NEWS

Cognition and behavior: Rare syndrome distinct from autism

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Genetic disorder: Cornelia de Lange syndrome is characterized by severe developmental delay, abnormal physical features and, in many cases, a diagnosis of autism.

Individuals with Cornelia de Lange syndrome (CdLS), a rare genetic disorder often accompanied by autism, have subtle differences in the nature of their social deficits compared with those who have autism alone, according to a report published 10 April in the *Journal of Child Psychology and Psychiatry*[1].

CdLS affects 1 in every 40,000 people. About half of individuals with the syndrome have a mutation in NIPBL, a protein that regulates chromosome structure and repairs damaged DNA. Individuals with CdLS have some combination of developmental disability, abnormal facial and physical features, short stature, epilepsy and autism.

In the new study, researchers looked in detail at the symptoms of 20 individuals with CdLS and compared them with those of 20 people who have classic autism. Of those with CdLS, 17 meet the criteria for an autism spectrum disorder, which includes **Asperger syndrome** and pervasive developmental disorder-not otherwise specified, on the Autism Diagnostic Observation Schedule, and 12 meet the criteria for classic autism.

However, people with CdLS are less likely to have **repetitive behaviors** than those with autism, the study found. They are more likely to make eye contact and to gesture when communicating,

and are more anxious than those with autism.

The results suggest that the social deficits associated with CdLS could be the result of social anxiety, which is also a prominent feature of **fragile X syndrome**. Diagnostic measures for autism alone may miss these subtle distinctions and make autism appear more similar to other genetic syndromes than it really is, the researchers say.

References:

1: Moss J. *et al. J. Child Psychol. Psychiatry* Epub ahead of print (2012) **PubMed**