What Is Schaaf-Yang Syndrome?

Basic SYS Facts

Schaaf-Yang syndrome (SYS) is a rare genetic disorder caused by a disruption of the *MAGEL2* gene. It affects males and females equally, as well as all races and ethnicities. Symptoms of SYS often include:

- Hypotonia
- Developmental delay
- Intellectual disability
- Sleep apnea
- Feeding difficulties
- Autism spectrum disorder
- Joint contractures

Schaaf-Yang syndrome shares many clinical features with another rare disorder: Prader-Willi syndrome, which occurs when the *MAGEL2* gene, along with several others, are missing or inactivated. Common features include hypotonia, feeding difficulties during infancy, global developmental delay/intellectual disability, and sleep apnea. However, patients with Schaaf-Yang syndrome have a relatively higher prevalence of autism spectrum disorder, and almost all of them manifest joint contractures, typically affecting the finger joints, but in more severe cases also involving knees and elbows.

Hope for the Future

Currently, there is no cure for SYS. The lives of people with SYS can be improved with early diagnosis and careful management of symptoms, but more effective therapies are needed. Research is being done around the world to better understand what causes SYS and how to improve the quality of life of those affected

Schaaf-Yang syndrome is generally not detected during prenatal testing, so families of children born with SYS are usually surprised by the diagnosis. Most parents are eager for answers about the immediate and the long-term effects of SYS and the prospects for successful treatment. While connecting with others dealing with SYS offers hope for families in the present, the research being conducted into SYS treatments offers them hope for the future.

FOUNDATION FOR PRADER-WILLI RESEARCH

The Foundation for Prader-Willi Research (FPWR) is a nonprofit organization made up of parents, families, and loved ones of people with Prader-Willi and Schaaf-Yang syndromes. We work with researchers and professionals to ***eliminate the challenges*** *of these syndromes through the advancement of research.*

To learn more, get involved, or make a donation, please visit us at www.fpwr.org