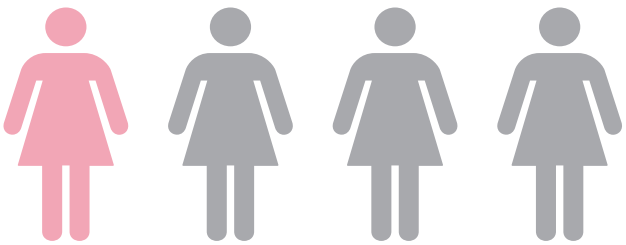
CARRIER SCREENING FOR ASHKENAZI JEWISH DISEASES

Ashkenazi Jewish carrier screening helps to identify carriers for inherited genetic conditions that occur frequently in individuals with Eastern European (Ashkenazi) Jewish ancestry.



According to The American College of Medical Genetics (ACMG), **1 in 4 Ashkenazi Jews is a carrier** of a genetic disorder.[1]

GenPath's Expanded Ashkenazi Jewish carrier panel screens for 47 disorders.The clinical presentations of the disorders on the panel are:

- Mostly severe, often with childhood onset and are inherited in an autosomal recessive manner.
- For some, clinically managed treatment can significantly improve quality of life.

Reasons for testing

- Testing will identify carriers of a genetic disorder, who are typically healthy individuals who show no sign of disease.
- Carriers, although healthy, can have children with the disease and its symptoms, if the partner is also a carrier.
- Some diseases cause physical and mental impairment, have a shortened lifespan or require lifelong treatment and management.

As Technology Evolves, So Does Our Carrier Testing

P659 Ashkenazi Jewish 18 Profile
(with carrier frequency rates)

Bloom Syndrome - 1/107	Joubert Syndrome - 1/92
Canavan Disease - 1/55	Maple Syrup Urine Disease (Type 1A & 1B)- 1/80
Cystic Fibrosis (Expanded CF) 1/24	Mucolipidosis (Type IV) - 1/96
Dihydrolipoamide Dehydrogenase Deficiency - <1/80	Nemaline Myopathy 2 - <1/108
Familial Dysautonomia - <1/52	Niemann-Pick Disease (Type A & B) - 1/90
Familial Hyperinsulinism - 1/67	Tay-Sachs Disease - 1/28
Fanconi Anemia, Type C - 1/127	Usher Syndrome Type 1F - 1/140
Gaucher Disease - 1/15	Usher Syndrome Type 3 - 1/140
Glycogen Storage Type IA (GSDIA) 1/71	Walker-Warburg Syndrome - 1/144

J293 Expanded Ashkenazi Jewish Profile
(Includes Ashkenazi Jewish 18 disorders plus below tests with carrier frequency rates)

Abetalipoproteinemia - 1/131	Hermansky-Pudlak Syndrome 3 1/235
Alport Syndrome, Autosomal Recessive - 1/183	Mitochondrial Complex I Deficiency - 1/291
Arthrogryposis, Mental Retardation, & Seizures - 1/373	Multiple Sulfatase Deficiency 1/320
Bardet-Biedl Syndrome 2 - 1/136	Osteopetrosis, Autosomal Recessive 1 - 1/350
Carnitine Palmitoyltransferase Deficiency, Type 2 - 1/43	Peroxisome Biogenesis Disorder 5A (Zellweger) - 1/123
Ciliary Dyskinesia, Primary 1 - 1/357	Phenylketonuria (PKU) - 1/225
Coenzyme Q10 Deficiency, Primary 7 - 1/151	Phosphoglycerate Dehydrogenase Deficiency - 1/280
Congenital Amegakaryocytic Thrombocytopenia (CAMT) - 1/55	Polycystic Kidney Disease, Autosomal Recessive (ARPKD) 1/108
Deafness, Autosomal Recessive 1A (GJB2) - 1/21	Retinitis Pimentosa 59 - 1/120
Dyskeratosis Congenita, Autosomal Recessive 5 - 1/204	Smith-Lemli-Opitz Syndrome 1/37
Ehlers-Danos Syndrome, Type VIIC - 1/501	Spastic Tetraplegia, Thin Corpus Callosum & Progressive Microcephaly - 1/118
Factor XI Deficiency (Hemophilia C) - 1/11	Spinal Muscular Atrophy (SMA) 1/41
Familial Hypercholesterolemia, Homozygous, LDLR-Associated 1/130	Tyrosinemia, Type 1 - 1/100
Familial Mediterranean Fever - 1/5	Wilson Disease - 1/100
Galactosemia - 1/127	

1. **Inheritance**
   Autosomal recessive
2. **Methodology**
   Genotyping by Next-generation sequencing (NGS)
3. **Test Code**
   P659-Ashkenazi Jewish 18 Profile
   J293-Expanded Ashkenazi Jewish Profile
4. **TAT**
   12-14 days
5. **Requirements**
   One full EDTA (Lavender top) tube or Oral rinse

Reference:

1. Carrier screening in Individuals of Ashkenazi Jewish Descent. ACMG Practice Guidelines. American College of Medical Genetics, and Genomics. Genet Med 2008 10:1:54-56.

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