Schinzel-Giedion Syndrome Foundation

Information for families

What is Schinzel-Giedion Syndrome (SGS)?

SGS is an ultra-rare neurodevelopmental genetic disorder that affects many body systems, including the brain.

It was first described by Dr. Schinzel *(Medical Genetics)* and Dr. Giedion *(Pediatric Radiology)* in 1978.

The genetic cause was identified in 2010.

It was originally diagnosed based on a child's physical appearance {e.g. distinctive facial features} and symptoms but now genetic testing is commonly used.

There are less than 150 children worldwide diagnosed with SGS; however, the true frequency is unknown.

Nymeria

Elisa

What causes SGS?

SGS is caused by a change or variant in the DNA sequence of the SETBP1 gene, also called a mutation or pathogenic variant.

The job of a gene is to create a protein to fulfill a certain role in the body.

The SETBP1 gene is important for normal development of the embryonic brain.

The SGS mutations affect the ability of the SETBP1 protein to break down by disrupting a region of the protein called the degron.

This results in SETBP1 accumulation in the cells which causes toxicity and abnormal development and function of the brain and potentially other organs.

"Connecting with other families caring for a child with SGS, and being able to ask questions and share my hopes and fears, has been such a comfort and support for me."

SGS Parent, United Kingdom.

What are the symptoms of SGS, and which medical providers should follow my child?

Neurologist (brain, spinal cord) **Neurodevelopmental delay, seizures.**

Pulmonologist (lungs) **Mucus build-up, breathing problems, pneumonia.**

Gastroenterologist (stomach and GI tract) **Constipation, reflux, feeding difficulties.**

Opthalmologist (eyes) **Vision problems** *(cortical visual impairment).*

Audiologist (hearing) **Hearing problems.**

Urologist (bladder) & **Nephrologist** (kidneys) **Kidney swelling** *(hydronephrosis),* **recurrent urinary tract infections.**

Consult a developmental/complex care pediatrician to follow your child and receive appropriate referrals to specialists.

Other notable symptoms and medical specialists your child may need to see:

Physical, Occupational, Speech & Respiratory Therapists – Low muscle tone

Sleep specialist – Abnormal sleep

Oncologist – Increased risk for certain cancers

Orthopedic surgeon – Club feet and scoliosis

Felicity

Visit our website
www.sgsfoundation.org

Join our
Patient Registry

Join our
Private Facebook Group

Email us
contact@sgsfoundation.org

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Can I have other children with SGS (recurrence risk)?

SGS is caused by a de novo variant, meaning it is not inherited from either parent but develops due to a spontaneous change in an egg or sperm.

The likelihood of having another child with SGS is very low. This is something to further discuss with your genetic counselor/ geneticist.

Theo

Where can I find out more?

Private SGS Family Facebook Group
Connect with other SGS families who can provide answers to your questions, and offer support and friendship **here**

Family Conferences
Attend to learn about new SGS research and share your experiences with other families: Interpreters provided for non-English speakers (advertised on Family Facebook group.)

SGS Patient Registry
Complete health and development surveys to help doctors and researchers better understand SGS (available in 8 languages) **here** or **email SGS Foundation** to schedule a Zoom meeting.

Research Initiatives
Volunteer to participate in research to find treatments for SGS (advertised on Family Facebook group).

Visit the SGS Foundation Website **here**

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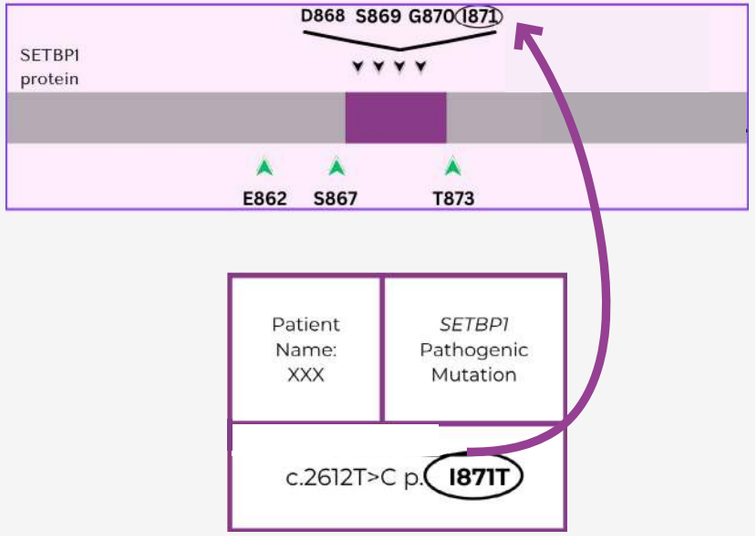
Which SETBP1 mutations/variants are associated with SGS?

Please refer to your child's genetic testing report to identify the specific mutation.
This can be located at the top of the report, where it names the SETBP1 gene and the p. (protein change) and c. (DNA change)

Classical SGS: caused by variants within the hotspot region of the SETBP1 gene which code for the degron (purple box with indicated black arrows).

Atypical SGS: caused by variants near the hotspot, not within it (green arrows); symptoms tend to be milder than with Classical SGS.



Example genetic report: SETBP1 mutation I871T has been identified, which is one of the mutations that causes Classical SGS.

Sadie

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