

# FACTS ABOUT HEALTH CONDITIONS CAUSED BY CHANGES IN THE *KAT6A* GENE

**This fact sheet contains information about the possible impact of a change (variant) in the *KAT6A* 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 *KAT6A* 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 *KAT6A* gene. This fact sheet does not relate to cancers that are due to acquired variants in the *KAT6A* gene (changes that occur after a person is born).



## Key points

- A change in the *KAT6A* gene will often cause intellectual disability, which may vary from mild to severe. Children may also have problems with talking and feeding
- Most changes in the *KAT6A* gene that cause health problems are not passed down (inherited) from either parent (called '*de novo*' changes)
- Rarely, the change in the *KAT6A* gene is 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 *KAT6A* gene. Support is available from a number of different organisations and services

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

- Lysine (K) acetyltransferase 6 A
- MOZ
- MYST3



## 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.



## About the *KAT6A* gene

Genes contain instructions that tell the body how to grow, develop and function. *KAT6A* ('cat-six-ay') is a **gene** that has a wide range of functions across the body. A change (**variant**) in *KAT6A* can affect a number of body systems, and can alter how other genes work. Each individual may have a unique variant in the *KAT6A* gene, and so each individual may be affected differently.

The *KAT6A* gene is found on chromosome 8. Usually *KAT6A*-related conditions are caused by a single spelling variation in the gene, which means the message is not read or received properly. In most individuals with *KAT6A*-related conditions, the change in the gene occurred when the baby was conceived (a new or '*de novo*' variant), and is not seen in the parents. In rare cases, the variant is passed from a parent to their child.

*KAT6A*-related disorders 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. *KAT6A*-related conditions are very rare. Less than 400 individuals are known to have the condition.



## What could a change in the *KAT6A* gene mean for my child?

A change in the *KAT6A* 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 a *KAT6A*-related condition may be slower than other children to reach early milestones (i.e. walking and talking). In particular, children with *KAT6A*-related conditions often have significant problems with their speech and language. Some individuals may learn to speak clearly even though they took longer than usual to learn as children. Others never learn to speak and may communicate through other means (e.g. body language, sign language).

It is common for children to have feeding problems and digestive (gastrointestinal) issues (e.g. constipation and/or reflux). Many children have problems with their eyesight, sleep disturbances, or differences in the structure of their heart. Individuals with *KAT6A*-related conditions often share some common facial features and many have ears that are positioned or shaped differently.

