What is Snyder-Robinson Syndrome?

Snyder-Robinson Syndrome (SRS) is an X-linked intellectual disability disorder caused by a mutation in the Spermine Synthase (SMS) gene. It occurs exclusively in males. Typical symptoms of SRS include:

- Intellectual Disability
- Developmental Delays
- Seizures
- Osteoporosis
- Hypotonia
- Scoliosis/Kyphosis
- Pulmonary / Respiratory Issues (Less Common)
- Cardiac Issues (Less Common)
- Renal Issues (Less Common)

SRS is the Only Known Disease Caused by an Imbalance of Polyamines
Spermine Synthase (SMS) Gene and the SMS Enzyme

- The SMS Gene Provides Instructions for Making an Enzyme Called Spermine Synthase
- This enzyme is an Essential Protein that Helps Convert Spermidine to Spermine
- Spermine is Thought to be Necessary for Normal Development and Function of the Brain, and Other Parts of the Body
- All Individuals with SRS have Deficient SMS Enzyme Activity
Historical Timeline

- 1969: First Reported Case by Drs. Russell Snyder and Arthur Robinson
- 1989: NIH Funding for GGC-Miami Research on X-linked Intellectual Disability
- 1991-2009: Visits with SRS Families
- 1996: Mapping of Gene Xp22.11
- 2003: SMS Gene Identification
- 2009: First Treatment Trial
- 2014: SRF Foundation Established
- 2015: Inaugural SRF Conference
- 2016: Funded 1 Research Fellowship; Collaboration with Penn Medicine for 2 Research Grants
The Snyder-Robinson Foundation (SRF)

- Six SRS Families Started the SRS Foundation in 2014
- Goals:
  - Raise Awareness
  - Advance Medical Research
- Through SRF Many Newly Diagnosed SRS Families are Connecting
- SRF is a 501c(3) Non-Profit Organization
Worldwide Distribution

Since 1969, Approximately 35 Males Have Been Identified in at Least Eleven Countries
Collaborative Research Plan

1. **Research Collaboration** – Collaborate with Penn Medicine for Research Grants – Through the Million Dollar Bike Ride (MDBR), Penn Medicine has graciously matched donations for Snyder-Robinson Research.

2. **Current Studies Include:**
   a) The Role of Mitochondria and ROS in Etiology of SRS
   b) Exploring why MSC with a dysfunctional SMS gene cannot become normal, mature osteoblasts
Contact Us

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Questions?