Williams Syndrome (WS)

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Course: Genetics II (MA 206)

Summer Class

Lecture 7

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Outline

- What Williams Syndrome
- Etiology and Genetic Basis
- Clinical Features of WS
- Treatment and Management
- What is Williams Syndrome

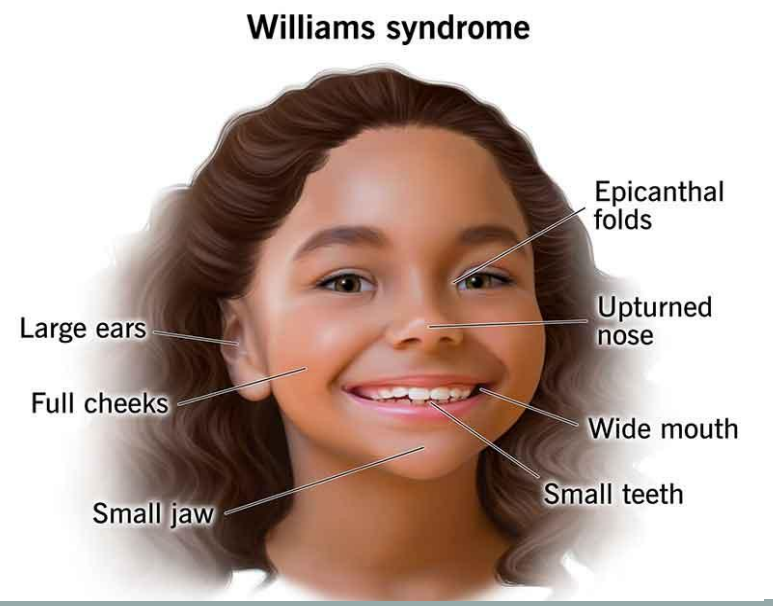
Objectives

- By the end of this lecture, students should be able to:
- Explain the genetic basis of cystic fibrosis (CF)
- Describe the molecular mechanism of CFTR protein dysfunction.
- Link CFTR mutations to clinical symptoms.
- Discuss current treatment strategies targeting CF mechanisms.

❑ What is Williams Syndrome (WS)?

- Is a rare **neurodevelopmental genetic disorder** that affects multiple systems in the body.

- It is caused by a microdeletion of genes on chromosome 7q11.23, which includes the **elastin (ELN) gene**.



❑ Williams Syndrome (WS):

- This disorder is characterized by:

✓ Distinctive facial features

✓ Cardiovascular problems

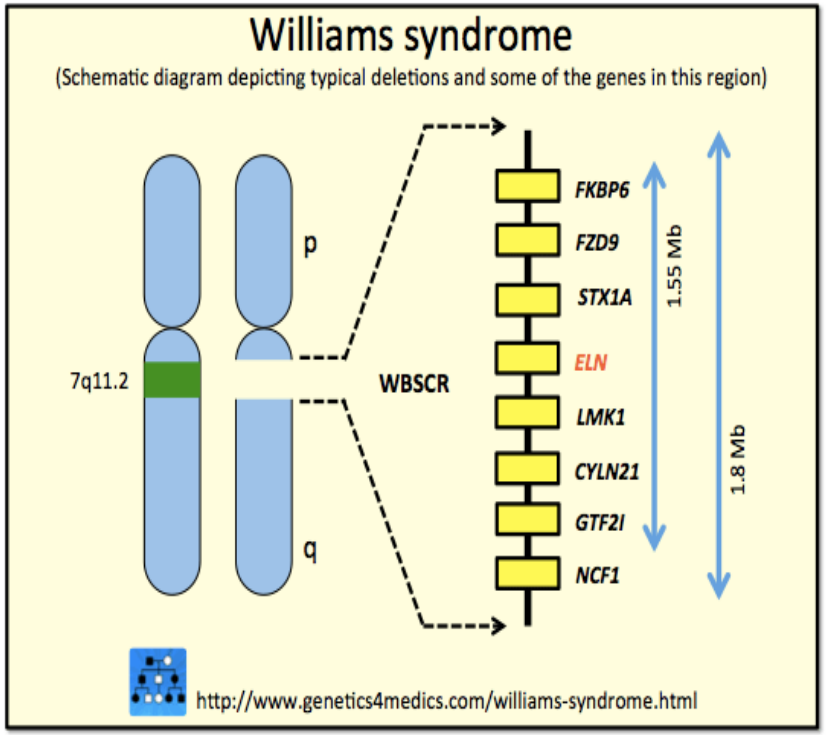
✓ Intellectual disabilities

✓ Unique personality traits.

