



Further Inform Neurogenetic Disorders

Health Issues in Rubinstein-Taybi Syndrome

Parents and carers have told us that being presented with a long list of health difficulties can be daunting. However, they also reported that having this list available can be reassuring and can help them to watch out for early warning signs. The key message here is that children with Rubinstein-Taybi Syndrome may be prone to some health difficulties; however, with appropriate support and regular health checks with an informed professional, a lot can be done to reduce the impact on an individual's well-being.

Eating behaviour and health

It has been estimated that 80% of young children with Rubinstein-Taybi Syndrome exhibit feeding difficulties, vomiting and failure to thrive (reduced growth development). For most infants, these feeding difficulties decrease after the first year of life.

While young children with Rubinstein-Taybi Syndrome tend to be underweight compared to other individuals their age, throughout childhood their weight tends to increase and obesity may become prominent within adolescence.

Gastro-oesophageal reflux

The above feeding difficulties mentioned may be aggravated by gastro-oesophageal reflux. Reflux is a health condition where stomach acid moves back up into the oesophagus (similar to heartburn); this can be painful for the individuals that experience it.

Keloids and cancer

In individuals with Rubinstein-Taybi Syndrome, the healing of wounds tends to be atypical where scar tissue forms irregularly and covers a larger region – these are referred to as 'keloids.'

Keloids have been reported to affect around one quarter of individuals with Rubinstein-Taybi Syndrome with emergence occurring during puberty. Keloids can be itchy and painful and occur despite very little damage to the skin.

Individuals with Rubinstein-Taybi Syndrome are also at risk of rare benign tumours; with one of the most common reported being tumours of the hair follicles. Another type of tumour reported has been of dental tissue.

Research conducted in the Netherlands of all individuals in the country with Rubinstein-Taybi Syndrome suggested that individuals are at heightened risk of cancer with the affected genes in the syndrome being

labelled 'cancer suppressor' genes. Types of cancer that individuals may be at heightened risk of include cancers of the brain, testicles and connective tissues, bone marrow, adrenal glands, gut and kidneys.

Dental and eye problems

Dental issues have been reported in Rubinstein-Taybi Syndrome which have included abnormal positioning of teeth, missing or additional teeth and talon cusps (masses on the back of the front teeth).

There have also been a variety of eye problems reported in Rubinstein-Taybi Syndrome. Most individuals are short-sighted and around half experience a sensitivity to light. Other issues include blocked tear ducts, drooping eyelids, misalignment of eyes and refractory errors. There are other issues that are less common such as holes in the eye structure, increased pressure in the eyes and involuntary movement of the eyes. Individuals may also have cornea abnormalities and cataracts (clouding of the lens of the eye).

Skeletal issues

Individuals with Rubinstein-Taybi Syndrome may experience curvature of the spine and problems where the head and spine meet. There have also been reports of dislocation of knee caps and problems with bones/joints of the leg and hip.

Bone fractures have been reported to be prevalent within Rubinstein-Taybi Syndrome.

Other health conditions

Individuals with Rubinstein-Taybi Syndrome may experience heart and kidney problems, extreme constipation and frequent respiratory infections. Heart problems from birth affect an estimated one-third of individuals with Rubinstein-Taybi Syndrome.

Nearly all males with Rubinstein-Taybi Syndrome have undescended testes but puberty onset is normal

There are a range of health issues that can be associated with Rubinstein-Taybi syndrome so if you are concerned that any of them are affecting the individual with Rubinstein-Taybi Syndrome that you know, please contact a medical professional for advice.