the orphan disease center

MILLION DOLLAR BIKE RIDE
RAISING MONEY FOR RARE DISEASE RESEARCH

Snyder-Robinson Syndrome (SRS) is a genetic condition caused by mutations in Spermine Synthase (SMS). SMS catalyzes the conversion of spermidine to spermine and the dysfunction of SMS results in altered elevated levels of spermidine and reduced levels of spermine in SRS. There is some evidence that SMS may have additional functions.
Clinical features of SRS include intellectual disability, seizures, developmental delay, kyphoscoliosis and osteoporosis with fractures in the absence of trauma, as well as defects in other organ systems. There is a wide range of severity among individuals with SRS. There is some evidence to suggest possible immune suppression and/or overactivation is present in some patients with SRS. Mouse models with alterations in SMS are available for research studies through The Jackson Laboratory.

Research focus area: One $74,691 grant is available for SRS. There is interest in new studies focused on understanding the pathophysiology or mechanisms by which mutations in SMS cause SRS including how they may affect the immune systems. Applications addressing treatment options are welcomed. These funds have been made available by Team SRS.