TIPS WHEN WORKING WITH SATB2 FAMILIES

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1 MAKE THEM FEEL HEARD

Due to the low prevalence of SAS, the resulting lack of knowledge of healthcare professionals has been shown to be a major issue in the highly specialised treatment process of rare diseases. In such cases, families often are the true experts on the disorder. Thus, it is important to ensure that the families' and parents' perspectives are heard and understood by putting yourself in their shoes.

Families interact with their child regularly and can provide valuable observations and insight regarding their child's behaviours and needs.

Families may have gone through difficult experiences. They may have already spent large amounts of money in attempt to get answers, been continuously referred to various health professionals over a prolonged period of time without receiving adequate support and guidance, and may have received much conflicting advice on their next steps, and as a result, have many questions.

Additionally, after receiving the diagnosis, they may feel isolated because of the limited resources available and may be struggling to cope with living with a rare disease like SAS.

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2 communicate

As parents and carers are experts on their own children, it is important to show parents and carers that you value their experiences, ideas, and take their concerns seriously.

We understand that with a rare disease like SATB2, there is so much uncertainty and it is difficult to provide evidence-based advice, but it is important to work with the family to provide the best possible support.

Based on a survey conducted with families, the items below are reflections of the kind of support families want and expect from the current healthcare system.

- Support through grief and next steps
- Going the extra mile to educate themselves and finding alternatives / opportunites
- Willingness to network and connect with other professionals who knew more about this rare disorder
- Deeper curiosity and desire to help
- Recognising that there may be issues that aren't explicit / visible that are apparent to the family

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3 ACCESS USEFUL RESOURCES FOR RARE DISEASES

1

Table 2. Useful resources for clinicians on the epidemiology, clinical features, diagnosis and management of rare diseases

Organisation name	Services provided*
Orphanet – the portal for rare diseases and orphan drugs (European-based organisation) www.orpha.net/consor/cgi-bin/index.php	• Information regarding rare diseases (searchable by disease or by symptom) • Directory of clinical trials and research studies • Directory of centres of expertise
Centre for Genetics Education www.genetics.edu.au/ www.genetics.edu.au/Professionals/genetics-in-general-practice	• Educational resources for clinicians, including disease fact sheets, guidelines on genetic testing • A specific resource for general practitioners • List of genetic services • List of genetic counselling services
OMIM – Online Mendelian Inheritance in Man: An online catalogue of human genes and genetic disorders www.omim.org	• Comprehensive database of genetic disorders providing information regarding: • disease characteristics • diagnosis • prevalence • genotype-phenotype correlations • associated genetic anomalies
National Institutes of Health, Office for Rare Diseases https://rarediseases.info.nih.gov	• Information regarding rare diseases • Genetic educational tools and fact sheets • Information regarding genetic testing and treatments

Australian Paediatric Surveillance Unit www.apsu.org.au
 	
www.inopsu.com
	• Information regarding selected rare childhood diseases including:
	  ◦ Australian incidence estimates
	  ◦ diagnosis
	  ◦ disease features and study summaries
	  ◦ national platform for the study of rare childhood diseases; international collaboration via the International Network of Paediatric Surveillance Units
	• Research on health services utility and health service costs and psychosocial and economic impacts on families living with rare disease

*Examples of services/information only. Please go to the organisation's website to find out more.

[1] Information accurate as of 2015

Retrieved from Australian Family Physician. (2015, September). Rare diseases are a 'common' problem for clinicians. https://www.racgp.org.au/afp/2015/september/rare-diseases-are-a-%E2%80%98common%E2%80%99-problem-for-clinicians/

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4 advocate, advocate, advocate. contributing on a national scale

- Increase knowledge and raise awareness of the epidemiology and impact of SAS on families, health professionals, and the SAS community in Australia.
- Improve healthcare for people with rare diseases through participation in initiatives that allow better access to diagnostic tests, new treatments and specialised services.
- Promote scientific and social research on rare diseases, such as SAS, through engagement in the development of national and international multidisciplinary research partnerships.
- If you have had experience working with SAS, provide educational resources and networking opportunities for health professionals to allow them to better identify and manage SAS.
- Support families affected by rare diseases such as SAS by facilitating the development of integrated peer support networks.
- Promote the development and funding of a national umbrella organisation addressing all aspects of rare diseases or organisations that specifically cater to SAS (e.g. SATB2 Gene Foundation Australia).

Retrieved from Australian Family Physician. (2015, September). Rare diseases are a 'common' problem for clinicians. https://www.racgp.org.au/afp/2015/september/rare-diseases-are-a-%E2%80%98common%E2%80%99-problem-for-clinicians/

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References

Alliance, C. P. (n.d.). All services and programs. Cerebral Palsy Alliance. https://cerebralpalsy.org.au/services/all-programs-and-services/

Australasian Cleft Lip & Palate Association Inc.(n.d.). General Cleft Lip & Palate Information.
https://www.cleft.org.au/members/education/

Australian Family Physician. (2015, September). Rare diseases are a ‘common’ problem for clinicians. https://www.racgp.org.au/afp/2015/september/rare-diseases-are-a-%E2%80%98common%E2%80%99-problem-for-clinicians/

Budych, K., Helms, T. M., & Schultz, C. (2012). How do patients with rare diseases experience the medical encounter? Exploring role behavior and its impact on patient–physician interaction. Health policy, 105(2-3), 154-164.

Elliott, E. J., & Zurynski, Y. A. (2015). Rare diseases are a 'common' problem for clinicians. Australian family physician, 44(9), 630-633.

SATB2 Gene Foundation. (2020, December 7). Medical information.
https://satb2gene.org/resources/medical-information/

SATB2 Gene Foundation. (2018). SATB2-Associated Syndrome.
https://satb2gene.org/resources/satb2-information-sheet/

Support professionals for people with autism. (2019, November 8). The Spectrum. https://thespectrum.org.au/autism-support-services/professionals/

Zarate YA, Kaylor J, Fish J. SATB2-Associated Syndrome. 2017 Oct 12. In: Adam MP, Ardinger HH, Pagon RA, et al., editors. GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2022. Available from: https://www.ncbi.nlm.nih.gov/books/NBK458647/

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