Pre-pregnancy

Jews at Risk of Passing on Certain Genetic Disorders; Screening Advised

By Eve Glicksman, Staff Writer

If you or your spouse come from a Jewish background, there is a greater risk of passing on certain genetic disorders to your children. These serious, sometimes fatal disorders can show up in families with no history of the disease. In couples where only one person is Jewish. In families where no one has practiced the religion for centuries. And sometimes, in people of other ethnicities.

If you're Jewish and planning to have a child, talk to your doctor about carrier testing. If a simple blood test shows that both you and your partner carry one of these genes, you may want to talk to a genetic counselor to help you understand and address the potential problems.

Ashkenazi or Sephardic?
Where your family came from affects which diseases your offspring may be at risk for. The tendency for Jews to marry other Jews within their communities led to a group of disorders common in European Jews. It also led to another cluster of disorders more common in Jews from African and Mediterranean regions.

In North America, 90 to 95 percent of Jews are of Ashkenazi descent. This means their ancestors are from central and Eastern Europe: Germany, Poland, Lithuania or Russia. Sephardic Jews have roots in Spain, Portugal, Italy, North Africa, India, Persia and the Balkans.

If you are Ashkenazi, there is a 1 in 5 chance you carry a gene for one of the following disorders. Next to the disease is the approximate carrier rate.

Gaucher disease	1 in 10 Ashkenazi Jews
Cystic fibrosis	1 in 25
Tay-Sachs disease	1 in 25
Familial dysautonomia	1 in 30
Canavan disease	1 in 40
Nieman-Pick disease	1 in 70
Fanconi anemia	1 in 90
Bloom's syndrome	1 in 100
Mucolipidosis IV	1 in 100

Sephardic Jews are more likely to carry a gene for thalassemia, a blood disorder, or for familial Mediterranean fever.

Being a carrier does not mean you have or will get the disease. But you could pass it on to your child if your mate also carries the gene.

What are the odds?
The only way to get one of these diseases is to inherit it. If only one parent carries the defective gene, the baby will not get the disease, but may inherit the gene and become a carrier. If both parents carry the gene:

- The odds are 50 percent that the baby will be healthy but carry the gene.
- There is a 1 in 4 chance that the child will have the disorder.
- Chances are 1 in 4 that a child will not have the disease or carry the gene.

Note that other ethnic groups are affected by these disorders, too. Some non-Jewish French-Canadians and Cajuns in Louisiana also have a higher risk for Tay-Sachs disease, for instance. A child is much less likely to inherit the disease if only one parent is Jewish, but it can happen.

Who should get tested for what?
While Gaucher disease is the most common of the Jewish-linked disorders, most people don't screen for it because it can be managed with treatment. For the fatal degenerative disorders like Tay-Sachs or Canavan, DNA carrier screening gives couples the chance to plan a prevention strategy if needed.

The American College of Obstetricians and Gynecologists recommends that doctors offer carrier screening to anyone with Ashkenazi Jewish roots prior to conceiving for:

- Tay-Sachs disease
- Canavan disease
- Cystic fibrosis
- Familial dysautonomia

One test does not screen for every disease. If any of the other Jewish-linked disorders are in your family, you may ask your doctor to get screened for that gene as well.

If only one person in the couple is Ashkenazi, that person should get tested first. If he or she is found to be a carrier, then the partner should get tested. If you are already pregnant, be aware that routine fetal tests do not rule out these relatively rare disorders unless you request it.

When both partners are carriers
If you and your partner are both found to be carriers, a genetic counselor can help you understand the risks to your child and help you weigh the pros and cons of actions you can take.

Your options may include:

- Artificial insemination of sperm or an egg donation from a noncarrier.
- Adoption of a child.
- In-vitro fertilization. Fertilized eggs can be screened and only healthy embryos implanted.
- Have an amniocentesis or chorionic villus sampling to see if the fetus has the disorder. Couples may choose to end the pregnancy.
- Ignore the risk. There is a 75 percent chance of having a healthy baby.

Some people think that Jewish-linked disorders are no longer a problem because they seldom hear about them. But the lower incidence is largely due to more Jewish couples taking preventive screening steps. Every generation produces carriers of the disease, so the risk never disappears. The only way to avoid the heartbreak of one of these fatal disorders is to be informed. Talk to your doctor about how you can reduce your risks.

Related Articles

- **Cystic Fibrosis Symptoms, Diagnosis and Treatment**
- **Deciding About Genetic Testing for Inherited Breast and Ovarian Cancers**
- **Introduction to Prenatal Tests**
- **Genetic Counseling: What to Expect**

External Sources

- **The Mount Sinai Medical Center for Jewish Genetic Diseases. Genetic diseases.. Accessed February 10, 2009**
- **Weinstein LB. Selected genetic disorders affecting Ashkenazi Jewish families. Family and Community Health. 2007;30(1):50-62.. Accessed February 10, 2009**
- **Chicago Center for Jewish Genetic Disorders. Jews and increased risk. . Accessed February 10, 2009**
- **March of Dimes. Tay-Sachs disease. . Accessed February 10, 2009**
- **American College of Gynecologists and Obstetricians. Cystic fibrosis: prenatal screening. . Accessed February 16, 2009**
- **ACOG Committee on Genetics. Prenatal and preconceptional carrier screening for genetic diseases in individuals of Eastern European Jewish descent. Obstetrics and Gynecology. 2004;104(2):425-428. .**

Accessed February 10, 2009

This article was reviewed and updated 08-01-2009.

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