Further Inform Neurogenetic Disorders

Key Facts

It is estimated that between 1 in 10,000 - 25,000 people in the population have Cornelia de Lange syndrome.

Most diagnoses are made based on the clinical features of the syndrome.

Cornelia de Lange syndrome is caused by a single genetic changeaffecting a single gene in each individual. However, the genetic change can be in different genes in different individuals.

Research studies have suggested that most individuals with Cornelia de Lange syndrome had a severe to profound intellectual disability. Some individuals with Cornelia de Lange syndrome have mild intellectual disabilities.

Some of the health difficulties in Cornelia de Lange syndrome may be particularly important for understanding behaviours that challenge.

Children and adults with Cornelia de Lange syndrome show higher levels of compulsive-like behaviours in comparison to other genetic syndromes.

While many children and adults meet the cut-off for Autism Spectrum Disorder (ASD) on standard assessments, the profile of ASD characteristics may be different in Cornelia de Lange syndrome.

Everyday living skills may be delayed among children with Cornelia de Lange syndrome but some do achieve a degree of independence in this area.

Many individuals with Cornelia de Lange syndrome experience social anxiety and are selectively mute.

Between 50-60% of individuals with Cornelia de Lange syndrome engage in some form ofself-injurious behaviour.

Overactivity seen in children and adults with Cornelia de Lange syndrome may be linked to pain and discomfort associated with gastro-intestinal difficulties.
