CORNELIA DE LANGE SYNDROME

Australasia

www.cdls.org.au

"These individuals light up every room with their cheeky smiles and curious eyes. They're loving, adventurous, and full of surprises — just when you think you've figured them out, they'll do something wonderfully unpredictable. You can't help but laugh at their clever tricks and admire their fearless spirit. They may challenge expectations, but that's only because they were never meant to fit into boxes. Don't underestimate them — they're here to explore, connect, and show the world just how much joy and magic they bring simply by being themselves."

Gemma, Vice President, CdLS Association,

What is Cornelia de Lange Syndrome?

It's all in the Genetics!

Cornelia de Lange Syndrome (CdLS) is a genetic disorder present from birth. It is usually due to an acquired change mutation in one of seven important developmental genes at or shortly after conception.

More about the condition:

Cornelia de Lange syndrome (CdLS) is a rare genetic disorder that causes physical, cognitive and behavioral differences. The signs and symptoms of the condition vary widely and range from mild to severe. No two individuals with the disorder have exactly the same combination of traits, but there are many similarities in their appearance and behaviour. The disorder affects many different parts of a person's body.

Typical features to name a few include:

- Delayed growth before and after birth.
- Head and facial differences.
- Hand and arm defects.
- Intellectual disability.
- Behavioural abnormalities
- Hearing Loss
- Gastrointestinal issues (chronic acid reflux)
- Seizures
- Heart Problems
- Feeding issues
- Communication delays

Gene Fact...

About 60% to 80% of all people with Cornelia de Lange syndrome have a mutation in the NIPBL gene. Mutations in the six other genes are less common. In 5% to 20% of people with the condition, the genetic cause is unknown.

How is CdLS diagnosed?

The diagnosis of CdLS is primarily a clinical one based on signs and symptoms observed through an evaluation by a physician, including a medical history, physical examination and laboratory tests. However, genetic testing can be helpful in confirming the clinical diagnosis and assessing which gene is involved.

How is Cornelia de Lange syndrome treated?

Treatment for Cornelia de Lange syndrome varies based on the individual specific symptoms. Because the condition can affect many different parts of their body, a team of healthcare and allied heath providers may help with treatment.

How many people have CdLS?

The occurrence of CdLS is estimated to be 1:10,000 and 1:30,000 live births

Its estimated that there are about 1000 families affected by CDLS in Australasia.
With the introduction of new technologies, more and more are being diagnosed yearly.

Therapy Intervention:

Therapy should be ongoing throughout an individuals life. Different therapies will address their delayed growth, intellectual disabilities and behavioral issues. Therapies may include:

- Physical therapy.
- Occupational therapy.
- Speech therapy.
- Psychotherapy.

In addition, they should be evaluated and monitored for certain functions and developmental delays throughout their life. These include:

- Hearing and vision screenings.
- Growth and psychomotor development.
- Heart and kidney function.
- Gastrointestinal function.

Awareness Day!

Cornelia De Lange Syndrome is celebrated on the second Saturday of May every year. This day was not observed until 1989 when it was established as a means to educate society about this condition, the tests, and the diagnostics. It was also set up in order to create awareness about this still not so known genetic syndrome. By letting others know about Cornelia De Lange Syndrome Awareness (CdLS), we hope to eliminate the stigma individuals and families have to endure and with more people having a dialogue about the syndrome.

10TH MAY 2025

Wear Purple!

Meet some of us....

Margot (1)
Loves snuggles + holding your hand

Nadiya (3)
Loves to giggle and smile

Teddy (3)
Loves Chocolate!

Louis (4)
Loves water play

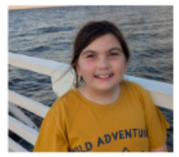
Rome (7)
Loves Harry Potter

Levi (7)
Loves the outdoors

Morgan (7)
Loves all things cricket

Penny (7)
Loves music and dancing

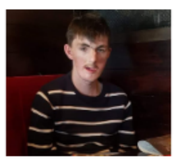
Darcy (8)
Loves riding his bike



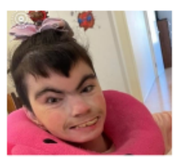
Chloe (9)
Loves Cricket and crafts

Brylea (20)
Loves to go to school

Chloe (20)
Loves shopping!



Danny (25)
Loves to collect keys



Kyah (28)

David (42)
Loves listening to music

Kaya (43)

FOLLOW US!

cdlsaus

Where can I get more information?

www.cdls.org.au
president@cdls.org.au

How can I help raise awareness?

Donate Follow Share

The Cornelia de Lange Syndrome Association (Australasia) Inc (CdLSA) is a family support organisation. We exist to support families, with emotional support, a community, and quality information so they can make informed decisions. We also work to assist the accurate diagnosis of CdLS and promote research into the cause and manifestations of the syndrome.

DONATE TODAY

www.paypal.com/AU/fundraiser/charity/3499743