Snyder-Robinson Syndrome (SRS) is a genetic condition resulting in the dysfunction of Spermine Synthase (SMS). SMS catalyzes the conversion of spermidine to spermine is the last step in the polyamine pathway and polyamine levels are altered in SRS. There is some evidence that SMS may have additional functions.

Clinical features include intellectual disability, seizures, developmental delay, and osteoporosis with fractures in the absence of trauma, along with defects in other organ systems. There is a wide range of severity among individuals with SRS.

Mouse models with alterations in SMS are available for research studies through The Jackson Laboratory.

One $68,840 grant is available for SRS. Research focus areas include new studies into understanding pathophysiology or mechanisms by which SRS causes disease, as well as corresponding treatment options.

There is particular interest in the following areas, however, other novel approaches are encouraged:

1. Examine whether the loss of the ability of the SMS protein to synthesize spermine accounts for all of the changes seen in SRS. Structural studies on the SMS protein have identified mutations leading to stable but inactive protein that could be used for such investigations.
2. Determine if an existing pharmaceutical and or other technique can rescue some or all of the phenotype in an SRS mouse model by reducing spermidine and/or resulting toxins in circulation and target tissues.
3. Develop an inexpensive and convenient assay for SMS activity.

These funds have been made available by Team SRS.