Learn

What is Snyder-Robinson Syndrome?

SRS Overview Slides

Snyder-Robinson Syndrome (SRS) is a rare genetic condition. SRS is an X-linked Intellectual Disability syndrome that affects only males. In a few cases SRS has resulted from a de novo mutation. SRS is characterized by intellectual disability, muscle and bone abnormalities, developmental delays, and sometimes other medical problems.

Individuals with SRS have delayed development evident in early childhood. The delays affect speech, mobility and cognition. Intellectual disability in a person with SRS can range from mild to severe. Some individuals with SRS develop speech, often late, and speech difficulties are common.

Most people with SRS are thin and have low muscle mass. Low muscle tone (hypotonia) typically becomes apparent in infancy, and the loss of muscle mass continues with age. People with SRS often have difficulty walking, some are not able to walk.

In those people who develop the ability to walk, most do so with an unsteady gait.

SRS causes skeletal problems, particularly thinning of the bones (osteoporosis) that starts in early childhood. Osteoporosis causes bones to be brittle and to break easily, often during normal activities and in the absence of trauma. In people with SRS, broken bones occur most often in the long bones of the arms and legs. Most affected individuals also develop an abnormal side-to-side and back-to-front curvature of the spine (scoliosis and kyphosis, often called kyphoscoliosis when they occur together). Affected individuals, in general, tend to be shorter than their peers and others in their family.

SRS is also associated with distinctive facial features, including a prominent lower lip; a high, narrow roof of the mouth or an opening in the roof of the mouth (a cleft palate); and differences in the size and shape of the right and left sides of the face (facial asymmetry). Other signs and symptoms may include neurological problems, seizures, and abnormalities of the genitalia and kidneys.

What Causes SRS?

SRS Tri-Fold Brochure

SRS is associated with alterations or mutations of the SMS gene on the X chromosome. The SMS gene contains the instructions for making an enzyme called spermine synthase. This enzyme is involved in the production of spermine, which is a type of small molecule called a polyamine. Scientists know that polyamines have many critical functions within cells. Studies suggest that these molecules play roles in cell growth and division, the production of new proteins, the repair of damaged tissues, the function of molecules called ion channels, and the controlled self-destruction of cells. Polyamines are thought to be necessary for normal development and function of the brain and other parts of the body.

The mutations in the SMS gene lead to a greatly reduced or entirely eliminated activity of spermine synthase, which decreases the amount of spermine in cells. A shortage of this polyamine clearly impacts normal development, including the development of the brain, muscles, and bones. Scientists do not yet know how this shortage results in the specific signs and symptoms of Snyder- Robinson syndrome. This is the subject of current medical research.

Historical Timeline:

First SRS Case

Drs. Russell Snyder and Charles Robinson Report the first Case of SRS

1969

NIH Funding

NIH Provides Funding for Greewood Genetic Center (GGC)-Miami for Research on X-Linked Intellectual Disability

1989

Visits with SRS Families

1991-2009

Mapping of Gene Xp22.11

1996

SMS Gene Identification

2003

First Treatment Trial

2009

SRS Foundation Established

Six Families Affected by SRS Come Together to form the Snyder-Robinson Foundation

2014

SRF Inaugural Conference

The SRF Brings Together Patients, Families, Physicians, and Researchers in the Same Room for the First Time

July 2015

Research Opportunities

SRF Awards the First Research Fellowship, and Joins with Penn Medicine to Award the First Two Research Grants for SRS Research

2016

For Further Reading…

- http://ghr.nlm.nih.gov/condition/snyder-robinson-syndrome
- http://www.ncbi.nlm.nih.gov/books/NBK144284
- http://www.plosone.org/article/info%3Adoi%2F10.1371%2Fjournal.pone.0110884#authcontrib
- http://rarediseases.info.nih.gov/gard/5615/snyder-robinson-syndrome/resources/1

- http://www.ncbi.nlm.nih.gov/pubmed/20556796
- http://www.genome.jp/dbget-bin/www_bget?hsa:6611+H00597
- http://hmg.oxfordjournals.org/content/early/2013/05/21/hmg.ddt229.short
- http://onlinelibrary.wiley.com/doi/10.1002/iub.1237/full
- http://articles.complexchild.com/feb2014/00531.html
- http://video214.com/play/FKV1KZjLpCMSX72V1qFrrg/s/dark
- https://globalgenes.org/raredaily/one-300-million-snyder-robinson-syndrome
- http://www.foxcarolina.com/story/27528797/clemson-researchers-work-to-change-dna-prevent-disease