcutaneous), causing pain and inflammation. Inflammation of the conjunctiva of the eyes and the gums is common and can be severe. Bloodwork shows normal phosphate levels. There is no cure for NFTC, but medical surveillance and care may help to improve some symptoms and overall condition of life. **Some individuals with gain-of-function variants in SAMD9 develop a different condition called MIRAGE syndrome that is often fatal in childhood; it is characterized by significant prenatal and postnatal growth restriction, frequent infections, genital and adrenal abnormalities, a specific type of bone marrow cancer (myelodysplastic syndrome (MDS)), and severe developmental delay and intellectual disability.**

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SEC23B	Congenital dyserythropoietic anemia, type II	Autosomal recessive	Attenuated disease	Section: What is... Congenital dyserythropoietic anemia (CDA) is an inherited disorder affecting the development and function of red blood cells. CDA, type II, also known as hereditary erythroblastic multinuclearity with a positive acidified serum test (HEMPAS), is the most common type of CDA and is caused by biallelic pathogenic variants in SEC23B. Individuals often present in early adulthood with yellowing of the skin and eyes, an enlarged liver and spleen, and gallstones. Anemia can be mild to severe, and in the third decade of life, most affected individuals begin to develop iron overload. The hallmark feature of the disease is evidence of unusually-shaped red blood cell precursors called erythroblasts in the bone marrow; erythroblasts may also have multiple nuclei. **Carriers of CDA, type II may have abnormal blood tests but are not thought to have symptoms of the disease. Treatment includes blood transfusions and iron chelation.**
SERPINA1	Alpha-1 antitrypsin deficiency	Autosomal recessive	Attenuated disease	Section: What does it mean to be a carrier? Individuals with an MS or SS combination usually produce enough alpha-1 antitrypsin to protect the lungs. **People with the MZ genotype have a slightly increased risk of impaired lung or liver function.** Individuals that carry one S or Z allele are at higher risk of having children with the more severe ZZ and SZ genotypes. Testing of reproductive partners is recommended for carriers of alpha-1 antitrypsin deficiency.
SLC12A6	Agenesis of the corpus callosum with peripheral neuropathy	Autosomal recessive	Other - alternate condition	Section: What is... Agenesis of the corpus callosum with peripheral neuropathy (ACCPN), also known as Andermann syndrome, is an inherited disorder affecting the brain and muscle movement. ACCPN is caused by biallelic pathogenic variants in SLC12A6. The hallmark features of this condition are absence of the brain tissue connecting the two halves of the brain and poor development of the nerves. Symptoms include weak muscle tone, muscle wasting, and progressive loss of sensation in the limbs; individuals are developmentally delayed and often lose the ability to walk independently by adolescence. Intellectual disability and issues of the nerves controlling the face and neck are common. There is no cure for ACCPN, but medical surveillance and care may help to improve some symptoms and overall condition of life. **Certain single pathogenic variants in SLC12A6 can cause Charcot-Marie-Tooth disease, type 2II (CMT2II), however, individuals with these variants are not carriers of ACCPN and are not at risk to have children with the disorder; carriers of ACCPN are not at risk for CMT2II and are unaffected.**
SLC37A4	Glycogen storage disease, type Ib / IIw	Autosomal recessive	Other - alternate condition	Section: How is...inherited Glycogen storage disease, type Ib is inherited in an autosomal recessive manner. Both parents must be carriers of a pathogenic variant in the gene in order to be at risk to have an affected child. This type of inheritance requires the presence of two copies of a pathogenic variant in the gene, one inherited from each carrier parent, for the child to have the genetic disease. There is a 1 in 4 chance that a child will inherit two pathogenic variants, one in each copy of the gene, and be affected when both parents are carriers. **Children with pathogenic variants in the same gene may have a different disorder called congenital disorder of glycosylation, type IIw which can be inherited in an autosomal recessive manner, or very rarely in an autosomal dominant manner. Autosomal dominant inheritance requires the presence of a pathogenic variant in one copy of the associated gene for a person to have the genetic disease. First-degree relatives (children, siblings, parents) of an individual with a pathogenic variant have a 50% or 1 in 2 chance to have that same variant.**

