A 600 kb deletion syndrome at 16p11.2 leads to energy imbalance and neuropsychiatric disorders
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This paper combined information from the 16p11.2 European Project and Simons Variation in Individuals Project to collect the largest group of individuals with 16p11.2 deletions studied so far. When possible, the researchers collected information on whether the 16p11.2 deletion was inherited from a parent or happened for the first time in the affected individual (de novo). Overall, 64% of deletions were de novo and 36% were inherited. Information was collected on 285 participants with the deletion; 71 of whom were evaluated at one of the clinical sites.

IQ was measured and compared to IQ scores in family members without the deletion. This allowed the researchers to control for other familial factors that could influence IQ. Individuals with the deletion had IQ scores on average 32 points lower than family members without the deletion, indicating that the 16p11.2 deletion causes problems with cognitive functioning. The overall average IQ was 76.1. 20% of participants with the deletion had intellectual disabilities with IQ<70. Verbal IQ was significantly lower than nonverbal IQ.

Autism spectrum disorders (ASDs) were of special interest to the researchers because of previous reports of an association between the 16p11.2 deletion and autism. 11% of individuals with the deletion who were evaluated for autism met diagnostic criteria. Of those without ASDs, 73% were found to have other psychological diagnoses including ADD/ADHD, behavioral disorders, and mood disorders. 83% of people with the 16p11.2 deletion needed speech therapy at some point.

The 16p11.2 deletion also seems to affect growth. Birth weight in people with 16p11.2 deletions was found to be below average. However, obesity was more common later in life. 50% of individuals with the deletion were obese by age 7 and 45% of adults with the deletion were morbidly obese (compared to 5% in the general population). Adults with the deletion were also overall shorter than average. Head size was larger than average in individuals with the 16p11.2 deletion and 17% had head sizes large enough to be diagnosed with macrocephaly.

24% of individuals with the deletion had epilepsy (seizures). The authors recommended that EEG should be performed if abnormal movements or behaviors suspicious for seizures are observed. About 5% of people with the deletion had paroxysmal dyskinesia syndrome, which causes episodes of uncontrollable movement which can be subtle.
Brain MRIs were performed in a group of people with the deletion and major abnormalities were uncommon.

16p11.2 deletions did not seem to be frequently associated major birth defects. The most common major structural problems included defects in the vertebrae and/or curvature of the spine in 20%, hearing loss in 11%, and heart defects in 6%. Several other medical problems and birth defects were reported in individuals with the 16p11.2 deletion; but since they were present in 5% or less of this population, they are likely either unrelated or represent a very small increased risk. The most common medical problems related to the 16p11.2 deletion are epilepsy and obesity.