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Genetic science is a rapidly developing field. Information about Jewish genetic diseases is updated frequently. For the most recent and detailed information visit:

JewishGeneticDiseases.org

Jewish Genetic Diseases

Jewish Genetic Disease Consortium

JGDC

There are different genetic concerns for people of Ashkenazi Jewish background (German, French or Eastern European), and people of Sephardic (Mediterranean) or Mizrahi (Persian/ Iranian or Middle Eastern) background. Regardless of the specific Jewish background, **all Jewish and interfaith couples should have preconception carrier screening for the Jewish genetic diseases.**

Ashkenazi

It is estimated that nearly 1 in 2 Ashkenazi Jews in the United States is a carrier of at least one of 38 Jewish genetic diseases.

19 disease panel

38 disease panel
includes diseases from panel of 19

Ashkenazi Jewish Genetic Disease	Carrier Frequency
Abetalipoproteinemia Severe malabsorption of dietary fats and fat-soluble vitamins causing failure to thrive, diarrhea, blood abnormalities (acanthocytosis), and stool abnormalities (steatorrhea). Later in childhood symptoms include poor muscle coordination, ataxia and retinitis pigmentosa.	1 in 180
Alport Syndrome, Autosomal Recessive Progressive loss of kidney function (hematuria, proteinuria) resulting in end-stage renal disease, sensorineural hearing loss, and eye abnormalities such as anterior lenticonus.	1 in 188
Arthrogryposis, Mental Retardation and Seizures Arthrogryposis, mental retardation, autism spectrum disorder, epilepsy, microcephaly, and hypotonia.	1 in 373
Bardet-Biedl Syndrome Features include retinitis pigmentosa, obesity, polydactyly, intellectual disability/developmental delay, renal problems, anosmia, genital abnormalities and male infertility. Other affected organs include the heart, liver and digestive system. There is variable age of onset and severity of symptoms.	1 in 107
Bloom Syndrome Poor growth, frequent infections, and possible learning disabilities. Increased predisposition for leukemia and cancers of the breast and colon.	1 in 134
Canavan Disease Progressive disease of central nervous system with no cure. Symptoms include seizures, regression of milestones, severe mental retardation and death in childhood.	1 in 55
Carnitine Palmitoyltransferase II Deficiency Characterized by recurrent episodes of myalgia and rhabdomyolysis causing myoglobinuria which may be triggered by exercise, stress, exposure to extreme temperatures, infections, or fasting. The first episode usually occurs during childhood or adolescence. This can damage the kidneys, in some cases leading to life-threatening kidney failure.	1 in 51
Congenital Amegakaryocytic Thrombocytopenia Pancytopenia, decreased bone marrow activity, and very low platelet counts.	1 in 55
Congenital Disorder of Glycosylation Ia Hypotonia, abnormal fat distribution, strabismus, developmental delay and failure to thrive appear in infancy. Other symptoms include elevated liver function tests, seizures, and pericardial effusion that could lead to death under 1 year of life due to multiple organ failure. Affected individuals who survive infancy may have intellectual disability, lethargy, temporary paralysis, neuropathy, kyphoscoliosis, ataxia, contractures and retinitis pigmentosa.	1 in 57
Cystic Fibrosis Thick mucous buildup in the lungs leading to breathing difficulty and infection, with no cure. Symptoms include poor digestion, male infertility and shortened life expectancy (into the 30s).	1 in 24
Dyskeratosis Congenita, Autosomal Recessive Abnormally growing and poorly growing fingernails and toenails, pigmentary changes on neck and chest. Symptoms include bone marrow failure, aplastic anemia and increased risk for leukemia. Increased risk for cancers of the head, neck, anus or genitals. Other features include pulmonary fibrosis, hair loss, osteoporosis, avascular necrosis of the joints, liver disease and short stature.	1 in 203

