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Schinzel-Giedion syndrome

Fact sheet

What is Schinzel-Giedion syndrome?

Schinzel-Giedion syndrome (SGS) is a rare genetic condition which causes neurodegeneration.[1] SGS is an autosomal dominant genetic condition. Autosomal dominant means that an individual needs a variant in one copy of the gene. In the case of SGS, individuals have a gain-of-fuction variant in the *SETBP1* gene. This gain-of-function variant causes too much of the SETBP1 protein to be produced.[2]
Loss-of-function variants in the *SETBP1* gene cause SETBP1 haploinsufficency disorder.[3,4]

Currently, there are no available treatments available for SGS. Most children with SGS pass away in the first decade of life.[1]

What are the associated health and medical conditions seen in Schinzel-Giedion syndrome?[1,2,5]

- Epilepsy: individuals experience seizures which are difficult to control with medication.
- Vision and hearing: individuals with SGS often have severe visual and hearing impairments.
- Motor disorders: many individuals have limited fine and gross motor skills, requiring assistance to move around (e.g., a wheelchair).
- Feeding difficulties: individuals often requiring enteral feeding (e.g., a G-tube).
- Intellectual disability: individuals usually have severe to profound intellectual disability and experience neurodegeneration, which may be referred to as childhood dementia.[6,7]
- Urogenital abnormalities: kidney and genital abnormalities are also seen in children with SGS.
- Respiratory tract issues and infections (e.g., pneumonias).
- Recurrent infections (e.g., urinary tract infections).
- Some individuals with SGS have cancer.

What is communication like for children with Schinzel-Giedion syndrome?

Children with SGS do not learn to speak, and typically use behaviours like facial expression, crying, and eye-contact to communicate, which are interpreted by communication partners like their parents. Regardless of an individual's age, language skills are similar to typically developing children younger than 3 months of age.

In relation to early language skills, some parents of children with SGS report that their child can perform the following skills:[5]

- Look when they hear a voice
- Look when you wave

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- Look when someone calls their name
- Make happy sounds
- Make sounds to get attention
- Make more than three different speech sounds
- Cry or fuss when they are uncomfortable

A few parents of children with SGS report that their child can: [5]

- Respond to the tone of someone else's voice
- Understand three basic gestures
- Follow simple directions
- Babble
- Make sounds or gestures to continue or stop an activity

How do communication skills change overtime in children with Schinzel-Giedion syndrome?

SGS causes neurodegeneration, but it is not yet known how communication skills change overtime in children with SGS.[6] Some parents of children with SGS report loss of communication skills, including losing the ability to smile, hum and vocalise. For most children with SGS, parents report fluctuating communication skills depending on seizure frequency and severity, and presence of other health concerns.[5]

What are social skills like for children with Schinzel-Giedion syndrome?

Socialisation skills typically include looking at another persons face and smiling. Children with SGS can express emotions, and often recognise familiar people, particularly by their voice. Most children respond when another person acts playful and show interest in objects around them.[5]

How can speech pathologists/therapists support children with Schinzel-Giedion syndrome?

There is no research on speech and language interventions that are specifically designed for individuals with SGS. At present, an individualised approach should be taken to assessment and management to ensure therapies are tailored to and optimised for each child.

Due to multiple, co-occurring medical features associated with SGS, it is important that these needs are considered when providing therapy. Likewise, vision and hearing impairment should be considered when communicating with individuals with SGS. For example, using tangible symbols and touch to support communication. Some children with SGS learn to use switches to trigger a cause and effect.

To support families as communication changes overtime, caregivers and support people around the child should be provided with communication partner training.[8] Communication partner training is frequently used in adult dementias, and may include strategies such as attending to non-verbal communication (e.g., facial expressions).

Likewise, environmental supports should be considered to support an individual's understanding (receptive language), reduce disorientation, and support social connection with others.[9] Environmental supports should also consider vision and hearing impairments as core features of SGS. For instance, using a combination of visual, auditory and tactile cues to communicate changes to routine, or reducing background noise.

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Assessment/evaluation

Important domains for a speech pathology assessment include:

- Feeding and swallowing abilities
- Augmentative and alternative communication (AAC), e.g., communication aids
- Assessment of the interaction style of a child with SGS and their communication partners
- Assessment of the appropriate environmental supports and practical communication needs of the child and their support people (e.g., parents, support workers)
- Working with other professionals, such as occupational and physiotherapists, to assess motor skills for access to augmentative and alternative communication (AAC), e.g., using switches.

The types of assessment tools used will vary depending on the child's individual profile and developmental age. The goal of assessment will be to understand the nature of an individual's support needs, then make recommendations for appropriate therapies. Intervention should be tailored to an individual's communication support needs and consider the progressive nature of the disease.

Further information and support:

- For more information on communication in SGS see the research paper or plain language summary
- More information on AAC: AAC Fact Sheet
- The Schinzel-Giedion Syndrome Foundation - a charity / non-profit patient advocacy organisation: https://sgsfoundation.org/
- Closed Family Support Group on Facebook: https://www.facebook.com/groups/46580394354

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