NTSAD CARRIER SCREENING TOOLKIT

Information on Testing and Support for Providers

National Tay-Sachs & Allied Diseases Association

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Carrier Screening 101: Start Here

1. **What is Carrier Screening?**
   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. **Resources and 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

Connect Your Patients to NTSAD

Founded in 1957, NTSAD is one of the oldest patient advocacy organizations. Today, we provide approximately 1,000 individuals and families with connection, resources, and individual support. We are a trusted source for impartial information that is carefully reviewed by expert clinicians and researchers of our Scientific Advisory Council. We host an Annual Family Conference and offer specialized programming to help individuals and families affected by rare disease. Supporting families is the center of everything we do.

ACMG/ACOG Guidelines

The ACMG (American College of Medical Genetics and Genomics) and ACOG (American College of Obstetricians and Gynecologists) provide guidelines for reproductive carrier screening. Currently they stratify diseases/genes to be included in carrier screens based on their population frequency, thus they may not include all ultra-rare diseases. More information can be found in https://pubmed.ncbi.nlm.nih.gov/38814327/

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

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