UNIQUE
UNDERSTANDING GENES & CHROMOSOMES

Okur-Chung neurodevelopmental syndrome (OCNDS)

(*CSNK2A1* variants or deletions)

rarechromo.org

This guide is designed to help families and healthcare professionals looking after people with Okur-Chung neurodevelopmental syndrome (OCNDS). It contains information about the cause of the condition, the ways in which it can affect people and suggestions about the help and management techniques that can benefit affected individuals.

What is Okur-Chung neurodevelopmental syndrome (OCNDS)?

Okur-Chung neurodevelopmental syndrome, also referred to as OCNDS, is a rare genetic condition associated with developmental delays, speech disorders or delays, and varying differences in brain function. As with many genetic conditions, individuals with OCNDS can be affected in different ways. Not everyone will have all possible features, and the severity of symptoms can vary from person to person, including among people with the same genetic change.

OCNDS is caused by either a variant (change) in the CSNK2A1 gene, or by a deletion (loss of one copy of the gene, or part of it). The deletion may occur as part of a larger change that affects the chromosome where the gene is located. OCNDS is also referred to as CSNK2A1-related neurodevelopmental disorder (CSNK2A1-NDD) due to the genetic cause. The genetic basis and causes of OCNDS are described in more detail later in this guide.

How common is Okur-Chung neurodevelopmental syndrome?

OCNDS is extremely rare with an estimated prevalence of 1 in every 100,000 live births. Currently (2026), about 51 individuals with a variant in the CSNK2A1 gene have been reported in the medical literature, but more have been diagnosed. The CSNK2A1 Foundation has a patient registry, which includes over 400 individuals with OCNDS as of July 2026. It is expected that more people will be diagnosed with this condition as awareness increases and genetic testing becomes more routine.

What features and symptoms do individuals with Okur-Chung neurodevelopmental syndrome have?

As is the case with many genetic conditions, children and adults with OCNDS can have a range of features and symptoms. As more people are diagnosed and information is shared, the range of features, and the likelihood of a child or adult having these features, will become clearer.

Common features

Most children with Okur-Chung neurodevelopmental syndrome have:

- Some degree of developmental delay, ranging from mild to severe
- Some degree of intellectual disability (ID) or learning difficulties (LD), ranging from mild to moderate
- Speech and language delay, or are non-verbal
- Sleep issues and sleep disorders
- Feeding difficulties, which usually resolve after babyhood or early childhood; some children will need to use a G-tube
- Gastro-oesophageal reflux (GERD/GORD)
- Constipation
- Characteristic facial features including short and round facial structure/broad nasal tip
- Low muscle tone (hypotonia)
- Joint hypermobility
- Minor ear and respiratory infections, which usually resolve during childhood
- Behavioural differences e.g. autistic spectrum disorder (ASD), ADHD, anxiety, tantrums, self-injurious behaviour, and differing movements such as hand flapping
- Small head size (microcephaly)
- Growth delay

- Motor delay
- Eye/vision anomaly
- Brain anomaly e.g. non-specific MRI abnormalities including delayed myelination, smaller anterior pituitary gland, and thin corpus callosum
- Seizures and/or epilepsy disorders
- Dental issues e.g. misaligned teeth, cavities, late teething, over-crowding, tooth-grinding, weak enamel
- Hernias

Other possible features include:

- Spinal curvature e.g. scoliosis, kyphosis
- Short stature
- Hip dysplasia
- High muscle tone (hypertonia)
- Large head size (macrocephaly)
- Anomalies of the hands and feet
- Skin condition e.g. eczema
- Anomalies of the kidneys and genitals (urogenital anomalies)
- Hormone (endocrine) anomalies
- Connective tissue anomalies
- Heart condition, which often resolves naturally
- Skeletal anomaly
- Hearing loss
- Cleft lip/palate or other palate anomalies
- Delayed bone age
- Overgrowth

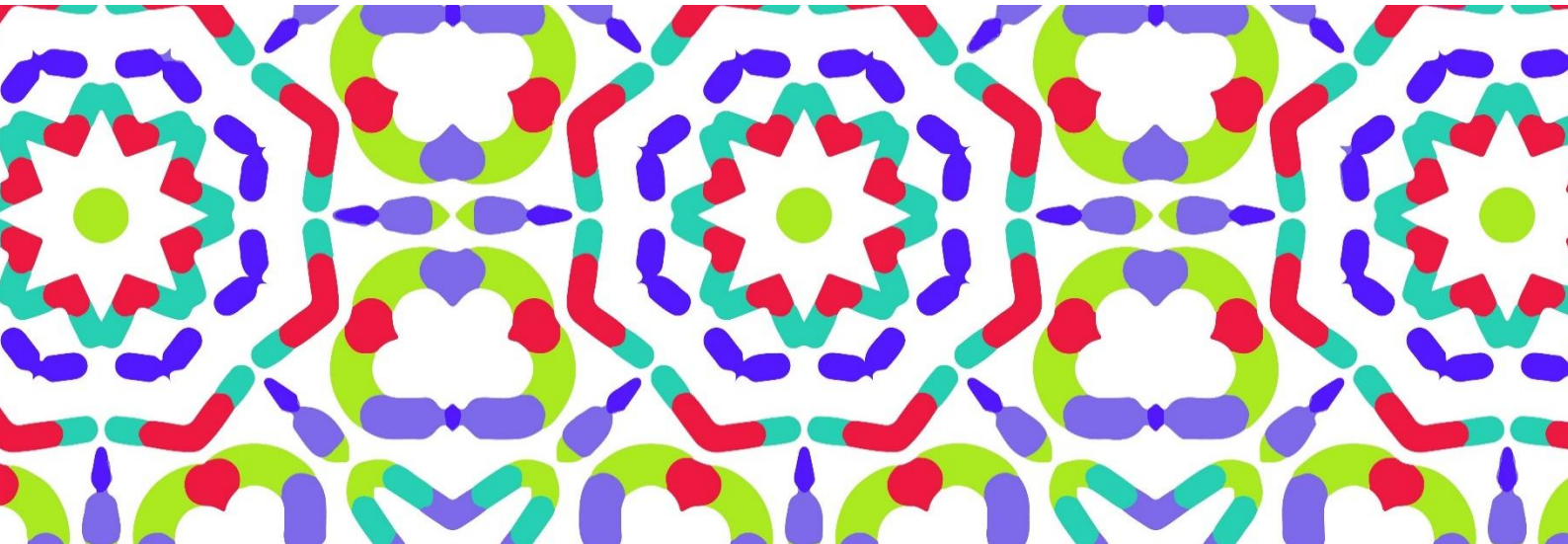
Pregnancy and birth

While most pregnancies are unremarkable and proceed without complication, some concerns during pregnancy have been reported. Only a small number of pregnancies have been described in the literature, so it is difficult to draw any firm conclusions. Published literature so far (2026) has reported one baby who showed slow growth in the womb, but the child also had an additional genetic diagnosis (Kratochwila 2025). Another baby was reported to have multiple heart conditions and sadly did not survive (Zhang 2025).

Most pregnancies go to term, but a few babies are born prematurely. Some babies show some signs of difficulty at birth, related to difficulties with feeding, jaundice, or a heart condition. Birth weights may be below average.

Appearance

Certain facial features are found more often in children with OCNDS than in other children. These features may mean that you see unexpected similarities between your child and others with OCNDS. The most common characteristic features include a round face, wide-set eyes, skin folds at the inner corner of the eye, a pointed chin, low-set ears, and a broad nose with a bulbous tip.



