FACTS ABOUT HEALTH CONDITIONS
CAUSED BY CHANGES IN THE *KMT2A* GENE

This fact sheet contains information about the possible impact of a change (variant) in the *KMT2A* 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 *KMT2A* 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 *KMT2A* gene. This fact sheet does not relate to cancers that are due to acquired variants in the *KMT2A* gene (changes that occur after a person is born).

Key points

- A change in the *KMT2A* gene can cause a health condition called Wiedemann-Steiner syndrome
- People with this condition usually have mild-to-moderate intellectual disability, are of short stature and sometimes have a small head (microcephaly)
- Changes (variants) in the *KMT2A* gene that cause health problems may be inherited from one 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 *KMT2A* gene. Support is available from a number of different organisations and services

Other names this condition may be referred to as

- Wiedemann-Steiner syndrome

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 *KMT2A* gene

Genes contain instructions that tell our body how to grow, develop and function. *KMT2A* is a **gene** that regulates other genes. Changes in the *KMT2A* gene can affect a number of developmental pathways in our bodies.

The *KMT2A* gene is found on chromosome 11. Usually *KMT2A*-related conditions are caused by a single spelling variation in the gene (called a **variant**), which means the message is not read or received properly.

In most cases of Weidemann-Steiner syndrome, the change in the gene occurs when the baby is conceived (a new or *'de novo'* change), and is not seen in the parents. In a small number of families the change was inherited from a less affected parent in an **autosomal dominant** manner.

Weidemann-Steiner syndrome is a **genetic condition**. 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. Changes in the *KMT2A* gene are one of the more commonly-identified causes of intellectual disability. The number of children with Wiedemann-Steiner syndrome worldwide is thought to be a few hundred, although it is likely there are more.

What could a change in the *KMT2A* gene mean for my child?

A change in the *KMT2A* 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 or within the same family.

Children with Weidemann-Steiner syndrome usually have a mild-to-moderate intellectual disability. They may be born small and feed poorly. Some children have problems with gastrointestinal reflux when they are babies. This can mean that they gain weight and grow more slowly than other children.

Some children are inflexible with their daily routine and have repetitive behaviours. They may be diagnosed with autism spectrum disorder (ASD).

In some children, Weidemann-Steiner Syndrome may be suspected because of an increase in body hair over the back or on the elbows. Children with Weidemann-Steiner syndrome may have distinctive facial features including widely spaced down sloping eyes, bushy eyebrows and a large nasal tip. This is most noticeable when they are babies or a young child. Some children have minor changes in the skeleton, kidney and heart.

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. They should see their paediatricians every 3–6 months while they are infants and then once every year through childhood, 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%)	• Early intervention, including speech therapy, occupational therapy and physiotherapy • Formal developmental assessment before starting school or by school counsellor for school age children • At least yearly checks by GP/paediatrician
Challenging behaviours (30-40%)	• Diagnosis and management by paediatrician as appropriate • Refer to psychologist and/or psychiatrist for even mildly challenging behaviour
Minor structural brain changes (60%) e.g. thinning of the nerves that connect the left and right sides of the brain (corpus callosum), or slower than usual development of the coating (myelin) around nerve cells (delayed myelination)	• Brain imaging is not usually needed
Seizures (20%)	• Standard investigations and treatments, including EEG and anti-epileptic medications
Feeding difficulties (60-70%)	• Investigation for swallowing difficulties or reflux where appropriate • Children with severe feeding problems may need to be referred to a paediatric gastroenterologist and may need a feeding (nasogastric or gastrostomy) tube
Constipation (60%)	• Constipation can be treated with a high fibre diet, adequate fluid intake and/or laxatives
Differences in the skeleton (50%)	• Around 15% of individuals are born with two or more bones of the upper spine joined together (Klippel Feil anomaly). These individuals should be watched for signs indicating that the nerve fibres of the spinal cord are affected (long tract signs) • A flexion and extension X-ray of the cervical spine, and possibly also an MRI, may be needed
Short stature (>50%)	• At least yearly review by GP/paediatrician • Consider investigation for growth hormone deficiencies if height is less than the third centile • There is some evidence that children respond to growth hormone even if there is no sign of growth hormone deficiency
Abnormal heart structure (cardiac malformations) (30%)	• Typically minor problems only • Non-urgent consultation with a cardiologist including a baseline ultrasound of the heart (echocardiogram)
Abnormal heart rhythm (<5%)	• Heart rhythm assessment (electrocardiogram [ECG])
Abnormal genital, kidney or urinary structure (genitourinary malformations) (50%)	• Baseline ultrasound of the kidneys
Visual (eyesight) problems (50%)	• Initial review by ophthalmologist for assessment of vision and squint (strabismus); yearly eye review by ophthalmologist • A squint (strabismus) may require patching or glasses • Long or near sightedness (refractive error) may require glasses
Hormone (endocrine) problems (20%) e.g. reduced growth hormone	• Assessment of growth hormone levels, thyroid function and bone density
Early signs of puberty (30%) e.g. armpit and pubic hair or body odour before the age of 8 years in females and 9 years in boys (premature adrenarche)	• No treatment is needed

Detailed management recommendations for healthcare professionals can be found in the **GeneReviews article**.

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 *KMT2A*-related condition. You can make these connections through:

- Umbrella groups (e.g. **Genetic Alliance Australia** and **Rare Voices Australia**)
- Condition-specific groups (e.g. **Wiedemann-Steiner 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 **Belongside Families**) 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 Wiedemann-Steiner syndrome

You can find further information about Weideman-Steiner syndrome by following the links below:

- Genetic and Rare Disease Information Center: **Weidemann-Steiner syndrome**
- National Orgnization for Rare Disorders: **Wiedemann-Steiner Syndrome**
- Unique: **Wiedemann-Steiner 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. 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 a *KMT2A*-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 **IAMRARE** and the **Wiedemann-Steiner syndrome data collection program** can help build further knowledge about this condition.

Information about current clinical trials can be found by searching the **Australian Clinical Trials** database, 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 **www.genetics.edu.au** and search for 'KMT2A'.