The Jnetics Clinic - Information Sheet

WHAT ARE JEWISH GENETIC DISORDERS (JGDs)?

These are genetic disorders which are more common in people who have Jewish ancestry relative to those in the general population. Tay-Sachs disease is the best-known example of a JDG, but research has identified a number of other serious disorders that can potentially be prevented through carrier screening.

WHICH JEWISH GENETIC DISORDERS ARE COVERED IN JNETICS' CARRIER SCREENING?

The Jnetics screening covers 48 of the most severe recessive genetic disorders that are relatively more common in either Ashkenazi or Sephardi/Mizrahi Jews. This total includes a common X-linked recessive disorder, Fragile X Syndrome, for which testing is optional for female clinic participants. A full list of the disorders covered by Jnetics screening can be found overleaf.

WHAT IS CARRIER SCREENING?

Carrier screening gives people the opportunity to find out if they are a 'carrier' of a genetic condition. **Carriers of recessive disorders are typically not affected themselves but are at increased risk of passing on a genetic condition if their partner is also a carrier of the same disorder.** Screening involves taking a saliva sample and testing it to see if there is a 'mutation' (a genetic fault or alteration) for a particular genetic disorder.

WHY HAVE CARRIER SCREENING?

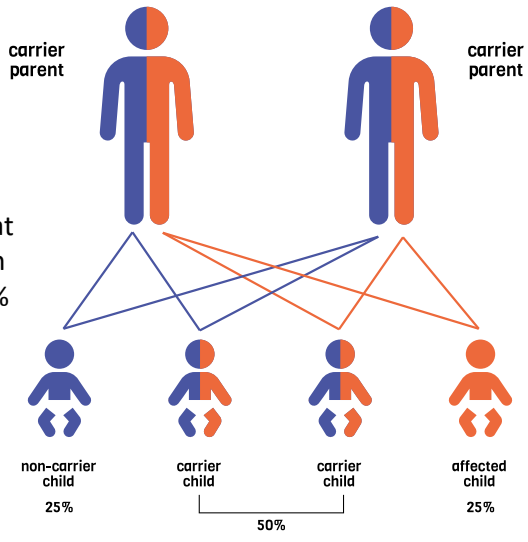
While Jewish genetic disorders vary in severity, those covered by Jnetics can all be severely debilitating and most cause shortened lifespan. At this time most lack any effective treatment but can be prevented through effective carrier screening. Though rare, these conditions are devastating and can typically occur even when there is no known family history.

1 in 5 people of Jewish descent are carriers of *at least one* of the main Jewish genetic disorders.

Unless screened carriers often go undetected until it is too late. The best time to screen for these conditions is before starting (or growing) a family as it gives people the greatest number of reproductive options available should they be at risk of having an affected child.

HOW ARE GENETIC DISORDERS PASSED ON?

The disorders covered in this screening programme (except for Fragile X Syndrome) follow an autosomal recessive inheritance pattern, shown in the diagram. This means that two copies of an altered gene must be present for the disorder to develop. The diagram demonstrates that if both parents are carriers of the same disorder (they each carry a single altered gene), then each child will have a 25% chance of inheriting two copies of the altered gene and being affected by the disorder. For every pregnancy, there is also a 50% chance that the child will be a carrier but not develop the genetic condition themselves.



WHO IS ELIGIBLE FOR JNETICS CARRIER SCREENING?

Testing is available for anyone aged 16 who has at least one Jewish grandparent.

DISORDERS COVERED BY THIS SCREENING TEST (see overleaf)

The disorders are listed below with at risk Jewish populations and associated carrier frequencies in brackets. Common symptoms and outlook of the disorders can be found on our website. 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.

