

OKUR-CHUNG NEURODEVELOPMENTAL SYNDROME (OCNDS) CARE RECOMMENDATIONS

NOTE:

This is for informational and educational purposes only and does not substitute professional medical advice or consultations with healthcare professionals. Please consult with your medical professional team for medical advice.

REFERENCES

Reports from the online community: [OCNDS Gene Reviews Chapter](#)

DEFINITION

OCNDS is a neurodevelopmental syndrome caused by variants ('mutations') in the CSNK2A1 gene, which OCNDS is located on Chromosome 20.

DIAGNOSIS

OCNDS is diagnosed through molecular genetic testing, more commonly through genetic blood tests such as Whole Exome Sequencing (WES) or Whole Genome Sequencing (WGS).

INHERITANCE

Everyone is born with two copies of the CSNK2A1 gene. OCNDS is an autosomal dominant condition which means that one copy of the gene is healthy, and the other copy has a mutation causing the syndrome. Typically, the mutation is *de novo* or "new" which means that it was not present in either parent.

FAMILY PLANNING

When OCNDS is *de novo* or new (meaning neither parent has the mutation), if the parents have more children, they have less than a 1% risk of having another affected child. Individuals with OCNDS have a 50% chance of passing it on to their children.

COMMON FEATURES

The impact of OCNDS varies in occurrence and severity between individuals. The table summarizes the main symptoms reported in publications about individuals with OCNDS. Developmental delay affects all areas of development, but language is more impaired than gross motor skills in most individuals. Please note that these symptoms may or may not be present for everyone with the diagnosis.



COMMON FEATURES OF OCNDS

Symptom	Potential Impact	Recommended Evaluation	Recommended Intervention
Microcephaly	Smaller head size	MRI if needed	None
Hypotonia	Trouble with: <ul style="list-style-type: none"> • Mobility • Activities of Daily Living • Loose/hyperextensible joints due to reduced muscle tone 	<ul style="list-style-type: none"> • Physical Therapy • Occupational Therapy 	<ul style="list-style-type: none"> • PT & OT • Physical medicine & rehab • Orthopedics
Speech Delays	Speech Delay	<ul style="list-style-type: none"> • Speech Evaluation • AAC evaluation 	<ul style="list-style-type: none"> • Speech Therapy • Alternative Communication Methods
Vision Difficulties	Strabismus & refractive error	Ophthalmologic assessment	Ophthalmologic assessment every 1-3 yrs
Feeding Challenges	Trouble chewing and swallowing, feeding refusal	Feeding Therapy (usually from an occupational or speech therapist)	Possible need for a NG-tube or G-tube
Gastrointestinal Challenges	Constipation	Monitor for ongoing feeding issues & signs/symptoms of constipation.	Stool softeners, prokinetics, osmoticagents, or laxatives as needed
Seizures	Varying types	Neurology (EEG) & head MRI	None
Global Developmental Delay	Affects all areas of development: language and motor skills	Developmental Pediatrics	Regular and frequent therapies
Sleep Challenges	Trouble falling asleep or staying asleep, sleep apnea	<ul style="list-style-type: none"> • Sleep disorder clinic • Sleep study 	None
Learning Disabilities	Challenges at school	Special Education Team	<ul style="list-style-type: none"> • Special accommodations • Hypotonia and communication support
Behavioral	<ul style="list-style-type: none"> • Stereotypic movements • Autism spectrum disorder • Aggressiveness and tantrums • ADD/ADHD 	Individualized Education Plan (IEP)	Environmental supports: least restrictive environment
Oral Structural Differences	Crooked (misaligned) teeth and cavities	Dentist or Orthodontics	<ul style="list-style-type: none"> • Dental treatments • Potential orthodontics treatment
Musculoskeletal	Scoliosis, kyphoscoliosis (less common)	Orthopedist	None
Heart Condition	Congenital heart defects	Cardiologist (ECG), Echocardiography	Regular cardiologist monitoring
Sensory Challenges	Autism	Applied Behavior Analysis (ABA)	Consultation with a developmental pediatrician

RECOMMENDED INTERVENTIONS FREQUENCY

Maximizing on therapies, especially through early development has been proven beneficial. Families are encouraged to access therapies without sacrificing their mental health and the quality of life of the entire family. We recommend that you connect with local disability groups to get a better understanding of available resources.

DEVELOPMENT DELAYS MANAGEMENT

Per Gene Reviews Article: [OCNDS Gene Review](#)

Consultation with a developmental pediatrician is recommended to ensure the involvement of appropriate community, state, and educational agencies and to support parents in maximizing quality of life.

The following information represents typical management recommendations for individuals with developmental delay in the United States; standard recommendations may vary from country to country.

