

A ROADMAP FOR HEALTHY HEREDITY

A genetic screening program piloted in L.A. could significantly reduce the prevalence of a debilitating disease that disproportionately affects Persian Jews.

HIBM: four letters that could spell out the name of a personal computer. Sadly, the acronym carries a much less innocuous meaning. HIBM stands for hereditary inclusion body myopathy, a devastating muscle disorder that causes progressive weakness of the arms and legs. HIBM's symptoms usually don't appear until a person is in her 20s or 30s. As muscles deteriorate, a person may lose their ability to walk altogether and become confined to a wheelchair. There is no known cure for the disease.

HIBM is extremely rare in the general population. But in the Persian Jewish population, this vicious genetic disease is so prevalent that it is a genuine concern for many families. In order to make genetic testing for the disease more efficient—and hopefully lower its incidence—the Medical Genetics Institute at Cedars-Sinai has launched a one-of-a-kind community program to screen for it, along with other conditions that also disproportionately affect Persian Jews—conditions that can have serious health consequences if left unchecked but are easily treated or prevented once screened.

Why this effort? Because genetic

testing means that couples may know whether their children would be at risk for HIBM before they are conceived. With that knowledge, comes the power for individuals to make informed decisions and potentially reduce the impact of a tragic disease for which there is no cure.


This pilot program is the brainchild of Dr. David L. Rimoin, director of the Cedars-Sinai Medical Genetics Institute, a force that's world-renowned for its research in the areas of genetic diseases and birth defects. "No one group has more mutations than another," explains Dr. Rimoin. "Every human being can carry five to 10 gene mutations that increase the risk for developing specific conditions. These mutations are simply more easily identifiable—because they are more common—in groups with a strong tradition of intermarriage," he adds.

This includes individuals in the Persian Jewish population. Their large numbers in Los Angeles made this group a prime target for the launch of the screening program. Approximately one in 20 Persian Jews carries the gene mutation responsible for HIBM and one in 400 couples could be having a child

affected by this condition.

Couples who plan to have children may want to know whether they both carry the gene mutation. If they do and decide to have children, in vitro fertilization and pre-natal screenings could help ensure that the trait is not passed on to their offspring.

Dr. Rimoin hopes to test at least 10,000 of the 50,000 or more Persian Jews in Southern California through this vital project. Within a few months of the program's launch last summer, 1,000 men and women had already come forward to be screened anonymously, and for free—thanks in part to a grant from the Jewish Community Foundation.

For Dr. Rimoin, the success of this initial effort should foster screenings in other communities as well as demonstrate the prevalence of these disorders. In doing so, the hope is to make genetic testing available in doctors' offices and encourage insurance companies to cover the cost of testing. 

DID YOU KNOW?

Like sickle cell anemia, Tay Sachs disease, and Cystic fibrosis, HIBM is an autosomal recessive disorder, meaning that the malfunctioning gene responsible for the disease is located on one of the autosomes—the chromosomes that are not sex chromosomes. "Recessive" means that two copies of the gene are necessary to have the trait, one inherited from the mother, and one from the father. A person who has only one recessive gene is said to be a "carrier" for the trait or disease, but they do not have any health problems from "carrying" one copy of the gene. Most people do not know they carry a recessive gene for a disease until they have a child with the disease, or are tested for it.

