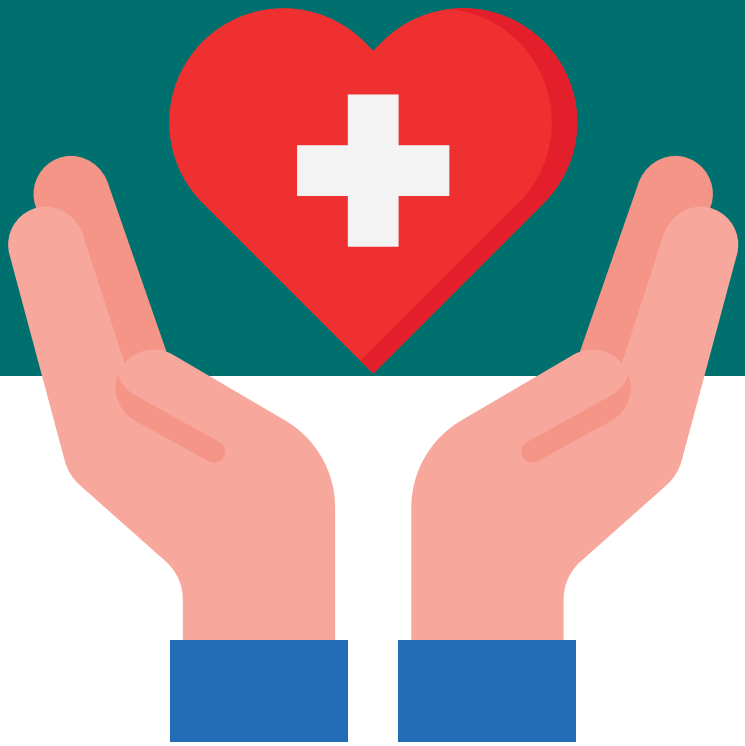
A guide for SATB2 ASSOCIATED SYNDROME

For Health professionals



We would greatly appreciate it if you could scan this QR code and provide us with feedback on the information packs!

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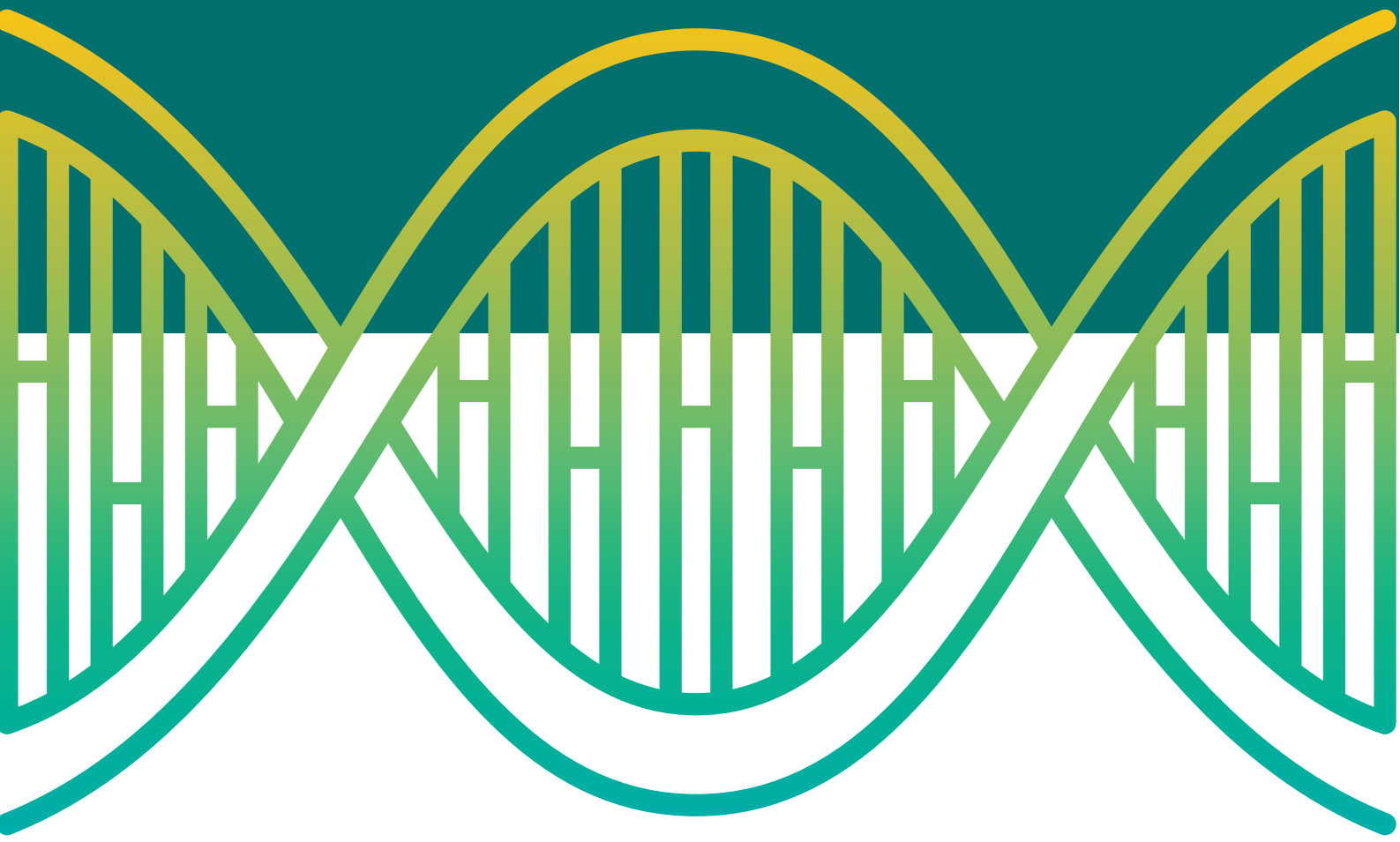
This project was completed by Aaron D'Hary, Fiorina Rayen, Han Xu & Pui Ying (Nicole) Yung as part of the requirements of the OCCP5239 Community Fieldwork Project Placement. It was supervised by Dalal D Baumgartner (SATB2 Connect) and supported by academic instructors at The University of Sydney.