Development

Gross and fine motor skills

Developmental delay has been reported in most children with OCNDS so far (Bagatelas 2025). The degree of delay ranges from mild to severe. Developmental milestones, including rolling, sitting, walking, playing with toys, using cutlery, using zips and buttons, and toilet training, are often delayed. However, there is a wide range of eventual ability, with some children acquiring mobility and other skills around the same age as typically developing children, while others show a more obvious delay. Low muscle tone (hypotonia) and/or loose (hypermobile) joints are common and may affect mobility. Some children may have an unusual gait when walking because of stiffness or balance issues (known as ataxia). Many benefit from early intervention with treatments or therapies, such as orthotics (e.g. insoles, braces, splints and callipers), occupational therapy (OT), and physiotherapy (PT).

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Families say …

“His physical abilities have declined massively, so he now requires full support in all areas. We need to dress him in the mornings and address his intimate care needs. He uses a walker to get around the house and a power wheelchair when we leave the house.”

“He has always required physiotherapy due to being delayed with all his gross motor skills, and then as time went, on he needed it due to losing all his gross motor skills. His physiotherapist has been incredible and supportive of his ever-changing needs, and when he broke his femur and had to relearn to walk, she never gave up on making sure he regained that skill.”

“The biggest impact has been our son’s loss of physical abilities. We had no inclination that this would occur, so it's been a massive life alteration to manage now having a child dependent on a wheelchair and full physical support. We have lost many opportunities we used to avail of as a family, our abilities to go places have become very restricted as many places don't cater for wheelchairs. His loss of physical ability has increased his need for support, meaning we now have to provide him with support that would be equivalent to a toddler, yet he has the learning ability of an older child. The impact on our whole family has been huge.”

Intellectual development and learning

Most children with OCNDS have intellectual disability (ID) or learning difficulties. ID ranges from mild to severe but is usually in the mild-moderate range, and many children have needed additional support with their learning. Early intervention can prove particularly beneficial, and formal testing to assess specific, individual needs is recommended.

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Individuals with OCNDS often benefit from tailored educational support, as learning needs can vary, and the diagnosis may be unfamiliar to many educators. Clear communication between families and schools is important for effective planning. The CSNK2A1 Foundation has developed educational resources to help families explain OCNDS and its potential impact in school settings. These include a brief slide set and a companion script that support discussions with teachers, special educational needs coordinators (SENCOs), and other education professionals.

CSNK2A1 Foundation educational resources information can be found here

These resources can help schools understand a child's individual strengths, challenges, and therapy needs and support planning for appropriate adjustments and interventions. They may be adapted for use within the UK education system, including the Education, Health and Care Plan (EHCP) process, and across mainstream or specialist settings.

> ***"At school something that worked well was small classes and one-to-one teaching assistants. A challenge we have had at school is anxiety to return after holidays and transition to a new setting."***

"He attends a special school, but we have frequent school refusals due to high anxiety levels. He does love school; he's very bright & loves learning but his anxiety can be overwhelming for him. A supportive team and visual schedule of his days have been a huge help for him. He does struggle with friendships; he doesn't quite understand them. He loves having friends but doesn't quite manage the stereotypical style of friendships."

"He was put into an "unknown syndrome" category at an early age since none of the genetic tests he had completed found anything. At 4 months of age, he began receiving Early Intervention services. I feel this was an important step in helping him to begin his development into what he could be. He received occupational, physical, and speech therapies (each 2x/week) until he reached preschool age at 3 years. He then transitioned into a 12:1+1 preschool setting for one year and then transitioned to an 8:1+1 setting for his second year of preschool. For K–1st grades, he was kept in an 8:1+1 setting through our local BOCES as his old home school district didn't have any placements that met his needs. Needing a smaller class size, this was appropriate at that time. Upon entering 2nd grade, we had moved to another school district, and he was placed in a 12:1+1, where he truly began to flourish. He remained in this setting until he transitioned into a BOCES 15:1 setting during 9th grade that focused more on life skills. He still maintained all levels of therapies throughout the years until he reached about 18–19 years of age, when he became more hesitant to attend sessions. We all agreed to stop since he was refusing to go. He remained in school until he reached the age of 21, then he aged out."

"Something that worked well for our son throughout his education was being in a smaller class setting that was more focused on his needs with less distractions. Getting into our local BOCES [Boards of Cooperative Educational Services] program was the biggest help for him since the placement focused more on life skills than academics. He was always a happy-go-lucky kid. We never met challenges until he was older and realized he was "different." The biggest challenge for us was him going to therapies."

"As my child got older one big challenge was the gap increasing between herself and her peers. One support that helped during that transition was moving from a mainstream to special school."

Speech and language

Children with OCNDS typically experience some degree of speech and language delay, and some may find it difficult to coordinate movement of their lips, jaw, and tongue to make the right sounds (apraxia of speech). The eventual range of achievement is broad, but some may remain non-verbal. Those who develop speech may achieve single words, short phrases, or basic sentences, and some go on to develop conversational skills and a broad vocabulary. Many parents believe that their child can understand a lot more than they can express.

An assessment by a speech therapist should be able to identify your child's specific difficulties, allowing regular therapy sessions tailored to your child's specific areas of need. Where individuals have no speech or very few words, Augmentative and Alternative Communication (AAC) methods, including pointing, pictograms, gestures, facial expressions, simplified sign language, and high-tech communication systems (aided communication) have enabled many to communicate their thoughts and needs.

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Feeding

Feeding issues in the new-born period are common. Low muscle tone may contribute to difficulties with swallowing, and some babies will suck weakly and may need high energy milks to encourage weight gain. Some babies also suffer from gastro-oesophageal reflux (GERD/GORD) (in which feeds return readily up the food passage), which may require treatment, including careful positioning for feeds, medication, nutritional supplements, or, in some cases, insertion of a nasogastric tube (NGT) or percutaneous endoscopic gastrostomy tube (PEG/G-tube). Aspiration (where fluid, food, or saliva enters the airway or lungs) has also been reported as an issue. Some children have benefited from attending a feeding clinic, where specialists can assess eating and drinking difficulties and provide advice on how to treat them.

Unique publishes a separate guide to Feeding

"A typical day looks like very little breakfast, get dressed, and then school or TV. Playing with baby dolls, watching YouTube on tablet. Grazing / snacking throughout the day, very small meals. Bedtime story and co-sleeping."

"He can eat independently but is unable to create any meals for himself."

Growth and stature

Many children with OCNDS described in the medical literature so far (2026) have short stature and a smaller than expected head size (microcephaly). In one case, a child's bone age was delayed compared to their actual (chronological) age (Martinez-Monseny 2019). Beyond infancy, height and weight may remain below average.

Behaviour

Children with OCNDS display behaviour consistent with their overall degree of developmental delay, and most have a happy disposition. Children with OCNDS are known to be affectionate, socially-oriented, and have a positive viewpoint. Some children have an autism spectrum disorder (ASD) diagnosis or traits. Other behaviours, including attention deficit hyperactivity disorder (ADHD), anxiety, self-harming, aggression, and obsessive-compulsive disorder (OCD) have also been reported.

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"When behaviour or regulation was hard, one thing that helped was playing favourite music and offering rewards."

"His anxiety is our biggest hurdle. He has severe anxiety and tends to not want to go far from home. He is content with familiar routines such as walking the dogs with his dad, going to the cinema, etc. We have to work consistently to manage his anxiety."

