Ashkenazi Jewish
Carrier Screening

1 in 5 Ashkenazi Jews

is a carrier for one or more genetic mutations that may threaten infant health.

What is Ashkenazi Jewish Carrier Screening?

The purpose of Ashkenazi Jewish Carrier Screening is to determine if a couple is at increased risk of delivering a child with a list of genetic conditions that occur with significant frequency within the Ashkenazi Jewish Population. Many of the conditions are severe and result in childhood death.

Like all carrier testing, both parents must test positive for one copy of the mutation of the same condition in order to have a 25% chance they will deliver a child that is affected with the disease. If the results show that a couple is at high risk, additional testing can be performed during pregnancy to see whether the baby is affected by the disease. If only one parent is a carrier, then there is a very low chance they will have an affected child, but the child may be a carrier.

Who should be tested?

Any patient of Eastern European or Ashkenazi Jewish descent, or who has a relative with one of the genetic conditions prevalent in the Ashkenazi Jewish population should be offered screening.

Having one grandparent of Eastern European or Ashkenazi Jewish descent is enough to qualify for screening.

Partners of parents that test positive should be screened as these diseases occur in all ethnicities, however in lower frequency.

When should testing occur?

Ideally, carrier testing of either partner of an Ashkenazi Jewish couple is initiated at a pre-conceptional counseling visit.

If that partner is positive, carrier testing is extended to the other.

If pregnant, testing is performed on both partners at the first prenatal visit to allow time for further genetic studies and counseling.

What conditions should be included?

The American Congress of Obstetricians and Gynecologists (ACOG) in 2004 published recommendations for carrier screening recommending the following:

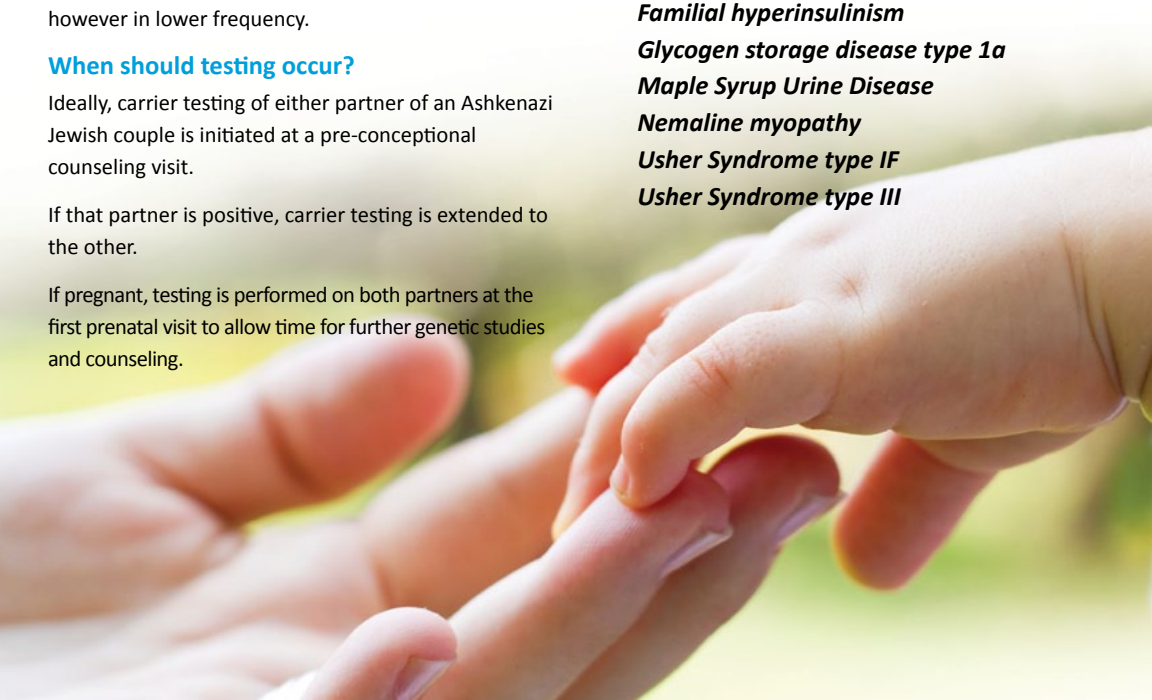
Tay-Sachs
Canavan Disease
Cystic Fibrosis
Familial Dysautonomia

In addition to the ACOG recommendations, in 2008 The American College of Medical Geneticists (ACMG) added the following conditions for carrier screening recommending the following:

Mucolipidosis IV
Niemann Pick Type A
Fanconi anemia group C
Bloom Syndrome
Gaucher Disease

These additional Jewish genetic conditions are also available through aMDx Laboratory Sciences:

Dihydrolipoamide dehydrogenase deficiency
Familial hyperinsulinism
Glycogen storage disease type 1a
Maple Syrup Urine Disease
Nemaline myopathy
Usher Syndrome type IF
Usher Syndrome type III



26842 Haggerty Road, Farmington Hills, MI 48331
855.293.AMDX (2639) fax: 248.848.1623
www.amdxlabs.com

Patient Assistance Program

The aMDx Patient Assistance Program is designed to assist patients who receive laboratory services from aMDx Laboratory Sciences. If a patient is financially challenged or struggling to pay co-payment, co-insurance or deductible amounts, aMDx encourages the patient to contact our Patient Assistance Hotline at 855-293-2639 extension 4.

aMDx Laboratory Sciences is a CLIA licensed clinical laboratory and is accredited by the College of American Pathologist (CAP). The information contained in this brochure is provided by aMDx Laboratory Sciences as an educational service for physicians and their patients.

AJPCarrierTesting 102711