AWARENESS FOR RUBINSTEIN-TAYBI SYNDROME

Rare Disease Day is celebrated the last day in February each year to raise awareness for the millions of individuals living with a rare disease.

World RTS Day is celebrated July 3rd each year. RTS families across the world celebrate their loved ones who have RTS.

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.

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

COMMON MEDICAL PROBLEMS

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

NEUROLOGICAL

Moderate to severe intellectual disability, seizures, structural abnormalities, tethered spinal cord, and 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, kyphois,or lordosis.

OPHTHALMOLOGY

Lacrimal Duct Obstruction, ptosis, strabismus, refractive errors, coloboma. Glaucoma 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 steeing) or surgical.

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