At this point in time, it is not possible to reverse or directly repair most gene changes that occur. 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 6 to 12 months 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
<b>Developmental delay/intellectual disability (&gt;90%), including low muscle tone (hypotonia) in infancy (~75%), speech and language problems (&gt;90%), and difficulty/delayed walking (common)</b>	<ul style="list-style-type: none"> <li>• At least yearly checks by GP/paediatrician</li> <li>• Early intervention, including speech therapy, occupational therapy and physiotherapy</li> <li>• Early intervention and special education programs</li> <li>• Consider a formal developmental assessment before starting school or by school counsellor for school age children</li> </ul>
<b>Autism spectrum disorder (~25%)</b>	<ul style="list-style-type: none"> <li>• Diagnosis and management by paediatrician as appropriate</li> </ul>
<b>Challenging behaviours (~40%)</b>	<ul style="list-style-type: none"> <li>• Diagnosis and management by paediatrician as appropriate</li> </ul>
<b>Feeding or swallowing difficulties (~70%)</b>	<ul style="list-style-type: none"> <li>• Investigation for swallowing difficulties or reflux where appropriate</li> <li>• Children with severe feeding problems may need to be referred to a paediatric gastroenterologist and may need a feeding (nasogastric or gastrostomy) tube</li> </ul>
<b>Reflux (~60%)</b>	<ul style="list-style-type: none"> <li>• Review and management by paediatrician</li> </ul>
<b>Constipation (~50%)</b>	<ul style="list-style-type: none"> <li>• Constipation can be managed with a high fibre diet, adequate fluid intake and/or laxatives</li> <li>• Intestinal malrotation (where the intestines do not move into the correct place before the baby is born) has been reported in a small number of children</li> </ul>
<b>Dental problems (~15–20%)</b>	<ul style="list-style-type: none"> <li>• At least yearly review by a dentist</li> </ul>
<b>Abnormal heart structure (cardiac malformations) (~50%)</b>	<ul style="list-style-type: none"> <li>• Consultation with a cardiologist including a baseline ultrasound (echocardiogram) of the heart. Intervention may be required (e.g. open heart surgery or cardiac catheterisation)</li> <li>• Yearly review by paediatrician/cardiologist</li> </ul>
<b>Abnormal kidney, genital or urinary structure/development (less common)</b>	<ul style="list-style-type: none"> <li>• Baseline ultrasound of the renal tract</li> <li>• If a testicle cannot be felt (palpated), a surgical consultation is required</li> </ul>
<b>Small head size (microcephaly) (~25–35%) or fusion of the skull bones too early (craniosynostosis) (~10–15%)</b>	<ul style="list-style-type: none"> <li>• Regular review of height, weight and head size (head circumference).</li> <li>• Craniosynostosis may require surgery</li> </ul>
<b>Brain structural malformations/developmental differences of brain (less common)</b>	<ul style="list-style-type: none"> <li>• Brain MRI if clinically indicated. Most structural problems do not require any specific treatment</li> </ul>
<b>Seizures (epilepsy) (~10%)</b>	<ul style="list-style-type: none"> <li>• Investigations and management as directed by paediatrician</li> </ul>
<b>Visual (eyesight) problems/vision loss (~50–60%)</b>	<ul style="list-style-type: none"> <li>• Initial review by ophthalmologist for assessment of vision and squint (strabismus)</li> <li>• Yearly eye review by ophthalmologist</li> <li>• A squint (strabismus) may require patching or glasses</li> <li>• Long- or near-sightedness (refractive error) may require glasses</li> </ul>
<b>Hearing loss/perforated eardrums (&lt;10%)</b>	<ul style="list-style-type: none"> <li>• Hearing assessment at birth and repeat as needed</li> </ul>
<b>Frequent infections (cause is currently unknown) (~50%)</b>	<ul style="list-style-type: none"> <li>• Respiratory infections, urinary tract infections, and ear infections should be monitored by a paediatrician</li> </ul>
<b>Disorders of sleep (~35%)</b>	<ul style="list-style-type: none"> <li>• Review by paediatrician. Consider consultation with sleep physician and/or sleep studies for diagnosis and ongoing management</li> </ul>
<b>Hormone (endocrine) problems (~10–15%) including hypothyroidism (~5–10%), pituitary abnormalities (~5%) and diabetes (~2.5%)</b>	<ul style="list-style-type: none"> <li>• Hypothyroidism should be tested for if suspected clinically</li> <li>• Type 2 diabetes should be screened for if suspected clinically</li> </ul>
<b>Floppy windpipe (larynx/trachea) (less common)</b>	<ul style="list-style-type: none"> <li>• May require review and/or investigation by a doctor specialising in treating ear, nose and throat problems (ENT) or a respiratory paediatrician</li> </ul>



## 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 *KAT6A*-related condition. You can make these connections through:

- Social media (e.g. closed Facebook groups: [KAT6A Foundation Facebook group](#))
- Umbrella groups (e.g. [Genetic Alliance Australia](#) and [Rare Voices Australia](#))
- Condition-specific groups (e.g. [KAT6A 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 *KAT6A*-related conditions

You can find further information about *KAT6A*-related conditions by following the links below.

- Unique: [KAT6A Syndrome](#)
- National Organization for Rare Disorders (NORD): [KAT6A 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.

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](http://www.genetics.edu.au) and search for 'KAT6A'.

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