FACTS ABOUT HEALTH CONDITIONS
CAUSED BY CHANGES IN THE *SETBP1* GENE

This fact sheet contains information about the possible impact of a change (variant) in the *SETBP1* 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 this 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 changes in the genetic code of the *SETBP1* 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 *SETBP1* gene. This fact sheet does not relate to cancers that are due to acquired changes in the *SETBP1* gene (changes that occur after a person is born) or to specific changes in the *SETBP1* gene that cause Schinzel-Giedion syndrome.

Key points

- A change in the *SETBP1* gene will usually cause intellectual disability, which may vary from mild to severe
- Changes (variants) in the *SETBP1* 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 change. Genetic counselling before any further pregnancies is recommended
- A separate condition, called Schinzel-Giedion syndrome (SGS), is caused by specific changes in the *SETBP1* gene. The information in this fact sheet is not relevant for individuals with Schinzel-Giedion syndrome
- Supportive management is available
- You and your family are not alone in adjusting to life with the diagnosis of a change in the *SETBP1* gene. Support is available from a number of different organisations and services

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 *SETBP1* gene

Genes contain instructions that tell our body how to grow, develop and function. *SETBP1* is a **gene** that plays an important role in the normal development of the body. Changes in the *SETBP1* gene can affect health and development, including how other genes work. The *SETBP1* gene makes a protein that is very active during brain development before birth. This protein helps nerve cells grow and divide and move to their proper location in the brain.

Most changes in the *SETBP1* gene cause less protein to be produced, which affects brain development. This usually causes intellectual disability without other major problems (non-syndromic intellectual disability).

In most individuals with *SETBP1*-related conditions, the change in the gene occurred when the baby was conceived (a new or *'de novo'* change), and is not seen in the parents. In some of the milder forms, the variant is passed to the child by a parent.

SETBP1-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. *SETBP1*-related conditions are very rare, affecting less than one in a million people.

What could a change in the *SETBP1* gene mean for my child?

A change in the *SETBP1* gene can affect children in different ways. Some are more severely affected than others. There may be a range of signs and symptoms even in children with the same genetic variant.

Children with *SETBP1*-related conditions may be slower to reach their physical milestones (sitting, crawling, walking, running), and have problems with learning. Some children have problems with their hearing and vision, and may be slow to speak. A small number of children experience seizures (epilepsy). Many children with *SETBP1*-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 every year or more often if needed. The list below 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.

Most common health problems	Management recommendations
Developmental delay/ intellectual disability	• Early intervention including speech therapy, occupational therapy and physiotherapy • Special education programs
Hearing loss	• Newborn screening hearing test to identify hearing loss that is caused by damage to the nerves in the ear (sensorineural hearing loss). 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
Visual (eyesight) problems	• Initial review by ophthalmologist for assessment of vision and squint (strabismus) • Yearly eye review by ophthalmologist • A squint (strabismus) may require patching or glasses
Seizures (epilepsy)	• Standard investigations and treatments, including EEG and anti-seizure medications.
Autism spectrum disorder/ challenging behaviours	• Diagnosis and management by paediatrician as appropriate • Allied health support may include psychologist, behavioural therapist, occupational therapist, speech therapist

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 *SETBP1*-related condition. You can make these connections through:

- Social media (e.g. closed Facebook groups such as **SETBP1 Friends and Family Facebook support group**)
- Umbrella groups (e.g. **Genetic Alliance Australia**)
- 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 **Kindred**) 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 *SETBP1*-related conditions

You can find further information about *SETBP1*-related conditions by following the links below.

- MedlinePlus: **SETBP1 disorder**
- National Organisation for Rare Disorders (NORD): **SETBP1 haploinsufficiency syndrome**
- **SETBP1 Society**

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.

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

To print more copies of this fact sheet and access links to the underlined topics, go to **https://www.genetics.edu.au/SitePages/SETBP1.aspx**

The recommendations in this fact sheet were current at the time it was written. This fact sheet should not replace a consultation with a specialist healthcare professional.

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