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My46 Trait Profile

Williams syndrome

Other Names: Williams-Beuren syndrome

Williams syndrome is a genetic condition with characteristic facial features and birth defects. It is caused by a deletion in one of the two copies of chromosome 7.

Characteristics of Williams syndrome

Williams syndrome is a multi-system genetic disorder with both medical and developmental features. A hallmark medical feature of Williams syndrome is blood vessel stenosis or narrowing. Although narrowing may occur anywhere, supravalvar aortic stenosis (SVAS), a narrowing above the aortic valve in the heart, is the most common site of narrowing. Other medical problems often seen in Williams syndrome vary by age, but may include elevated calcium (hypercalcemia), feeding difficulties, constipation, reflux, diabetes, and contractures. Though the majority of individuals with this condition have mild to moderate intellectual disability, affected individuals often have a unique pattern of relative strengths and weaknesses. For instance, most individuals with Williams syndrome have a friendly, social personality with relatively strong language skills. Visual-spatial tasks (i.e., activities that require understanding of spatial relations among objects; e.g., reading a map, and driving), however, tend to be rather difficult for most individuals with Williams syndrome. Behavioral problems such as attention deficit hyperactivity disorder (ADHD) and anxiety are also very common in this condition. Individuals with Williams syndrome have a characteristic facial appearance; in children, these include a round upturned nose, wide mouth, small widely spaced teeth, stellate irises (a star pattern in the colored part of the eye visible in light colored eyes), and puffiness around the eyes.

Diagnosis/Testing

Suspicion of the diagnosis of Williams syndrome can be raised by a doctor based on clinical features alone. However, the diagnosis is usually confirmed with genetic testing. Williams syndrome is caused by a deletion (i.e., missing piece) of approximately 26-28 genes on one of the two copies of chromosome 7 in each cell. This deletion is too small to detect on a standard chromosome analysis, so specialized genetic testing must be used. The most widely used diagnostic test is the FISH (flourescence in situ hybridization) test. The FISH test looks for the presence or absence of one of the genes in the middle of the Williams syndrome deletion, called the ELN gene. Other tests, such as microarrays (also known as oligoarrays, SNP arrays or arrayCGH) can also confirm the diagnosis of Williams syndrome. In almost all individuals with Williams syndrome, the size of the deletion is exactly the same.

Management/Surveillance

There is significant variability in the type, severity, and number of medical problems a person with Williams syndrome develops. This also means individuals with Williams syndrome may differ in their needs for medical interventions (such as heart surgery) and supportive therapies. Additionally, some medical conditions may develop or progress over time, so ongoing monitoring is recommended. In general, individuals with Williams syndrome should see a cardiologist regularly and have periodic blood work to monitor calcium levels, thyroid, kidney, and blood sugar status.

Developmentally, all individuals with Williams syndrome benefit from extra support and therapies (especially speech) while in school and will continue to need some level of support throughout their lives. Several guidelines have

been published with care recommendations for both children and adults with Williams syndrome.

Mode of inheritance

Williams syndrome is inherited in an autosomal dominant pattern. This means inheriting one deletion is typically enough for an individual to be affected and show signs of Williams syndrome. The deletion can be inherited from an affected parent, but most often it occurs brand new (de novo) in an affected child.

Risk to family members

The risk to family members depends on whether or not the individual with Williams syndrome has a parent with the deletion. If a parent also has the deletion, the risk for that parent to have another child with the deletion is 50% with each pregnancy. If a parent does not have the deletion, the risk of other siblings being affected is very low.

Special considerations

Although a rare occurrence, there is an increased risk for complications with anesthesia use among individuals with Williams syndrome. Therefore, an individual with Williams syndrome should see a cardiologist, have routine lab work, and meet the anesthesiologist prior to having surgery.

Resources

The Williams Syndrome Association
http://www.williams-syndrome.org
Genetics Home Reference: Williams syndrome
http://ghr.nlm.nih.gov/condition/williams-syndrome
Canadian Association for Williams Syndrome
http://caws.sasktelwebhosting.com

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