What is SRS?

What is Snyder-Robinson Syndrome?

SRS is a rare genetic disorder, characterized by intellectual disability and developmental delays. These delays may affect speech, mobility, and cognition. SRS may present muscle and bone abnormalities, and other challenges with normal development. Many people with SRS are thin and have low muscle mass. SRS causes skeletal problems, particularly osteoporosis. Neurological problems are common, including seizures that begin in childhood. SRS may also present additional health challenges, such as kidney or respiratory issues.

Who is Affected?

Snyder Robinson occurs only in males. It is associated with alterations, or mutations of the SMS gene, on the X chromosome. Because of this mutation, the body does not produce certain enzymes, such as polyamines, like it should. Polyamines are necessary for normal development, and function of the brain and other parts of the body. SRS is an ultra rare condition. Globally, about 35 infected males have been identified, although actual occurrences are probably greater. SRS has been discovered in at least eight countries worldwide.

What are we Doing About It?

In 2014, the Snyder Robinson foundation was established, by six SRS families, to promote awareness, and advance medical research, related to SRS. The foundation held its inaugural conference in 2015, and raised enough funds for its first medical research fellowship in 2016. The foundation has an established board of directors, and a medical and scientific advisory group.

How Can I Help?

There are many ways for you to get involved. Donations to the Snyder Robinson Foundation, help to fund scientific research, medical advancements, and therapy options for Snyder Robinson Syndrome. For more information, contact us at michael.raymond@snyder-robinson.org, or visit us at www.snyder-robinson.org.