"Fortunately for us, we didn't face any behavior or emotional challenges. Our son was diagnosed with ADHD and was medicated at one point, but then he didn't like the way it made him feel so we stopped the medication."

Autism

Community reports from the CSNK2A1 Foundation indicate that almost one third of individuals with OCNDS have autism spectrum disorder (ASD) (private communication CSNK2A1 Foundation; Bagatelas 2025). Autism is a neurodevelopmental condition. Children with OCNDS who show autism-related features may benefit from therapies commonly used for autism, such as applied behaviour analysis (ABA), which is individualized to each child's strengths and needs. A developmental paediatrician can help guide behaviour strategies and, when appropriate, prescribe medications such as those used for ADHD. For more severe behavioural concerns, including aggression or destructive behaviours, evaluation by a paediatric psychiatrist may be helpful.

"The challenges that affect our family the most are anxiety, not sleeping in own bed, incontinence, autistic behaviour, such as inflexible / rigid routines, and avoidance of anything new or different."

Sleep

Most children with OCNDS have some form of sleep difficulty, ranging from issues with initiating and maintaining sleep, the need to sleep in the same room or bed as a family member, insufficient quantity of sleep, daytime drowsiness, and severely disrupted sleep patterns. Some children also have unusual breathing patterns during sleep (sleep apnoea).

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Puberty

There is limited information available about puberty in children with OCNDS. We do know that some children appeared to go through puberty as expected. Some individuals with OCNDS experience delayed puberty,

which can be associated with low sex hormone levels, or other endocrine-related symptoms, such as undescended testicles (Bagatelas 2025). Some families of children with genetic disorders and behavioural or learning difficulties can be particularly concerned about their daughters' ability to cope with menstruation, and for some, discussing menstrual regulation options with a paediatrician may be beneficial.

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Adulthood

Experiences of adulthood are likely to vary considerably and will depend on many factors. These include the level of any language disorders or intellectual disabilities, possible ongoing medical concerns, and improvements in early intervention and therapies/treatments.

Adults with OCNDS have varying levels of independence. Some have gone on to have families of their own and manage to run their own households and work with significant help from a spouse. Most continue to live with their parents or in supported settings, such as a group/residential care home, with caregivers who can provide support. A few may live independently but require support from family or friends with certain tasks. Levels of employment, and the nature of employment, varies but some do undertake a form of paid or unpaid employment. Several adults with OCNDS only discovered that they had a CSNK2A1 variant or deletion when their child was diagnosed.

Unique publishes a separate guide to Transition

"Our son transitioned very easily. Once he aged out of school at 21, he was able to start using Self-Direct services. With this, he is able to hire an aide and get 40 hours/week to work on goals. He does volunteer work with Meals on Wheels and our local food pantry. He goes to the gym a couple times a week and also has equine therapy 1x/week. He loves chatting with his friends and going out with them to do things. Currently, he still lives at home."

Medical concerns

The following medical concerns have been found in children and adults with OCNDS. Not all individuals with OCNDS will be affected by a given feature or condition.

Eyes and sight

Problems with eyes and vision are common in children with OCNDS. A wide range of conditions have been reported, and an individual may have more than one vision or eye-related concern.

The most common known concerns include: a slight change in eye shape that can lead to blurry vision (astigmatism); a squint (strabismus), where one eye or both turns inward, outward, up, or down, which may be treated with patching, glasses, exercises, or surgical correction; short or long-sightedness (myopia and hypermetropia), which can usually be corrected by glasses; a “lazy eye” (amblyopia), which can be a consequence of a constant squint in one eye; and droopy upper eyelids (ptosis).

Less common conditions that may be observed include: difficulty closing the eyelids completely (lagophthalmos), which can lead to dry and irritated eyes; widely-spaced eyes (hypertelorism) or eyes that are slightly slanted; uncontrolled eye movements (nystagmus); underdevelopment (hypoplasia) of the optic nerve; glaucoma (an eye condition that leads to progressive damage to the optic nerve); cataracts (when the usually clear lens of the eye develops cloudy patches); Duane anomaly (misalignment of the eyes that varies with gaze direction); and retinal dystrophy (a range of chronic and progressive eye conditions affecting vision).

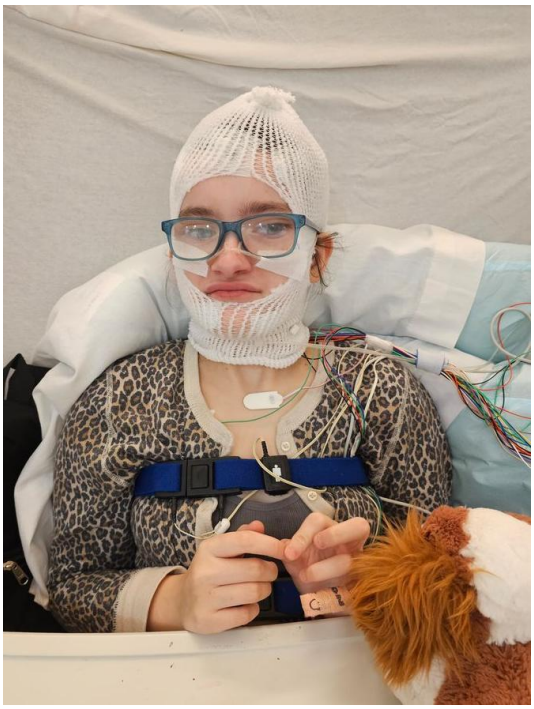
Skin

Some children with OCNDS have a skin condition (e.g. eczema) where the skin becomes red, itchy, and inflamed. Your doctor should be able to recommend self-care techniques, emollients, and other treatments that may help to relieve symptoms.

Benign (non-cancerous) skin lesions known as pilomatricomas, pilomatrixoma, calcifying epithelioma of Malherbe, or trichomatricoma, have been found in a few people with OCNDS. Pilomatricomas are harmless, but occasionally they may burst and release a white and yellow chalky fluid. Very occasionally, they can become sore and inflamed if they become infected, so picking and squeezing them should be avoided. Where necessary, pilomatricomas may be surgically removed.

Other skin conditions that have been reported include skin tags that either fall off naturally or can be surgically removed, and cutaneous hemangioma (an unusual build-up of blood vessels on or under the surface of the skin that can look like a red-coloured birthmark).

Seizures



About one third of children with OCNDS experience some form of seizure (a sudden and unexpected change in the electrical activity in the brain)(Chung 2022, Bagatelas 2025). Depending on the part(s) of the brain affected, symptoms vary, but include temporary confusion, uncontrollable jerking movements, and loss of consciousness or awareness. Age of onset can vary considerably, while seizures may be isolated to a single incident or occur more regularly. More than one type of seizure may be present in the same individual.

Electroencephalograph (EEG) and video telemetry (video EEG) are medical tests that can be used to measure and record the electrical activity of the brain and are tools that, when used alongside other tests, can help diagnose the type of seizure experienced.

Seizures can cause a lot of worry for families and can be frightening to observe, but in some cases, they self-resolve or resolve with medical treatment. Valproic acid, Lamotrigine, Levetiracetam, Carbamazepine, Topiramate, and Benzodiazepines are commonly used for seizure control. If your child has a seizure for the first time, it is important to remove nearby hazards so they can't hurt themselves and to contact a medical professional. Most individuals with OCNDS and seizures control the seizures using existing medications. A few patients still have seizures despite taking medication(s).