Ashkenazi Jewish Genetic Disease	Carrier Frequency
Ehlers-Danlos VIIC Hypermobility, easy bruising, fragile skin, and blue sclera.	1 in 248
Familial Dysautonomia Autonomic nervous system disorder (e.g. swallowing, sweating, pain sensitivity). Increased risk for pulmonary (e.g. pneumonia) and gastrointestinal complications.	1 in 31
Familial Hyperinsulinism Inability to stop insulin production leading to seizures, poor muscle tone, poor feeding and breathing difficulty (newborn and children). Treatment options include glucose infusion, insulin release-reducing drugs, and/or surgical removal of portions of the pancreas. (Focal or localized, disease is present in 1-2% of children who inherit a single paternal mutation).	1 in 68
Fanconi Anemia C Bone problems (short stature, bone marrow failure, etc), predisposition to leukemia and possible learning disability/mental retardation.	1 in 100
Fragile X Syndrome X-linked condition. Features include mental retardation, behavioral problems (autistic-like features, etc.), and characteristic facial features in affected males. Affected females may also have milder clinical manifestations. Premutation carrier females are at increased risk for premature ovarian insufficiency; whereas premutation carrier males are at increased risk for Fragile X-associated tremor/ataxia syndrome.	1 in 115
Galactosemia Feeding difficulties, lethargy, failure to thrive, jaundice, and bleeding within a few days after birth. Increased risk for sepsis and shock, developmental delay/intellectual disability, and cataracts. Managed by dietary restrictions	1 in 172
Gaucher Disease Enlargement of spleen and liver, blood abnormalities (anemia, easy bruising, impaired clotting, etc), and bone problems (joint pain, bone fractures, etc). Variable age of onset and severity of symptoms. Successful Enzyme Replacement Therapy exists, reducing or reversing symptoms.	1 in 15

Ashkenazi Jewish Genetic Disease	Carrier Frequency
Glycogen Storage Disease Ia Biochemical abnormalities such as very low glucose level leading to delayed growth/development presenting in infancy. Symptoms include enlarged spleen, gastrointestinal problems, recurrent infection, and pancreatitis. Managed by dietary restrictions.	1 in 64
Joubert Syndrome 2 Neurological disorder with brain malformations leading to developmental delay, mental retardation, breathing difficulty, ataxia, failure to thrive, retinal degeneration, and renal dysfunction.	1 in 110
Lipoamide Dehydrogenase Deficiency (E3) Variable age of onset and severity of symptoms including fatigue, episodes of decompensation, severe neurological decline and sometimes death. Managed by dietary restrictions.	1 in 107
Maple Syrup Urine Disease Ib Neurological impairment in infants including poor suck, irritability, lethargy and potential lapse into coma after ingesting dietary protein. Delay in diagnosis can cause impaired intellectual development. Managed by dietary restrictions.	1 in 97
Mucolipidosis IV Severe neurodegenerative condition leading to abnormalities of the cornea and retina, inability to walk or speak.	1 in 89
Multiple Sulphatase Deficiency Accumulation of sulfatides, sulfated glycosaminoglycans, sphingolipids and steroid sulfates causing neurologic deterioration with mental retardation, skeletal anomalies, organomegaly, and ichthyosis.	1 in 320
Nemaline Myopathy Progressive disease causing muscle weakness, delayed motor milestones, and feeding/respiratory difficulty potentially leading to death in infancy.	1 in 168
Niemann-Pick Disease Severe neurodegenerative condition leading to loss of brain function, enlargement of liver and spleen and shortened life expectancy (2-3 years).	1 in 115

Ashkenazi Jewish Genetic Disease	Carrier Frequency
3-Phosphoglycerate Dehydrogenase Deficiency Microcephaly, psychomotor retardation, and seizures.	1 in 280
Polycystic Kidney Disease, Autosomal Recessive Cyst development in the kidneys causing kidney enlargement and can lead to kidney failure. Symptoms include cysts in the liver, hypertension, hematuria, recurrent urinary tract infections, kidney stones, and an increased risk for aneurysms. This condition is often lethal early in life.	1 in 107
Retinitis Pigmentosa 59 Childhood loss of night vision developing into peripheral blind spots and later, leading to tunnel vision and blindness.	1 in 118
Smith-Lemli-Opitz Syndrome Characteristic facial features, microcephaly, intellectual disability, and behavioral problems (e.g. autism). Abnormalities of the heart, lungs, kidneys, gastrointestinal tract, fingers/toes and genitalia are also common. Variable severity of symptoms.	1 in 36
Spinal Muscular Atrophy Severe and progressive weakness of the voluntary muscles affecting breathing, swallowing, head/neck control, walking and crawling. Variable onset and severity, with shortened lifespan for those with onset in infancy.	1 in 41
Tay-Sachs Disease Progressive disease of the central nervous system leading to loss of coordination, seizures, difficulty swallowing, poor pulmonary function, blindness, paralysis, severe mental retardation and shortened life expectancy (3-5 years). **Tay-Sachs enzyme analysis using blood must be done in addition to DNA for complete screening.**	1 in 27
Tyrosinemia I Tyrosine aminotransferase deficiency that can affect the eyes, skin, and mental development. Symptoms include photophobia, painful skin lesions on the palms and soles, and intellectual disability.	1 in 150

