Autism Spectrum Disorder in Cornelia de Lange Syndrome

The three core characteristics of Autism Spectrum Disorder (ASD) include the following:

- Poor or unusual social interaction skills
- Delayed development or difficulties in verbal and non-verbal (gestures, pointing, showing etc) communication.
- The presence of repetitive behaviours.

Recent studies have shown that characteristics associated with Autism Spectrum Disorder occur in Cornelia de Lange syndrome.

People with Cornelia de Lange syndrome are more likely than people with Cri du Chat syndrome (who have a comparable level of ability and receptive language skills) to reach the cut-off scores for autism on an observational assessment of characteristics associated with autism. The diagnostic cut-off scores indicate the level at which individuals with a confirmed diagnosis of autism who do not have Cornelia de Lange syndrome would be expected to score. Approximately 60% of people with Cornelia de Lange syndrome score on this assessment at a level that would be expected in individuals with autism.

This does not mean that all individuals with Cornelia de Lange syndrome should receive a diagnosis of autism. Rather, it suggests that autistic-like characteristics are common in individuals with Cornelia de Lange syndrome and that some people with Cornelia de Lange syndrome might be considered to fall within the autistic spectrum.

Impairments in communication

Impairments in communication are the most prominent autistic-like characteristic associated with Cornelia de Lange syndrome. Research suggests that both verbal and nonverbal communication skills are compromised.

Social interaction skills

Social interaction skills might also be compromised in Cornelia de Lange syndrome. Unlike in autism, social difficulties in people with Cornelia de Lange syndrome are likely to take the form of social anxiety.
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Original research article: the prevalence of ASD in Cornelia de Lange syndrome

Original research article: Characteristics of ASD in Cornelia de Lange syndrome

Accessible research summary on the Cornelia de Lange syndrome Foundation USA website