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Gene	Condition Name	Inheritance Pattern	Risk Category	Example Report Language
SLC4A11	Corneal dystrophy and perceptive deafness syndrome	Autosomal recessive	Other - alternate condition	Section: How is...inherited CDPD is inherited in an autosomal recessive manner. Both parents must be carriers of a pathogenic variant in the gene in order to be at risk to have an affected child. This type of inheritance requires the presence of two copies of a pathogenic variant in the gene, one inherited from each carrier parent, for the child to have the genetic disease. There is a 1 in 4 chance that a child will inherit two pathogenic variants, one in each copy of the gene, and be affected when both parents are carriers. **Children with pathogenic variants in the same gene may have a different disorder called Corneal Dystrophy, Endothelial 2 which can be inherited in an autosomal recessive manner, or very rarely in an autosomal dominant manner. Autosomal dominant inheritance requires the presence of a pathogenic variant in one copy of the associated gene for a person to have the genetic disease. First-degree relatives (children, siblings, parents) of an individual with a pathogenic variant have a 50% or 1 in 2 chance to have that same variant.**
SLC6A8	Creatine transporter defect, SLC6A8-related, X-linked / Cerebral creatine deficiency syndrome	X-linked	XL - symptomatic female	Section: What does it mean to be a carrier? **Some carrier females of CCDS1 have mild intellectual disability, learning difficulties, and behavioral issues, while others are asymptomatic.** The risk for a female carrier to have a male child affected with CCDS1 is increased.
SPR	Sepiapterin reductase deficiency	Autosomal recessive	Attenuated disease	Section: What does it mean to be a carrier? **Carriers may have a mild form of SRD, but only one case has been reported, and most carriers have no symptoms**. In both cases, the risk to have a child affected with SRD is increased. Testing of reproductive partners is recommended for carriers of SRD.
TAFAZZIN	Barth syndrome	X-linked	XL - symptomatic female	Section: What does it mean to be a carrier? There are generally no signs or symptoms associated with being a carrier for Barth syndrome; **in rare cases, female carriers experience mild symptoms.** However, the risk to have a child affected with Barth syndrome is increased.
TERT	Dyskeratosis congenita spectrum disorders	Autosomal recessive	Attenuated disease	Section: What does it mean to be a carrier? **Some carriers may have symptoms of the condition, as specific variants in TERT are associated with autosomal dominant DC.** The risk to have a child affected with DC is increased. Testing of reproductive partners is recommended for carriers of DC.
TMC1	Nonsyndromic hearing loss and deafness (DFNB) 7	Autosomal recessive	Attenuated disease	Section: What does it mean to be a carrier? **Carriers of DFNB7 may have hearing loss, but most carriers have no symptoms.** Clinical correlation and consultation with a healthcare provider is recommended. In both cases, the risk to have a child affected with DFNB7 is increased. Testing of reproductive partners is recommended for carriers of DFNB7.
TREX1	TREX1-related disorders	Autosomal recessive	Attenuated disease	Section: What does it mean to be a carrier? **Carriers for TREX1-related disorders may develop symptoms associated with SLE, AGS1, chilblain lupus, and autosomal dominant retinal vasculopathy with cerebral leukodystrophy.** Clinical correlation and consultation with a healthcare provider is recommended. In general, the risk to have a child affected with TREX1-related disorders is increased. Testing of reproductive partners is recommended for carriers of TREX1-related disorders.

Personal Carrier Risk Genes: All data and example report language are reported by Baylor Genetics®. 1/23/26

Gene	Condition Name	Inheritance Pattern	Risk Category	Example Report Language
TSHR	Congenital hypothyroidism, TSHR-related	Autosomal recessive	Attenuated disease	Section: What does it mean to be a carrier? **Carriers for congenital hypothyroidism, TSHR-related may be at increased risk for familial gestational hyperthyroidism during pregnancy and nonautoimmune hyperthyroidism, but most carriers have no symptoms.** Clinical correlation and consultation with a healthcare provider is recommended. In both cases, the risk to have a child affected with congenital hypothyroidism, TSHR-related is increased. Testing of reproductive partners is recommended for carriers of congenital hypothyroidism, TSHR-related.
USH1C	Usher syndrome, type 1C	Autosomal recessive	Other - one variant issue	Section: What does it mean to be a carrier? **Carriers of a specific variant, c.667G>T (p.Gly223Cys), have been reported to be at risk for nonsyndromic hearing loss; however, there are generally no signs or symptoms associated with being a carrier for USH1C.** Additionally, the risk to have a child affected with USH1C is increased. Testing of reproductive partners is recommended for carriers of USH1C.
WAS	Wiskott-Aldrich syndrome, X-linked	X-linked	XL - symptomatic female	Section: What does it mean to be a carrier? There are generally no signs or symptoms associated with being a carrier for WAS-related disorders; **some female carriers may have low platelets and rarely, female carriers have significantly low platelets and immunodeficiency.** The risk to have a child affected with WAS-related disorders is increased.
WNT10A	Odonto-onycho-dermal dysplasia / Schopf-Schulz-Passarge syndrome	Autosomal recessive	Attenuated disease	Section: What does it mean to be a carrier? **Carriers with a heterozygous pathogenic or likely pathogenic variant in the WNT10A gene may be at risk of developing mild symptoms of Odontoonychodermal dysplasia / Schopf-Schulz-Passarge syndrome.** Clinical correlation is recommended.
ZIC3	X-linked heterotaxy-1	X-linked	XL - symptomatic female	Section: How is...inherited X-linked heterotaxy-1 is inherited in an X-linked recessive manner. This type of inheritance requires the presence of one copy of a pathogenic variant in the gene located on the X-chromosome for males to be affected. When a female carries this variant on one of her X chromosomes, male children have a 50% risk to be affected, and female children have a 50% risk to be carriers. **Females who carry one copy of this variant may also be affected but show milder symptoms.**