What is Ashkenazi Jewish Carrier Testing?

Ashkenazi Jewish (AJ) Carrier Testing is a way of telling if you are a carrier of a misprint for one of the autosomal recessive disorders that occurs more commonly in the AJ population. Please see Genetics Primer below. These conditions are not unique to AJ individuals, meaning they also occur in individuals who are not of AJ descent but at a much lower frequency.

A Genetics Primer

Each person has ~25,000 pairs of genes in each cell on the body and 2 copies of every gene in each of our cells. Our genes carry the necessary information to direct our growth and development, starting at conception. This information is in the form of a genetic code, known as DNA. In this brochure, we discuss several genetic disorders, each caused by a misprint in the DNA of the one single gene. These genetic disorders are thus called single gene disorders. Another word often used to describe a DNA misprint is a “mutation”. We inherit one copy of every gene from each of our parents. In a child, if both copies of a gene have a mutation (misprint), this can result in a serious autosomal recessive condition. Each parent would be considered a “carrier” of one copy of the mutation. The other copy has the correct DNA code. A carrier of a recessive misprint (mutation) has no symptoms of the disorder and is not at risk for developing the disorder. Only when both parents who carry a misprint in the same gene both pass the misprint onto their child, then their child will be affected with the disorder in question.

Who is Offered AJ Carrier Testing?

All individuals of AJ descent can be offered carrier testing. In general, if one grandparent of an individual is of AJ descent, carrier testing is offered. Even if you have had Tay-Sachs carrier testing in the past, having repeat carrier testing is recommended as additional conditions have been added to the panel of AJ disorders tested. Couple screening is recommended, even if only one member of the couple or one grandparent is known to be AJ, and ideally before starting a family, so that if a carrier couple is identified all reproductive options can be explained.

What is the Purpose of AJ Carrier Testing?

If both members of a couple are found to be carriers of a misprint in the same gene, then formal genetic counselling would be offered to explain what this means and what reproductive options are available. If an AJ individual is found to be a carrier for one of the disorders for which screening is available and the partner of that individual is not of AJ descent, carrier testing may still be available to the individual who is not of AJ descent, depending on the disorder in question.

Why Would I Want the Test?

Some couples wish to know if they are both carriers for a specific condition so when starting a family, they would know if there is a higher risk for one of the disorders screened for and learn the reproductive options available to them. Others do not wish to know.

Do I Have to Have the Test?

No, carrier testing is entirely voluntary. However only adults can ask for the test. Testing requires a blood sample and is ordered by a physician.

Who Will Get the Results?

Only your ordering physician and you will receive the results. This information will not be shared with any third party without your permission.

Where is Carrier Testing Available and How Can I Get More Information?

Contact your health care provider for information how to access carrier testing in your area. Policies may differ in different centres. Click on the links found on the back page of this brochure to connect to websites of partner advocacy groups and professional organizations. These websites contain important information about AJ diseases and how to access genetic counseling centres and carrier testing in your area. You can search for a genetics clinic in your area through Search Genetic Clinics and the CAGC website. In Quebec, information can be accessed through the Montreal Jewish Genetic Disorders Fund (MJGDF) and testjgd.com and for the rest of Canada the website whatsinyourgenes.com provides very useful material. Alternatively, please call the NCJWC head office at 1-866-625-9274 or email info@ncjwc.org.

Note: There is a core group of 3 disorders (Tay-Sachs disease, Canavan disease, Familial Dysautonomia) for which AJ carrier testing is offered in Canadian centres but carrier testing for 4 other autosomal recessive disorders described below, may be offered depending on your geographic location. All these disorders follow an autosomal recessive pattern of inheritance. Carriers of a single copy of the genetic misprint in any one of these genes do not show signs or have symptoms of disease.

Tay-Sachs disease (TSD)

A degenerative disorder of the central nervous system caused by the deficiency of an enzyme Hexosaminidase A leading to progressive accumulation of a lipid material in cells of the brain. A previously normally developing child begins to lose milestones around 7 months of age. There is relentless deterioration, no known effective treatment and the life expectancy of an affected child is greatly reduced.

Canavan disease (CD)

A disorder of the nervous system known as a form of leukodystrophy, CD affects the myelin sheath of nerves. Myelin is the protective covering of nerves that allows the proper transmission of nerve impulses. Affected children after a short period of normal development lose motor milestones, muscle tone is very poor and seizures often develop.

