

Characteristics of Autism Spectrum Disorder in Cornelia de Lange Syndrome

Joanna Moss¹, Patricia Howlin², Iliana Magiati³ and Chris Oliver¹

¹The Cerebra Centre for Neurodevelopmental Disorders, University of Birmingham

²King's College London, ³University of Singapore

Background

- ◆ Individuals with autism spectrum disorder (ASD) show impairments in communication and social interaction skills and a range of repetitive behaviours and restricted interests.
- ◆ ASD characteristics have been reported to occur in a number of genetic syndromes including Cornelia de Lange syndrome (CdLS).
- ◆ As has been identified in many genetic syndromes, the presentation of ASD characteristics in CdLS is considered to be different to that of individuals with ASD who do not have a genetic syndrome.

Aims of the project

- ◆ To estimate the prevalence of ASD characteristics in individuals with CdLS.
- ◆ To describe the presentation of ASD characteristics in individuals with CdLS.
- ◆ To evaluate similarities and differences in the presentation of ASD characteristics (frequency/severity) in individuals with CdLS and individuals with ASD who do not have a genetic syndrome.

Methods

- ◆ Twenty individuals with CdLS were matched to twenty individuals with ASD on chronological age, receptive language skills and adaptive behaviour skills.
- ◆ Individuals with CdLS and ASD were observed during a direct assessment of ASD characteristics.
- ◆ This observational assessment is typically used to help to diagnose ASD.

Results

- ◆ 85% of individuals with CdLS scored above the cut off for ASD and 65% of individuals with CdLS scored above a more stringent cut off for autism.
- ◆ Individuals with CdLS showed similar levels of impairments to the ASD group in overall communication and social interaction skills.
- ◆ Individuals with CdLS showed fewer repetitive behaviours than individuals with ASD. In particular they showed fewer behaviours related to sensory interests.
- ◆ The CdLS group also showed less stereotyped speech and less impaired use of gestures and eye contact compared to those with ASD but they showed higher levels of anxiety than the ASD group.

Conclusions and future directions

- ◆ ASD related characteristics are common in CdLS with at least 3 in every 5 individuals showing some of these traits.
- ◆ There are some broad similarities across the groups with regard to the nature of ASD characteristics but there are also some areas of subtle difference, particularly in the domain of repetitive behaviour and with regard to levels of anxiety.
- ◆ These differences will be important to take into account when considering appropriate interventions for individuals with CdLS who show ASD characteristics.

Full reference: Moss, J., Howlin, P., Magiati, I. and Oliver, C. (In press). Characteristics of Autism Spectrum Disorder in Cornelia de Lange Syndrome. *Journal of Child Psychiatry and Psychology*.

The Research described in this report was funded by the Cornelia de Lange Syndrome Foundation and Cerebra

More information? Go to www.birmingham.ac.uk/cnnd or find us on facebook:
<https://www.facebook.com/pages/The-Cerebra-Centre-for-Neurodevelopmental-Disorders/230197213724784?sk=wall>