



Further Inform Neurogenetic Disorders

Key Facts about Rubinstein-Taybi Syndrome

It is estimated that 1 in every 100,000 to 125,000 people have a diagnosis of Rubinstein-Taybi Syndrome (RTS).

Rubinstein-Taybi Syndrome can also be known as Broad thumb-hallux Syndrome. This is because physical characteristics of the syndrome include broad thumbs and toes.

In most cases, Rubinstein-Taybi Syndrome is caused by a genetic change to a single gene. Rubinstein-Taybi Syndrome can also be caused by a genetic change to another single gene while for some individuals the genetic cause is unknown.

Research has suggested that most individuals with Rubinstein-Taybi Syndrome have a moderate intellectual disability. Some individuals may have a greater degree of disability and some may be mildly affected.

While research has suggested that 60% of individuals with Rubinstein-Taybi Syndrome may show autistic-like behaviours, the profile of these characteristics may differ from the standard characteristics of autism.

Individuals with Rubinstein-Taybi Syndrome may experience mood changes as they grow older which may include temper outbursts and anxiety.

Sleep difficulties may be experienced by individuals with Rubinstein-Taybi Syndrome such as sleep apnoea.

Health issues may occur in Rubinstein-Taybi Syndrome. These may include heart, kidney, skeletal, dental and eye problems. Gastro-oesophageal reflux (similar to heartburn) may occur. Nearly all males have undescended testes.

Individuals with Rubinstein-Taybi Syndrome have been described as having friendly and happy personalities where social contact and interest is common.

Hyperactivity, impulsivity and repetitive behaviours have been reported in some individuals with Rubinstein-Taybi Syndrome.

The majority of individuals with Rubinstein-Taybi Syndrome will develop some independence but may struggle with mobility.