

More Than Results

Jewish Heritage Carrier Screening

The term Jewish Heritage disorders refers to a broad group of genetic disorders that are found at greater incidence, compared to the general population, among individuals of Jewish descent. Ashkenazi Jews can trace their ancestry to Western, Central, and Eastern Europe, and are more likely to be carriers of certain disease-associated genes.¹

The common genetic disorders often considered for Jewish heritage carrier screening during pregnancy include

- **Tay-Sachs disease** – *a neurodegenerative disease often resulting in death by age 4.*²
- **Canavan disease** – *a progressive leukodystrophy often resulting in death by age 10.*^{3,4}
- **Cystic fibrosis** – *a disorder of pulmonary and pancreatic function.*^{5,6}
- **Familial dysautonomia** – *a sensory neuropathy characterized by gastrointestinal and cardiac dysfunction; about half of patients survive beyond age 30.*^{7,8}
- **Fanconi anemia** – *characterized by pancytopenia, congenital malformations, and a predisposition to malignancy.*⁹
- **Niemann-Pick disease** – *a neurodegenerative disease often resulting in death by age 3.*¹⁰
- **Gaucher disease** – *Type I-adult onset, characterized by bone and organ disease. Treatment is available. Types II and III are characterized by onset in infancy or childhood and also involve the central nervous system.*¹¹

- **Bloom syndrome** – *associated with small stature and a predisposition to malignancy from chromosome breakage.*¹²
- **Mucopolysaccharidosis Type IV** – *a disease of psychomotor retardation and ophthalmological abnormalities. Prognosis and life expectancy beyond age 40 are unavailable.*¹³

The American College of Obstetricians and Gynecologists (ACOG) and the American College of Medical Genetics (ACMG) recommend screening for gene carrier detection for Tay-Sachs, Canavan disease, and cystic fibrosis for couples in which at least one member is Ashkenazi Jewish, and the couple is planning a pregnancy or is pregnant.¹⁴ In addition to these tests, ACOG recommends that couples of Ashkenazi Jewish ancestry be offered carrier screening for familial dysautonomia.¹⁵ ACOG recognizes that some Ashkenazi Jewish individuals may be interested in additional carrier screening and highly sensitive tests are available for Fanconi anemia, Niemann-Pick A disease, Bloom syndrome, mucopolysaccharidosis IV, and Gaucher disease.¹⁵ All of these inherited diseases are autosomal-recessive, meaning both parents must be carriers to have an affected child. When both parents are carriers, there is a 25% risk with each pregnancy that the baby will inherit both genes and be affected.¹⁴

For more information about carrier screening and Jewish heritage, call 800-345-GENE.

Available Jewish Heritage Screening Tests

Disorder	Test Number	Ashkenazi Jewish Carrier Frequency 2,4,5,8,9,10,11,12,13	Ashkenazi Jewish Detection Rate 2,4,6,8,9,10,11,12,13	Specimen Requirements (See <i>Directory of Services</i> for full details.)
Tay-Sachs Disease* Enzyme, Leukocytes	511246	1/30	>99%	10 mL whole blood, yellow-stopper (ACD-A) tube
Tay-Sachs Disease Enzyme, serum (Not for use in pregnant women and those taking birth control pills)	510412	1/30	>99%	3 mL frozen serum
Tay-Sachs Disease DNA mutations: 1278TATC, 1421+G→C, G269S, R247W	510404	1/30	>95%	7 mL whole blood, lavender-stopper (EDTA) tube
Canavan Disease DNA mutations: Y231X, E285A	511147	1/40	>97%	7 mL whole blood, lavender-stopper (EDTA) tube
Cystic Fibrosis** DNA mutations (listed below)	480533	1/25	>97%	7 mL whole blood, lavender-stopper (EDTA) tube or buccal swab
Ashkenazi Jewish Carrier Panel (includes Tay-Sachs leukocytes, Canavan, CF, and familial dysautonomia)	333561	(See individual tests.)	(See individual tests.)	18 mL whole blood, yellow-stopper (ACD-A) tube and whole blood, lavender-stopper (EDTA) tube
Familial Dysautonomia DNA mutations: IVS20(+6T→C), R696P	511352	1/30	>99.5%	7 mL whole blood, lavender-stopper (EDTA) tube
Fanconi Anemia Type C DNA mutation IVS+4A→T	511212	1/89	>99%	7 mL whole blood, lavender-stopper (EDTA) tube
Niemann-Pick Disease (Types A and B) DNA mutations: L302P, R496L, fsP330, ΔR608	511329	1/90	>95%	7 mL whole blood, lavender-stopper (EDTA) tube
Gaucher Disease DNA mutations: N370S, L444P, 84GG, IVS2	511048	1/14	>95%	7 mL whole blood, lavender-stopper (EDTA) tube
Bloom Syndrome DNA mutation 2281del6ins7	512145	1/104	>97%	7 mL whole blood, lavender-stopper (EDTA) tube
Mucopolipidosis Type IV DNA mutations: IVS-2A→G, 511del6434	511386	1/100	>95%	7 mL whole blood, lavender-stopper (EDTA) tube

* Leukocyte enzyme testing for Tay-Sachs disease is recommended by ACOG for women who are pregnant or taking birth control pills.

** CF mutations: G85E; delta1507; R553X; 711+1G>T; 3659delC; R117H; deltaF508; R560T; 1078delT; 3849+10kbC>T; I148T; V520F; R1162X; 1717-1G>A; 3876delA; R334W; G542X; W1282X; 1898+1G>A; 3905insT; R347P; S549N; N1303K; 2184delA; R347H; S549R; 394delTT; 2789+5G>A; A455E; G551D; 621+1G>T; 3120+1G>A

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