SUMMARY TABLE OF GENETIC DISORDERS PREVALENT AMONG PEOPLE OF ASHKENAZI JEWISH ANCESTRY

Disorder	Mode of Inheritance	Description	Carrier Frequency[1] among Ashkenazi Jews
Bloom syndrome	Autosomal recessive[2]	A condition characterized by poor growth, increased skin sensitivity to sunlight, infections and a predisposition to cancer. Shortened lifespan – usually before the age of 30 and cancer related.	1 in 100
Breast and ovarian cancer (BRCA 1 & 2)[3]	Autosomal dominant[4] (with reduced penetrance[5])	A disease in which cells grow and reproduce themselves abnormally causing cancerous tumours (in the breast area for breast cancer and in one or both ovaries for ovarian cancer). These tumours spread to other sites in the body where they begin to grow and replace normal tissue, forming new tumours.	1 in 40
Canavan disease	Autosomal recessive	A disorder where affected children show normal early development but then experience progressive neurological deterioration of physical and mental capabilities. Shortened lifespan (typically fatal between early childhood through to late teens).	1 in 40-57
Non-Classical Congenital Adrenal Hyperplasia	Autosomal recessive	This is a hormonal disorder characterized by early signs of puberty namely excessive hair growth, increase in height and acne. It can also involve decreased fertility as well as menstrual problems in females. The non-classical form is much milder than the classical form of CAH, and can be treated effectively using steroid hormones.	1 in 3
Cystic fibrosis[3]	Autosomal recessive	A progressive disorder that causes the body to produce thick, sticky mucus in the lungs and digestive system. Symptoms vary but may include frequent respiratory infections, poor weight gain, and progressive lung damage. Shortened lifespan (typically fatal around the age of 40).	1 in 25-29
DYT1 generalised dystonia	Autosomal dominant[4] (with reduced penetrance[5])	A neurological movement disorder characterised by sustained and involuntary muscle contractions or muscle spasms. It usually has an early onset (children or young adults), with abnormal muscle contractions beginning in the lower or upper limbs and then often spreading to other parts of the body, causing physical disability, often with associated pain. Sympton severity varies and treatments are available. Lifespan is not affected.	1 in 1000 - 3000
Factor XI deficiency	Autosomal recessive (that is partially dominant[6])	A bleeding disorder characterized by easy bruising, and abnormal bleeding after dental work, surgery, and injury. In women it can also cause heavy or prolonged bleeding during menstrual periods and after childbirth. Although symptoms vary widely, the condition is usually mild and can be treated effectively.	1 in 12
Familial dysautonomia	Autosomal recessive	A progressive disorder that causes the autonomic and sensory nervous systems to malfunction affecting a range of bodily functions including temperature and blood pressure regulation, swallowing, tear production, pain sensitivity and response to stress. Shortened lifespan (50% live to 40 years).	1 in 30

Disorder	Mode of Inheritance	Description	Carrier Frequency[1] among Ashkenazi Jews
Familial Mediterranean fever	Autosomal recessive	A disease characterized by recurring episodes of painful inflammation in the abdomen, chest, or joints. It occurs predominantly in Sephardic Jews, though people of Ashkenazi Jewish origin can also be affected – often with a milder form of the condition. Symptoms can be easily treated in the vast majority of patients.	Varies among Ashkenazi and Sephardi populations From 1 in 5 to 1 in 135
Fanconi anaemia (type C)	Autosomal recessive	A disorder characterised by a reduced production of all types of blood cells and an increased risk of cancer. Symptoms include birth defects, short stature, skin discolouration, fatigue, hearing loss and reduced fertility. Shortened lifespan (typically fatal by 30 years).	1 in 89
Gaucher disease (type 1)	Autosomal recessive	Lysosomal storage disorder caused by an enzyme deficiency that can result in anaemia, low platelet count, easy bruising and bleeding, some form of bone disease and enlarged liver/spleen. Range and severity of symptoms varies greatly and can be treated. Lifespan tends to be unaffected.	1 in 10-15
Glycogen Storage disease (type 1A)	Autosomal recessive	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.	1 in 70
Mucolipidosis IV	Autosomal recessive	A progressive disease affecting the brain and nervous system and characterised by significant delays in motor and cognitive development, low muscle tone, and progressive visual problems. Mild to severe progressive developmental delay – shortened lifespan (ranging from 1-45 years).	1 in 100 - 125
Niemann-Pick disease (type A)	Autosomal recessive	A progressive disorder caused by a deficiency of an enzyme responsible for breaking down a specific fat in the body. Fat accumulates in various organs, brain and nervous system causing rapid neurological decline. Shortened lifespan (typically fatal by 2-5 years).	1 in 90
Tay-Sachs disease	Autosomal recessive	A neurodegenerative disorder characterised by normal birth and development until 3-6 months then rapid and progressive brain and nervous system deterioration. Classical infantile form has a shortened lifespan (typically fatal by age 4).	1 in 25 - 30

Notes

1. Carrier frequency is the number that describes how many people in a population are carriers (have one altered copy of a gene and one normal copy of the same genes) for a genetic disorder.

2. Recessive disorders require both parents to carry a specific disorder-related mutation and pass it on to their child for the disorder to be inherited.

3. Cystic fibrosis and breast/ovarian cancer (BRCA 1 & 2) have specific mutations that are more prevalent among Ashkenazi Jews. Cystic fibrosis, however, is no more common in the Jewish community than in the general population; and breast/ovarian cancer (BRCA 1 & 2) may be only slightly more common among people of Ashkenazi origin.

4. Dominant disorders require only one copy of a specific disorder-related mutation to be passed onto their child for the disorder to be inherited.

5. A disorder has reduced penetrance when only a percentage of people with the mutation will ever develop the disorder.

6. This means that a person can show symptoms even when they inherit only one abnormal factor XI gene, however, they are likely to be affected more if they inherit two abnormal factor XI genes, one from each parent.