FACTS ABOUT HEALTH CONDITIONS

CAUSED BY CHANGES IN THE *POGZ* GENE

This fact sheet contains information about the possible impact of a change (variant) in the *POGZ* gene on your child and family. You can talk about the information in this fact sheet with your paediatrician or GP (family doctor). The links in the fact sheet may help you move forward with family life beyond receiving this rare diagnosis.

This fact sheet relates to health conditions that are due to small variants in the genetic code of the *POGZ* gene. These changes were identified by a genomic (DNA) test. It does not provide information about conditions caused by chromosome deletions or duplications that involve the *POGZ* gene.

Key points

- Health conditions caused by changes (variants) in the *POGZ* gene are called *POGZ*-related conditions. They may also be referred to as White Sutton syndrome
- Most children will have some form of intellectual disability and many have autism spectrum disorders. It is common for children to have a small head size
- Variants in the *POGZ* gene that cause health problems may be inherited from a parent or may be a new (*'de novo'*) change in a child. This means that future children may also have this variant. Genetic counselling before any further pregnancies is recommended
- Supportive management is available
- You and your family are not alone in adjusting to life with the diagnosis of a change in the *POGZ* gene. Support is available from a number of different organisations and services

Other names this condition may be referred to as

- White Sutton syndrome
- *POGZ*-related condition

When a rare condition has been diagnosed

For some families, receiving a genetic diagnosis is a relief. Others may feel overwhelmed and sad. It is very common to have a mixture of thoughts and feelings about the news, and your hopes and expectations for the future may shift and change over time.

While experiences may be shared, individuals and families can respond in different ways and have different information and support needs. Many parents describe an ongoing process of adjusting to a different focus and finding ways to celebrate their child's gains made in their own way and time. It is very important to remember that the diagnosis is only one of many things that make your child unique.

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About the *POGZ* gene

The *POGZ* (Pogo transposable element derived with ZNF domain) **gene** contains instructions that tell our body how to grow, develop and function. The *POGZ* gene is thought to be important in controlling how genetic instructions are used by a person's cells and is particularly important in the brain.

The *POGZ* gene is found on chromosome 1. Usually *POGZ*-related conditions are caused by a single spelling variation in the gene, which means the message is not read or received properly.

Individuals with a *POGZ*-related condition may have a number of different features, which can vary even within the one family. Whether the features of the condition are mild or more severe often depends on the particular type of gene change (**variant**) and its location within the *POGZ* gene (known as the genotype).

In most individuals with *POGZ*-related conditions, the change occurred when the baby was conceived (a new or *'de novo'* variant), and is not seen in the parents. In some of the milder forms, the variant is passed to the child from a parent.

POGZ-related conditions are **genetic conditions**. This means that the condition was not caused by anything the mother or father did before the baby was conceived, during pregnancy or birth, or after the baby was born. *POGZ*-related conditions are rare, affecting less than 100 families worldwide.

What could a change in the *POGZ* gene mean for my child?

A change in the *POGZ* gene can affect children in different ways. Some are more severely affected than others, and there may be a range of signs and symptoms even in children with the same genetic variant.

Children with *POGZ*-related conditions may be slower to reach their physical milestones (sitting, crawling, walking, running) and have problems with learning. They may also be slower than other children to learn to speak or understand what others are saying (language delay).

Many children grow slowly, and more often are slow to grow taller rather than to put on weight. Some have autism spectrum disorder and/or are hyperactive. A small head size (microcephaly) is often seen. Many children with *POGZ*-related conditions have similar facial features.

At this point in time, it is not possible to reverse or directly repair this gene change. It is also not possible to accurately predict the level of care your child will require through to adulthood. Your child's individual needs and strengths will become more obvious over time, which will help with planning for the future.

Your child's development may be helped through early use of therapy services such as physiotherapy and treating symptoms if/when they arise. It is likely that many different health professionals will be involved in caring for your child. Your paediatrician or GP will arrange referrals to other health professionals as needed and help with applications for service funding through the **National Disability Insurance Scheme (NDIS)**.

Good communication with the health professionals caring for your child is important to establish common goals, trust and shared responsibility. We encourage you to ask questions and express your concerns as the primary carer for your child.

Management recommendations

As many health or developmental problems are not immediately obvious, your child will need to be checked by their paediatrician at diagnosis and then seen at least once every year or more often if needed. The list on the next page includes many of the common problems, but others may arise. If you have any concerns about your child's health, please speak with your family doctor (GP) or paediatrician.