Ages 0-3 years: Referral to an early intervention program is recommended for access to occupational, physical, speech, and feeding therapy as well as infant mental health services, special educators, and sensory impairment specialists. In the US, early intervention is a federally funded program available in all states that provides in-home services to target individual therapy needs.

Ages 3-5 years: In the US, developmental preschool through the local public school district is recommended. Before placement, an evaluation is made to determine needed services and therapies and an individualized education plan (IEP) is developed for those who qualify based on established motor, language, social, or cognitive delay. The early intervention program typically assists with this transition. Developmental preschool is center based; for children too medically unstable to attend, home-based services are provided.

[Early Intervention Contact Information by State | CDC](#)
[State Waivers List | Medicaid](#)

All ages: Consultation with a developmental pediatrician is recommended to ensure the involvement of appropriate community, state, and educational agencies (US) and to support parents in maximizing quality of life. Some issues to consider:

IEP services:

- An IEP provides specially designed instruction and related services to children who qualify.
- IEP services will be reviewed annually to determine whether any changes are needed.
- Special education law requires that children participating in an IEP be in the least restrictive environment feasible at school and included in general education as much as possible, when and where appropriate.
- Vision consultants should be a part of the child's IEP team to support access to academic material.
- PT, OT, and speech services will be provided in the IEP to the extent that the need affects the child's access to academic material. Beyond that, private supportive therapies based on the affected individual's needs may be considered. Specific recommendations regarding type of therapy can be made by a developmental pediatrician.
- As a child enters the teen years, a transition plan should be discussed and incorporated in the IEP. For those receiving IEP services, the public school district is required to provide services until age 21.

A 504 plan (Section 504: a US federal statute that prohibits discrimination based on disability) can be considered for those who require accommodations or modifications such as front-of-class seating, assistive technology devices, classroom scribes, extra time between classes, modified assignments, and enlarged text.

Developmental Disabilities Administration (DDA) enrollment is recommended. DDA is a US public agency that provides services and support to qualified individuals. Eligibility differs by state but is typically determined by diagnosis and/or associated cognitive/adaptive disabilities.

Families with limited income and resources may also qualify for supplemental security income (SSI) for their child with a disability.

SPECIAL ACCOMMODATIONS

Communication:

Consider evaluation for alternative means of communication (e.g., augmentative and alternative communication [AAC]) for individuals who have expressive language difficulties. An AAC evaluation can be completed by a speech-language pathologist who has expertise in the area. The evaluation will consider cognitive abilities and sensory impairments to determine the most appropriate form of communication. AAC devices can range from low-tech, such as picture exchange communication, to high-tech, such as voice-generating devices. Contrary to popular belief, AAC devices do not hinder verbal development of speech, but rather support optimal speech and language development.

Assistive Technology:

The field of assistive technology has come a long way and can expand abilities. Some examples include communication devices, communication buttons, speech to text, typing, and read/write programs.

Motor Support:

Physical therapy is recommended to maximize mobility and to reduce the risk for later-onset orthopedic problems. Consider use of durable medical equipment and positioning devices as needed (e.g., wheelchairs, walkers, bath chairs, orthotics, adaptive strollers). Occupational therapy is recommended for difficulty with fine motor skills that affect adaptive function such as feeding, grooming, dressing, and writing.

Special Diet:

Infant feeding difficulties are common and manifest as poor suck in early infancy or difficulty transitioning to solid foods later in infancy. Feeding and swallowing difficulties may require gastrostomy tube placement. Constipation is common.

Dental:

Crooked (misaligned) teeth and cavities are common symptoms of OCNDS.

ALTERNATIVE THERAPIES

These are interventions that our families have tried and recommended.

- 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
- Floor time Therapy
- Vision Therapy
- Communication Technology

Specialist our families have tried and recommended:

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

REGIONAL AMBASSADOR PROGRAM

Our regional ambassadors are responsible for building a supportive OCNDS community within their assigned region. Regional ambassadors connect with families as they navigate the social and emotional challenges of diagnosis, treatment, and management. To best support our global community, OCNDS Regional Ambassadors span across regions and languages including English, German, Norwegian, Spanish and Dutch. You can connect with your local community and access local resources as well as participate in virtual and in-person regional events. Email info@csnk2a1foundation.org to learn more.

USEFUL RESOURCES

[OCNDS FAQ](#)
[Parent Booklet](#)
[One Pager](#)
[Business Card](#)

Website: www.csnk2a1foundation.org/


Facebook: [@csnk2a1](#)


Instagram: [@csnk2a1_foundation](#)


LinkedIn: [CSNK2A1 Foundation](#)

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