Jewish Genetic Disorders Information Sheet

In almost every ethnic, racial or demographic group certain genetic diseases occur at higher frequencies among their members than in the general population. Almost all the conditions listed below are transmitted in an autosomal recessive fashion. This means that a couple can only have a child with the disorder if both parents are "carriers" of the trait for that disorder. Carriers usually have no symptoms of the condition. Carrier tests are performed on blood samples. If both parents carry a trait for the same condition, during pregnancy their fetus can be tested for the disease by sampling the placenta or the amniotic fluid. Carrier tests cannot entirely eliminate the possibility of being a carrier, but a negative test makes the chance very low.

Ashkenazi Jewish Background

There are several conditions that are more common in the Ashkenazi Jewish population. If one member of the couple is Jewish and the other is not, the Jewish parent can be tested first. If her/she is positive for the trait, the non-Jewish parent can then be tested. For some of these conditions, testing a non-Jewish individual is less reliable.

Bloom syndrome results in poor growth and poor resistance to infection. There is a high rate of cancer from which individuals usually die before age 30.

Canavan disease is characterized by progressive loss of white matter in the brain. Infants appear to be normal at birth but are later found to be hypotonic (floppy), fail to achieve head control and become developmentally delayed by 5-8 months. Features include an enlarged head, mental retardation, seizures and feeding difficulties. Death usually occurs within the first two decades of life.

Familial Dysautonomia is a disorder that results from the abnormal development of the nervous system, particularly the sensory and autonomic systems. The autonomic nervous system controls involuntary functions, such as swallowing, temperature and blood pressure regulation. Individuals with FD cannot regulate these functions. Other common manifestations are indifference to pain, inappropriate perception of heat and taste, excessive sweating, fluctuating blood pressures, gastrointestinal problems, poor speech and motor coordination.

Fanconi anemia (Type C) is a disorder which causes children to have anemia, short stature, learning disabilities or mental retardation. There may also be birth defects of the limbs, heart or kidneys. Risk for leukemia and early death is increased.

Gaucher disease (Type 1) can be very mild or severe. The absence of the enzyme glucocerebrosidase leads to the progressive build up of a fatty substance in the liver, spleen and bone marrow. Children and adults may have nosebleeds, anemia, an enlarged liver and spleen, bone pain, and easily broken bones. Because the severity is so variable and very effective treatment is available, decisions about testing are difficult.

Mucolipidosis IV is a disorder where children appear normal at birth but develop signs of central nervous system deterioration during the first year of life. Motor and mental retardation are usually mild to moderate, and are slowly progressive. Individuals with ML IV currently range from 1 to 30 years of age. Prognosis beyond this age and life expectancy are not known.

Niemann-Pick disease (Type A) is a severe neurodegenerative disorder of infancy. Affected babies exhibit an enlarged liver and spleen, poor growth and progressive physical and mental deterioration. Death usually occurs in early childhood (before age 5), due to infection.

Tay-Sachs disease is characterized by the onset of severe mental and developmental retardation during the first four to eight months of life. An early sign of the disease is the cherry-red spot, an unusual abnormality in the retina of the eye. The involvement of the central nervous system progresses rapidly and affected children become totally debilitated. Death usually occurs in early childhood. The Tay-Sachs gene is also more common among French Canadians from eastern Quebec and Cajuns from southern Louisiana.

Dystonia is an autosomal dominant disorder (an individual only needs one copy of an abnormal gene to be affected) characterized by sustained, twisting muscle spasms. With time, the frequency and duration of these spasms increase, leading to joint contracture and progressive disability. Children of an individual affected with Dystonia have a 50% chance of inheriting the gene mutation that causes the disease, but symptoms of Dystonia occur in only 30% of the individuals who inherit the gene mutation.

Caucasian Background

Cystic fibrosis (CF) is common among Caucasians, particularly those of northern European origin. It occurs with the same frequency in the Jewish and non-Jewish populations. The disease causes lung infections, difficulty breathing, and problems with bowel function, weight gain and growth. Children frequently need to be hospitalized, have physical therapy and take several medications. The average lifespan is 30 years. One in 2,500 Caucasian newborns has cystic fibrosis and 1:25 Caucasians carries the CF trait.

Mediterranean (Greek, Italian), Sephardi Jewish, Southeast Asian, African

Beta-thalassemia is a disorder that causes severe anemia in children, requiring frequent blood transfusion. Children may grow poorly, have bone deformities or fractures, and develop heart failure from their disease. Carrier frequency varies with ethnic group.[i]

[i]Sources:
The University of Chicago Hospitals, Women's Programs, Carrier Screening (Information Sheet), 2000.
What You Should Know About... Jewish Genetic Diseases, National Foundation for Jewish Genetic Diseases, Inc., 250 Park Avenue, Suite 1000, New York, NY, 10177

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Vital Statistics

	Inheritance Pattern	Outlook	Treatment	Carrier Frequency	Carrier Screening	Prenatal Testing
Bloom Syndrome	Recessive	Individuals can be functional	For symptoms	1 in 110	Yes	Yes
Canavan Disease	Recessive	Fatal, from early childhood through early teens	None	1 in 38	Yes	Yes
Dystonia	Dominant	Individuals can be functional	For symptoms	1 in 900	Yes	Yes
Familial Dysautonomia	Recessive	50% live to 30 years of age	For symptoms	1 in 30	Yes	Yes
Fanconi Anemia (Type C)	Recessive	Onset ranges from 17 months to 22 years - increased risk for cancer	For symptoms	1 in 89	Yes	Yes
Gaucher Disease (Type 1)	Recessive	Varies from mild to debilitating	Enzyme replacement therapy	1 in 10	Yes	Yes
Mucolipidosis IV	Recessive	Developmental delays may be progressive from mild to severe	For symptoms	1 in 100	Yes	Yes
Niemann - Pick Disease (Type A)	Recessive	Fatal by 3-5 years of age	None	1 in 70	Yes	Yes
Tay-Sachs Disease	Recessive	Fatal in early childhood	None	1 in 26 to 1 in 30	Yes	Yes

Sources:

Jewish Genetic Disorders Program – Factbook, Division of Genetics, Children's Memorial Hospital, 2300 Children's Plaza, Box #59, Chicago, IL, 60614

Carrier Testing for Genetic Conditions in Eastern European (Ashkenazi) Jewish People, Stamford Hospital, Center for Maternal-Fetal Medicine, P.O. Box 9317, Stamford, CT, 06904

What You Should Know About...Jewish Genetic Diseases, National Foundation for Jewish Genetic Diseases, Inc., 250 Park Avenue, Suite 1000, New York, NY, 10177