Disorders Covered on Jnetics' Carrier Test

The table below lists the disorders covered on Jnetics' carrier test.

- Below each name there is a brief description, followed by the relevant at-risk population(s) and associated carrier frequencies
- Disorders in blue are relevant to the Ashkenazi population, disorders in green are relevant to the Sephardi/Mizrahi population, and disorders in orange are relevant to both groups.
- Please note, optional testing for Fragile X is only available at the Jnetics Clinic.

ABCC8-related Hyperinsulinism
A disorder characterised by abnormally high insulin production leading to episodes of low blood sugar. Episodes can cause serious complications and death if not managed properly. Symptoms and their severity vary. Life-threatening if not managed properly but, with appropriate treatment, affected individuals can lead normal lives. For more information, click here.
Ashkenazi Jewish: 1 in 44
Asparagine Synthetase Deficiency
A severe neurological disorder associated with developmental delays, small head and seizures. Symptoms begin in utero or at birth and the disorder is usual fatal in infancy. For more information, click here.
Iranian Jewish: 1 in 80*
Acute Infantile Liver Failure
A disorder characterised by temporary liver failure and associated symptoms such as jaundice, vomiting, impaired ability for blood to clot and various liver problems. Patients who survive the initial liver failure can recover and show normal development. For more information, click here.
Yemenite Jewish: 1 in 34
Bardet-Biedl Syndrome
A condition associated with progressive worsening of vision, childhood obesity, extra digits on the hands and feet, kidney problems, reduced function of testes or ovaries, and learning disabilities. For more information, click here.
Ashkenazi Jewish: 1 in 107
Bloom Syndrome~
A condition characterised by increased risk of infections and cancer, as well as poor growth and increased sensitivity to light. There is no cure; treatment involves managing and monitoring the risk of cancer. Lifespan varies but is typically up to 30 years due to cancer. For more information, click here.
Ashkenazi Jewish: 1 in 134
Canavan Disease~
A progressive disease of the nervous system. Symptoms often develop in infancy and can include low muscle tone, large head size, intellectual disability and seizures. Life expectancy varies, but Canavan disease is typically fatal in the second decade. There is no treatment. For more information, click here.
Ashkenazi Jewish: 1 in 55
Carnitine Palmitoyltransferase II Deficiency
A disorder associated with an inability of the body to use certain fats for energy. Three different forms: (1) Myopathic form - least severe, good prognosis, characterised by muscle degeneration and weakness, triggered particularly by fasting. (2) Severe infantile form- fasting leads to metabolic disorders which may lead to sudden death in infancy. (3) Lethal neonatal form - fasting leads to metabolic disorders and abnormal body structure, almost always lethal in the first months of life. For more information, click here.