19 May 2022

WHAT IS SATB2-ASSOCIATED SYNDROME ?



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what is satb2-associated syndrome (sas)?

SATB2 Associated Syndrome is a rare genetic condition with only 25 individuals diagnosed in Australia. It is also known as Glass Syndrome or 2q33.1 mutation.

Signs & Symptoms

- Cleft palate and other craniofacial abnormalities
- Feeding difficulties
- Speech difficulties (absent or delayed)
- Dental abnormalities, including large teeth
- Gross and fine motor delay
- Hyperactivity
- Emotional regulation difficulties
- Epilepsy and seizures
- Continence
- Sleep issues
- Brain and bone abnormalities (low bone density)
- Growth delays
- Intellectual disability
- Gut health issues

Summary of the Most Common Clinical Findings in 76 Individuals with SATB2 Associated Syndrome*

Finding	% of Affected Individuals [1]
Developmental delay / intellectual disability	100%
Speech delay	95%
Craniofacial dysmorphism	89%
Dental anomalies	72%
Behavioral issues	55%
Cleft palate	50%
Abnormal brain MRI	49%
Micrognathia	42%
Hypotonia	42%
Feeding difficulties	39%
Growth restriction	34%
Skeletal anomalies	32%

*Information accurate as of 2017

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what is satb2-associated syndrome?

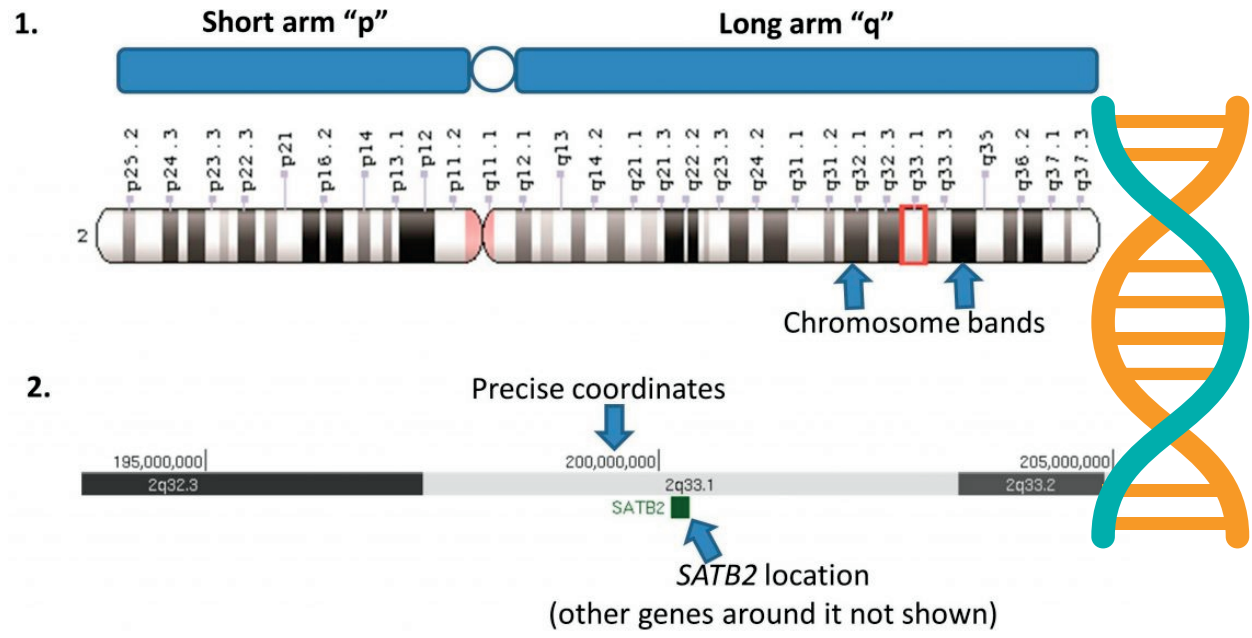
Causes

SATB2 Associated Syndrome occurs when there are changes to the SATB2 gene, such as:

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

SATB2 Associated Syndrome 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.

It is very unlikely another child will also have SATB2 Associated Syndrome.



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