NEWS

Genetics: Genes near Williams region linked to autism traits

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Deletion differences: People with deletions extending beyond the Williams syndrome region have severe versions of the disorder's characteristic facial features, such as skin folds on the upper eyelid and a short nose.

Genes near the chromosomal region implicated in **Williams syndrome** are involved in **epilepsy** and autism-like behaviors, according to a study published 12 June in the *European Journal of Human Genetics*[1].

Individuals with Williams syndrome lack a large part of chromosome 7 called **7q11.23**. They are **markedly friendly and talkative**, but also share many features with people who have autism: They **often experience anxiety**, cognitive deficits and difficulty forming or maintaining close relationships.

In people with Williams syndrome, the deleted region usually spans about 25 genes. Some individuals, however, are missing more or fewer genes.

The recent study focused on four people with Williams syndrome who have these unusual deletions — two who are missing more genes than is typical, and two fewer.

The researchers matched up detailed genetic information about the region each person lacks with their diagnostic and medical history, including intelligence tests, behavioral problems, health

problems and **characteristic facial features**. They used these data to draw correlations between genes in or near the Williams syndrome region and particular traits.

Their findings suggest that the genes HIP1 and YWHAG, which are missing in the individuals with larger deletions, are involved in more severe intellectual disability, epilepsy, and autism-linked behaviors such as hyperactivity, obsessive behavior and social communication problems. This corroborates the results of earlier studies in people and lab animals.

The genes FZD9and BAZ1B, which are usually deleted in Williams syndrome, may be involved in intellectual disability and the characteristic facial features associated with Williams syndrome, respectively. The two participants with smaller deletions carry these genes and have milder facial features and less intellectual disability than those lacking the genes.

HIP1 plays an important role in neuron death and YWHAG in cell communication. FZD9codes for a receptor protein expressed in the hippocampus, the brain's long-term memory center, and BAZ1Bfor an enzyme implicated in craniofacial development.

References:

1: Fusco C. *et al. Eur. J. Hum. Genet.* Epub ahead of print (2013) **PubMed**