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ntsad research review **friday, january 18, 2013**

Carrier Screen Technology Study

At the 2011 Annual Family Conference in Boston, families volunteered blood samples to study the efficacy of NextGen sequencing to detect Tay-Sachs carriers.

This study is an important step toward a testing method that detects all carriers regardless of heritage without the technical limitations of enzyme assay. Here is summary of their findings:

1. 51 out of 74 participants had positive enzyme results (46 were carriers, 5 had Late-Onset Tay-Sachs).

2. 42 of the 51 people who had abnormal enzyme results were found to have a pathogenic mutation (well-known DNA abnormality in the Tay-Sachs gene), 8 had variants of unknown significance (a DNA code change with unknown meaning), and one had the HEXA pseudoallele (decreased enzyme on the test that does not cause disease).

3. NextGen sequencing plus common deletion screening of HEXA found a disease causing mutation, pseudoallele, or variant of unknown significance in 100% of the people who were found to be carriers via enzyme testing or personal report. Detected two carriers missed by enzyme alone, and detected mutations in seven of the 51 enzyme positive individuals that would not be detected on common DNA mutation panels provided by commercial labs.
Click **here** to download abstract.

What is NextGen sequencing?

NextGen stands for Next Generation. This new technology allows for quick and accurate sequencing of entire genes for abnormal changes.

Conclusion

Our data suggest that NextGen sequencing and common deletion testing in combination with enzyme analysis may be used as an efficient screening technology for people of all backgrounds, and will provide a more sensitive (better) test than enzyme analysis alone or a mutation panel with enzyme.

Enzyme assay screening remains the gold standard. Click **here** to download NTSAD's position statement on Tay-

Sachs carrier screening.

Searching for a Cure

NTSAD Issues Request for Research Proposals

Every year NTSAD's Research Initiative solicits proposals for innovative research projects to fund in the area of neurodegenerative disorders. The announcement, otherwise known as Request for Proposals (RFP), is now available.

The grant proposals are due **February 8, 2013**. Funding awarded will be up to $50,000 for one year. Please share this RFP with anyone you know that would be potentially interested in applying for a grant. Click here for full details.

The Research Initiative has awarded over $2 million to 46 projects.

The impact of these grants has grown as several grantees have collectively received over $10 million in funding from NIH.

give light and people will find the way.
~ ella baker

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