Paper in the Journal of Child Psychology and Psychiatry indicates that forthcoming changes to diagnostic criteria for Autism could lead to exclusions for many individuals with genetic syndromes

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The prevalence of autism spectrum disorder (ASD) has been reported to be comparatively high in individuals with a range of genetic syndromes. It is often reported that the presentation of ASD characteristics in these genetic syndromes differs (in very subtle ways) to that observed in individuals with ASD. According to current diagnostic guidelines this might be classified as 'atypical autism'. Forthcoming changes to these guidelines may result in many individuals with genetic syndromes who currently meet diagnostic criteria for 'atypical autism' to be excluded, with inevitable consequences for access to appropriate resources and intervention.

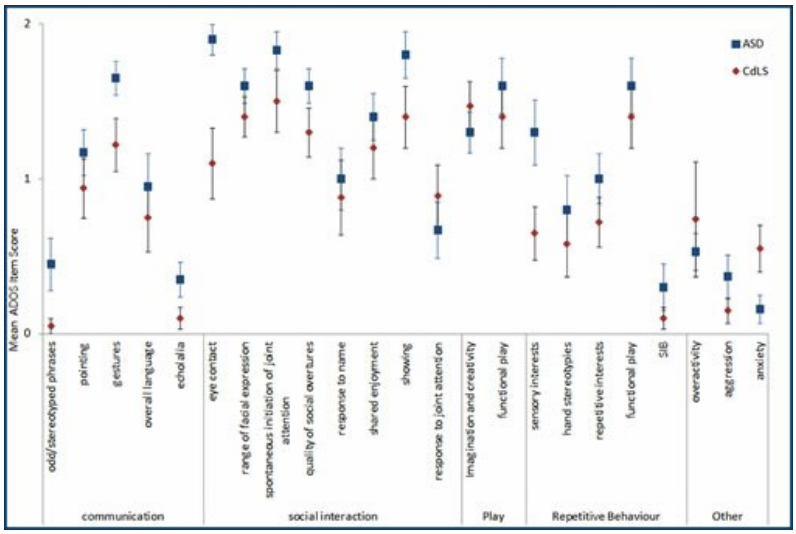
This paper evaluates the prevalence and nature of ASD characteristics in individuals with Cornelia de Lange syndrome (caused by deletions on chromosome 5 in 50% of cases and chromosomes X or 10 in 5% of cases) in comparison to a matched contrast group of individuals with idiopathic ASD. The prevalence of ASD characteristics in Cornelia de Lange syndrome is reported to be between 50% and 60%.

The findings confirm shared characteristics of ASD phenomenology between individuals with Cornelia de Lange syndrome and those with idiopathic ASD at a broad level, indicating that the needs of these groups might be similar . However, subtle differences in the manifestation of these characteristics are identifiable at a more refined level of investigation. Specifically, fewer ASD specific repetitive behaviours (particularly sensory related behaviours), greater use of eye contact and gestures, less stereotyped speech and higher levels of anxiety were identified in Cornelia de Lange syndrome compared to individuals with idiopathic ASD.

The findings contribute to an expanding evidence base identifying atypicalities in ASD related characteristics among individuals with genetic syndromes, which may have important implications regarding understanding and conceptualisation of ASD within this population. The findings also have significant clinical implications for assessment and intervention for ASD in individuals with genetic syndromes and these will need to be considered following the forthcoming changes to diagnostic guidelines for ASD.

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*.



Mean item scores on the Autism Diagnostic Observation Schedule in twenty individuals with Cornelia de Lange syndrome and a matched contrast group of individuals with idiopathic ASD

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