

Schinzel-Giedion syndrome: Communication, feeding and motor skills in 16 individuals

Key terms

Schinzel-Giedion syndrome, *SETBP1*, speech, language, communication

What this research is about



Communication impairments are associated with changes in the *SETBP1* gene.

People with *SETBP1*-haploinsufficiency disorder do not have enough *SETBP1* protein. These people have mild to moderate communication difficulties.

People with Schinzel-Giedion syndrome have too much of the *SETBP1* protein. Communication has been studied in *SETBP1*-haploinsufficiency disorder but not Schinzel-Giedion syndrome.

We wanted to understand communication, feeding and motor skills in Schinzel-Giedion syndrome. This research helps us to better understand Schinzel-Giedion syndrome.

What the researchers did



- 16 children with Schinzel-Giedion syndrome took part.
- Parents completed online surveys and video call assessed health and development, feeding, adaptive behaviour and communication.

What the researchers found



- Most people with Schinzel-Giedion syndrome have communication skills that are similar to children less than 3 months old.
- Some people with Schinzel-Giedion syndrome cry and make happy sounds.
- Some people with Schinzel-Giedion syndrome can respond to things around them, like hearing their name and waving.
- A few people with Schinzel-Giedion syndrome

have relatively stronger communication, feeding and motor skills than other people with Schinzel-Giedion syndrome.

- Several parents of people with Schinzel-Giedion syndrome report a loss of skills overtime

What this means for people with Schinzel-Giedion syndrome and their families



- Clinicians should be aware of the communication, motor and feeding skills typical of people with Schinzel-Giedion syndrome
- Tailored therapies are important to support communication, feeding and motor skills
- Intervention should adapt to the vision and hearing impairments that many people with SGS have
- Communication interventions should include communication partner training and environment adaptations

Learn more here: Morison, L.D., Summerfield, N., Bradley, D., van Bon, B.W., & Morgan, A.T. (2025). Schinzel-Giedion syndrome: communication, feeding and motor skills in 16 individuals. *Neurogenetics* 26, 64, 1-13. <https://doi.org/10.1007/s10048-025-00846-3>