❑ Etiology and Genetic Basis

- **Cause:** A deletion of approximately **26-28 genes** on **chromosome 7q11.23.**

- Key gene affected: ELN (**elastin gene**).

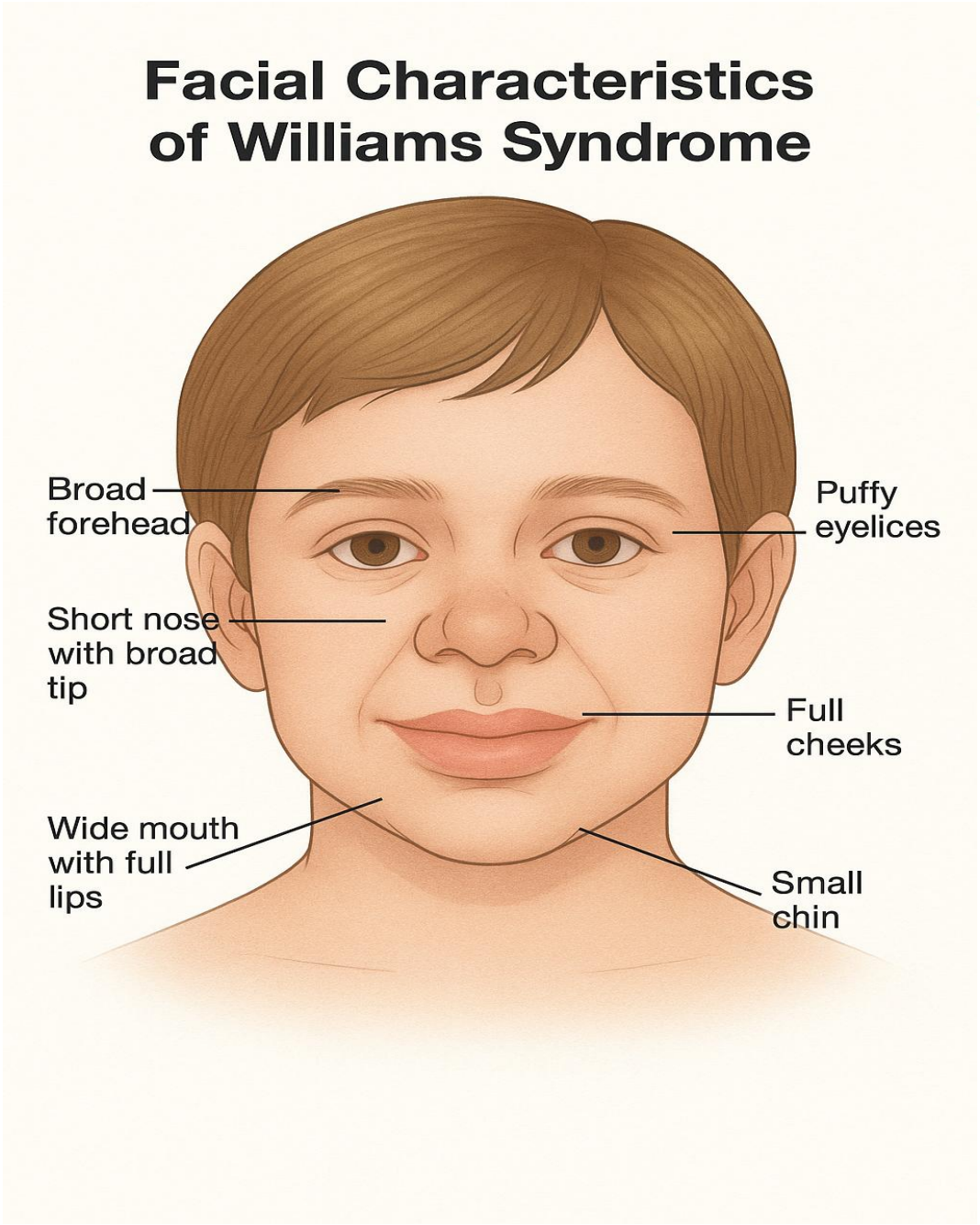


- **Inheritance:** Typically sporadic, but it can be inherited in an **autosomal dominant** manner.

