

What is your role in this journey?



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.

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What is your role in this journey?

Importance of your role

Based on research from the Australian Paediatric Surveillance Unit, people living with rare disorders face significant challenges when navigating the healthcare system, such as:

- Diagnostic delays
- Lack of available treatment
- Difficulty finding the right health service

It is important to recognise the perspectives of families with members that have a rare disease such as SATB2 Associated Syndrome.

Many of these families feel isolated, under-supported and often face economic hardship due to the rarity of the condition. With this rarity comes an imbalance in support and access to services needed. As a result, it is important to recognise the role you have in these families' journeys as they continue to support their family members and children. Within this role, health professionals from various professions help lay the foundation for families, through assessments and therapy to develop NDIS plans and access necessary services that would be scarcely available given the rarity of their genetic disorder.

What is your role in this journey?

Medical Health Professionals

Geneticist

Individuals with SATB2 need your expertise in genetic testing and your understanding of test results as parents may have been bounced around the healthcare system. So, getting a formal diagnosis may provide a clear diagnosis and some certainty for families and individuals with SATB2, especially since it is a rare disease.

Neurologist

Individuals with SATB2 need MRI and EEG scans because seizures are a common symptom of SATB2. Families and parents may require further explanation related to behaviours reminiscent in ADHD, ASD and OCD.

Psychiatrist

Individuals with SATB2 may require behavioural assessments as these individuals tend to exhibit social difficulty, hyperactivity and emotional regulation difficulties. Neuropsychological evaluations may be necessary given that learning and intellectual disabilities are common for people with SATB2.

What is your role in this journey?

Medical Health Professionals

Paediatrician

Individuals with SATB2 may be assessed and referred to developmental paediatricians because they tend to exhibit developmental impairments related to sleep, hyperactivity language and learning.

Paediatric Ophthalmologist

Individuals with SATB2 need visual acuity assessments and visual function exams which will assist with Individualised Education Plans (IEP), and for assistance in informing materials necessary for social and daily living.

What is your role in this journey?

Allied Health Professionals

Genetic Counsellor

Families and individuals with SATB2 need your expertise in navigating the genetic condition, from analysis to diagnosis to living life with this rare disease, because of the barriers families may have faced with the healthcare system and the uncertainty associated with being diagnosed with a rare disease.

Psychologist

Individuals with SATB2 may need behaviour support assessments which informs support plans and cognitive and behavioural therapy. Furthermore assessments from psychologists are important in informing NDIS plans.

Occupational Therapist

Individuals with SATB2 require functional assessments to inform physical, cognitive, sensory, communication capacities and ADLs. These assessments inform NDIS reports which can promote their access to assistive technology and home modifications.

Physiotherapist

Individuals with SATB2 require your support to develop and improve their physical and motor skills, as well as your guidance on the use of mobility aids and equipment. These interventions may be similar to your patients with cerebral palsy and ASD.

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Allied Health Professionals

Speech Therapist

Individuals with SATB2 need your expertise in swallowing, drinking and communication difficulty assessments. AAC and speech therapy are important as communication difficulties are common in people with SATB2.

Dietician

Individuals with SATB2 may need your advice on improved and balanced nutrition. This is because individuals with SATB2 may have sensitivities towards certain foods, which may be similar to the needs of your clients with ASD.

Social Worker

Families and individuals with SATB2 require your support with case management and service coordination, counselling and coping with adjustment to a disability, and accessing various methods of financial assistance.

what is your role in this journey?

multidisciplinary team

craniofacial (cleft) team

Individuals with SATB2 have palatal abnormalities, including cleft palatal and high arched palate, as well as dental issues such as large teeth, so they require the team's expertise and comprehensive management of all craniofacial abnormalities and associated conditions.

other professionals

special educator

Individuals with SATB2 need your expertise in developing programs that address the individual's learning needs in order to help with their socialisation and play skills, independence relating to self-care and emotional skills, communication development, literacy and numeracy, and thinking skills. Their needs may be similar to your clients with ASD, ADHD and intellectual disabilities.

behavioural therapist

Individuals with SATB2 need your expertise in examining challenging behaviours that fall outside of the age-appropriate range and reduce behaviours of concern.

Support worker

Individuals with SATB2 require a range of support, including indoor personal care, education, therapy, social interactions and community access and participation.

