

# A pocket Guide to understanding SATB2-Associated syndrome for health professionals

## SATB2 Associated Syndrome (Glass Syndrome, 2Q33.13) is an extremely rare genetic developmental disorder

### What are the causes of SATB2 Associated Syndrome?

SATB2-Associated Syndrome (SAS) is associated with mutations to the 2q33.1 gene, such as:

- Mutations (code misspellings)
- Deletions (code pieces missing)
- Duplications (extra code pieces).

SAS is associated with the mutation of the 2q33.1 gene that is associated with head, palate and brain development. As a result of this mutation people diagnosed with SAS experience craniofacial abnormalities and behavioural and motor developmental delay.



### SATB2 stands for



- S**evere speech anomalies
- A**bnormalities of the palate
- T**eeth anomalies
- B**ehavioural issues with or without bone or brain anomalies
- 2** Age of onset before 2 years of age

### Signs and symptoms of SAS?

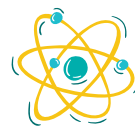
- Cleft palate and other craniofacial abnormalities
- Speech and feeding difficulties
- Dental abnormalities
- Gross and fine motor delay
- Hyperactivity
- Emotional regulation difficulties
- Epilepsy and seizures
- Continence
- Sleep issues
- Brain and bone abnormalities
- Growth delays
- Intellectual disability
- Gut health issues



### What are the associated disorders of SATB2 Associated Syndrome?

- Autism Spectrum Disorder (ASD)
- Obsessive Compulsive Disorder (OCD)
- Hyperactive behaviours similarly found in Attention Deficit Hyperactivity Disorder (ADHD)
- Cerebral Palsy (CP)

### How can you help individuals and families living with SAS?



Engage and contribute to SAS research through your field of expertise



Assist families with navigating through complex NDIS processes and provide sufficient evidence for NDIS funding applications



Empathise and make them feel heard as these families have very limited resources and support



Raise awareness on SAS and advocate for SAS communities

### Common treatment services associated with SAS

#### Medical Practitioners

General Practitioners  
Paediatricians  
Cardiologists  
Geneticists  
Dentists



#### Allied Health Professionals

Speech Therapists  
Psychologists  
Occupational Therapists  
Physiotherapists  
Genetic Counsellors  
Social Workers



#### Others

Case Manager  
Support Coordinator  
Behavioural Therapists  
Special Educators



### For additional resources



Contact SATB2 Connect or visit their website at [satb2.org.au](https://satb2.org.au)

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