❖ Clinical Features of WS:

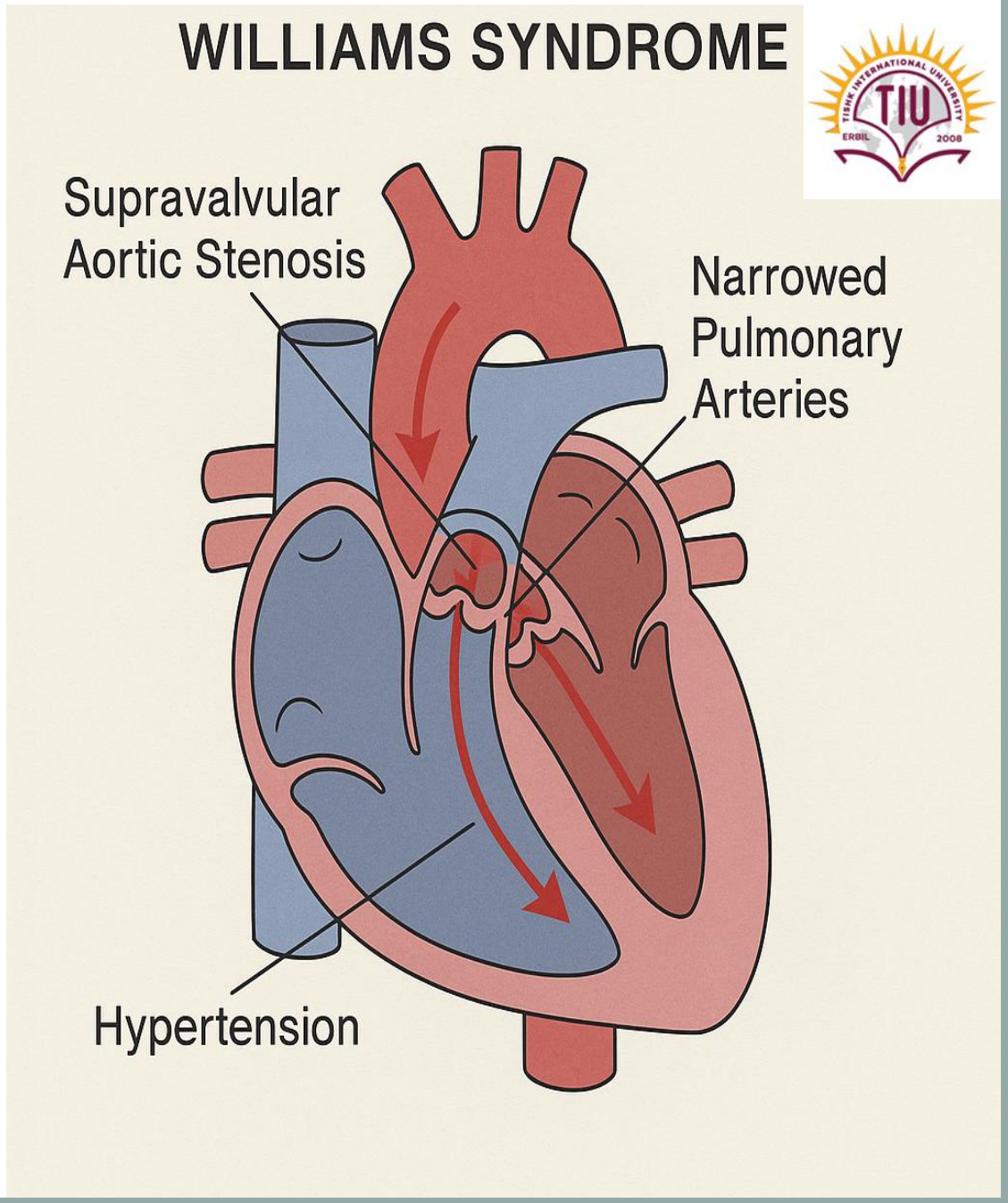
i. **Facial Characteristics**:

- Wide mouth
- Full lips
- Small chin
- Puffy eyes
- "Elfin-like" appearance.