Ashkenazi Jewish: 1 in 51
Congenital Amegakaryocytic Thrombocytopenia
A disorder characterised by an inability to make platelets, which are part of the blood and help with clotting and preventing bleeding. This can lead to bruising, life-threatening bleeds and increased risk of certain blood cancers. Treatment includes blood transfusions and stem cell transplant. For more information, click here.
Ashkenazi Jewish: 1 in 55
Congenital Disorder of Glycosylation Ia
A condition affecting many body systems. Symptoms and age of onset varies. Symptoms can include low muscle tone, abnormal distribution of fat and developmental delays. Treatment is mostly symptoms-based, such as medication to prevent seizures. Many babies affected by the disorder die before birth. For more information, click here.
Ashkenazi Jewish: 1 in 57
Costeff Optic Atrophy Syndrome
A neurological disorder associated with vision loss beginning in infancy, and movement disorder beginning in childhood. Life expectancy is unknown, but affected individuals have lived into their 30s. For more information, click here.
Iraqi Jewish: 1 in 50
Cystic Fibrosis~
A progressive disorder associated with build-up of thick mucus in the lungs and digestive system. Leads to symptoms including chronic respiratory infections, lung damage, poor growth, pancreatic issues and infertility in males. With treatment, lifespan can approach 50+ years. For more information, click here.
Ashkenazi Jewish: 1 in 24 | **General population: 1 in 32**
Cystinosis
A disorder characterised by build-up of an amino acid called cystine, leading to damage of the kidneys or eyes and sometimes other organs including the brain and liver. Medications can help the disorder by lowering the levels of cystine in the body. Kidney transplant may also be part of treatment. Life expectancy to 50 years or beyond can be achieved with proper treatment. For more information, click here.
Moroccan Jewish: 1 in 100*
Dihydrolipamide Dehydrogenase Deficiency
A disorder characterised by Life-threatening problems with metabolism, leading to abnormalities with the brain or liver beginning in infancy. Typically, there will be periods of time with symptoms and periods without symptoms. Many affected children don not survive the first few years of life. For more information, click here.
Ashkenazi Jewish: 1 in 107
Familial Dysautonomia~
A disorder of the nervous system, associated with symptoms including inability to regulate body temperature and respond to pain. No cure; treatment focusses on managing symptoms. Average life expectancy is about 40 years. For more information, click here.
Ashkenazi Jewish: 1 in 31
Fanconi Anaemia C~
Associated with impaired bone marrow function, physical abnormalities, organ defects and increased risk of cancer. Typically fatal by around age 30. There is no cure. For more information, click here.
Ashkenazi Jewish: 1 in 99
Fanconi Anemia, Complementation group A
A disorder associated with impaired bone marrow function, physical abnormalities, organ defects and increased risk of cancer. Typically fatal by around age 30. For more information, click here.
Moroccan Jewish: 1 in 100* | **Indian Jewish: 1 in 27***
Fumarase Deficiency
A condition affected the brain and other parts of the nervous system. Symptoms include low muscle tone, developmental delays, and failure to thrive. Life expectancy is typically only a few months, but few individuals have lived into early adulthood. For more information, click here.
Ashkenazi Jewish: 1 in 99*

Galactosemia
A condition affecting the body's ability to process a type of sugar called galactose. Symptoms in infants include feeding difficulties, lethargy, and failure to thrive, developmental delay and cognitive impairment. Can be managed with dietary restriction. For more information, click here.
Ashkenazi Jewish: 1 in 127
Glycogen Storage Disease 1a~
A condition characterized by abnormally low blood sugar levels, enlarged liver and kidneys, and impaired growth – that results from the build-up of stored glycogen in the body. The main symptoms can be treated with strict dietary management. For more information, click here.
Ashkenazi Jewish: 1 in 64
Glycogen Storage Disease Type II/Pompe Disease
A disorder caused by a build-up of glycogen in the body's cells, which leads leads to damage of muscle cells and other cell types. Early onset form is associated with severe heart and respiratory problems and death in infancy occurring if there is no intervention. Later onset forms are not associated with heart issues, but individuals experience respiratory problems and muscle weakness. For more information, click here.
Ashkenazi Jewish: 1 in 76* | **Sephardi (unspecified population(s): ****
Glycogen Storage Disease Type III
A condition associated with a build-up of glycogen in cells, leading to impairment in the function of the liver, muscles and other tissues. Low blood sugar begins in infancy and some affected individuals develop liver failure and muscle weakness later in life. Treatment involves strict diet monitoring. For more information, click here.
Moroccan Jewish: 1 in 37
Glycogen Storage Disease Type VII
A disease associated with the inability of muscle cells are unable to break down the complex sugar glycogen. Severity and symptoms are the disorder can vary, with severest forms causing death before the age of one. Other severe symptoms include low muscle tone, heart disease and breathing difficulties. The late-onset form of the disease is less severe. For more information, click here.
Ashkenazi Jewish: 1 in 100-120*
Infantile Cerebral Cerebellar Atrophy
A severe progressive condition associated with swallowing difficulties, failure to thrive beginning in infancy, microcephaly (small head size) which gets worse over time, high muscle tone, intellectual disability and seizures. Life expectancy is not currently known. For more information, click here.
Bukharian/Kurdish Jewish: 1 in 20*
Joubert Syndrome 2
A disorder characterised by problems with the development of the brain, leading to symptoms such as low muscle tone, developmental delays and intellectual disabilities. Treatment focuses on managing symptoms. For more information, click here.
Ashkenazi Jewish: 1 in 92
Maple Syrup Urine Disease type 1B
A condition associated with the inability to process certain amino acids (the building blocks of proteins). Symptoms include poor feeding, vomiting, lethargy, and developmental delays. Life-threatening if not treated. Prognosis varies depending on symptom severity and early diagnosis. For more information, click here.
Ashkenazi Jewish: 1 in 97
Megalencephalic Leukoencephalopathy with subcortical cysts
A disorder impacting brain development and function, characterised by an enlarged brain with fluid-filled spaces. Other features include irregular muscle coordination, muscle stiffness, developmental delays, intellectual disability and seizures. Prognosis varies between individuals. For more information, click here.
Libyan Jewish: 1 in 40*
Metachromatic Leukodystrophy
A disorder associated with progressive loss of brain functions and motor skills, such as the ability to walk. Incontinence, seizures, paralysis and blindness develop over time. Symptoms can begin in early childhood or

