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Join the Conversation.



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ABOUT US

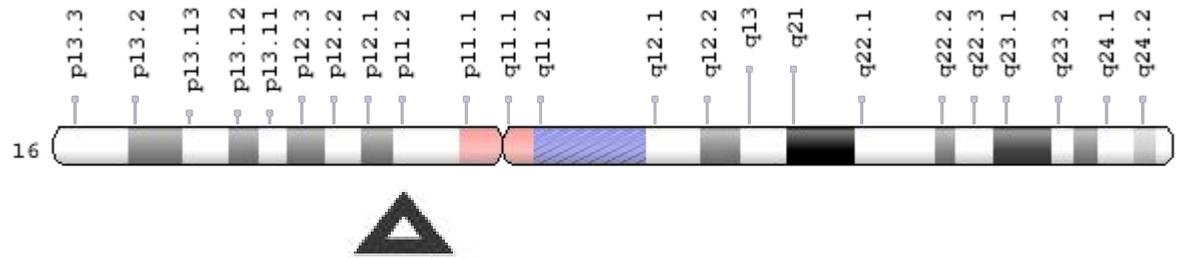
Chromosome Disorder Outreach provides support and information to anyone diagnosed with a rare chromosome change, rearrangement or disorder. CDO actively promotes research and a positive community understanding of all chromosome disorders.

CDO is a 501c3 organization founded in 1992.

16p11.2 microduplication

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Common clinical features for this disorder can include psychiatric illnesses (in particular, autism and schizophrenia), developmental delay, neurological defects, and lower BMI caused by metabolic issues. Additional common diagnoses include intellectual disability (ID), developmental coordination disorder, speech sound disorder, behavioral disorder, Attention Deficit Hyperactivity Disorder (ADHD) in children, and anxiety in adults.



The 16p11.2 deletion and duplication are among the most frequent genetic causes of autism spectrum disorder (ASD) and schizophrenia. However, the severity of ASD and schizophrenia vary greatly from person to person.

In the development of cognitive ability, verbal IQ typically improves with age, but nonverbal IQ does not. Development of adaptive abilities (determined by an individual's abilities in communication, socializing, motor, and daily living skills) tend to increase with age. While behavioral disorders are typical for children with the duplication, these tendencies typically diminish with age.

The most common neurologic defects include hypotonia (low muscle tone), weakness, unprovoked seizures, tremor (involuntary movements), microcephaly (abnormally small head), and tics (sudden, rhythmic twitching). Other issues that were observed include hyperreflexia (overactive reflexes), hyporeflexia (weakened reflexes), café au lait spots (flat, brown birthmarks), sacral dimples (indentation in the skin of the lower back), abnormalities of eye convergence (inability of the eyes to point in the same direction), and dysrhythmia (abnormal rhythm of the brain or heart).

Duplication carriers have an eightfold risk of being underweight and consistently have lower BMIs. Duplication carriers are shown to have smaller appetites when compared to the average, and are not shown to eat in response to stress.