CARRIER SCREENING TOOLKIT

Information on Testing and Support for Patients and Families

NTSAD

National Tay-Sachs & Allied Diseases Association

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Carrier Screening 101: Start Here

1. **What Is Carrier Screening and When is it Usually Ordered?**
https://ntsad.org/support-for-families/carrier-screening/#carrier-screening

2. **Who Should Get Carrier Screening?**
https://ntsad.org/support-for-families/carrier-screening/#who-should-get-carrier-screening

3. **Why Carrier Screening Is Important**
https://ntsad.org/support-for-families/carrier-screening/#why-carrier-screening-is-important

4. **How to Start the Carrier Screening Process**
https://ntsad.org/support-for-families/carrier-screening/#how-to-start

5. **Selecting a Carrier Screening Panel**
https://ntsad.org/support-for-families/carrier-screening/#selecting-a-carrier-screening-panel

6. **How to Find a Genetic Counselor**
https://ntsad.org/support-for-families/carrier-screening/#find-a-counselor

7. **Understanding Your Results**
https://ntsad.org/support-for-families/carrier-screening/#understanding-your-results

8. **Family Connection**
https://ntsad.org/support-for-families/carrier-screening/#family-connection

9. **Frequently Asked Questions**
https://ntsad.org/support-for-families/carrier-screening/#FAQ

Find a provider at the NSGC and ACMG websites.

Comparing Carrier Screening with Other Types of Genetic Screenings

CARRIER SCREENING

- Parents are tested individually or sequentially
- Ideally before pregnancy, though can be competed during pregnancy if not previously done
- Tells you whether you carry certain genetic variants that could be passed to a child

PRENATAL SCREENING

- Fetus is tested
- During pregnancy
- Tells you whether a pregnancy has an increased risk for certain conditions
- This may be a non-invasive blood test that provides a risk assessment to be confirmed by prenatal diagnostic testing

PRENATAL DIAGNOSTIC TESTING

- Fetus is tested
- During pregnancy
- Tells you whether a specific genetic condition is present
- Often done when a pregnancy is known to be at risk for a specific condition(s)

NEWBORN SCREENING

- Newborn is tested
- Shortly after birth
- Tells you whether a newborn may have certain conditions that benefit from early detection

Start a Conversation with your Healthcare Provider: Questions to Ask

1. Do you routinely order carrier screening for your patients and, if so, what laboratory/company do you use, and when do you order? Are they typically expanded carrier screening panels or ethnicity-based panels?
2. What conditions/genes are included? Does the panel include testing for Tay-Sachs, Canavan, GM1 gangliosidosis, and Sandhoff disease (their respective genes *HEXA, ASPA, GLB1,* and *HEXB*)?
3. Who can help me understand my results if I have questions or receive a positive result?
4. If I am a carrier, should I share this information with other family members, and how should I do that?
5. If testing while pregnant: Will you order carrier screening for both myself and my partner simultaneously to avoid a time delay?

Find NTSAD's Carrier Screening Resources at:
https://ntsad.org/support-for-families/carrier-screening/

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