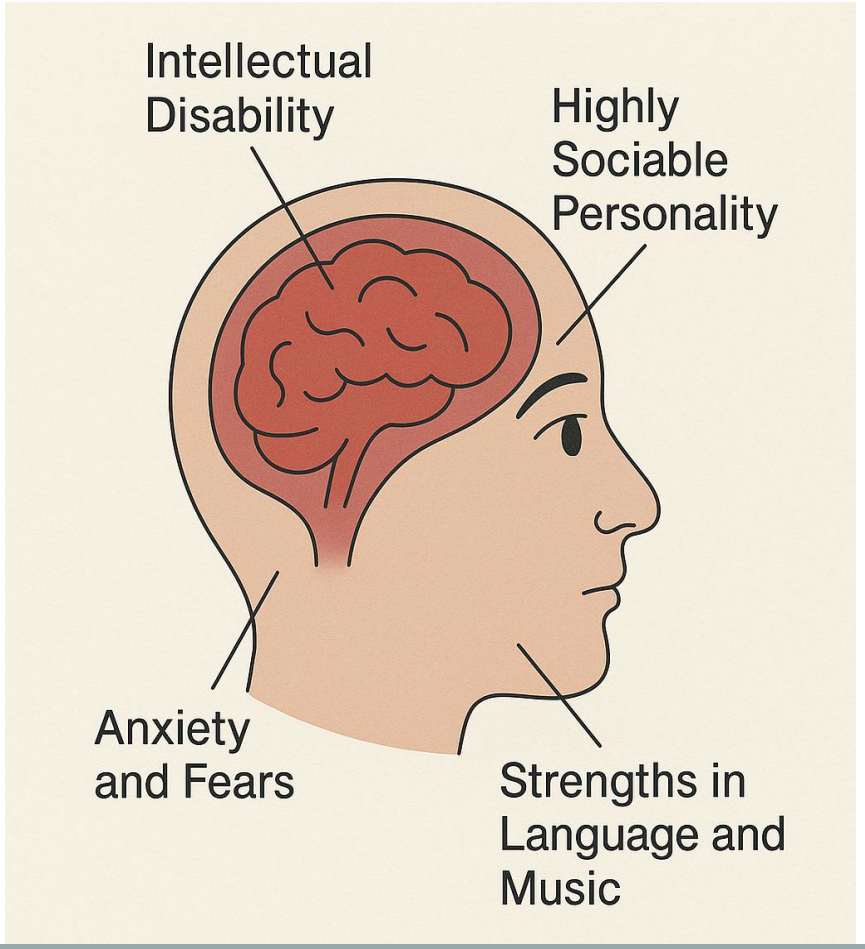
ii. **Cardiovascular Abnormalities**:

- Supravalvular aortic stenosis (SVAS)
- Hypertension



iii. Neurological and Cognitive Profile:

- Mild to moderate intellectual disability.
- Strong verbal and auditory memory skills.
- Poor visual-spatial skills



iv. Behavioral Traits:

- Highly social and friendly.
- Anxiety and attention deficit.
- Increased empathy

❖ Other Medical Issues:

- Hypercalcemia in infancy.
- Dental abnormalities.
- Gastrointestinal problems.
- Delayed growth and puberty

❑ Multisystem Effects:

System	Mechanism
Cardiovascular	Loss of ELN → vascular stiffness, SVAS
Nervous system	Gene deletions → abnormal neural development
Endocrine	Dysregulated calcium metabolism
Behavior	GTF2I deletion → altered social cognition & anxiety

❑ Diagnosis

1. Physical examination:

- ✓ Based on characteristic features.

2. Genetic testing:

- ✓ FISH
- ✓ Chromosomal microarray.

3. Echocardiogram:

- ✓ To assess heart anomalies.

❑ Management and Treatment

- There is no cure for Williams Syndrome, but symptoms can be managed:

✓ Cardiovascular monitoring

✓ Speech therapy, and educational support

✓ Behavioral therapy for attention and anxiety

✓ Calcium level management during infancy

References

1. Wallace DC. "Mitochondrial DNA mutations in disease and aging." Environmental and Molecular Mutagenesis, 2010.

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3. Vafai SB, Mootha VK. "Mitochondrial disorders as windows into an ancient organelle." Nature, 2012.

Thanks