Possible health problems (% of children affected)	Management
Developmental delay/intellectual disability (>90%), including low muscle tone (hypotonia) (80%)	• At least yearly checks by GP/paediatrician • Early intervention, including occupational therapy and physiotherapy
Speech delay (>90%)/absence of speech (40%)	• Speech therapy to help with language development • Speech aids
Flexible (hyperflexible) joints (60%)	• Physiotherapy and/or occupational therapy may be helpful
Autism spectrum disorder (40–70%)	• Diagnosis and management by paediatrician as appropriate
Attention-deficit/hyperactivity disorder (ADD/ADHD) (30%)	• Diagnosis and management by paediatrician as appropriate
Gastrointestinal reflux/feeding difficulties (10–40%)	• Investigation for swallowing difficulties or reflux where appropriate • Review and management by paediatrician
Constipation (30%)	• Constipation can be treated with a high fibre diet, adequate fluid intake and/or laxatives
Slow growth (30%)	• At least yearly review by GP/paediatrician • Consider screening for hypothyroidism and growth hormone deficiency
Brain structural malformations/ developmental differences of the brain (10–20%)	• Brain MRI if clinically indicated. Most structural problems do not require any specific treatment
Small head (microcephaly) (10–40%)	• Regular review of height, weight and head size (head circumference). No treatment is needed
Epilepsy (20%) **Many seizure types are possible**	• Standard investigations and treatments, including EEG and anti-epileptic medications • Consultation with a paediatric neurologist if seizures are difficult to control
Visual (eyesight) problems (50%)	• Yearly eye review by ophthalmologist • A squint (strabismus) may require patching or glasses • Long- or near-sightedness (refractive error) may require glasses
Hearing problems (up to 60%)	• Newborn screening hearing test to identify sensorineural hearing loss (caused by damage to the nerves in the ear). Treatment with hearing aids and/or cochlear implant • Additional hearing assessment if there are concerns about hearing loss, as conductive hearing loss (when problems with the ear prevent sound from passing into it) may occur over time

Resources, support and connecting with others

You may find it helpful to connect with other people who have personal experience of day-to-day life with a child who has a *POGZ*-related condition. You can make these connections through:

- Social media (e.g. closed Facebook groups)
- Umbrella groups (e.g. **Genetic Alliance Australia** and **Rare Voices Australia**)
- Condition-specific groups (e.g. the **White Sutton Syndrome Foundation**)
- Groups for individuals with common symptoms that may have many different causes (e.g. intellectual disability, hearing loss, autism).

Many organisations (e.g. **Carers NSW** and **Reframing Disability**) can also offer general advice and support in caring for a family member with long-term needs.

It is important to know that you are not alone on this journey

More information about *POGZ*-related conditions

You can find further information about *POGZ*-related conditions by following the links below.

- MedlinePlus: **POGZ gene**
- MedlinePlus: **White-Sutton syndrome**

For more information about genetic conditions and to find your local Clinical Genetics services, visit the **NSW Centre for Genetics Education**.

Family planning

Genetic conditions are sometimes passed from a parent to their child. Even if the same variant was not found in one of the parents, it is still possible to have another child with the same condition. The chance of this happening is 1–2%.

If you are thinking about having more children, it is recommended that you talk with your local **Clinical Genetics service**. Some people may choose to have **genetic testing** before or during a pregnancy. Specialised health professionals such as **genetic counsellors** can advise you on your options.

You can also speak with your GP about options for **reproductive genetic carrier screening**. When planning a family, it is best to explore your options before becoming pregnant.

Research, registries and clinical trials

Some people with rare conditions are able to participate in **research**, which may be of benefit to your child. This may investigate how a particular variant causes health problems or it may be a clinical trial testing new treatments. Sharing information about your child's signs and symptoms through registries such as **FaceMatch** and **IAMRARE** can help build further knowledge about this condition. The White Sutton Syndrome Foundation have an online **Patient-Powered Registry**, which is collecting information from parents.

Information about current clinical trials can be found by searching the international databases **ClinicalTrials.gov** or **EudraCT**.

To print more copies of this fact sheet and access links to the underlined topics, go to **www.genetics.edu.au** and search for 'POGZ'.

This fact sheet should not replace a consultation with a specialist healthcare professional.