ABCC8-related Hyperinsulinism	
Ashkenazi Jewish: 1 in 44	
Asparagine Synthetase Deficiency	
Iranian Jewish: 1 in 80*	
Acute Infantile Liver Failure	
Yemenite Jewish: 1 in 34	
Bardet-Biedl Syndrome	
Ashkenazi Jewish: 1 in 107	
Bloom Syndrome~	
Ashkenazi Jewish: 1 in 134	
Canavan Disease~	
Ashkenazi Jewish: 1 in 55	
Carnitine Palmitoyltransferase II Deficiency	
Ashkenazi Jewish: 1 in 51	
Congenital Amegakaryocytic Thrombocytopenia	
Ashkenazi Jewish: 1 in 55	
Congenital Disorder of Glycosylation Ia	
Ashkenazi Jewish: 1 in 57	
Costeff Optic Atrophy Syndrome	
Iraqi Jewish: 1 in 50	
Cystic Fibrosis~	
Ashkenazi Jewish: 1 in 24	General population: 1 in 32
Cystinosis	
Moroccan Jewish: 1 in 100*	
Dihydrolipamide Dehydrogenase Deficiency	
Ashkenazi Jewish: 1 in 107	
Familial Dysautonomia~	
Ashkenazi Jewish: 1 in 31	
Fanconi Anaemia C~	
Ashkenazi Jewish: 1 in 99	
Fanconi Anemia, Complementation group A	
Moroccan Jewish: 1 in 100*	Indian Jewish: 1 in 27*
Fumarase Deficiency	
Ashkenazi Jewish: 1 in 99*	
Galactosemia	
Ashkenazi Jewish: 1 in 127	
Glycogen Storage Disease 1a~	
Ashkenazi Jewish: 1 in 64	
Glycogen Storage Disease Type II/Pompe Disease	
Ashkenazi Jewish: 1 in 76*	Sephardi (unspecified population(s): **
Glycogen Storage Disease Type III	
Moroccan Jewish: 1 in 37	
Glycogen Storage Disease Type VII	
Ashkenazi Jewish: 1 in 100-120*	
Infantile Cerebral Cerebellar Atrophy	
Bukharian/Kurdish Jewish: 1 in 20*	
Joubert Syndrome 2	
Ashkenazi Jewish: 1 in 92	
Maple Syrup Urine Disease type 1B	
Ashkenazi Jewish: 1 in 97	

Megalencephalic Leukoencephalopathy with subcortical cysts	
Libyan Jewish: 1 in 40*	
Metachromatic Leukodystrophy	
Yemenite Jewish: 1 in 50*	
Mitochondrial Complex I Deficiency	
Jews from Caucasus: **	
Mucolipidosis IV~	
Ashkenazi Jewish: 1 in 100	
Nemaline Myopathy 2	
Ashkenazi Jewish: 1 in 108	
Niemann Pick Disease (Type A) ~	
Ashkenazi Jewish: 1 in 115	
Omenn Syndrome	
Iraqi Jewish: **	
Polycystic Kidney Disease, Autosomal Recessive	
Ashkenazi Jewish: 1 in 107	
Primary Ciliary Dyskinesia	
Ashkenazi Jewish: 1 in 113*	
Progressive Cerebello-Cerebral Atrophy 1	
Moroccan/Iraqi Jewish: 1 in 44*	
Progressive Cerebello-Cerebral Atrophy 2	
Moroccan Jewish: 1 in 37*	
Smith-Lemli-Opitz Syndrome	
Ashkenazi Jewish: 1 in 36	
Spinal Muscular Atrophy	
General population: 1 in 54	Ashkenazi Jewish: 1 in 67
Tay Sachs Disease~	
Ashkenazi Jewish: 1 in 25	Moroccan/Iraqi Jewish: 1 in 110-140*
Tryosinemia Type 1	
Ashkenazi Jewish: 1 in 150	
Usher Syndrome Type IIIA	
Ashkenazi Jewish: 1 in 120	
Usher Syndrome Type 2	
Ashkenazi Jewish: 1 in 35*	Iranian Jewish: 1 in 60*	Iraqi Jewish: 1 in 50-100*
Usher Syndrome Type IF	
Ashkenazi Jewish: 1 in 72	
Walker-Warburg Syndrome	
Ashkenazi Jewish: 1 in 150	
Wolman Disease/Cholesteryl Ester Storage Disease	
Iranian Jewish: 1 in 27-45*	
Zellweger Syndrome Spectrum (PEX6 related)	
Yemenite: 1 in 18*	
Zellweger Syndrome Spectrum (PEX2 related)	
Ashkenazi Jewish: 1 in 123	

Fragile X Syndrome ***	
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. Only available for opt-in by women participating via the Jnetics Clinic.