



OKUR-CHUNG NEURODEVELOPMENTAL SYNDROME (OCNDS)

What is Okur-Chung Neurodevelopmental Syndrome?

Okur-Chung Neurodevelopmental Syndrome (OCNDS) (OMIM # 617062) is a rare genetic disorder first identified in 2016. Everyone with OCNDS has some degree of developmental delay and/or differences in brain function.

How many individuals are diagnosed with OCNDS?

Over 350 individuals worldwide have been diagnosed with OCNDS so far. This number is expected to increase with the increasing utilization of WES in other regions of the world.

What causes OCNDS?

OCNDS is caused by heterozygous mutations in the CSNK2A1 gene on chromosome 20.

What type of mutations are seen in OCNDS?

Missense (most common), frameshift, stop-gain, splice site, and whole or partial gene deletion mutations occur in OCNDS. We don't yet know if the severity of the clinical findings is correlated with different mutation types.

How is OCNDS inherited? Is there any recurrence risk?

CSNK2A1 mutations are new in the individual with OCNDS (de novo) and are not present in either parent. The risk for parents to have another child with OCNDS in a future pregnancy is ~1% because there is a small chance that one of the parents has additional egg or sperm cells with the CSNK2A1 mutation. If an individual with OCNDS has children, there is a 50% risk of passing the CSNK2A1 mutation to their children.

Is there any gender bias?

OCNDS affects both males and females.

Is there a disease organization?

You can contact CSNK2A1 (OCNDS) Foundation at <https://www.csnk2a1foundation.org/>

How can I reach out to other families?

You can connect with families via their Facebook group page <https://www.facebook.com/groups/524315764434784/?ref=bookmarks>



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
- Motor delay (i.e., walking)
- Intellectual disabilities, learning disabilities, autism spectrum disorder traits
- Behavioral challenges such as tantrums, hand flapping, and other stereotypic movements
- Sleep problems due to disrupted circadian rhythm
- Neurologic problems such as low muscle tone (hypotonia), clumsy movements, small head (microcephaly), epilepsy (seizures), gait abnormalities
- Nonspecific structural abnormalities in the brain
- Short stature; often time without growth hormone deficiency
- Feeding difficulties starting from birth; Reflux (heartburn), constipation
- Minor infections of the ears and lung
- Crooked (misaligned) teeth and cavities
- Hypermobility, Hernias, Hip dysplasia
- Vision issues such as strabismus, near/far sightedness, astigmatism
- Minor skeletal deformations in vertebrae

Is there a cure or treatment for OCNDS?

There is not a definitive cure for OCNDS yet; however, specific measures should be taken for associated findings:

- Speech therapy, including assistive communication, starting at 12 months of age
- Intellectual and behavioral assessments with appropriate educational support
- Physical and occupational therapy for motor delays
- Monitor and evaluate for epilepsy and gait difficulties
- Monitoring growth and use of G-tubes if there are persistent feeding issues
- Evaluate vision, teeth, and immune system function
- For more detailed recommendations, you can visit the OCNDS chapter on GeneReviews at <https://www.ncbi.nlm.nih.gov/books/NBK581083/>