Journal of Down Syndrome & Chromosome Abnormalities

OPEN ACCESS Freely available online

Perspective

Causes and Symptoms of the Contiguous Gene Syndrome

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DESCRIPTION

Williams's Syndrome (WS) is a relatively rare hemizygous micro deletion disorder that affects 1 in every 7 people. The mispairing of low-copy DNA repetitive elements during meiosis causes WS. Most people with WS have a similar size deletion, which results in the loss of one copy of 25-27 genes on chromosome 7q11. Cases identified by a cardiologist are characterized by Supravalvular Aortic Stenosis (SVAS) along with a similar presentation of persistent growth failure, distinctive facial appearances, developmental delays, and an overly-friendly personality. Case reports describing individuals with both hypercalcemia and SVAS, as well as other features shared by both phenotypes, suggested that these seemingly unrelated presentations were variations of the same condition and same phenotype Williams-Beuren Syndrome (WBS) is a rare neurodevelopmental disorder caused by a mutation on chromosome 7 (7q11.23). With an estimated prevalence of 1 in 7,500-10,000 live births, the deletion is composed of approximately 26-28 genes and commonly includes one allele of the elastin gene. Laboratory tests such as Fluorescence *in situ* Hybridization (FISH) and array comparative genomic hybridization are examples of modern diagnostic methods Individuals with WS frequently exhibit a distinct set of medical, developmental, and social-emotional characteristics. Almond-shaped eyes, a stellate pattern in the iris, high and prominent cheekbones, and a flat nasal bridge with an upturned nose, full lips, a broad mouth, and abnormal dentition are all distinguishing features. Individuals with WS may have cardiovascular abnormalities, hypertension, hypercalcemia, and other symptoms. The strongest evidence for genotype-phenotype association is for ELN, the gene encoding elastin, which is responsible for the vascular and connective tissue features of WS, as well as the *GTF2I* and *GTF2IRD1* transcription factor genes have been linked to cognitive ability, social functioning, and anxiety. The deletion of *BAZ1B, LIMK1, STX1A*, and *MLXIPL* also has phenotypic consequences, but more research is needed to understand the mechanism by which these deletions contribute to clinical outcomes. In parts of the world where technological advances, such as chromosomal microarray, allow clinicians to make the diagnosis of WS without formally suspecting it, earlier intervention by medical and developmental specialists is possible. All cardinal features of WS have significant phenotypic variability, but the specific sources of this variability are unknown. Further research to identify the factors causing these differences may lead to mechanism-based rather than symptom-based therapies, which should be a high research priority.

Diagnosis

Williams's Syndrome is a contiguous gene syndrome, which means that all of the deleted genes "line up" within the "critical region" of 26-28 genes in Williams syndrome. Two DNA tests can be used to determine whether or not a person has Williams's syndrome. The FISH and Microarray test. Almost every person with Williams's syndrome will benefit from therapeutic intervention to help them overcome developmental delays, joint problems, fine motor issues, and other symptoms. A physician can assist to determine the need for therapy or can refer a therapeutic service provider. It is critical to begin therapies as soon as possible to reap the greatest benefit.

Causes

Neonates born with Williams' Syndrome lack certain genes. The symptoms they experience are determined by the genes they lack. Someone born without the *ELN* gene, for example, will have heart and blood vessel problems. Before the sperm and egg combine to form the baby, the genes are usually missing. Babies inherit genetic deletion from a parent who has the condition in a small number of cases, but it is usually a random disorder in the genes.

Symptoms

Symptoms included in Williams's Syndrome are Elevated calcium level in the blood, Sleep problems, ear infections or loss of hearing, unsteady walk and endocrine abnormalities like diabetes in adulthood, early puberty and hypothyroidism and manifest itself in various parts of the body, including the face, heart, and other organs.

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Received: 02-Sep -2022, Manuscript No. JDSCA-22-19535; **Editor assigned:** 06- Sep -2022, Pre QC No. JDSCA-22-19535 (PQ); **Reviewed:** 20- Sep -2022, QC No. JDSCA-22-19535; **Revised:** 27-Sep-2022, Manuscript No. JDSCA-22-19535 (R); **Published:** 04-Oct 2022, DOI: 10.35248/2472-1115.22.08.209.

Citation: Kelley R (2022) Causes and Symptoms of the Contiguous Gene Syndrome. J Down Syndr Chr Abnorm. 8: 209.