adolescence. In general, the earlier symptoms start, the more rapidly the disease progresses. For more information, click here.
Yemenite Jewish: 1 in 50*
Mitochondrial Complex I Deficiency
A disorder affecting important intracellular structures known as mitochondria. Symptoms begin in first year of life and usually results in death within 2-3 years due to respiratory failure. Other symptoms can include difficulty feeding, failure to thrive, poor muscle tone and problems with movement and balance. For more information, click here.
Jews from Caucasus: **
Mucolipidosis IV~
A neurodegenerative condition that is characterised by significant psychomotor and cognitive development delay, visual impairment, and poor muscle tone. Most reported cases have been severe, with onset in the first year of life, but some milder cases have also been seen. Treatment focusses on managing symptoms and preventing secondary complications. For more information, click here.
Ashkenazi Jewish: 1 in 100
Nemaline Myopathy 2
A disorder primarily affecting skeletal muscle. Muscle weakness which typically worsens over time and can lead to problems with feeding and breathing. There is no cure. Treatment involves managing complications of the disease. The age at which symptoms start, severity of symptoms and the prognosis varies between individuals. For more information, click here.
Ashkenazi Jewish: 1 in 108
Niemann Pick Disease (Type A) ~
A severe neurodegenerative condition in which a harmful amount of a fatty substance accumulates in various organs, causing rapid deterioration and death in infancy. Symptoms appear by about 6 months of age and include loss of brain function and enlargement of the liver and spleen. For more information, click here.
Ashkenazi Jewish: 1 in 115
Omenn Syndrome
A type of severe combined immunodeficiency (SCID), resulting in no immune protection from bacteria, viruses and fungi, leading to persistent, serious infections. Standard treatment is bone marrow or cord blood stem cell transplant. The disorder is fatal if left untreated. For more information, click here.
Iraqi Jewish: **
Polycystic Kidney Disease, Autosomal Recessive
A disorder causing cysts on the kidneys and linked to recurrent infections and high blood pressure. Many infants with the disease die hours or days after birth due to breathing difficulties. Children with the disease require careful monitoring to ensure adequate growth, medication can be used to control high blood pressure. Many individuals require dialysis and kidney transplant as the disease progresses. For more information, click here.
Ashkenazi Jewish: 1 in 107
Primary Ciliary Dyskinesia
A disorder impacting the respiratory system, associated with increased respiratory infections, leading to chronic lung inflammation. Treatment involves managing infections and inflammation, some patients may require lung transplantation. For more information, click here.
Ashkenazi Jewish: 1 in 113*
Progressive Cerebello-Cerebral Atrophy 1
A neurological disorder associated with symptoms begin in first year of life and include intellectual disability, developmental delay, and microcephaly (small head size) which gets worse over time. Many children with the condition live only into infancy or childhood, though some have lived into adulthood. Also known as pontocerebellar hypoplasia type 2D. For more information, click here.
Moroccan/Iraqi Jewish: 1 in 44*
Progressive Cerebello-Cerebreal Atrophy 2

A neurodegenerative disorder beginning in infancy, characterised by intellectual disability, microcephaly (small head size) which gets worse over time, and seizures. Also known as pontocerebellar hypoplasia type 2E. For more information, click here.
Moroccan Jewish: 1 in 37*
Smith-Lemli-Opitz Syndrome
A condition associated with an impairment in the body's ability to make cholesterol leading to hypotonia, failure to thrive and birth defects such as cleft palate. There is currently no cure, but cholesterol supplements may improve symptoms. Prognosis depends on severity of symptoms. For more information, click here.
Ashkenazi Jewish: 1 in 36
Spinal Muscular Atrophy
A severe neurodegenerative disorders associated with progressive degeneration of cells in the spinal cord and brainstem, resulting in muscle weakness and degeneration of muscle mass. Life expectancy varies, but severe forms of the disorder can cause fatality in infancy or childhood. Genetic testing for SNM1 can be extremely complex. For more information, click here.

