

Similarities and Differences of CdLS and Autism

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Many syndromes, such as Cri du Chat, Fragile X and Rett, present with a range of impairments in the areas of communication, behavior, and emotion. Individuals with Cornelia de Lange Syndrome (CdLS) also present with impairments in these three areas. These are the areas that highlight the hallmark diagnostic symptoms for Autism Spectrum Disorder. For an individual to receive a diagnosis of autism, the following symptoms must be displayed:

- qualitative impairment in social interaction;
- qualitative impairment in communication; and
- restricted repetitive and stereotyped patterns of behavior, interests and activities, as well as delays or abnormal functioning, with onset prior to age three, in (at least one of the following) social interaction, language as used in social communication, or symbolic or imaginative play.

Some of the symptoms of autism that also occur in individuals diagnosed with CdLS include developmental, language and social development delays. The range of delay varies across individuals. Specifically, individuals diagnosed with CdLS are described as having receptive and expressive language delays with a significant discrepancy in these language skills.^{3, 4} A similar discrepancy is described in individuals on the Autism Spectrum.

Previous studies suggest a strong association between CdLS and Autism Spectrum Disorder.¹ For example, Berney and colleagues¹ reported that 53 percent of the 49 CdLS individuals in their study displayed autism symptoms and these associations were not attributed to the level of intellectual disability.⁷

However, in the area of social interaction, the overlap in presentation across the CdLS and autism groups may be less clear. Individuals with CdLS may present with increased anxiety, selective mutism, and shyness² that is not found in the autism group.



In the Moss, Howlin, Magiati & Oliver article,⁵ the CdLS group demonstrated a greater capacity to use eye contact and gestures in comparison to the autism group suggesting different social skills impairments across the groups.

Further research is needed into the nature of the social, communicative and emotional impairments in CdLS compared to individuals with Autism Spectrum Disorder with similar intellectual disability. Not only are we interested in what ways these groups present in a similar fashion, but also in what ways the CdLS group presents differently than the autism group. These studies could aid in the delineation of different subtypes of individuals with CdLS along with specific treatment strategies that would be more beneficial for the specific subgroups.

¹ Berney T. P., Ireland M. & Burn J. (1999). Behavioral phenotype of Cornelia de Lange Syndrome. *Archives of Disease in Childhood*, 81, 333–6.

² Collis, L., Oliver, C., & Moss, J. (2006). Low mood and social anxiety in Cornelia de Lange syndrome. *Journal of Intellectual Disability Research*, 50, 791–800.

³ Cornish, K., & Munir, F. (1998). Receptive and expressive language skills in children with Cri du-Chat syndrome. *Journal of Communication Disorders*, 31, 73–81.

⁴ Goodban, M. T. (1993). Survey of speech and language skills with prognostic indicators in 116 patients with Cornelia de Lange Syndrome. *American Journal of Medical Genetics*, 47, 1059–63.

⁵ Moss, J., Howlin, P., Magiati, I., & Oliver, C. (2012). Characteristics of autism spectrum disorder in Cornelia de Lange syndrome. *Journal of Child Psychology and Psychiatry*, 53, 883–891.

⁶ Moss, J., Oliver, C., Berg, K., Kaur, K., Jephcott, G., & Cornish, K. (2008). Prevalence of autism spectrum phenomenology in Cornelia de Lange and Cri du Chat syndromes. *American Journal on Mental Retardation*, 113, 278-291.

⁷ Oliver, C., Arron, K., Berg, K., Burbidge, C., Caley, A., Duffay, S., Hooker, M., & Moss, J. (2005). A comparison of Cornelia de Lange, Cri du Chat, Prader-Willi, Smith-Magenis, Lowe, Angelman and fragile X syndromes. *Genetic Counseling*, 13, 363–381.

