Putting A Spotlight On...

Smith Magenis Syndrome

What is Smith Magenis Syndrome?

- Smith-Magenis syndrome (SMS) is a complex developmental disorder that affects multiple organ systems of the body. The disorder is characterized by a pattern of abnormalities that are present at birth (congenital) as well as behavioral and cognitive problems. It is a rare condition that occurs in 1 in 15,000-25,000 people.

What are the signs/symptoms?

- Symptoms include:
  - Distinctive facial features
  - Skeletal malformations
  - Varying degrees of intellectual disability
  - Speech and motor delays
  - Sleep disturbances
  - Self-injurious or attention-seeking behaviors

What is Smith Magenis Syndrome a result of?

- Smith Magenis Syndrome is a result of missing genes on chromosome 17, or by changes in the RAI1 gene on chromosome 17. This genetic change happens around the time of conception.

What are the complications of Smith Magenis Syndrome?

- ear problems – for example, chronic ear infections or hearing loss
- dental problems
- seizures
- heart and kidney defects
- retinal detachment
- chronic constipation

References:

- https://rarediseases.org/rare-diseases/smith-magenis-syndrome/
- raisingchildren.net.au