Seizure types may include:

Absence seizure: A change in behaviour, as if the child 'switches off', sometimes with staring, eyelid flickering, or lip smacking. Absences are very brief, often lasting less than half a minute.

Generalised tonic clonic: At the onset of a seizure, the abnormal electrical activity involves both sides of the brain. The seizure involves a phase of stiffening, followed by jerking.

Myotonic: Seizure involving a temporary loss of muscle control.

Atonic: Seizure involving stiffening of the muscles.

Myoclonic-atonic: Seizure involving jerky or shock-like contraction of muscles, followed by a loss of tone so someone standing up falls to the ground.

Infantile spasm: Type of seizure usually occurring in clusters in babies between 3-10 months. Seen most often when a baby wakes and may be obvious or subtle.

Complex partial seizure: Type of seizure that starts in one area of the brain and causes impaired awareness, often with staring, unresponsiveness, or automatic movements.

Simple partial seizure: Type of seizure that starts in one area of the brain and does not impair awareness, with symptoms such as unusual sensations, movements, or emotions.

Genitals

Minor anomalies of the genitals in boys have occasionally been reported, including undescended testes (cryptorchidism). These anomalies can also be seen in children without OCNDS. If necessary, most can be corrected with surgery.

Females are much less likely to be affected. One condition that has been reported, rarely, is when the inner lips of the vulva stick together (labial adhesions).

Kidney and urinary tract

A few babies are born with minor anomalies of the kidneys and/or urinary tract. Reported anomalies include a duplicated renal collection system and a kidney that is not found in its expected position (ectopic kidney). “Kidney stretching” (pelvicaliectasia – a dilation of part of the kidneys) has also been reported.

Breathing

Issues reported in OCNDS include sleep apnoea (when there is an abnormal breathing pattern during sleep).

Joints

Joint anomalies are a known feature of OCNDS. These include extremely loose (hyper-mobile) joints (elbows, wrists, knees, hips), which mean babies and children can move their limbs into positions others find impossible. While this may not cause issues, hyper-mobility is sometimes associated with pain and stiffness in the joints and muscles, joints that dislocate (come out of position) easily, and injuries, including sprains. Children with very loose joints may need physiotherapy, massage, or additional braces (supports, splints) before they are able to walk. In some cases, joints are unusually tight and may require surgery and tendon lengthening to extend their range of movement. Some children have a degree of hip dysplasia, in which the hip joints are easily dislocated. This may be apparent at birth or develop later. In either case, it is treated with splinting and, if necessary, immobilisation in plaster and possibly surgery.

Heart

Heart condition(s) have been reported in individuals with OCNDS so far (2026). These can be present at birth (congenital) or develop later in life. Suspected heart problems in children can be diagnosed using tests like an electrocardiogram (ECG) (recording the electrical activity of the heart), echocardiogram (ultrasound scan of the heart), or chest X-ray. The types of heart condition(s) are variable but include anomalies affecting the size and structure of the heart muscle and valves.

Some of these conditions are relatively minor and resolve naturally in time. Medical treatment may be necessary for others, and some may require surgery. Most often, these are congenital (present at birth) heart conditions and include dilation of the aorta and atrial septal defect. A hole between the top two chambers of the heart has been reported in a few individuals. Additionally, one individual was reported to have Tetralogy of Fallot, a condition that is a combination of four heart anomalies.

Brain

Many children with OCNDS have a structural brain anomaly, which can be detected by an MRI (magnetic resonance imaging) or a CT (computerised tomography) scan of their brain. The observed changes vary but include mostly nonspecific anomalies.

Examples include:

Delayed myelination: the protective sheath around nerves in the brain develops slower than expected.

Smaller anterior pituitary gland: part of the pituitary gland, which produces and releases hormones, is smaller than expected.

Thin corpus callosum: the white matter that connects the two brain hemispheres is not as thick as expected.

Reduced volume of the cerebellar vermis: a structure that separates the two hemispheres in the brain near the back of the head has a lower volume than expected.

Simple gyral pattern: a reduced number of ridges on the brain and shallow grooves.

Cerebral atrophy: a reduced brain volume due to loss of neurons.

Mega cisterna magna: an enlarged area behind the cerebellum, a part of the brain that controls movement and coordination.

Lesion on the pineal gland: rare growths on a gland at the centre of the brain that produces melatonin, which controls sleep-wake cycles.

Duplication of the pituitary gland: having an extra pituitary gland, a major hormone producing gland.

Rathke cleft cysts: benign, fluid-filled sacs near the pituitary gland.

Absent olfactory bulbs: the neural structures above each nasal cavity, that process smells, are missing.

Constipation

Constipation is common among children with OCNDS and can be related to low muscle tone, little exercise, a low-bulk diet, and small fluid intake, among other reasons that are not fully understood. It is important that possible causes are discussed with a health visitor or doctor, who may recommend adapting diet or giving stool softeners or laxatives.

Hormones

Children with OCNDS may have conditions that affect their hormones. Conditions that have been reported so far (2026) include early (precocious) puberty, polycystic ovarian syndrome, an underactive thyroid (hypothyroidism), growth hormone deficiency, and delayed bone age.

Hands and feet

Children with OCNDS occasionally have anomalies of the hands and feet. Among these are fingers or toes that curve inwards (clinodactyly), fingers or toes that are unusually short (brachydactyly) or fused (syndactyly), and underdeveloped (hypoplastic) or misshapen nails. Some children may benefit from massage, orthotics, and physiotherapy. Treatment is tailored to the individual child, and in some cases, surgical correction will best enhance eventual mobility.

Spine

Some babies are born with or develop a spinal curvature, either a sideways curve of the spine (scoliosis), a rounding of the upper back (kyphosis), or kyphoscoliosis (a combination of kyphosis and scoliosis). The curvature can be treated with physiotherapy and exercises, or a support brace or surgery may be needed.

Teeth

Dental concerns are very common in children with genetic disorders. A number of issues have been described by parents of children with OCNDS (Ming 2024), including unusual dental development; an unusual size of the jaw, leading to overcrowding or widely-spaced teeth; feeding difficulties and delayed eating and chewing activity; unusually thin, weak enamel (enamel hypoplasia); and tooth grinding (bruxism), which can prematurely wear down the enamel. Teeth may emerge late, and milk teeth may be late to fall out. A high standard of dental care is important to minimise damage by decay and erosion (by grinding). Children and adults may also benefit from specialist hospital dental services and may require treatment under general anaesthetic.

Unique publishes separate guides to Looking after your child's teeth and Teeth: common concerns

Hernias

Some babies may be born with a hernia, where an organ or fatty tissue pushes through a weak spot in a surrounding muscle or tissue. These include instances of umbilical (at or near the belly button) and inguinal (inner groin) hernias.

Palate

Anomalies of the palate (roof of the mouth), ranging from those that may be invisible to the casual onlooker, such as a high/arched palate, to more obvious conditions, such as a cleft palate, have been reported. Anomalies of the palate, particularly clefting, can cause difficulties in feeding, hearing, teething, and speech production. As well as helping aesthetically, surgical repair eases these problems and may eliminate them altogether.