Familial Dysautonomia (FD)

A disorder of the autonomic nervous system affecting certain types of nerve cells leading to varying degrees of symptoms such as absent tears, vomiting crises and less perception of pain and temperature. Symptoms can begin in infancy. Intelligence is not affected.

Fanconi Anemia type C (FANCC)

A cancer susceptibility disorder often associated with developmental abnormalities in major organ systems, short stature and bone marrow failure.

Bloom syndrome (BS)

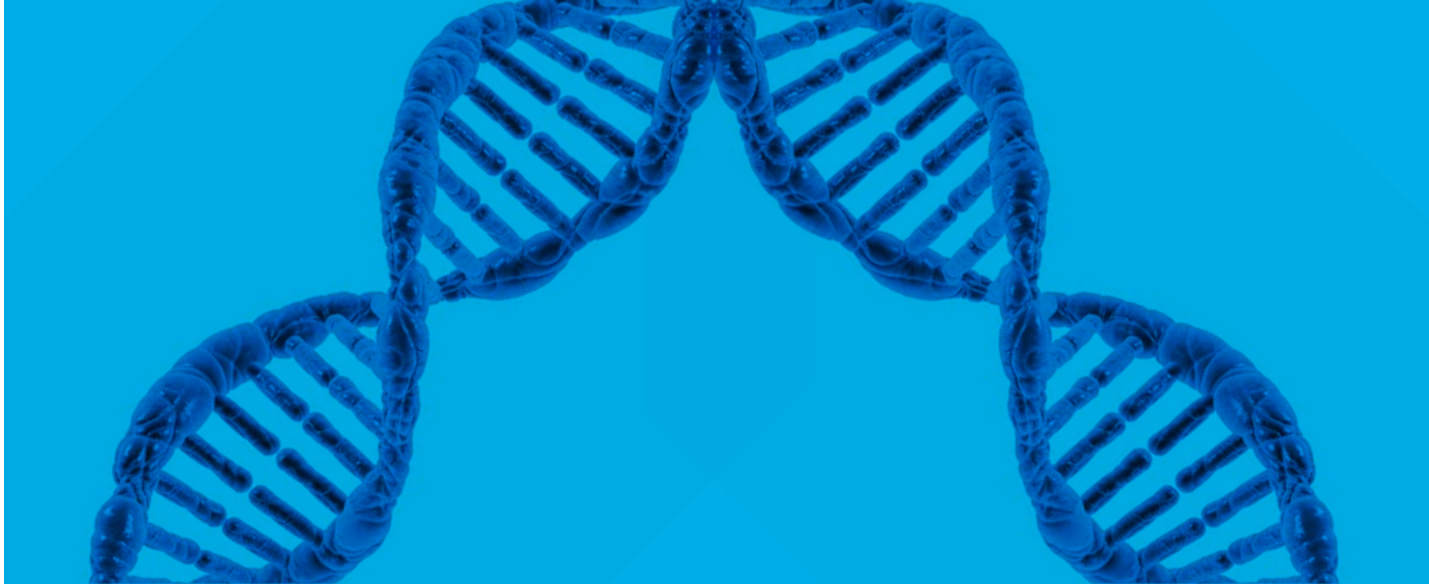
A disorder characterized by short stature, distinct facial features, a characteristic skin rash, increased sensitivity to the sun and an increased risk of diverse cancers.

Mucolipidosis type IV (ML4)

Affected children usually manifest signs and symptoms in the first year of life and have delayed motor and cognitive development, progressive deterioration including visual impairment.

Niemann-Pick disease type A (NPA)

A severe degenerative disorder of the nervous system with progressive deterioration in motor and cognitive skills. Affected children also have enlarged livers and spleens.



Acknowledgments & Resources

Jacob's Ladder
jacobsladder.ca

Montreal Jewish Genetic Disorders Fund

testjgd.com

Fanconi Canada

fanconi.ca

Familial Dysautanomia

fdmontreal.ca

NCJWC
ncjwc.org

whatsinyourgenes.com

Canadian Association of Genetic Counsellors

CAGC - ACCG
cagc-accg.ca

Search Genetic Clinics https://cagc-accg.ca/?page=225

ASHKENAZI JEWISH CARRIER TESTING

Information about carrier testing for Genetic Disorders in the AJ population—2017 Update

National Council of Jewish Women of Canada

ncjwc.org