**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 160 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.

**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/](https://www.ncbi.nlm.nih.gov/books/NBK581083/)

---

Is there a disease organization?
You can contact CSNK2A1 (OCNDS) Foundation at [https://www.csnk2a1foundation.org/](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](https://www.facebook.com/groups/524315764434784/?ref=bookmarks)

Is there a disease organization?
You can contact CSNK2A1 (OCNDS) Foundation at [https://www.csnk2a1foundation.org/](https://www.csnk2a1foundation.org/)