Temperature regulation

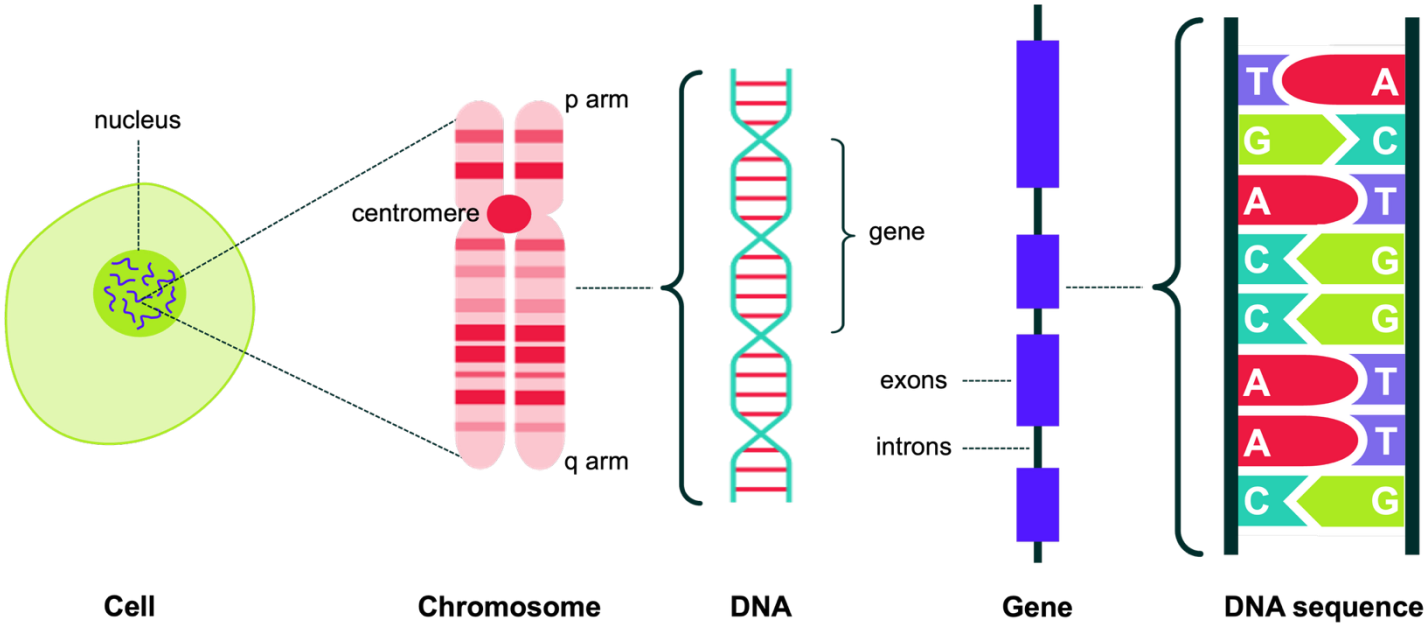
A few families have reported that their child has trouble regulating body temperature. This can range from a low body temperature to excessive sweating of their hands and feet (private communication CSNK2A1 Foundation).

This section includes some more complicated scientific terms and concepts — don't worry if it's a lot to take in, you can always come back to this section later if you want to.

Genes and chromosomes

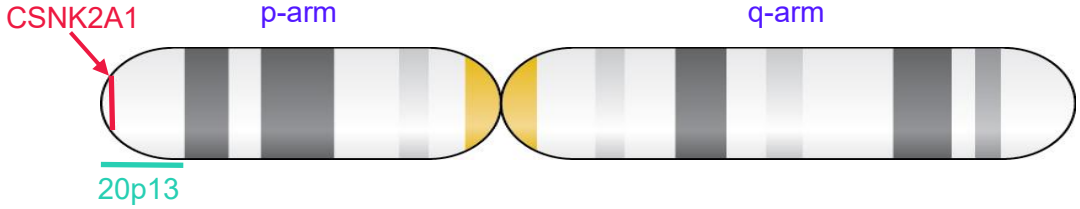
Genes are instructions that have important roles in our growth and development. They are made of DNA and are incorporated into organised structures called chromosomes. Chromosomes therefore contain our genetic information. We typically have 46 chromosomes, with one set of 23 inherited from each parent. Chromosomes are located inside our cells, the building blocks of our bodies. In people with genetic conditions, one or more of their genes don't instruct the body as we would expect, which can lead to changes in how their body works.

DNA is made up of building blocks called "bases" or "nucleotides." There are four DNA bases which can be abbreviated to the letters A, C, G, and T. These DNA bases are paired up in the DNA structure into "base-pairs." The full sequence of our DNA is over three billion base-pairs long.



OCNDS is caused by specific changes (known as pathogenic variants) to the DNA sequence of a gene called CSNK2A1. CSNK2A1 is an abbreviation of the gene's full name, *casein kinase 2 alpha 1*. OCNDS can also be caused by the partial or entire loss of the CSNK2A1 gene due to a chromosome deletion, which leaves only one working copy of the gene. This disrupts typical development and can lead to OCNDS. The CSNK2A1 gene is located in the short p arm of chromosome 20 in a region called 20p13, as shown in the image below.

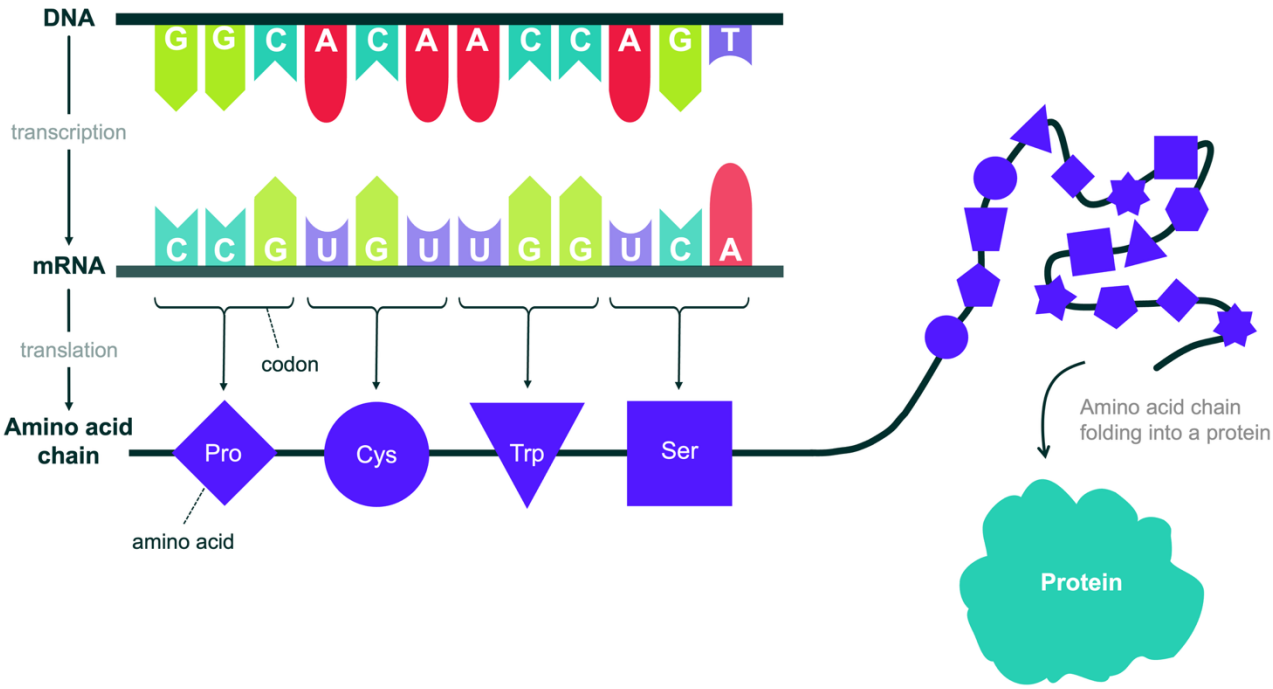
Chromosome 20



We have two copies of chromosome 20 in our cells, so we also have two copies of the CSNK2A1 gene. In OCNDS, only one copy of the CSNK2A1 gene is affected; the second copy is fully functional. This is known as autosomal dominant, since all numbered chromosomes are called autosomes, and genetic conditions that occur when only one copy of an autosomal gene is affected are known as dominant.

