

Steps to Take Now Continued



— Visit Faces of OCNDS to see children like yours.
<https://loom.ly/nkfTz1s>

View the Worldwide Cases of OCNDS Map where you will discover that our syndrome spans the globe.
https://loom.ly/_wKR7g



Follow
Us on:



Read our latest newsletter for up to date information.

<https://loom.ly/b8Mw7Bc>



Invest in a cure by donating
<https://loom.ly/gPV5CAQ> or holding a fundraiser or purchasing CSNK2A1 Foundation merchandise
<https://loom.ly/Zar4XYE> to support meaningful OCNDS research.



www.csnk2a1foundation.org



Welcome to our community!



What is OCNDS and CSNK2A1?

Okur-Chung Neurodevelopmental Syndrome (OCNDS) is an ultra rare genetic disorder first identified in 2016.

- Everyone with OCNDS has some degree of developmental delay and/or differences in brain function.
- OCNDS is caused by a mutation in the CSNK2A1 gene which is located on Chromosome 20. The gene CSNK2A1 creates a protein called CK2 which plays a crucial role in development. A mutation in this gene disrupts typical development.
- Currently there are over 150 people worldwide with this ultra rare genetic condition. (2021)

People with OCNDS are...



Steps to Take Now

As you are navigating this new diagnosis, it can be overwhelming and difficult to know where to begin. Here are steps you can take to get informed, connected and make an impact today!



Stay-up-to date on the latest OCNDS news by **joining our Contact Registry.**

<https://loom.ly/ckeWg4o>



Register with Simons
Searchlight to participate
in the natural history study.

**SIMONS
SEARCHLIGHT**

<https://www.simonssearchlight.org>



Watch our conference, informational,
research update, and awareness videos.

<https://loom.ly/TVcFKEM>



Download and distribute the

What is Okur-Chung Neurodevelopmental Syndrome - CSNK2A1 Foundation

information page to your family, doctors, friends, teachers, and service providers.

<https://loom.ly/qRlBDlw>



Participate in the family zoom calls and connect with families around the world. Ask questions, get new ideas and check for upcoming events. <https://loom.ly/yvg8IWc>



People with OCNDS enjoy...



What are the symptoms of OCNDS?

The most common symptoms of OCNDS are below. We are still determining how frequently these occur and at what ages they commonly occur or resolve.

- Speech delay/inability to speak
- Global developmental delay (GDD)
- Learning disabilities
- Autism spectrum disorder traits
- Behavioral challenges such as tantrums, hand flapping, other stereotypic movements
- Feeding difficulties starting from birth
- Reflux (heartburn)
- Constipation
- Neurological problems such as Hypotonia (low muscle tone, clumsy movements), Microcephaly (small head), Epilepsy (seizures)
- Crooked (misaligned) teeth and cavities
- Hyper-mobility, Hernias, Hip Dysplasia
- Vision issues such as Strabismus, Near/Far sightedness, Astigmatism
- Minor skeletal deformations in vertebrae
- Structural abnormalities in the brain



Is there a cure or treatment for OCNDS?

There is no definitive cure but research is underway. Assessments and therapies recommended include

- Speech therapy including assistive communication starting at 12 months of age
- Intellectual and behavioral assessments with appropriate educational support
- Physical and occupational therapy
- Monitor growth and use of G-tubes if there are persistent feeding issues
- Evaluate vision, teeth, hearing, and immune system function



Is there any biological sex bias?

OCNDS affects each gender equally.

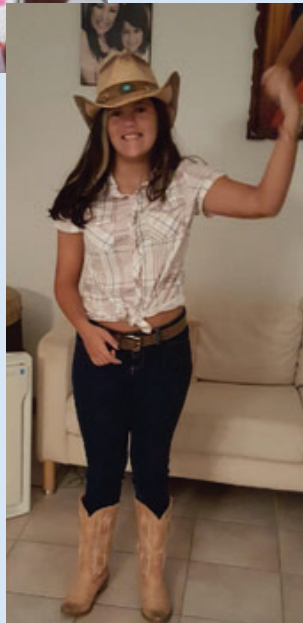
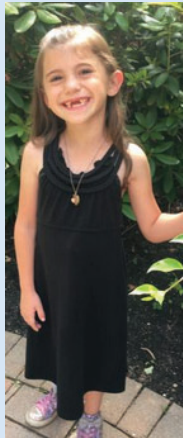
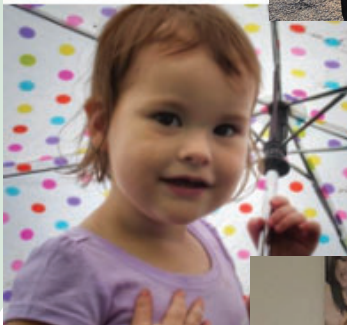
Parents have found these therapies to be helpful:

- Applied Behavior Analysis Therapy (ABA)
- Speech/Language Pathology / Speech Therapy
- Occupational Therapy
- Physical Therapy / Physiotherapy
- Early Intervention
- Early Intensive Behavioral Intervention
- Behavioral Therapy
- Music Therapy
- Equine Therapy
- Floortime Therapy
- Vision Therapy
- Communication Technology



Parents have found these specialists to be helpful:

- Geneticist
- Feeding / Nutritionist
- Dentist / Orthodontist
- Pediatric Gynecologist
- Ophthalmologist
- Immunologist
- Cardiologist
- Orthopedist
- Neuromuscular
- Neurologist
- Epileptologist
- Endocrinologist
- Audiologist
- Ear Nose & Throat



Participate in Research

Because we are rare we all need to participate in research opportunities to develop a standard of care. <https://www.simonssearchlight.org>



What is Simons Searchlight?

Leading scientists, doctors, and families have come together to form Simons Searchlight. Together, they are driving science forward. The plan is to:

- Collect detailed medical and behavioral histories along with blood and saliva samples.
- Synthesize the information you provide and share results with families.
- Freely share data and samples with qualified researchers.
- Connect participants around the world.
- Promote better understanding of these genetic changes.



Why Join?

You can play a meaningful part in the quest for more answers. As part of the Simons Searchlight community, you can:

- Gain clarity on your or your family member's diagnosis.
- Partner with some of the best minds in science.
- Get updates on the latest research findings.
- Connect with others who share your diagnosis.
- Contribute to advancements that will also help future families.

