Awareness for Rubinstein-Taybi Syndrome

RARE genetic disorder that occurs in 1 in 100,000 to 300,000 people worldwide

GENETICS
Usually the result of a de novo mutation, meaning there is no record of familial incidence. RTS can be the result of mutations in the CREBBP or EP300 genes. Deletions in Chromosome 16 are also responsible.

About HALF of current RTS cases do not have these particular mutations, so diagnosis is based on clinical information and available medical records for the patient.

PHYSICAL APPEARANCE
Clinical diagnosis is based on distinct physical characteristics, including:
- Broad thumb and great toe
- Angulated thumbs
- Highly Arched Eyebrows
- Long eyelashes
- Downslanting eyes
- Broad Nasal Bridge
- Small Head
- High Arched Palate

The grimacing or unusual smile with nearly complete closing of the eyes is almost universal.

GROWTH
Average length at birth, but drop well below average within the first few months of life. Children with RTS are short throughout life and do not seem to hit the usual pubertal growth spurt. Average male height is 5’ and average female height is 4’10”.

LANGUAGE
90% of children have significant speech delays. A small percentage use sign language exclusively, while many use a combination of sign language, speech, and augmentative devices. Most require formal speech therapy.

DEVELOPMENT
All children with RTS have developmental delays, but the degree of limitation varies widely. All children develop at their own time.

<table>
<thead>
<tr>
<th>Skill</th>
<th>Avg (mos)</th>
<th>Range (mos)</th>
<th>Typical (mos)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Rolled Over</td>
<td>7</td>
<td>2-24</td>
<td>2-5</td>
</tr>
<tr>
<td>Sat Up</td>
<td>11</td>
<td>6-30</td>
<td>5-8</td>
</tr>
<tr>
<td>Crawled</td>
<td>15</td>
<td>8-30</td>
<td>7-10</td>
</tr>
<tr>
<td>Walked</td>
<td>30</td>
<td>15-54</td>
<td>11-15</td>
</tr>
<tr>
<td>Rode Trike</td>
<td>68</td>
<td>42-216</td>
<td>36-48</td>
</tr>
<tr>
<td>First Word</td>
<td>25</td>
<td>6-57</td>
<td>9-13</td>
</tr>
<tr>
<td>Word phrases</td>
<td>65</td>
<td>24-156</td>
<td>14-24</td>
</tr>
<tr>
<td>Toilet Trained</td>
<td>63</td>
<td>30-216</td>
<td>24-27</td>
</tr>
</tbody>
</table>

COMMON MEDICAL PROBLEMS
Medical issues vary widely among children, but commonly reported are:

NEUROLOGICAL
Moderate to severe intellectual disability, seizures, structural abnormalities, high/low muscle tone.

DENTAL
Small mouth and jaw can lead to overcrowding of teeth, dental cavities, high incidence of talon cusps.

EAR, NOSE, & THROAT
Frequent ear infections, mild hearing loss, enlarged tonsils and adenoids, floppy laryngeal wall.

MUSCULOSKELETAL
Surgery for angulated thumbs is recommended before age 2 to prevent complications in functional dexterity. Surgery on angulated great toes only when it affects walking or shoes. Hypermobility can be an issue in adolescence. Some children are also at risk for scoliosis, kyphosis, or lordosis.

OPHTHALMOLOGY
Lacrimal Duct Obstruction, ptosis, strabismus, refractive errors, coloboma. Glaucma may be present at birth or early in life which can lead to blindness if undetected.

CARDIOLOGY
1/3 have congenital heart defect. 65% have a single defect, while 35% have two or more defects or complex formation.

GASTROENTEROLOGY
Reflux, vomiting, constipation, in the first year 80% of children have feeding difficulty requiring a nasogastric or gastrostomy feeding tube. Feeding difficulties often resolve over time and many develop a voracious appetite.

RESPIRATORY
Recurrent respiratory infections, aspiration, obstructive sleep apnea, tracheostomy, pulmonary hypertension, and recurrent pneumonia.

GENITOURINARY
Almost all boys have incomplete or delayed descent of testicles. Hypospadias is seen in 11%, Renal anomalies are present in about 50% and may increase likelihood of urinary tract infections. Kidney abnormalities may also be present.

DERMATOLOGIC
22% of individuals have keloids and 16% have issues with hypertrophic scarring. Can be result of minimal trauma (bee stinging) or surgical.

Facts from:
- rubinstein-taybi.com
- omim.org/entry/180849
- ghr.nlm.nih.gov/condition/rubinstein-taybi-syndrome
- rtsuk.org