Protein-coding genes

Most genetic conditions are caused by changes to genes that provide instructions for making proteins; these are called protein-coding genes. CSNK2A1 is one of thousands of these protein-coding genes. Proteins are molecules that are made up of long chains of chemicals called amino acids and play critical roles in the body. This is because proteins are essential for lots of processes, such as breaking down food, moving our muscles, and growing the body's organs and making sure they function properly. To make these amino acids, a copy of the DNA sequence is created, called messenger RNA (mRNA). mRNA uses the same A, C and G bases as DNA, but the "T" bases are swapped to "U" bases. Within a gene, each three-letter sequence (codon) of mRNA bases provides the instructions for one amino acid. Changes to the amino acid sequence can affect the function of the protein.



The role of the CK2 protein

The CSNK2A1 gene sequence is used to make a protein called CK2 (Casein Kinase II). This protein is important because it regulates daily functions of cells in the body by controlling the cell cycle, regulating DNA repair, and facilitating other key cellular processes throughout the body. Scientists are still working to understand exactly how changes in the CSNK2A1 gene lead to OCNDS. Current research suggests that these genetic changes may reduce how well the CK2 protein works or change how the protein interacts with other proteins in the cell. In some cases, the change may also cause the protein to gain new or altered activity. The exact effect may depend on where the genetic change occurs within the gene.

Different types of genetic changes

The specific type of genetic change in the CSNK2A1 gene is an important factor that could influence an individual's symptoms and features. Research suggests that different types of genetic changes can affect the CK2 protein in different ways, which might account for the variability in symptoms and features, or their severity, in different people with OCNDS.

Gene deletions

In some individuals, one entire copy of the CSNK2A1 gene is missing (deleted). This means the body has only one functioning copy of the gene to produce the CK2 protein. This is called haploinsufficiency, and having half the expected amount of protein is not enough for typical development.

Unique publishes a separate guide to deletions and microdeletions

Gene sequence variants

People with OCNDS are more commonly found to have a gene sequence variant, as opposed to a deletion of the entire gene. Such variants include:

- Missense variants: Where a gene variant results in one amino acid being replaced by another, producing an altered protein. These are the most common variants found in people with OCNDS. Rather than causing the CK2 protein to work more, or less, many missense variants appear to change *how* the protein works, changing cellular processes that involve the CK2 protein. This is sometimes described as a neomorphic effect, meaning the protein gains a new or altered function; sometimes this is called a novel (new) gain-of-function.
- Nonsense variants: Where a gene variant leads in an amino acid change that results in a premature "stop" signal, which ends protein production too early and leads to a shorter, incomplete (truncated) protein
- Frameshift variants: Where the DNA coding sequence is shifted by the addition or deletion of individual DNA bases, which means the incorrect amino acid sequence is used to produce the protein, or a premature "stop" signal is introduced, and a shortened protein is formed
- Splice-site variants: Where changes in the DNA sequence of a gene at specific sites called "splice-sites" disrupt how the gene is processed, resulting in significant changes to the amino acid sequence of a protein or reduced protein production. (Before mRNA is translated into a protein, sections are removed, these sections lie between splice sites)

Some variants reduce CK2 activity, some change the protein's function, and others interfere with the unaffected protein made from the second copy of the gene (this is known as a dominant-negative effect). Because of this complexity, researchers do not usually classify CSNK2A1 variants as simply "gain of function" or "loss of function." Instead, OCNDS is best understood as a disorder of *altered* protein function.

Unique publishes a separate guide to single gene disorders

Genetic tests

OCNDS can be identified by a type of genetic test called sequencing (e.g. whole exome sequencing (WES) or whole genome sequencing (WGS)). Gene deletions can also be identified by a sequencing test but are more commonly found using a different type of genetic test called a chromosome microarray (CMA, e.g. arrayCGH or SNParray).

Unique publishes separate guides to DNA sequencing, arrayCGH and SNParrays

Genetic test results

The results of genetic (genomic) testing are likely to be given to you by your geneticist, a genetic counsellor, or the clinician who ordered the test. Depending on the test that was carried out, someone with OCNDS might have results that look like the following example:

An example result of a DNA sequencing test (e.g. WES or WGS), that can identify gene variants, is shown here for the CSNK2A1 gene:

c.593A>G p. (Lys198Arg) in the CSNK2A1 gene NM_001895.4

c.593	signifies the base pair position of the change within the gene sequence
A>G	signifies the gene sequence change: the A nucleotide has been replaced by a G nucleotide
p. Lys198Arg	signifies the change to the protein: the amino acid lysine (Lys) has been replaced by the amino acid arginine (Arg) at position 198 in the sequence of amino acids that make up the protein
CSNK2A1	signifies the gene that is affected
NM_001895.4	this is the specific "reference sequence," or blueprint, of the gene that scientists use to identify the location of the variant

The result of a chromosome microarray (CMA) test (e.g. arrayCGH or a SNParray), that can identify deletions, is shown here for a deletion within band 20p13:

arr[GRCh37] 20p13 (61,001_1,388,506)x1 dn

arr	signifies the analysis was by array (arr) comparative genomic hybridisation (cgh)
GRCh37	signifies human genome build GRCh37. This is the reference DNA sequence that the base pair numbers refer to. As more information about the human genome is found, new "builds" of the genome are made, and the base pair numbers may be adjusted. This means base pair positions change depending on the assembly used
20p13	This shows the chromosome involved is chromosome 20 and the position of the deletion is in the p-arm in band 13
61,001_1,388,506	signifies the coordinates of the DNA that has been deleted. The base pairs between 61,001 and 1,388,506 have been deleted. Subtract the first long number from the second and you get the number of base pairs that have been deleted, in this case 1,327,505 or approximately 1.3 million base pairs
x1	means there is one copy of these base pairs, not two—one on each chromosome 20—as you would normally expect, so this a deletion
dn	means *de novo*. The biological parents' chromosomes have been checked, and no deletion or other chromosome change has been found in band 20p13. The deletion is very unlikely to be inherited and has almost certainly occurred for the first time in this family with this child mat here would mean that the deletion has been inherited from the biological mother pat here would mean that it has been inherited from the biological father

Unique publishes a separate guide to Interpreting Genetic Test Results

Our son wasn't diagnosed until he was 13. We didn't know what we were looking for. We knew it was something genetic but had no diagnosis for a long time. I wish we'd had some platform of information to work from when he was diagnosed. Even now, a year later, we're very shy on information pertaining to how he'll develop going forward."

"We knew almost immediately at birth that he had differences. It took 14 years of many doctor visits and genetic testing before we got the official diagnosis of Okur-Chung neurodevelopmental syndrome. One of our geneticists had said early on, "Mom, you may never find an answer." That stayed with me throughout the years and was a phrase that I held onto and was okay with. There was no stone unturned, or road traveled in looking for an answer. Finally, in 2017 we got our answer. Although our son was older when he was diagnosed (14 years). Life before and after diagnosis has basically stayed the same; we just finally had a diagnosis for his differences."

