Feeding Disorders and Growth in Williams Syndrome

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Williams (Williams Beuren) Syndrome
Overview

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Williams syndrome

Multi-system disorder: distinctive pattern

- Eating and speech dysfunction
- Mental retardation
  - specific strengths and weaknesses
  - characteristic personality and behavior
- Hyperacusis & sensorineural hearing loss
- Characteristic phenotype
  - distinctive subtle facial appearance
  - short stature with relatively short limbs
- Low tone and lax joints
- Soft skin
- Arterial narrowing
Williams syndrome

Cause:
- deletion of ±28 contiguous genes
- long arm of one chromosome 7 (del 7q11.23)

Incidence: 1: 8000

Multi-ethnic
Some of the ±28 genes that are deleted are very important for the fetal development of the brain:

- **GTF2IRD1** (general transcription factor 2 I repeat domain-containing 1) - may be related to mental retardation
- **LIMK1** (LIM kinase 1) - ? visuo-spatial
- Many other genes

Some genes have specific roles in other organs – for example

- **ELN** (elastin) - arteries, skin,
  - elasticity of gastro-intestinal tract & bladder
Williams syndrome

Multisystem disorder

- Brain
- Arteries & heart
- Gastrointestinal tract
- Renal (kidneys & bladder)
- Skin
- Endocrine
- Growth

Specific problems occur at each age:

- Infancy
- Childhood
- Early adolescence
- Late adolescence
- Adulthood
Williams syndrome: neonate & infant

Early infancy

Two most common medical problems:

- **Failure to thrive:**
  - Poor feeding – hypotonia
  - Vomiting - gastro-esophageal reflux
  - Irritability – colic

- **Murmur – narrow arteries**
  - Supravalvar aortic stenosis (SVAS)
  - Peripheral pulmonic stenosis (PPS)

- **Dysmorphism esp facial**
Williams syndrome: neonate & infant

- Fullness around the eyes
- Lacy blue irises
- Flat nose bridge
- Upturned nares
- Bulbous nose tip
- Full lips – pouty lower lip
- Small chin
- Full cheeks
- Soft skin
- Low tone
Williams syndrome: feeding problems

Infancy & Childhood - many problems are inter-related:

**CNS:** • cognitive problems • developmental delays • low tone

**CVS:** • cardio-vascular

**GI:** • feeding difficulties: failure to thrive
• gastro-esophageal reflux (GER)
• constipation → diverticula
• irritable and crying incessantly: “colic”

Hypercalcemia

Tactile defensiveness
Williams syndrome: mid-childhood

2nd year onwards

- Irritability and vomiting diminishes/resolves
- Hypercalcemia resolves
- Hypotonia persists - drooling
- Teeth: Malocclusion
  - Small and absent teeth
- Persistent feeding problems:
  - Delayed ability to chew and swallow coarser textured foods
- Constipation persists
Williams Syndrome: growth & puberty

- Linear growth
  - Short stature is common
  - Usually not evident at birth: 5-50%ile.
  - Manifests in early childhood:
    - Large proportion < 5%ile
    - Some - normal range compared with general population and midparental height
  - Pubertal growth spurts - average 2 years earlier than healthy control children
  - Adult heights – 10-15 cm lower than control
Williams syndrome: growth
Weight and length – often below 5th centile for general population – causes concern

General population: <5%
Specific growth charts for WS: 25%

Weight, Females
Stature, Females
Williams Syndrome: Cognition

Global Developmental Delay

- Unique profile of strengths and weaknesses

Relative Strengths

- Speech and language – delayed until approximately 3 years
  - flowery, emotional (prosody),
  - good grammar
- Short term (working) memory
- Facial recognition

Weaknesses

- Visuo-spatial motor cognition
- Mathematics
- Understanding social actions
Williams syndrome: Personality and Behavior

- Infancy = irritable → childhood = pleasant
  Very sociable; inappropriately friendly to strangers
- Hyperactive
- Distractable
- Hyperacusis (and sensorineural hearing loss)
- Perseveration and obsessions
- Anxiety – generalized and anticipatory
- Sleep - disturbed architecture
- Musical appreciation (but not higher ability)
- Empathy
- Do not perceive social cues
Feeding Disorders in Williams Syndrome

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Introduction

- Feeding and swallowing problems
- Impact of medical conditions on development
- Evaluation and treatment
Conditions Contributing to Feeding and Swallowing Disorders in Williams Syndrome

- Cardiovascular Disease
- Neurological Abnormality/Hypotonia
- Gastrointestinal Disorders
- Failure to Thrive
- Developmental Delay

“Williams syndrome is associated with oral motor delay and feeding difficulty in infancy and early childhood”.

*Morris & Mervis, Sept, 2000*
### Medical Problems of Infants with Williams Syndrome

<table>
<thead>
<tr>
<th>Problem</th>
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<tr>
<td>Failure to Thrive</td>
<td>81</td>
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<tr>
<td>Feeding difficulty</td>
<td>71</td>
</tr>
<tr>
<td>Colic</td>
<td>67</td>
</tr>
<tr>
<td>Constipation</td>
<td>43</td>
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<tr>
<td>Vomiting</td>
<td>40</td>
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<tr>
<td>Chronic otitis media</td>
<td>38</td>
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1988
Identified Feeding Problems

- Disordered sucking in infancy
- Inefficient oral motor patterns
- Delay in chewing skill acquisition
- Limited volume (oral intake)
- Poor growth
- Dysphagia
Conclusions

Experience with Williams Syndrome has provided consistent information that emphasizes the need for:

- Early diagnosis
- Comprehensive medical management
- Appropriate assessment and intervention of feeding and swallowing function