

Health Policy

NEWSLETTER

Preventing Jewish Genetic Diseases in Philadelphia and Nationally

It is estimated that each person is a carrier for approximately five to 15 recessive mutations in his or her genes. Since all of our genes come in pairs – one from our mother and one from our father – a mutation in one copy is compensated for by the other copy. Therefore, a carrier for a recessive genetic disease is a healthy person. There are usually no outward signs of one's carrier status. However, if a carrier for a recessive disease mates with someone who is also a carrier for **the same** recessive disease, then there is a 1 in 4 chance, with each pregnancy, of having a child affected with that disease.¹ Too often, people find out they are carriers only after a child is born with a rare recessive disease.

There are many ethnic groups at higher risk for certain genetic diseases. This may be due to intermarriage, geographic isolation, or – as is the case for African Americans, who are at higher risk for Sickle Cell disease – carrier status offered protection against acquired diseases more common in their geographical location. Jews of Eastern European descent, known as “Ashkenazi Jews,” are at increased risk to be carriers for several genetic diseases, many of which are also found in the general population. These diseases are severe and many are life-threatening. **One in five** Ashkenazi Jews is a carrier for at least one of **18** diseases² for which carrier screening is recommended in the Ashkenazi Jewish population (Table 1).

There are no cures for any of these diseases. Many lead to early death or shortened lifespans, and require rigorous daily medical management. Only one of these diseases, Gaucher disease, has an effective treatment. However, they can all be prevented through carrier screening prior to pregnancy (pre-conception screening). Medically accurate screening is available with a simple blood test.

Screening programs have been initiated across the country in order to reduce the probability of babies being born with any of these life-threatening and preventable Jewish genetic diseases (JGD). The Victor Center for the Prevention of Jewish Genetic Diseases was founded in 2002 by Albert Einstein

Health Care Network in partnership with Lois Victor, a mother who lost two children to a Jewish genetic disease. The Victor Center partners with college campuses, clergy, healthcare professionals and communities to build awareness, educate and provide ongoing access to comprehensive genetic education, counseling and screening services.

Since the Victor Center was founded in Philadelphia, Centers have been established at The Floating Hospital for Children in Boston and at the University of Miami Miller School of Medicine. Partnerships are underway in cities throughout the United States including Atlanta, Pittsburgh, Minneapolis, Tulsa and Omaha.

The screening program of the Victor Center is modeled on the community-based screening movement for Tay-Sachs disease. During the 1970s, Jewish and medical communities galvanized around screening for Tay-Sachs disease and created a powerful grassroots movement that eventually became standard medical practice. As a result, the number of Ashkenazi Jewish babies born with Tay-Sachs disease has decreased by more than 90%.³ However, many additional diseases have been identified as having significant carrier frequency in the Ashkenazi Jewish population since Tay-Sachs screening was started. The Victor Center is working to raise awareness amongst physicians of the availability and need for screening for all 18 diseases through outreach to physician practices, and providing education at conferences held by major medical organizations (e.g., the National Society of Genetic Counselors, American College of Medical Genetics and the American College of Obstetrics and Gynecology).

The Victor Center approach focuses on prevention as a fundamental component of social change. While research continues to search for a cure for devastating inheritable diseases such as Tay-Sachs, Canavan, Familial Dysautonomia and others, the best way to ensure that couples have the greatest number of reproductive options and that Jewish children are born free of these diseases, is to educate our young adults about the diseases and enable them to know their carrier status. There

Table 1

Genetic Diseases Associated with Ashkenazi Jews

Bloom Syndrome
 Canavan Disease
 Cystic Fibrosis
 Dihydroipoamide Dehydrogenase Deficiency
 Familial Dysautonomia
 Familial Hyperinsulinism
 Fanconi Anemia Type C
 Gaucher Disease
 Glycogen Storage Disease Type 1A
 Joubert Syndrome
 Maple Syrup Urine Disease
 Mucopolidosis IV
 Nemaline Myopathy
 Niemann-Pick Disease Type A
 Spinal Muscular Atrophy
 Tay-Sachs Disease
 Usher Syndrome Type IF
 Usher Syndrome Type III

are only two ways to find out if you are a carrier: through a simple blood test or by having an affected child. Education and awareness must be followed by screening if we are to have any success in eradicating these diseases.

The Victor Center is currently involved in several major projects, including: a pilot education and screening campaign in Atlanta with the support of the Marcus Foundation; development of a webinar to teach Rabbis how to discuss JGDs with their congregations and young couples;

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working with OB/GYN practices to create a toolkit for educating both doctors and patients about Jewish genetic diseases; creating a guide to ensure appropriate screening; and launching a video featuring Matisyahu, a Jewish reggae rapper, educating people about Jewish genetic diseases.

The Victor Center recently completed a three-year project focused on raising awareness of Jewish genetic diseases on college campuses. With funding from the Centers for Disease Control and Prevention (CDC), the Victor Center hired a social

marketing company to conduct research on the best practices for educating college students about Jewish genetic diseases and to empower students to create a comprehensive awareness campaign on college campus. The guide and campaign materials, known as the “1 in 5” campaign, were field tested at six campuses across the country last spring. These materials are currently being rolled out at campuses nationwide. The Victor Center plans to undertake a similar social marketing process to identify and create the best practices for educating young professionals. ■

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