This fact sheet describes Autism Spectrum Disorder (ASD) and includes a discussion of the symptoms, causes and available testing.

**In summary**

- Autism spectrum disorder (ASD) is a single diagnosis that has replaced the previous subdivisions of 'classical' autism, Asperger syndrome, and pervasive developmental delay not otherwise specified (PDD-N.O.S)
- ASD is ranked level 1,2 or 3, reflecting a spectrum from mild to more severe symptoms
- ASD is mainly characterised by:
  - ◊ deficits in social communication and
  - ◊ fixated interests and repetitive behaviour.
- The cause of ASD is still currently unknown for the majority of individuals.

## WHAT IS AUTISTIM SPECTRUM DISORDER?

Around 1 in 200 Australians are affected by Autism Spectrum Disorder (ASD). Males are four times more likely to be affected by ASD, with 8 in 1,000 males being affected, compared to 2 in 1,000 females. The male to female ratio is even more pronounced for the milder end of the ASD spectrum, where males are seven times more likely to be diagnosed than females. In the last twenty years there has been a large increase in the number of children diagnosed; probably due to a broadening of the criteria for diagnosis and better diagnostic methods for ASD rather than a true increase in the number of people affected.

Most people with ASD have it as an isolated issue however there are several genetic conditions where ASD-like features may be present. These include:

- Fragile X syndrome
- Rett Syndrome
- Tuberous Sclerosis

## WHAT ARE THE CHARACTERISTICS OF ASD?

ASD is a lifelong, non-progressive neuro-developmental condition. Symptoms are usually apparent by the age of 30 months. Early diagnosis, therapeutic treatment and early intervention in childhood helps to improve outcomes.

There is no conclusive evidence to support that early childhood vaccinations play any role in the development of ASD.

Children with ASD can have a range of difficulties, particularly with social communication and repetitive behaviours.

They may:

- take longer to learn and understand language
- have difficulty understanding social norms
- appear disinterested in social interaction
- have repetitive behaviours
- be upset by changes in routine
- have sensory issues, such as being sensitive to sound or texture.

## WHAT CAUSES ASD?

Research suggests that both genes and the environment are important factors in the development of ASD, however in over 75% of cases, the cause is still unknown.

Identical twin studies suggest that, where one twin is diagnosed with ASD, there is a very high chance that the other twin will also be diagnosed with ASD, indicating a genetic predisposition to ASD. However, the other twin does not always develop the condition, which suggests that there may be environmental factors that contribute to the chance that ASD will develop.

Current research suggests that between 600-1200 genes may be involved in the development of ASD, but genetic causes are not always identified in people with ASD. For many individuals it may be that a number of different genetic variants contribute to the range and severity of their symptoms.

## HOW IS ASD INHERITED?

For the majority of cases of ASD, both genetic factors and environmental factors are involved in the cause of the condition. The combination of genetics and external factors is called multifactorial inheritance (See figure 46.1).

Multifactorial inheritance refers to the pattern of inheritance of certain conditions due to a combination of both genetic and other factors that may include internal factors such as ageing, and exposure to external environmental factors such as diet, lifestyle, and exposure to chemicals or other toxins.

Multifactorial conditions do not always develop despite the presence of a genetic mutation which increases the person's risk. The reason for this **incomplete penetrance** of the condition is most likely due to the interaction between the information in the gene mutation with the information in other genes and with other 'environmental' factors.

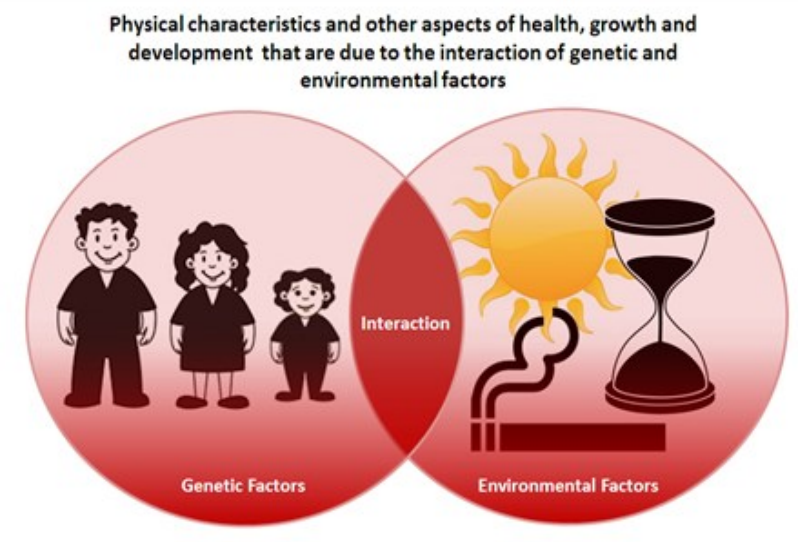


***Figure 46.1:*** *A diagrammatic representation of the interaction between genetic and environmental factors*

## GENETIC CONDITIONS WHERE AUTISTIC-LIKE FEATURES MAY BE PRESENT

As mentioned above, most people with ASD will have it as an isolated issue without any other features or symptoms. A small percentage of people however will have ASD due to a single gene mutation and the features of ASD form part of a collection of features, or a syndrome.

These conditions are called monogenic neurodevelopemental conditions and may be characterised by a distinctive pattern of learning, behavioural or physical features.

Three of the more commonly diagnosed single-gene causes of ASD are Fragile X syndrome, Rett syndrome and Tuberous Sclerosis. A specialist genetic clinic can identify whether ASD is a part of one of these conditions or not.

## IS THERE ANY TESTING OR TREATMENT AVAILABLE FOR ASD?

If an individual is showing signs and symptoms of ASD, a developmental paediatrician will usually offer 'first line' testing to look for genetic causes including chromosomal microarray and fragile X testing. Other testing may be offered depending on the child's specific symptoms.

For many individuals and their family, no genetic cause is identified and therefore it is difficult to predict the chance that another family member will be affected by ASD.

If the family is concerned about the cause of their child's ASD, or would like more information about the chance of recurrence in the family, a genetic counselling consultation may be recommended to provide current information, discuss genetic testing options and information about the chance of the condition affecting future children.

It is anticipated that more comprehensive genetic testing for individuals with ASD will be available in the future, for example screening a number of individuals' genes at one time using 'next generation' genetic testing.

Currently there is no cure for ASD but early diagnosis and therapeutic intervention in childhood helps to improve outcomes. The treatment is primarily educational with an emphasis on preventing and reducing difficult behaviours.