Preconception screening is covered by most insurance companies. Find guidance on what questions to ask your provider about coverage at atljewishgenescreen.org.

We recommend screening before becoming pregnant and before every subsequent pregnancy as testing for new diseases becomes available.

There are three ways to get screened:

- **Your physician or OB/GYN.**
- **Karen Grinzaid, MS, CGC, Atlanta Jewish Gene Screen genetic counselor, at Karen@atljewishgenescreen.org.**
- **Atlanta Jewish Gene Screen community screenings. Find an upcoming screening in your area at atljewishgenescreen.org.**

Please also visit **www.jewishgeneticanswers.org** to ask genetics experts about Jewish genetic diseases and how they can be prevented.

Download the **FREE Gene Screen App** on **iTunes**.
Find us on Facebook and Twitter.

atlanta jewish gene screen

www.atljewishgenescreen.org
404 778 8516
Karen@atljewishgenescreen.org

1 in 5

Ashkenazi Jews

is a carrier

for at least one

of 19 current,

preventable

genetic diseases...

a glossary

Disease	Approx. Carrier Frequency	Symptoms	Current Treatment Available
Bloom Syndrome	1 in 100	Poor growth, frequent infections, childhood leukemia, lymphoma, gastrointestinal cancers. Lifespan limited.	Chemotherapy provided for cancers and transfusions may be needed for low cell counts, antibiotics for infections. No cure available.
Canavan Disease	1 in 40-57	Progressive neurologic deterioration, large head, seizures, lifespan often limited to childhood.	Medications can reduce seizures and provide comfort. No cure available. Gene therapy research ongoing.
Cystic Fibrosis	1 in 26*	Lung disease, failure to thrive, normal IQ, variable severity with most requiring lifelong medical care and reduced life expectancy	Antibiotics can reduce/treat lung infections. Pancreatic enzymes can help alleviate gastrointestinal symptoms. Lung transplant may be required. No cure available.
Dihydrolipoamide Dehydrogenase Deficiency (DLD)	1 in 96	Episodes of recurring vomiting and abdominal pain with lactic acidosis, seizures, hypoglycemia, neurologic impairment. Blindness, coma, intellectual decline if untreated.	Special formula and low protein diet can limit symptoms. Hospitalizations often required. No cure available.
Familial Dysautonomia (FD)	1 in 30	Nervous system dysregulation (vomiting, sweating, decreased pain sensitivity, abnormal regulation of blood pressure and temperature), learning disabilities, variable life expectancy.	Medication and supportive care available to reduce symptoms and manage illnesses. No cure available.
Familial Hyperinsulinism (FHI)	1 in 66	Mild to severe low blood sugar if left untreated causing seizures which can lead to coma and intellectual disability.	Medications can be used to increase blood sugar. Surgical resection of part of the pancreas may be required. No cure available.
Fanconi Anemia type C	1 in 89	Congenital malformations of thumbs, kidneys, bone marrow failure, childhood leukemia, intellectual disability, lifespan limited.	Chemotherapy provided for cancers and transfusions may be needed for low cell counts. Hematopoietic stem cell transplant possible.
Gaucher Disease type 1	1 in 15	Anemia, low platelets, nosebleeds, bone pain and fractures, normal IQ, variable severity, onset in early childhood to adulthood.	IV Enzyme therapy replaces missing enzyme. Oral therapy available. Alleviates symptoms, not a cure.
Glycogen Storage Disease type 1A (GSD1A)	1 in 71	Hypoglycemia, hepatomegaly, seizures, kidney disease, bleeding, intellect can be affected by frequency of seizures.	Frequent feedings of glucose can prevent low blood sugar and seizures. Possible enzyme therapy soon. Liver transplant available.
Joubert Syndrome	1 in 92	Hypotonia, abnormal rapid breathing, rotary nystagmus, variable developmental delay.	No treatment at this time. No cure available.
Maple Syrup Urine Disease (MSUD)	1 in 81	Intellectual disability, seizures, coma with uncontrolled diet. Disease exacerbated with acute illness.	Special formula and low protein diet can limit symptoms. Hospitalizations may be required. Liver transplant available.
Mucolipidosis IV	1 in 122	Psychomotor retardation, loss of motor skills, and retinal degeneration leading to blindness. Severity is variable and lifespan may be limited to early adulthood.	Medications can reduce symptoms and provide comfort. No cure.
Nemaline Myopathy (NM)	1 in 108	Weakness and poor muscle tone. Respiratory failure can lead to early death.	No treatment at this time. No cure.
Niemann-Pick Disease type A	1 in 90	Poor growth, liver failure, blindness, and progressive neurologic decline. Lifespan limited to early childhood.	Medications can reduce symptoms and provide comfort. No cure available.
Spinal Muscular Atrophy	1 in 41*	Progressive muscle weakness, normal IQ, variable expression with onset from infancy to adulthood. Lifespan limited in early onset disease.	Some children require mechanical ventilation. Medications can reduce symptoms and provide comfort. No cure available.
Tay-Sachs Disease	1 in 25	Normal at birth with progressive neurologic deterioration, loss of sight and hearing, seizure, lifespan limited to early childhood. Late onset form may be detected.	Medications can reduce seizures and provide comfort. No cure available.
Usher Syndrome type 1F	1 in 141	Congenital deafness and progressive blindness. Motor delays.	Visual aids, hearing aids and cochlear implants for improved vision and hearing. No cure.
Usher Syndrome type III	1 in 107	Progressive hearing loss (moderate to severe) and vision loss with blindness by adulthood. Normal IQ. Variable severity.	Visual aids, hearing aids and cochlear implants for improved vision and hearing. No cure.
Walker-Warburg Syndrome	1 in 149	Congenital muscular dystrophy; brain and eye malformations causing seizures and blindness. Lifespan limited to early childhood.	No treatment at this time. No cure.

** CF and SMA occur at the same frequency in the Ashkenazi and non-Ashkenazi Jewish population.*