Ashkenazi Jewish Genetic Disease	Carrier Frequency
Usher IF Profound hearing loss and retinitis pigmentosa.	1 in 147
Usher III Postlingual onset of moderate to severe hearing loss and variable onset and severity of retinitis pigmentosa.	1 in 120
Walker-Warburg Severe disease of the brain (seizures, developmental delay, mental retardation), muscles (weakness, feeding difficulty) and eyes (blindness). There is a shortened life expectancy (less than 3 years).	1 in 120
Wilson Disease Copper accumulation in the liver (causing jaundice, fatigue, loss of appetite, and abdominal swelling), brain (causing nervous system and psychiatric problems), and eyes (causing Kayser-Fleischer rings and restricted ability to gaze upwards) with variable onset.	1 in 70
Zellweger Syndrome Demyelination of white matter causing hypotonia, feeding problems, hearing loss, vision loss, and seizures. Other affected organs include the liver, heart, kidneys and bones and there is a shortened life expectancy.	1 in 172

Disease information and carrier frequency developed by Icahn School of Medicine at Mount Sinai Genetic Testing Laboratory.

Please be aware that there are many laboratories offering Ashkenazi Jewish genetic disease screening with panels varying from 19 to 38 diseases. You should discuss these options with your physician or genetic counselor.

Individuals may also opt to do expanded carrier screening to include disorders not necessarily more common in the Ashkenazi Jewish ancestry (Pan Ethnic Panels).

Sephardic/Mizrahi

There is no single preconception carrier-screening panel for people of Sephardic or Mizrahi background. Carrier screening is dependent upon country of origin. People of Sephardic or Mizrahi background should seek genetic counseling.

Sephardic/Mizrahi Jewish Genetic Disease	Carrier Frequency
Alpha-Thalassemia A blood disorder that affects the production of the alpha protein in hemoglobin leading to anemia and insufficient supply of oxygen throughout the body. There are two types of the disease: Hb Bart syndrome (more serious) is usually fatal to the fetus or newborn and HbH (less serious) can be managed by blood transfusions if necessary. Many HbH patients require no treatment. Screening is done by a complete blood count (CBC) followed by DNA analysis if warranted.	1 in 5 to 1 in 100 Moroccan Jews 1 in 4 to 1 in 13 Yemeni Jews 1 in 7 to 1 in 40 Iraqi Jews 1 in 80 Kurdish Jews
Ataxia Telangiectasis Characterized by a progressive loss of the ability to control movement, weakened immune system, and increased risk for cancer. There is supportive treatment for individuals with severe frequent infections and respiratory issues.	1 in 80 North African Sephardic Jews
Beta-Thalassemia (B-T) A blood disorder that affects the production of the beta protein in hemoglobin leading to anemia and insufficient supply of oxygen throughout the body. There are two types of the disease: B-T Major (Cooley's Anemia) is the most serious and B-T Intermedia which is less serious. Treatment may include blood transfusions causing a build-up of iron, requiring chelation therapy. Screening is done by complete a blood count (CBC) followed by DNA analysis if warranted.	From 1 in 5 to 1 in 7 Sephardic and Mizrahi Jews
Corticosterone Methyloxidase Type II Deficiency Severe form may cause infantile seizures, dehydration, shock and possible death. Milder form (more common) causes growth failure, short stature, blood pressure irregularities, weakness, dizziness and salt craving. Disease is treatable when diagnosed early.	1 in 30 Persian Jews
Costeff Optic Atrophy (also known as Type III 3-methylglutaconic Aciduria) A progressive neurological disorder resulting in vision loss, unsteady gait, spasticity (rigid, jerky muscle movement) and loss of ability to walk. Symptoms appear in childhood and there is shortened life expectancy.	1 in 10 Iraqi Jews (also appears in Persian Jews)
Cystic Fibrosis Thick mucous buildup in the lungs leading to breathing difficulty and infection, with no cure. Symptoms include poor digestion, male infertility and shortened life expectancy (into the 30s).	1 in 26 Sephardic/ Mizrahi Jews
Familial Mediterranean Fever An inflammatory disorder that involves recurrent short episodes of fever and inflammation in the lining of the abdomen, chest cavity, lungs, heart and joints. If untreated, this can lead to kidney failure. Patients who receive early and regular treatment can have a normal lifespan.	1 in 3 to 1 in 7 Persian, Iraqi and Mediterranean Jews
Familial Tumoral Calcinosis Condition of excessive calcium deposits that begins in infancy with skin lesions that progress into calcified tumors, eye and gum inflammation.	1 in 92 Yemeni Jews
Glucose-6-Phosphate Dehydrogenase Deficiency (G6PD) A condition in which red blood cells break down when the body is exposed to certain drugs, foods, infections or stress. Sometimes referred to as Favism, as the condition can be triggered in some patients by fava beans. An X-linked condition (with a different inheritance pattern than the other, autosomal-recessive diseases). Symptoms can be minimized by avoidance of trigger foods and drugs.	1 in 2 Kurdish Jewish males have the condition and 1 in 2 females are carriers
Hereditary Inclusion Body Myopathy (HIBM)/ GNE Myopathy Progressive muscular dystrophy with onset in the late teens, early twenties or later. Variable disease manfestation may result in being wheelchair-bound at about age 40 or milder weakness, presenting late in life.	1 in 10 to 1 in 20 Persian Jews

