PREMIER GENOMICS

JEWISH GENETIC DISEASES CARRIER SCREENING

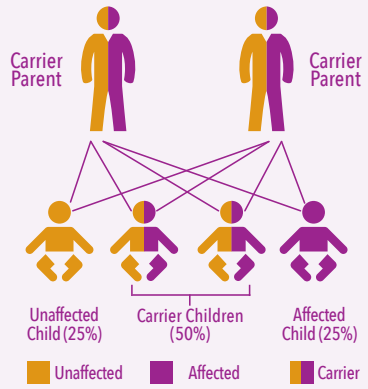
1 in 3 Ashkenazi Jews is a carrier of at least one Jewish genetic disease that may threaten infant health.

The American Congress of Obstetricians and Gynecologists (ACOG) recommends that individuals of Eastern European Jewish (Ashkenazi) ancestry be offered carrier screening for certain Jewish genetic diseases. Jewish genetic diseases are autosomal recessive disorders. When both parents are carriers of a specific genetic disease, there is a 1 in 4 (or 25%) chance with each pregnancy that they will have an affected child.

Screening results inform patients of their carrier status and can help guide decision-making for patients who are pregnant, planning a pregnancy, or considering fertility options.

Genetic carrier screening among Jewish and interfaith couples can be an important and empowering family planning tool for patients and their consulting physicians.

Autosomal Recessive Inheritance



Disease	Carrier frequency*	Detection rate*	Residual risk*	Clinical characteristics	Clinical considerations	ACOG[1]	ACMG[2,3]
BLOOM SYNDROME	1 in 134	95%	1 in 1980	Growth problems, low body fat, short stature, reflux, increased infections, facial rash, learning disabilities, lung problems, increased cancer risk, infertility in men, reduced fertility in women		✓	✓
CANAVAN DISEASE	1 in 55	99%	1 in 4900	Muscle weakness, developmental delay, mental disability due to destruction of the myelin sheath		✓	✓
CYSTIC FIBROSIS	1 in 23	96%	1 in 651	Breathing problems, lung infections due to the production and accumulation of thick mucus in organs		✓	✓
FAMILIAL DYSAUTONOMIA	1 in 31	99%	1 in 3100	Identified in newborn, poor muscle tone, nerve cell deterioration is progressive		✓	✓
FANCONI ANEMIA GROUP C	1 in 100	99%	1 in 8800	Impaired bone marrow, short stature, physical and organ malformations, leukemia and other cancer risks		✓	✓
GAUCHER DISEASE TYPE 1	1 in 15	90%	1 in 140	Bone pain, fracture, bruising, enlarged liver and spleen, anemia		✓	✓
MUCOLIPIDOSIS IV	1 in 89	95%	1 in 2500	Condition affecting nerve development, mobility, vision, speech		✓	✓
NIEMANN-PICK DISEASE TYPE A	1 in 115	95%	1 in 1780	Lysosomal storage disease; enlarged liver and spleen, growth and developmental delay, low muscle tone, rigidity, lung disease		✓	✓
TAY-SACHS DISEASE	1 in 27	92%	1 in 480	Progressive neurological disorder; muscle weakness, cherry red spot in the eye, seizures, blindness, early death; in a small number, less severe, presenting in adulthood		✓	✓

Abbreviation: ACMG, American College of Medical Genetics and Genomics.
*For Ashkenazi Jewish population only.

Condition affects life expectancy

Early intervention may improve outcomes

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JEWISH GENETIC DISEASES TESTING PROCESS

THE PROCESS	The DNA collection process is simple and requires only an in-office buccal (cheek) swab with our sample collection kit. UPS will pick up your patient samples and deliver them directly to our lab (labels and instructions are included in each kit). Dedicated account services are always available to answer questions and manage requests.
THE TURN-AROUND TIME	10 to 15 days
THE REPORT	Upon completion of DNA extraction and evaluation at our advanced laboratory facility, a comprehensive report is generated and uploaded to a secure portal with dedicated physician log-in and downloading capabilities. Physician-to-physician consultation is also available with our Medical Director or our Genetic Counselor. Monograph is available upon request.

CONTACT YOUR REPRESENTATIVE OR REACH US DIRECTLY AT OUR EMAIL ADDRESS BELOW TO OBTAIN SAMPLE TEST REPORTS AND DNA SAMPLE COLLECTION KITS FOR THIS OR ANY OF OUR OTHER ADVANCED GENETIC TESTING PANELS.

Jewish Genetic Diseases Carrier Screening	*BRCA1/2* Genetic Testing
Fragile X Syndrome Genetic Screening	Hereditary Colon Cancer/Lynch Syndrome Genetic Testing
Cystic Fibrosis Genetic Carrier Screening	Pharmacogenetics Testing

ABOUT PREMIER GENOMICS

Premier Genomics is committed to advancing the field of personalized genetic medicine by offering cutting-edge genetic screening services to help practitioners and their patients in pursuit of tailored treatment and optimized, personalized health care. We work together with patients and their insurance providers to help ensure that access to these important genetic tests does not icause patients financial hardship.

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Premier Genomics | www.PremierGx.com | dnatest@premiergx.com
250 Ed English Drive, Building 3, Unit D, Shenandoah, TX 77385
CLIA-accredited, CAP-certified clinical laboratory

References: 1. American Congress (formerly College) of Obstetricians and Gynecologists (ACOG) Committee on Genetics. ACOG Committee Opinion No. 442: Preconception and prenatal carrier screening for genetic diseases in individuals of Eastern European Jewish descent. *Obstet Gynecol.* 2009;114(4):950-953. **2**. Gross SJ, Pletcher BA, Monaghan KG; Professional Practice and Guidelines Committee. American College of Medical Genetics and Genomics (ACMG) Practice Guidelines: Carrier screening in individuals of Ashkenazi Jewish descent. *Genet Med.* 2008;10(1):54-56. **3.** Watson MS, Cutting GR, Desnick RJ, et al. Cystic fibrosis population carrier screening: 2004 revision of American College of Medical Genetics mutation panel [published corrections appear in *Genet Med.* 2004;6(6):548 and *Genet Med.* 2005;7(4):286]. *Genet Med.* 2004;6(5):387-391.