

## RUBINSTEIN –TAYBI SYNDROME

### Key facts

- Rubinstein –Taybi Syndrome is a rare chromosomal abnormality which creates a specific pattern of physical features and developmental disabilities.
- Occurs during a baby's development in the womb.
- This is a random condition and is non-hereditary (with the exception of those with Rubinstein-Taybi have a 50% chance of producing a child with the syndrome).
- Rubinstein –Taybi Syndrome occurs in between 1 in 100,000 and 1 in 125,000 births. Occurs in males and females with equal frequency.
- Can have feeding difficulties, particularly in the early months, eg difficulties in breast feeding, gastroesophageal reflux, vomiting (sometimes projectile), and constipation.
- Dental problems are common.
- Can develop a voracious appetite in late childhood (similar to Prader-Willi Syndrome).
- Can experience cardiac defects (most of which can be corrected or treated).
- May have a high pain threshold.
- Behaviour – can be stubborn and display rocking, spinning and hand-flapping, (more commonly in early years).
- Increased risk of cancer (especially benign and malignant tumours; leukaemia and lymphoma).
- Can suffer from sleep apnoea.
- Can suffer from keloid formation (overly aggressive healing response to a wound).

### Physical Appearance

- Beaked nose
- Highly-arched palate
- Downward-slanting eyes and heavy or highly-arched eyebrows
- Broad thumbs and big toes (sometimes angulated, which can be corrected surgically)
- Smaller than average height
- In males - undescended testicles

### Aspects of development that can be affected:

- Ranging from mild to severe learning disability.
- A small percentage will not develop speech, so sign language should be introduced.
- Can have eye defects e.g. Squint, tear duct obstruction, cataracts and glaucoma.
- Lax muscle tone.

### School implications to consider

#### **Strengths**

- Usually friendly

#### **Considerable weaknesses**

- Short concentration span
- Reluctant to accept changes in routine
- Possible hearing loss
- Diminished vision
- Mild to severe learning difficulties

### Treatment

- A range of treatments are available to reduce some of the possible symptoms, but there is no cure.
- SALT

### Useful websites

<http://rtsuk.org/>

The UK based Rubinstein –Taybi Syndrome Support group. The website contains lots of information and links.