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NTSAD

Topic of the Week

Knowledge is power.
- Frances Bacon

Friday, December 5, 2014

Carrier Screening...The Road to Prevention

Here, at NTSAD, we receive calls and emails from individuals and couples seeking advice about carrier screening. Common questions include: What do I do? What should I ask my doctor? I'm a carrier and my partner's results came back inconclusive; what do we do next? Where should I go to be screened?

Providing guidance and answers to questions about carrier screening is an important facet of NTSAD's mission. Alongside the experts on our Scientific Advisory Committee, we are also thankful to have partnerships with groups such as the Chicago Center for Jewish Genetics, Jewish Genetic Disease Consortium, and the Victor Centers at Einstein Medical Center in Philadelphia and at Tufts Medical Center in Boston. They are invaluable resources as we direct couples and individuals, *of any ethnic background*, in the right direction in the carrier screening process.

A Unique Collaboration in Chicago

The Center for Jewish Genetics in Chicago has partnered with JScreen to offer DNA sequencing for rare genetic diseases including Tay-Sachs and Canavan. The $99 sequencing is done by using a saliva sample. If unknown variants are reported, a follow-up enzyme assay (test) is conducted to determine carrier status. NTSAD strongly recommends that any DNA test is partnered with an enzyme test as it is the gold standard in identifying Tay-Sachs carriers.

Contact the NTSAD office at (617) 277-4463 if you or someone you know is interested in this program.

An Invaluable Partnership

NTSAD's current President, Shari Ungerleider, has honored her son Evan's memory by working tirelessly on raising awareness through prevention. She has played an important role in the history of the Jewish Genetic Disease Consortium (JGDC) and continues her commitment on a daily basis. NTSAD's membership in the JGDC directly aligns with our mission to raise awareness and secure a future free of rare genetic diseases.

The Consortium "unites its member organizations so that [they] may jointly strengthen public education and awareness and appropriate carrier screening for all persons of Jewish heritage, whether Ashkenazi, Mizrahi, Sephardic, as well as couples of interfaith marriage. Through its Medical Grand Rounds Program, Clergy Education Program and Jewish Community Program, the JGDC educates physicians, rabbis, Jews of all backgrounds and interfaith couples about Jewish genetic diseases."

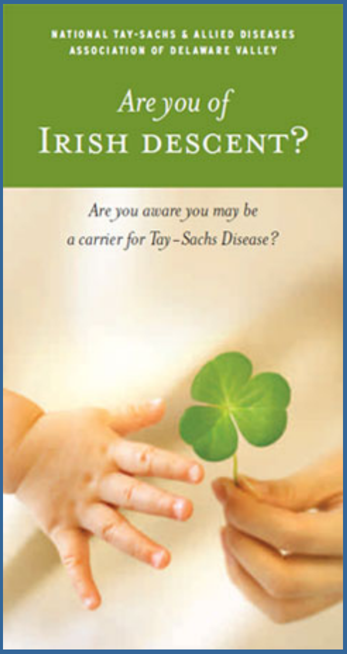
A Mission and a Vision

Victor Centers
for Jewish Genetic Diseases

Just as those founding families did when they created NTSAD in 1957, Lois Victor honored the memories of her two children by founding the Victor Center for the Prevention of Jewish Genetic Diseases in 2002. She remains committed to a future where families will not know the heartache of losing children to rare genetic diseases. NTSAD works with the Victor Centers in raising awareness and guiding individuals and families as they seek a healthy future for their families.

The Victor Centers in Boston, Miami and Philadelphia offer carrier screening and counseling to individuals and families.

Looking for the Four-Leaf Clover



NATIONAL TAY-SACHS & ALLIED DISEASES
ASSOCIATION OF DELAWARE VALLEY

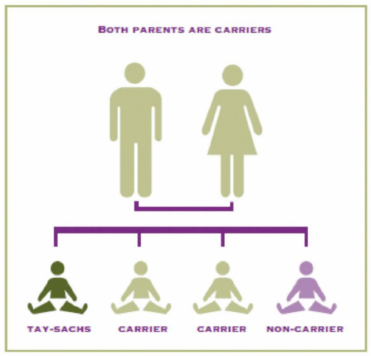
Are you of
IRISH DESCENT?

Are you aware you may be
a carrier for Tay-Sachs Disease?

One big misconception of Tay-Sachs is that it is solely a "Jewish Genetic Disease." The NTSAD family can quite definitively say it is not. There are other ethnic groups that have a higher carrier rate than the general population. For instance, there is indication that Irish Americans have a higher risk of being carriers of the Tay-Sachs gene. Adele Schneider and her team at Einstein Medical Center are currently studying the carrier rates within the Irish American community to determine what that rate is today.

The NTSAD Delaware Valley chapter has funded this study and NTSAD's New York Area Fund will soon support this important work. To date, close to 300 individuals have been screened. The goal is to have at least 1,000 participants in the study. The study is accepting samples from across the country from people who have at least three (3) Irish grandparents.If your family is of Irish ancestry, please consider being a part of this study. Email irish@tay-sachs.org for more information.

Know Your Risk. Be Tested. Be Sure.



Tay-Sachs Carrier Rates

1 in 27 Ashkenazi Jews of Eastern European descent
1 in 27 French-Canadians and Cajuns
1 in 50 to 1 in 190 Irish Americans
1 in 250 of the general population

1 in 35 Ashkenazi Jews is at risk of being a carrier for the Canavan disease gene.

The carrier statistics for Sandhoff, GM1 and the adult onset versions of these disease are not clear but we do know these diseases can affect any family in any generation. It is NTSAD's hope to see a day when universal screening is available for all of these diseases.

Supporting NTSAD Makes a Difference!

We welcome gifts to support our many efforts to fund research, care for families and their loved ones. Gifts can be made through our website here. *Thank you* for supporting NTSAD and giving hope to all our families!

Contact Us

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