FACTS ABOUT HEALTH CONDITIONS
CAUSED BY CHANGES IN THE *MECP2* GENE

This fact sheet contains information about the possible impact of a change (variant) in the *MECP2* 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 *MECP2* 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 *MECP2* gene.

Key points

- Changes in the *MECP2* gene cause Rett syndrome
- Rett syndrome is a rare condition that causes intellectual and physical disability
- As the *MECP2* gene is found on the X chromosome, girls are much more likely to have Rett syndrome than boys
- Some of the major signs of Rett syndrome include problems with speech and fine motor skills, and distinctive hand movements
- In most cases, the change in the *MECP2* gene occurs after the baby is conceived (a *'de novo'* change) and is not passed to the child from the parents
- Some changes (variants) in the *MECP2* gene 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 *MECP2* 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 *MECP2* gene

Genes contain instructions that tell our body how to grow, develop and function. *MECP2* (pronounced "meck-pea-two") is short for methyl CpG binding protein 2. The *MECP2* **gene** is responsible for controlling other genes. If the *MECP2* gene is not working properly, other genes may turn on or stay on at the wrong times. A child with Rett syndrome usually develops normally in the first few months of life, until the *MECP2* gene needs to play a bigger role in the child's development. At this time, the signs and symptoms of Rett syndrome start to be seen.

The *MECP2* gene is found on the X chromosome. Because females have two copies of the X chromosome and males have only one, X-linked conditions affect males and females differently. Most variants in the *MECP2* gene are so severe that males do not survive. Sometimes the males survive but are much more affected than females. Occasionally a family is identified with a variation in the *MECP2* gene that has a minor effect on the child. This causes variable intellectual disability in males, but does not affect their mothers. These children would not be described as having Rett syndrome.

In most individuals with Rett Syndrome, the change in the gene occurred when the baby was conceived (a new or *'de novo'* variant), and is not seen in the parents.

Rett 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. Rett syndrome is very rare, affecting around 1 in every 10,000–23,000 female babies (varying rates are seen in different countries).

What could a change in the *MECP2* gene mean for my child?

A change in the *MECP2* 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 Rett syndrome usually have a period of normal early development followed by a period where their development regresses (goes backwards). They develop unusual hand movements. Most have difficulty in carrying out motor movements such as walking, and also with speech and communication.

Seizures are common in children with Rett syndrome. Irregular breathing patterns, sleeping problems and feeding difficulties are also common.

At this point in time, it is not possible to reverse or directly repair variants of the *MECP2* gene. 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, occupational and speech therapies, 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.

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Management recommendations

As many health or developmental problems are not obvious straight away, your child will need to be checked by their paediatrician at diagnosis and then seen yearly, 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.

Possible health problems (% of children affected)	Management
Developmental delay/intellectual disability (100%)	• Early intervention, including speech therapy, occupational therapy and physiotherapy • Special education programs
Difficulty walking (50%), abnormal walking (100%)	• Physiotherapy and/or occupational therapy
Problems communicating (100%)	• Speech therapy to help with language development • **Eye gaze and augmentative communication approaches**
Teeth grinding	• Yearly review by dentist
Feeding difficulties	• Speech therapy • Consultation with a dietician • Children with severe feeding problems may need a feeding (nasogastric or gastrostomy) tube
Constipation (common)	• Constipation can be treated with a high fibre diet, adequate fluid intake and/or laxatives • Discussion with GP and/or paediatrician if required
Curved spine (scoliosis) (75%)	• Initial review by paediatrician • Referral to spinal surgeon if progressing
Abnormal heart rhythm (long QT syndrome)	• Yearly heart rhythm checks (electrocardiogram) • Avoidance of certain medications
Seizures (60–90%)	• Standard investigations and treatments, including EEG and anti-epileptic medications • Consultation with paediatric neurologist if seizures are difficult to control
Sleeping problems (70–80%)	• Review by paediatrician • Consider referral to a sleep physician if severe symptoms

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 Rett syndrome. You can make these connections through:

- Social media (e.g. closed Facebook groups such as the **Rett Syndrome Association of Australia**)
- **Rett Syndrome Association of Australasia** website
- Umbrella groups (e.g. **Genetic Alliance Australia** and **Rare Voices 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. You can find Information about specialised Rett syndrome clinics from the **Rett Syndrome Association of Australia**.

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

More information about Rett Syndrome

You can find further information about Rett Syndrome by following the links below.

- **Rett Syndrome.org** website
- MedlinePlus: **Rett Syndrome** or ***MECP2*-related conditions**
- National Organization for Rare Disorders (NORD): **Rett 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 can be passed from a parent to their child. Since the *MECP2* gene is on the X chromosome, conditions related to changes in the *MECP2* gene are known as **X-linked** conditions.

If the change in the *MECP2* gene was not passed to the child by their mother, the chance of having another child with the same condition is very low (1–2%). If the change in the *MECP2* gene was passed to the child by their mother, there is a 1 in 2 chance that the mother will have another affected child.

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 discuss your options with you.

You can also speak with your GP about options for **reproductive genetic carrier screening**, a genetic test that looks for changes in many different genes.

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 **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/MECP2.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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