Why did this happen?

When children are conceived, their parents' genetic material (DNA) is copied in the egg and sperm that makes a new child. The biological copying method is not perfect, and random changes occur in the genetic code of all children that are not seen in the DNA of their parents. This happens naturally and is not due to the parents' diet, environment, or lifestyle. Most of these DNA changes have no obvious effect. But in rare instances, these random DNA changes can lead to health issues or affect development.

When such a random change disrupts the function of the CSNK2A1 gene, then a child will have OCNDS. In almost all individuals with OCNDS identified so far (2026), the genetic change was a random (or "*de novo*") change, meaning the change occurred for the first time in that family in the affected individual. Very rarely, one parent may have the same change (or variant) in some of their egg or sperm cells and pass it on to their child (this is known as germline mosaicism). However, it is important to recognize that no one should be blamed for variants in their DNA, and no parent is at fault when a new DNA change occurs in their child.

Can it happen again?

The possibility of having another child affected by a rare gene disorder depends on the genetic code of the parents. In almost everyone reported with OCNDS so far (2026) the genetic alteration has been found to be *de novo*, which means neither parent was found to have the same CSNK2A1 gene change as their child, and neither parent was found to have a chromosomal rearrangement that might have resulted in a CSNK2A1 deletion in their child. Therefore, the chance of having another child with OCNDS is usually less than 1% in these cases. There are a few reports of inherited cases of OCNDS in the literature.

One reason why there is some residual chance of recurrence is due to the rare phenomenon called germline mosaicism that was mentioned above. This is when a parent carries a genetic change, but it is limited to some of their egg or sperm cells. The genetic change would not, therefore, be detected in the parents' blood tests.

Unique publishes a short general guide to Mosaicism

In families where the CSNK2A1 variant has been inherited from a parent, the possibility of having another child, either a girl or a boy, with OCNDS rises to 50% (1 in 2) in each pregnancy. However, the effect on the child's development, health, and behaviour cannot be reliably predicted. Your genetics centre should be able to offer counselling before you have another pregnancy.

If your child with a CSNK2A1 variant goes on to have children of their own, the chance of passing on the variant to their child is 50% in each pregnancy. Your child's ability to look after their own child is very likely to be closely related to their own learning ability and behaviour.

A clinical geneticist or genetic counsellor can provide specific advice for each family about the chance of having future children with OCNDS.

Unique publishes separate guides to Planning your next child, Prenatal genetic testing and diagnosis, A clinical genetics appointment and Supporting siblings of children with a rare genetic condition

The CSNK2A1 Foundation also has a family planning informational resource.

The CSNK2A1 Foundation family planning resource information can be found here

Can Okur-Chung neurodevelopmental syndrome be cured?

There is currently no cure for OCNDS. The condition results from a genetic change that affects early development. However, early diagnosis is valuable, as it allows for appropriate monitoring, supportive care, and interventions that can improve health, development, and quality of life.

Management

No clinical practice guidelines for OCNDS have been published (2026). The following suggestions have been provided by clinicians, who have personal experience managing/treating individuals with OCNDS, to improve quality of life and reduce complications.

Children and adults with OCNDS benefit from care coordinated by a multidisciplinary team. For children, this typically includes a clinical geneticist and paediatrician/developmental paediatrician who can oversee care, monitor development and behaviour, and coordinate referrals. Adults may transition to care led by a primary care provider with specialist involvement as needed.

Supportive therapies should be individualised and may include physical therapy, occupational therapy, speech and language therapy, and behavioural or developmental interventions when indicated.

Specialist involvement should be guided by symptoms and may include evaluations by neurology, psychiatry or developmental paediatrics, endocrinology, cardiology, ophthalmology, audiology, gastroenterology, urology, nephrology, sleep specialists, and dentists or orthodontists.

Immediately following diagnosis

Following a diagnosis of Okur-Chung neurodevelopmental syndrome (OCNDS), an evaluation should be carried out to identify which features of the condition are present and how they may affect the child or adult. This helps guide appropriate medical care, therapy supports, and ongoing monitoring.

The following assessments are recommended:

- A physical examination to assess growth (including height, weight, head circumference, and growth velocity), muscle tone (hypotonia), coordination, feeding concerns, constipation, and other physical features that may be associated with OCNDS
- A developmental and behavioral assessment to evaluate developmental delay, speech and language development, intellectual disability, autism spectrum features, ADHD, behavioral concerns, and educational support needs. Early referral for physical, occupational, speech, and behavioral therapies is recommended
- A feeding and nutritional assessment to monitor feeding difficulties, reflux, swallowing safety, poor weight gain, and nutritional status. Feeding therapy and, in some cases, gastroenterology evaluation may be helpful
- A growth and endocrine evaluation to assess for short stature, poor growth, or possible partial growth hormone deficiency. Referral to endocrinology may be considered if growth concerns are present
- A cardiac assessment if there are symptoms or clinical findings suggestive of a congenital heart defect, although heart differences are not among the most common features of OCNDS
- A renal evaluation if clinically indicated, since renal anomalies and pelviectasis (dilation or widening of the part of the kidney where urine collects) have been reported in some individuals
- An ophthalmology evaluation to assess vision concerns, strabismus or other eye findings. Ongoing ophthalmology follow-up every 1–3 years may be recommended

- An immunology evaluation if there is a history of frequent or unusual infections, since hypogammaglobulinemia has been reported in some individuals
- A sleep assessment to evaluate sleep disturbances, disrupted sleep patterns, or circadian rhythm concerns, which are common in OCNDS

Genetic counselling for affected individuals and their families is also recommended to discuss inheritance, recurrence risk, family planning options, and connection to family support resources. OCNDS is usually caused by a new (*de novo*) genetic change, but in some families, it can be inherited in an autosomal dominant pattern.

Supportive care

How a person with OCNDS is supported is likely to require co-ordinated care by a team of specialists, which may include:

Paediatrician – a doctor who specialises in the physical, mental, and social health of children from birth to young adulthood.

Neurologist – a doctor who specialises in conditions of the brain, spinal cord, and nervous system.

Cardiologist – a doctor who specialises in heart conditions.

Endocrinologist – a doctor who specialises in hormones and their effect on the body.

Urologist – a doctor who specialises in diagnosing and treating conditions affecting the urinary system.

Nephrologist – a doctor who specialises in conditions affecting the kidneys.

Surgeon – a doctor who is specially trained to perform medical operations.

Ophthalmologist – a doctor who specialises in conditions affecting the eyes.

Audiologist – a health care professional who diagnoses, treats, and helps manage a condition that involves hearing or balance.

Occupational therapist (OT) – a health care professional who uses activities to aid self-management of a condition and can provide equipment.

Physiotherapist (PT) – a health care professional who uses exercise, movement, manual therapy, education, and advice to help with the body's strength and mobility.

Speech and language therapist (SALT) – a health care professional who helps with speech, language, communication, and sometimes, feeding/swallowing difficulties.

Psychiatrist – a doctor who specialises in mental health.

Specialist nurses and/or other healthcare professionals may need to systematically and comprehensively plan a child or adult's treatment.

Treatments and therapies

Early intervention can prove particularly beneficial and formal testing to assess specific, individual needs is recommended. An individualized education plan (IEP) in the US, education, health and care plan (EHCP) in the UK, or equivalent document in other countries, may be issued after a child has undergone an assessment, to help ensure that the educational, health, and social provisions deemed necessary to support the child's needs are delivered.

