

The symptoms, progression and severity of SRS does not appear to vary from patient to patient. Affected children have a “gestalt” consisting of facial dysmorphism with a prominent lower lip, an asthenic build, low muscle mass, kyphoscoliosis and speech abnormalities. Males with SRS have low muscle tone (hypotonia) at birth. Symptoms appear early, especially the facial features. Developmental milestones are also not met early in life. The developmental delay progresses such that many boys with SRS have some motor disability. Osteoporosis develops which can result in numerous fractures without a causative event. Seizures are not common and severity varies. Snyder-Robinson syndrome is a rare X-linked intellectual disability disorder and as such it is difficult to estimate its prevalence. As SRS has been identified in patients located in the United States, South America and Europe, it is likely not restricted to any ethnic population geographical locale. Many XLID disorders present with hypotonia very early in life. However, SRS is the only human disorder related to an abnormality in polyamine biosynthesis.