# Diagnosis of Rubinstein-Taybi Syndrome

A genetic diagnosis of Rubinstein-Taybi Syndrome, where the genetic abnormality causing the syndrome is identified is only confirmed in approximately 65-70% of cases. To obtain a genetic diagnosis, genetic testing would need to be completed where a sample of blood or saliva is taken and is tested for the genetic changes known to cause Rubinstein-Taybi Syndrome.

In the remainder of cases, where a genetic diagnosis cannot be obtained, a clinical diagnosis is obtained from either a paediatrician, GP or clinical geneticist.

A clinical diagnosis of Rubinstein-Taybi Syndrome relies on identifying clinical features of the syndrome such as intellectual disability, a distinctive facial appearance and broad thumbs and toes which can be noted clinically or observed using x-ray techniques.