

# WHY SEE A GENETIC COUNSELOR?



If your child has **Okur-Chung Neurodevelopmental Syndrome (OCNDS)**, a genetic counselor can help you understand the condition and make informed decisions about your family's future. Genetic counselors are healthcare professionals with specialized training in genetics and counseling. They can provide valuable guidance, support, and resources for families with genetic conditions.

## WHAT CAN A GENETIC COUNSELOR DO FOR YOU?

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### Help You Understand Genetic Testing Results

Genetic testing results can be complex and difficult to interpret. Genetic counselors are **experts in interpreting these results** and can help answer any questions you might have. They break down the technical aspects of genetic testing and explain **what the results could mean for** your child and/or your family.

02

### Education about OCNDS

Genetic counselors can help you understand:

- ✓ What **symptoms** to watch for or how the genetic result may explain certain symptoms in your child.
- ✓ **Therapies** and **monitoring strategies** to support your child's health.
- ✓ The **genetics** of OCNDS and the potential **implications** for other family members.

03

### Facilitate Testing for Other Family Members

In some cases, a genetic variant associated with OCNDS may have **health implications for other family members**, such as siblings or parents who may carry the same genetic variant.

Genetic counselors can guide you in determining **which family members may benefit from testing** and can help you coordinate that testing

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### Discuss the Chance of Having Another Child With OCNDS

If you are planning future pregnancies, a genetic counselor can help you understand the likelihood of having another child with OCNDS **based on your family's genetic testing results**.

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### Explore Reproductive Options

If you are planning a future pregnancy, a genetic counselor can help guide you through various **reproductive options such as** testing during pregnancy, preimplantation genetic testing (PGT), egg/sperm donation, and adoption. If you're interested in learning more about these options, you can find more information on our family planning and reproductive options handout:

<https://www.csnk2a1foundation.org/family-planning-resource>

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## Provide Emotional Support and Guidance

Genetic counselors are trained to help families **understand** and **adapt to the psychological and medical impacts** of a genetic diagnosis like OCNDs.

Whether it's the anxiety of uncertain results, grief after a diagnosis, or navigating complex family decisions, genetic counselors provide a **safe space** to process these emotions and can offer strategies for coping.

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## Discuss Resources, Support Groups, and Research Opportunities

Genetic counselors can:

- ✓ Help **make referrals** to other healthcare specialists who may be helpful for your child's care or for parental support.
- ✓ **Connect you with local and online support groups** for families affected by OCNDs (e.g., the [CSNK2A1 Foundation](#)) and other genetic conditions.
- ✓ Keep you informed about **research studies** that may become available for individuals with OCNDs. One ongoing research opportunity is through **Simons Searchlight**, which studies genetic conditions like OCNDs that are linked to autism, with the goal to improve clinical care and treatment. Learn more about this research opportunity at: <https://clinicaltrials.gov/search?cond=CSNK2A1>

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## Help Keep You Up to Date as New Information is Discovered

Some families find it helpful to **regularly follow-up** with a genetic counselor, geneticist, or another similar provider to stay connected as new information about OCNDs emerges.

In some cases, genetic testing reveals a **Variant of Uncertain Significance (VUS)**. This means that a genetic change has been found, but **its effect on health is unclear**. This can be particularly challenging for families because the implications of a VUS are not immediately known. Regular follow-up with a genetic provider can be especially valuable for these families, since new information about the VUS may become available over time.

## HOW CAN YOU FIND A GENETIC COUNSELOR?

### ASK YOUR HEALTHCARE PROVIDER:

If you or your child is seeing a doctor or specialist for OCNDs, ask them to refer you to a genetic counselor.

### GENETIC COUNSELING DIRECTORY:

If you are located in the United States or Canada, the National Society of Genetic Counselors offers a directory to find a genetic counselor near you:

<https://findageneticcounselor.nsgc.org/>