FACTS ABOUT HEALTH CONDITIONS

CAUSED BY CHANGES IN THE *AUTS2* GENE

This fact sheet contains information about the possible impact of a change (variant) in the *AUTS2* 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.

Key points

- A change in the *AUTS2* gene will usually cause developmental delay, intellectual disability, small head size and short stature
- Changes (variants) in the *AUTS2* gene that cause health problems usually occur as a new *('de novo')* change in a child, or may be inherited from a parent. 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 *AUTS2* gene. Support is available from a number of different organisations and services

Other names this condition or gene may be referred to as:

- *AUTS2* syndrome
- Activator of transcription and developmental regulator
- Autism susceptibility candidate 2
- *KIAA0442* gene

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 *AUTS2* gene

Genes contain instructions that tell our body how to grow, develop and function. *AUTS2* (autism susceptibility candidate 2) is a **gene** that is important for the normal development of brain cells (neurons) and their correct positioning within the brain. Changes **(variants)** in the *AUTS2* gene can affect health and development.

The *AUTS2* gene is found on chromosome 7. Usually *AUTS2*-related conditions are caused by a single spelling variation in the gene, which means the message is not read or received properly. In some individuals, a section of the chromosome that includes the *AUTS2* gene (called 7q11.2) is missing. This is known as a deletion.

In around 8 out of 10 individuals (80%) with *AUTS2*-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 one parent in an **autosomal dominant** manner.

AUTS2-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. *AUTS2*-related conditions are very rare, with less than 100 individuals reported so far worldwide.

What could a change in the *AUTS2* gene mean for my child?

A change in the *AUTS2* 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.

Common features include learning difficulties, autism spectrum disorder and intellectual disability, which varies in severity from borderline to severe. Feeding difficulties, short stature and small head size (microcephaly) are also common. Many children have changes in muscle tone including low tone (hypotonia) or high tone (hypertonia). A small number of children have seizures. Children with *AUTS2*-related conditions are often described as being happy, easy-going and sociable.

Many children with *AUTS2*-related conditions have similar facial features. These include highly arched eyebrows, widely spaced eyes with droopy eyelids (ptosis), small mouth and a small jaw (micrognathia).

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 each 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 unusually low or high muscle tone (hypotonia or hypertonia) (>50%)	• Early intervention, including speech therapy, occupational therapy and physiotherapy • Consider a formal developmental assessment before starting school or by school counsellor for school age children • At least yearly checks by GP/paediatrician
Autism spectrum disorder or autistic/obsessive traits (70%)	• Diagnosis and management by paediatrician as appropriate
Attention-deficit/hyperactivity disorder (ADD/ADHD) (up to 50%)	• Diagnosis and management by paediatrician as appropriate
Feeding difficulties (>80%)	• Investigation for swallowing difficulties or reflux where appropriate
Short stature (50–60%)	• Measurement of the child's height at least once each year
Abnormal heart structure (cardiac malformations) (<10%)	• Non-urgent consultation with a cardiologist including a baseline ultrasound (echocardiogram) of the heart • Typically minor problems only, including patent foramen ovale or atrial septal defect
Genital structural differences (up to 40%)	• If the testicles cannot be felt (palpated), a surgical consultation is required
Minor structural brain malformations (20–30%)	• Brain imaging is not usually needed, and no treatment is required
Seizures (epilepsy) (<20%)	• Investigations and management as directed by paediatrician
Small head size (microcephaly) (60%)	• Regular review of height, weight and head size (circumference). No treatment is needed

Further information about the *AUTS2* gene for healthcare professionals is available at **Orphanet**.

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 an *AUTS2*-related condition. You can make these connections through:

- Social media (e.g. closed Facebook groups such as **7q11.22 Chromosomal Deletions & AUTS2 Syndrome**)
- Umbrella groups (e.g. **Genetic Alliance Australia** and **Rare Voices Australia**)
- Condition-specific groups
- 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

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. If a parent has the same gene change, there is a 50% chance (1 in 2) that future children will also be affected. 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. In this case, the chance of having another child with an *AUTS2*-related condition 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 gene 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 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 'AUTS2'.

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