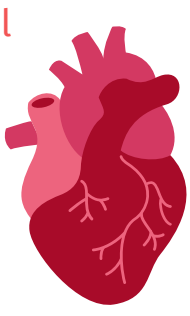
WILLIAMS SYNDROME

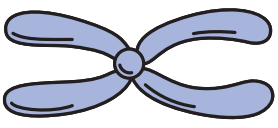
What is it?

Williams syndrome, also known as Williams-Beuren syndrome, is a rare genetic developmental disorder that affects various parts of the body. It is characterized by mild to moderate intellectual challenges, unique personality traits, distinctive physical features, and cardiovascular problems.

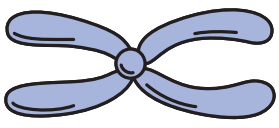
Symptoms

- Children with Williams syndrome can have delays in reaching developmental milestones, including learning, talking, sitting or walking, and socializing. The condition is marked by outgoing personalities, attention problems, anxiety, and phobias.
- Physical characteristics include large ears, upturned nose, full cheeks, epicanthal folds, wide mouth, small jaw, and a short stature.
- Other symptoms may involve chronic ear infections, dental abnormalities, endocrine abnormalities, elevated calcium levels, farsightedness, scoliosis, sleep problems, and connective tissue abnormalities.
- Cardiovascular issues, particularly supravalvular aortic stenosis (SVAS), are common as well and may lead to shortness of breath, chest pain, and heart failure.





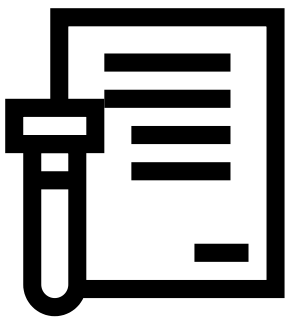
Cause



- Williams syndrome is caused by the deletion of genetic material from chromosome 7. The deleted region includes 25 to 27 genes, with ELN, GTF2I, GTF2IRD1, and LIMK1 among the affected genes. ELN gene loss is linked to connective tissue abnormalities and cardiovascular disease, while loss of GTF2I, GTF2IRD1, and LIMK1 contributes to cognitive difficulties and unique behavioural characteristics.
- The majority of cases are not commonly inherited. Chromosomal alteration is usually the result of a random event that occurs during the formation of reproductive cells, though the risk increases if a parent has an inversion chromosomal change in the region of chromosome 7 associated with Williams syndrome.

Diagnosis

Diagnosis is typically made during infancy or early childhood through physical examination, genetic testing, and assessment of characteristic features. Additional tests may include EKG or echocardiogram to check the heart, blood pressure monitoring, and tests for blood and urine to identify kidney symptoms.



Treatment

While there is no cure for Williams syndrome, management focuses on treating symptoms. Cardiovascular issues may require surgical intervention or other treatments determined by a cardiologist, and early intervention programs, therapies, and special education are recommended for developmental delays. Regular medical evaluations are essential to monitor and manage associated health problems.

References

- Cleveland Clinic [Internet]. Williams Syndrome; [reviewed 2021 Dec 10; cited 2024 Feb 19]. Available from: https://my.clevelandclinic.org/health/diseases/15174-williams-syndrome
- MedlinePlus [Internet]. Bethesda (MD): National Library of Medicine (US). Williams syndrome; [updated 2022 May 31; cited 2024 Feb 19]. Available from: https://medlineplus.gov/genetics/condition/williams-syndrome/

By: Faith Zou

The Vagus Project
Neuroscience Foundation