



CSNK2A1 FOUNDATION

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 patients are diagnosed with OCNDS?

Around 50 patients worldwide have been diagnosed with OCNDS so far. This number is expected to increase with 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?

Both missense, splice site, and whole or partial gene deletion mutations occur in OCNDS. We do not 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 de novo mutations or are new in the individual with OCNDS 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 on the CSNK2A1 mutation to their children.

Is there any gender bias?

OCNDS affects both males and females.

How can I reach out to other families?

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

Is there a disease organization?

Yes. Visit www.csnk2a1foundation.org

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 the birth.
- Reflux (heartburn), constipation
- Neurologic problems such as low muscle tone (hypotonia), clumsy movements, small head (microcephaly), epilepsy (seizures)
- 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
- Structural abnormalities in the brain

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
- Physical and occupational therapy
- Monitor growth and use of G-tubes if there are persistent feeding
- Evaluate vision

How can I learn about the future studies?

You can contact the researchers Dr. Wendy Chung and Dr. Volkan Okur on the following website
www.csnk2a1gene.com