

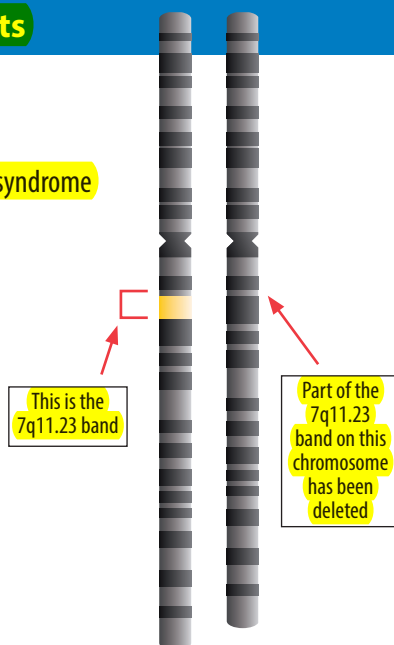
WILLIAMS SYNDROME

Patient and Clinical Presentation

Pediatrician ordered FirstStep^{Dx} for 5-year-old male with developmental delay

FirstStep^{Dx} Results

- 1.6 Mb deletion of chromosome 7q11.23
- Suggestive of Williams syndrome



Williams Syndrome Characteristics

- Supravalvular aortic stenosis (SVAS) and renal artery stenosis
- Intellectual disability
- Failure to thrive
- Hypotonia
- Hypercalcemia
- "Cocktail Personality"
- Incidence of 1 in 7,500-10,000 births
- AAP healthcare supervision guidelines published

AAP Management Guidance*

SPECIAL CONSIDERATIONS

- No multivitamins
- Diligent use of sunscreen
- Periodic cardiovascular evaluations
- Screen for hypertension
- Establish a medical home

LABORATORY EXAMS

- Yearly urinalysis
- Thyroid every 4 years
- Total calcium at least every 3 years
- Urinary calcium-creatinine ratio every 2 years
- Initial ultrasonographic exam of bladder and kidneys

ANNUAL EXAMS

- Williams syndrome specific growth charts
- Developmental exam
- Orthopedic issues
- Pediatric cardiology evaluation
- Inguinal hernia
- Review feeding issues
- Ophthalmologic evaluation
- Hearing and vision
- Constipation
- Blood pressure in both arms
- Femoral pulse
- Early puberty

TREATMENT

- Physical, speech and language, occupational, and sensory integration therapies
- Discuss treatment options for anxiety and ADHD
- Anesthesia complications, consult pediatric anesthesiologist
- Education placement assessment and planning

* American Academy of Pediatrics Committee on Genetics. 2001. Health Care Supervision for Children With Williams Syndrome. *Pediatrics* 107: 1192-1204.

Case Study Summary

- Idiopathic clinical diagnosis due to high variable expression (average age of diagnosis is 3.6 years)
- Identification of genetic diagnosis led to significant changes in clinical management
- Genetic counseling sessions focused on identifying strengths of children with Williams syndrome and the risk of recurrence