General population: 1 in 54	**Ashkenazi Jewish: 1 in 67**

Tay Sachs Disease~
A severe progressive neurological disorder associated with loss of brain functioning in infancy, which gets worse over time and usually leads to death in early childhood. Symptoms include muscle weakness, seizures, and blindness. For more information, click here.

Ashkenazi Jewish: 1 in 25	**Moroccan/Iraqi Jewish: 1 in 110-140***

Tryosinemia Type 1
A condition characterized by a build-up of an amino acid called tyrosine in the blood. This leads to failure to thrive, softening of bones, liver and kidney disease and problems with the nervous system. If diagnosed early symptoms can be managed by diet restrictions. For more information, click here.
Ashkenazi Jewish: 1 in 150
Usher Syndrome Type IIIA
A disorder characterised to progressive hearing loss from childhood or early teens, requiring hearing aids by mid-to-late adulthood. Night blindness begins during adolescence and progresses to severe vision loss by midlife. Chance of balance problems later in life. For more information, click here.
Ashkenazi Jewish: 1 in 120
Usher Syndrome Type 2
A disorder characterised by moderate to severe hearing loss at birth. Hearing aids are usually beneficial. Decreased night vision occurs by adolescence, progressing to severe vision loss by midlife. For more information, click here.

Ashkenazi Jewish: 1 in 35*	**Iranian Jewish: 1 in 60***	**Iraqi Jewish: 1 in 50-100***

Usher Syndrome Type IF
A condition associated with profound hearing loss or deafness from birth. Hearing aids are usually not beneficial, but may be a candidate for cochlear implants. Decreased night vision starts by age 10, progressing to severe vision loss by midlife. Symptoms also include severe balance problems from birth. For more information, click here.
Ashkenazi Jewish: 1 in 72
Walker-Warburg Syndrome
A severe neurological disorder associated with a smooth brain surface, eye abnormalities, feeding difficulties, seizures, low muscle tone and intellectual disability. Usually fatal in the first year of life. Also referred to as muscular dystrophy-dystroglycanopathy type A4. For more information, click here.
Ashkenazi Jewish: 1 in 150
Wolman Disease/Cholesteryl Ester Storage Disease
A severe disorder characterised by the body's inability to properly breakdown and use fats and cholesterol. Therefore, fats and cholesterol build up, particularly in the liver, leading to liver and spleen enlargement, low muscle town, vomiting, diarrhoea, developmental delay, poor absorption of nutrients from food. For more information, click here.
Iranian Jewish: 1 in 27-45*

Zellweger Syndrome Spectrum (PEX6 related)	
Spectrum of disorders, caused by degeneration of nerve cell coverings, with overlapping signs and symptoms such as low muscle town, feeding problems, hearing and vision loss, seizures, skeletal abnormalities and distinctive facial abnormalities. The severe form of the disease is typically fatal in infancy. Milder forms progress slower and can be variable, but life expectancy is still reduced. For more information, click here.	
Yemenite: 1 in 18*	
Zellweger Syndrome Spectrum (PEX2 related)	
Spectrum of disorders, caused by degeneration of nerve cell coverings, with overlapping signs and symptoms such as low muscle town, feeding problems, hearing and vision loss, seizures, skeletal abnormalities and distinctive facial abnormalities. The severe form of the disease is typically fatal in infancy. Milder forms progress slower and can be variable, but life expectancy is still reduced. For more information, click here.	
Ashkenazi Jewish: 1 in 123	
Fragile X Syndrome ***	
An X-linked disorder and the most common inherited cause of intellectual disability. The severity of intellectual disability varies between individuals and is generally more severe in males. Life span is usually normal. For more information, click here.	
Ashkenazi Jewish: 1 in 115	***General Population: 1 in 151***

~disorder was covered in Jnetics test prior to launch of new test in September 2021

*exact carrier frequency unknown but condition described is some Jewish families from named population

**disorder is severe and often included in Jewish carrier tests but carrier data is sparse.

***a variable and complex X-linked recessive disorder- opt in available for women participating in The Jnetics Clinic