Treatment will depend on the specific features and symptoms experienced by the person with OCNDS but may include:

Physiotherapy for mobility maximisation, which usually includes basic hands-on exercises to reduce the risk of more complicated orthopaedic (muscle and bone usage) issues later in life.

Occupational therapy for struggles with basic essential skills (such as feeding, grooming, dressing, writing, etc.), which can include fine motor skill development and adaptive strategies to address poor understanding of fundamental behaviours and coordination principles.

Speech therapy for speech delays, which can include frequent interventions to work towards independent verbal communication and/or substitutional techniques.

Behavioural therapies, including applied behaviour analysis (ABA) and other behavioural or developmental interventions, to support communication, adaptive skills, and behaviour regulation. Therapy is individualized to the person's strengths and needs and may be particularly helpful when autism-related features, emotional regulation challenges, or repetitive behaviours are present.

Medications may be prescribed, such as osmotic agents (laxative form) to treat gastrointestinal challenges and antiepileptic drugs to treat seizures.

Diet, such as a high-fibre diet or stool softeners and/or laxatives may be recommended to help relieve constipation. Some may benefit from enemas when symptoms are particularly severe.

Surgery, such as corrective jaw surgery to improve teeth alignment and overall function of the mouth.

Surveillance

It is recommended that the following evaluations are carried out to monitor an individual's existing symptoms, how they respond to care and treatment, and whether any new symptoms emerge over time:

- Regular dental check-ups
- Consider a palate review (particularly if there are feeding difficulties or speech concerns)
- Check position of testes in boys
- Consider an annual cardiac review
- Consider a neurologic review (including MRI and EEG, if indicated by seizures)
- Consider a clinical sleep study
- Consider a skeletal review
- Consider an abdominal ultrasound and review
- Developmental and behavioural monitoring, including evaluation for autism spectrum disorder if there are concerns related to social communication, behaviour, or sensory processing. Consider behavioural or psychiatric evaluation for attention, emotional regulation, anxiety, or aggressive behaviours
- Speech and language surveillance, including formal speech and language evaluation. Consider assessment for augmentative and alternative communication (AAC) if expressive communication is limited or delayed
- Motor and functional surveillance. Consider occupational therapy (OT) evaluation for fine motor skills, sensory processing and daily living skills. Consider physical therapy (PT) evaluation for gross motor development, strength, balance, and mobility

Research into new treatments for Okur-Chung neurodevelopmental syndrome

The genetic change that causes OCNDS affects development of the brain and other parts of the body before birth. Therefore, a complete cure is unlikely, even in the future, since the brain has already formed by the time a diagnosis is made. However, research into improved treatments and management for various features of OCNDS, like autism, is ongoing. In addition, although OCNDS is a relatively rare condition, the CSNK2A1 gene is the subject of a lot of research.

Ongoing OCNDS research at a glance – supported by the CSNK2A1 Foundation

Understanding how OCNDS works

CK2 and SYNGAP1 connection: Studying how changes in CK2 affect another important brain protein (SYNGAP1) and how this may lead to seizures and learning difficulties.

Cilia and brain development: Looking at how OCNDS developments may disrupt tiny "antenna-like" structures on cells (cilia) that are important for brain growth.

Tracking symptoms and daily life

Speech and language study: The first detailed look at how speech and language differ in people with OCNDS to guide therapy and future trials.

Sleep and body rhythms: Using surveys and lab models to understand sleep problems and test medicines that may improve sleep and daily rhythms.

Building tools for research and treatment

OCNDS knowledgebase: Creating a central online resource that organises genetic and medical information to help families, doctors, and researchers.

Nasal swab study: Collecting RNA from nasal cells to learn how OCNDS affects the body and to predict possible drug treatments.

Drug repurposing screen: Testing more than a thousand existing medicines in fly models to quickly find new treatment options.

New research models

Mouse models: Studying learning, memory, brain activity, and molecular changes of OCNDS in genetically altered "model" mice.

Patient-derived neurons: Growing nerve cells from patient stem cells to accurately associate genetic changes with symptoms.

Data integration: Comparing results across mouse and human models to choose the best treatment strategies to test next.

How it all fits together

These projects build the research "toolbox," identify new markers of the disease (such as speech and sleep), and test possible treatments. Together, they move us closer to better care, clinical trials, and ultimately, a cure for OCNDS. To learn more, please visit the **CSNK2A1 Foundation**.

Details of other clinical trials related to a particular condition or gene can be found at **ClinicalTrials.gov** and **EU Clinical Trials Register**.

Families say ...

"If you just received a diagnosis, I would tell you to join the monthly family zoom calls to connect with other parents in the OCNDS community and sign up to Simons Searchlight, the natural history longitudinal study."

*"I think the best thing that the (*CSNK2A1*) Foundation has provided me is ... that connection and that support and that family ... I'm just really grateful that if you had to be diagnosed with a rare syndrome, like one that's got a Foundation that's got ... such strength behind it is just very, very lucky."*

"I've really treasured because all her milestones … you know, walking, talking, breathing, eating, moving, sitting, running, jumping … you take all those things for granted. And so, yeah, I was always very excited when she completed something or did something. I was always so thankful."

"It's not just about learning math or science … we're able to teach her things that improve her quality of life." (Educator)

"She is capable. Don't write her off yet."

"Due to his care needs being high we always need an adult familiar with him to be with him, at all times. He is funny & loving, and we have some great movie days or gaming days."

"If you just received an OCNDS diagnosis, reach out to other families. Because there isn't much information on this syndrome, just chatting with others who are going through the same thing as you is comforting. There is an amazing Facebook group, CSNK2A1 *Foundation, that was created by a parent whose daughter is one of the first to be diagnosed. You will find a lot of support from fellow families as well as more information to help with understanding this syndrome."*

"Although none of us expected to be raising a child with such a rare syndrome, I am honored to be the parent of a young man who is part of history and research. Being able to write this chapter in his book is such a blessing. Being able to reach out to the two scientists/geneticists who discovered this syndrome has been very comforting, knowing they stand with us. OCNDS has become a giant family and continues to grow. For sure, the information they are finding is what has helped us, and the continuing research will continue to further help understanding."

Sources

The information in this booklet is drawn from the published medical literature and information from the **CSNK2A1 Foundation**. The first-named author and publication date for articles in the medical literature are given to allow you to look for the abstracts or original articles on the internet in **PubMed**.

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For a full list of OCNDS-related publications, please visit the **published research** webpage at the CSNK2A1 Foundation.

Note: an asterisk indicates articles which are “open access” and available to everyone at **PubMed**.

Inform Network Support

Rare Chromosome Disorder Support Group
The Stables, Station Road West, Oxted, Surrey, RH8, 9EE, UK
Tel: +44(0)1883 723356
help@rarechromo.org I rarechromo.org

Join Unique for family links, information and support:

Become a member

Please help us to help you!

Unique is a charity without government funding, existing entirely on donations and grants.
If you can, please make a donation:

Donate via our website

Websites, Facebook groups and other links:

- CSNK2A1 Foundation
- CSNK2A1 Facebook group
- CSNK2A1 Foundation LinkedIn

This information guide is not a substitute for personal medical advice. Families should consult a medically qualified clinician in all matters relating to genetic diagnosis, management and health. Information on genetic changes is a very fast-moving field and while the information in this guide is believed to be the best available at the time of publication, some facts may later change.

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