INTRODUCTION:
Rubinstein–Taybi syndrome (RSTS) was first reported by Greek orthopedic surgeons Michail et al. in 1957 as the broad thumb-hallux syndrome in “a new case of congenital malformations of the thumbs absolutely symmetrical” and then later in 1963 was described by American physicians, Rubinstein and Taybi, who gave their names to this syndrome, after a more detailed study on seven children with this syndrome. They described a new symptom-complex characterized by broad thumbs and great toes, facial dysmorphism, mental retardation and a group of congenital malformations.

Rubinstein–Taybi syndrome (RSTS) or 6p13.3 deletion syndrome is a rare autosomal dominant genetic disorder with a birth prevalence of 1:100,000 –125,000. Male and female individuals are affected at equal rates.

It is a multisystem developmental disorder characterized by prenatal and postnatal restriction, microcephaly, dysmorphic features, broad thumbs and toes, and intellectual disability. Features include distinctively broad and/or angled fingers and toes, growth and development delay, speech delay, intellectual disability, craniofacial dysmorphism, feeding difficulties, recurrent respiratory infections and urogenital anomalies. In some people, the skin, cardiac or respiratory system may also be affected. Symptoms associated with RSTS vary greatly from person to person.

CASE REPORT:
A 13-year old male, 2nd born to non-consanguineous parents, came to the Department of Pediatrics, Sree Balaji Medical College and Hospital, Chennai with the complaints of undescended testis since birth. The child was born at term via normal vaginal delivery, weighing 2.5 kg and cried immediately after birth with no history of perinatal asphyxia. He has history of delayed development of both motor function and speech with intellectual disability. He has had an episode of seizure at one and half year of age. His elder sister is normal.

On examination, he was found to have:
- Short stature (Height - 112 cm, < -3 SD according to WHO)
- Microcephaly (Head circumference - 48.5 cm)
- Intellectual disability: Moderate (IQ - 46)

Dysmorphic facial features- high arched eyebrows, downward slanting of palpebral fissures, strabismus, ptosis, long eyelashes, beaked nose with short low columella, deformed ear pinna, high arched palate, thin upper lip, everted lower lip, malocclusion and multiple caries of teeth, micrognathia

Broad thumb and first toe, flat foot
Micropenis, undescended testis, hirsutism

The child has had a past history of corrective surgeries performed for glaucoma and strabismus in his childhood at the age of 3 years.

The genetic work-up of the child could not be done owing to family's financial constraints.
Genitourinary:
Male infants with RSTS may have abnormalities of the genitourinary tract including cryptorchidism, hypospadias and shawl scrotum. In addition, infants with RSTS may have hypoplastic or absent kidneys, repeated infections of the urinary tract, renal stones, hydronephrosis, vesicoureteral reflux. In some cases, duplication of the kidneys and/or ureters may also be present.

Cardiac:
Congenital heart disease is seen in one-third of the patients. Patent ductus arteriosus is the most common congenital heart defect present in infants with RSTS. They may also have atrial septal defect, ventricular septal defect, coarctation and stenosis of the aorta, pulmonic stenosis and bicuspid aortic valve.\(^3\)

Respiratory:
The lungs may be divided into small extra lobulations. Laryngeal walls may be weak or easily collapsible, potentially resulting in swallowing or breathing difficulties like sleep apnea. This can also cause difficult intubation.

Behavior:
Individuals with RSTS often exhibit a short attention span, decreased tolerance for noise and crowds, impulsivity, and moodiness. Autistic behaviors are common.

Malignant potential:
Some persons with RSTS appear to be more prone to develop neural, developmental and hematological origin tumors including meningioma, pilomatrixoma, rhabdomyosarcoma, neuroblastoma, medulloblastoma, oligodendrogloma, leioyosarcoma, seminoma, odontoma, choristoma, and leukemia than the general population.\(^{3,4}\) However, this is somewhat controversial as one recent study found only an increased risk for meningiomas and pilomatrixomas, but not for malignancies in general.

The prognosis for children with RSTS is generally good, but it may vary due to the range and severity of the health problems that may be present. Most patients have developmental delay and intellectual disability but most of the 6-year old children are able to learn to read. Life expectancy generally does not seem to be affected, except in children with complex cardiac defects. Malignancies and respiratory infections are the most common causes of death. Survival rates in general are good and there are many reports of adults with RTS.

REFERENCES: