CAPRIN1-related NDD

UNDERSTANDING GENES & CHROMOSOMES

What is *CAPRIN1*-related neurodevelopmental disorder?

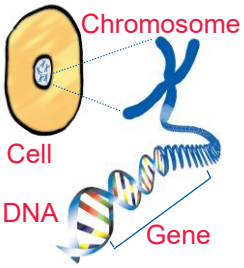
CAPRIN1-related neurodevelopmental disorder (NDD) is a rare genetic condition that causes learning difficulties or intellectual disability, speech and language difficulties and developmental delay. Children with this condition have also been diagnosed with autism spectrum disorder (ASD) and attention deficit hyperactivity disorder (ADHD) and a few less common features have been identified.

CAPRIN1-related NDD is also known as NEurodevelopmental Disorder with Language impairment, Autism and Attention Deficit-hyperactivity disorder (NEDLAAD).

What causes *CAPRIN1*-related NDD?

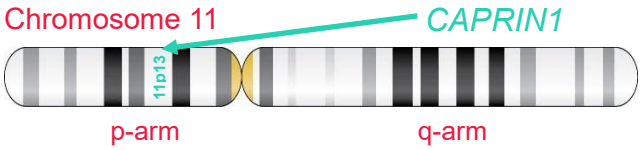
CAPRIN1-related NDD is caused by specific changes (known as pathogenic variants) in a gene called *CAPRIN1.* Most of these variants cause a loss of function (LOF) of this gene.

Gain of function variants are associated with a different condition, a child-onset neurodegenerative condition (known as CONDCAC), which is much less common.



Genes and chromosomes

The *CAPRIN1* gene is located on the short 'p' arm of chromosome 11 in a region called 11p13 (see below).



CAPRIN1 is an abbreviation of the gene's full name, cell cycle-associated protein 1. The *CAPRIN1* gene codes for the Caprin-1 protein, which has many different roles in our cells, including the development, maintenance, and function of the brain's nerve cells, called neurons.

Features and symptoms

As is common with many genetic conditions, children and adults with *CAPRIN1*-related NDD can have a range of symptoms and features. As more people are diagnosed, and information is shared, the range of symptoms and features, and the likelihood of a person having these features, will become clearer.

The following developmental and medical concerns have been reported in people with *CAPRIN1*-related NDD, but no one person will have all the features listed and each person with this condition can have different developmental and medical concerns.

Common features

- Speech and language delay: speech may be slow to develop or absent
- Learning difficulties or intellectual disability: may range from absent to severe, challenges in school and with everyday tasks are expected
- Autism spectrum disorder (ASD): challenges with social skills and communication, with repetitive behaviours
- ADHD (attention deficit hyperactivity disorder): difficulty focusing, sitting still, or controlling impulses
- Memory problems

Other possible features

- Gross and/or fine motor delays: trouble with crawling, walking, or using hands
- Seizures or epilepsy
- Feeding or swallowing problems in infancy or childhood
- Hearing difficulties
- Balance and coordination issues
- Low muscle tone (hypotonia)
- Unusual facial features
- Other behavioural challenges, e.g. anxiety
- Sleep disturbance
- Gastrointestinal (digestive) problems in infancy or childhood

Therapies and support

Although no clinical practice guidelines have been published, the care of a person with *CAPRIN1*-related NDD is likely to require regular follow-ups and coordinated care by a multidisciplinary team of specialists, which may include a paediatrician, neurologist and geneticist, together with the following therapy and support:

- Speech and language therapy (can also help with swallowing difficulties)
- Occupational therapy (to help with daily tasks like eating, dressing, and playing)
- Physical therapy (to help with strength, balance, and coordination)
- ABA therapy (applied behaviour analysis, to help with behaviour and learning)
- AAC tools (augmentative and alternative communication, tools for children who are non-verbal)

- Education plans and tailored support in mainstream or special educational needs (SEN) schools
- Medication to help manage focus, anxiety, mood, or seizures

How common is CAPRIN1-related neurodevelopmental disorder?

CAPRIN1-related NDD is very rare. Currently (2025), fewer than 50 people, mostly children, with this diagnosis have been reported in the medical literature. It is expected that more people will be diagnosed with this condition as awareness increases and genetic testing becomes more routine. It is important to know that the most severely affected children are likely to be the first identified, so initial findings may not represent the possible spectrum of symptom severity.

Why did this happen?

When children are conceived, random rare changes occur in the genetic material of the egg and/or sperm that make a new child. Such changes are part of the child's genome but they are not seen in the DNA of their parents. This happens naturally and is not due to any lifestyle, dietary or environmental factors. No one is to blame and nobody is at fault. Such changes happen to everyone but it's only when particular genes or specific parts of a chromosome are affected that there is an impact on health and/or development.

In most children diagnosed with CAPRIN1-related neurodevelopmental disorder so far, the change in the *CAPRIN1* gene occurred by chance in that child (this is known as a *de novo* variant) and was not found in their parents. Very few children are known to have inherited a *CAPRIN1* variant from an affected parent.

Can it happen again?

The chance of having another child affected by a rare gene disorder depends on the genetic code of the parents. If the change in the *CAPRIN1* gene has been shown to be *de novo*, meaning neither parent was found to carry it, the chance of having another child with *CAPRIN1*-related NDD is very low. The reason there is still a small chance is due to something called germline mosaicism, which is where the gene variant can be found in a few eggs or sperm, but is not found in the rest of the body's cells. A clinical geneticist or genetic counsellor can give you specific advice for your family.

Can it be cured?

CAPRIN1-related NDD cannot be cured at the present time; however, knowing the diagnosis means that appropriate monitoring and treatment can be put in place, especially for features of medical, behavioural, and educational concerns.

Families say .....

" When we first received the diagnosis, it felt overwhelming and isolating. We wish we had known that we weren't alone and that other families were going through a similar journey. It helped us to focus on our child's strengths and celebrate every milestone, no matter how small. Finding support from other families and researchers has made the road feel less lonely and has given us hope for the future ahead. "

" One thing we wish we had known at the start is how important it is to trust your instincts as a parent. Therapies, interventions and advice can feel overwhelming, but only you know your child best. It helps to find support where you can and to remember that progress looks different for every child and that's ok. "

" The diagnosis, the term and all the related information felt like a foreign language at first. It helped realising that we didn't have to understand and know everything straight away, we were and are still learning as we go along which doesn't feel scary anymore. We realised that our child's future is not defined by a label. They have their own personality, their own pace and their own potential. "

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
rarechromo.org

Websites & Facebook groups:
caprin1foundation.org/
facebook.com/profile.php?id=61574778207863

Join *Unique* for family links, information and support: rarechromo.org/join-us/

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