FACTS ABOUT HEALTH CONDITIONS
CAUSED BY CHANGES IN THE *TCF20* GENE

This fact sheet contains information about the possible impact of a change (variant) in the *TCF20* 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 changes (variants) in the genetic code of the *TCF20* gene. These changes were identified by a genomic (DNA) test. This fact sheet does not provide information about conditions caused by chromosome deletions or duplications that involve the *TCF20* gene.

Key points

- A change (variant) in the *TCF20* gene will usually cause developmental delay/intellectual disability, autism spectrum disorder (ASD), and/or attention deficit/hyperactivity disorder (ADD/ADHD). Some children will have minor differences in facial appearance, and a small number will develop epilepsy
- Variants in the *TCF20* gene that cause health problems are usually new (*'de novo'*) changes in a child. Rarely, the variant 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 *TCF20* gene. Support is available from a number of different organisations and services

Other names this condition may be referred to as:

- Developmental delay with variable intellectual impairment and behavioural abnormalities
- *TCF20*-related disorders

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 *TCF20* gene

Genes contain instructions that tell our body how to grow, develop and function. *TCF20* is a **gene** that controls how other genes are read and interpreted (transcribed), and affects a wide range of processes in the body. Changes (**variants**) in the *TCF20* gene affect development and learning.

The *TCF20* gene is found on chromosome 22. Usually *TCF20*-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 *TCF20* gene is missing. This is called a deletion.

In most individuals with *TCF20*-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 altered gene is passed to the child from a parent.

TCF20-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. *TCF20*-related conditions are very rare, and no information about the number of individuals with the condition is available at the moment.

What could a change in the *TCF20* gene mean for my child?

A change in the *TCF20* gene can affect children in different ways. Some are more severely affected than others, and there may be a range of signs and symptoms even in children with the same genetic variant. This variability in signs and symptoms is greater than is seen in most other genetic conditions.

The most common features of *TCF20*-related conditions are delayed development, mild-to-moderate intellectual disability, ASD, ADD/ADHD, other behavioural issues, and subtle differences in facial appearance.

At this point in time, it is not possible to reverse or 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 low muscle tone (hypotonia) (~60%) and speech delay (60%)	• 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 • Special education programs
High muscle tone (hypertonia) (5%) or muscle weakness and tightness (~5%)	• Orthopaedics/rehabilitation/physiotherapy/occupational therapy, including stretching to help avoid fixed tightness of the muscles (contractures) and falls
Unsteadiness (ataxia), abnormal walking (gait disturbance), balance issues, poor coordination, tremor (~50%)	• Physiotherapy may be helpful
Autism spectrum disorder (ASD) (~65%)	• Diagnosis and management by paediatrician as appropriate
Attention deficit/hyperactivity disorder (ADD/ADHD) (~60%)	• Diagnosis and management by paediatrician as appropriate
Challenging behaviours including anxiety (~20%), aggression (~10%), and food seeking behaviour (~3%)	• Diagnosis and management by paediatrician as appropriate
Feeding difficulties (~10%)	• Children with severe feeding problems may need to be referred to a paediatric gastroenterologist
Constipation (~30%)	• Constipation can be treated with a high fibre diet, adequate fluid intake and/or laxatives
Differences in the skeleton (~30%). This can include flat feet and funnel chest (pectus excavatum)	• Initial review by paediatrician • X-rays or other imaging may be helpful for diagnosis and management if skeletal differences are considered a possibility
Curved spine (scoliosis) (~15%)	• Usually mild and may develop over time • Yearly review by GP/paediatrician until the child has finished growing
Early (premature) fusion of the joints (sutures) of the skull (craniosynostosis) (~5%)	• Poor head growth or an unusually-shaped (asymmetric) skull should be investigated by a paediatrician or craniofacial surgeon
Large stature, large head size, or overweight (~5%)	• Management by paediatrician as appropriate
Brain structural malformations/ developmental differences of brain (~25%)	• Brain MRI if clinically indicated • Most structural problems do not require any specific treatment
Seizures (epilepsy) (~10%)	• Standard investigations and treatments, including EEG and anti-epileptic medications. Review by a paediatric neurologist if seizures are difficult to control
Disorders of sleep (~25%)	• Review by paediatrician; consider referral to sleep physician if severe symptoms
Visual (eyesight) problems (~10%). Problems can include near-sightedness (myopia), squint (strabismus), and a change in the shape of the cornea in the eye (keratoconus)	• Initial review by ophthalmologist for assessment of vision and squint (strabismus)

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 *TCF20*-related condition or a similar genetic condition. You can make these connections through:

- Social media (e.g. closed Facebook groups such as the **TCF20: Simons Searchlight Community)**
- 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

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 about 1%.

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**.

You may like to discuss this research with your child's healthcare team.

To print more copies of this fact sheet and access links to the underlined topics, go to **www.genetics.edu.au** and search for 'TCF20'.

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