Sephardic/Mizrahi Jewish Genetic Disease	Carrier Frequency
Limb Girdle Muscular Dystrophy 2B Slowly progressive muscle weakness that may have differences in severity, even within the same family. Onset generally between age 12 and 39, beginning with the lower extremities, progressing to upper limb weakness often over ten years. At least half of affected individuals may lose the ability to walk after their 40s.	1 in 10 to 1 in 25 Jews from Libya, Yemen and the Caucasus
Metachromatic Leukodystrophy A metabolic storage disorder resulting in progressive loss of neurological and movement abilities. Onset of the disease occurs at about 12-18 months leading to death in childhood.	1 in 4 Yemeni and Habbani Jews
Polyglandular Syndrome A condition that causes multiple hormones to malfunction, including adrenal and parathyroid hormones causing immune problems. Fertility issues and hair loss may occur.	1 in 40 Persian Jews
Pseudocholinesterase Deficiency People with this condition are unable to metabolize certain anesthetic drugs. These drugs stays active in the system much longer than in an unaffected individual. Patients may experience a temporary paralysis of the respiratory muscles and require a period of mechanically-assisted breathing. Individuals of Persian and Iraqi Jewish heritage may consider a blood test to determine if they are affected by this condition in order to avoid trigger anesthetics.	1 in 10 Persian and Iraqi Jews
Spinal Muscular Atrophy Severe and progressive weakness of the voluntary muscles affecting breathing, swallowing, head/neck control, walking and crawling. Variable onset and severity, with shortened lifespan for those with onset in infancy.	1 in 10 Egyptian Jews
Wolman Disease A metabolic disease resulting from an inability to process cholesterol. This disease occurs in infancy with failure to thrive, anemia, vomiting, diarrhea and adrenal gland deposits which are visible on imaging studies. Death usually occurs in the first year of life.	1 in 45 Persian and Bakharan Jews

The **Jewish Genetic Disease Consortium** (JGDC) increases awareness about Jewish genetic diseases and encourages timely and appropriate carrier screening for all persons who have any Jewish heritage, as well as couples of interfaith marriage. The JGDC is an alliance of non-for-profit organizations sharing the common goal of combating Jewish genetic diseases. While each JGDC member organization has its own individual mission, the JGDC unites these organizations so that we may jointly strengthen public education and awareness and appropriate carrier screening for all persons of Jewish heritage, whether Ashkenazi, Mizrahi, Sephardic, as well as couples of interfaith marriage.

Through its Medical Grand Rounds Program, Clergy Education Program and Jewish Community Program, the JGDC educates physicians, rabbis, Jews of all backgrounds and interfaith couples about Jewish genetic diseases. The goal is to decrease the incidence of Jewish genetic diseases and assure healthy Jewish families by increasing preconception carrier screening rates and promoting the understanding of reproductive options available to carrier couples.

Jewish Genetic Disease
Consortium

JGDC

Member Organizations

Bloom's Syndrome Foundation
Canavan Foundation
FD Now
Genetic Disease Foundation
Mathew Forbes Romer Foundation
Mucolipidosis Type 4 Foundation
National Gaucher Foundation
National Tay-Sachs & Allied Diseases Association
Sephardic Health Organization for Referral and Education

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For more information visit

JewishGeneticDiseases.org

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