**Important points**

- The autism spectrum disorders (ASDs) represent a group of developmental conditions characterised by impairments in communication and social interactions, restricted, repetitive and stereotyped behaviour and anxiety and compulsions.
- ASDs include ‘classical’ autism, Asperger syndrome and pervasive developmental delay not otherwise specified (PDD – N.O.S).
- The vast majority (about 90% of cases) are of unknown cause. There is no conclusive evidence to support any role in the development of autism as a result of the measles, mumps and rubella vaccine, illness experienced by the mother during pregnancy and the child’s diet early in life.
- There are a number of indications to suggest that the interaction between the information in one or more faulty genes and unknown ‘environmental factors’ plays a major role in the development of autism.
- Research is continuing with the aim of identifying the gene(s) involved in autism and developing genetic tests:
  - Genetic testing is not available for ‘classical’ autism.
  - A number of different genetic conditions including fragile X syndrome and Rett syndrome are characterised by autistic-like features.
  - Genetic testing is available for these conditions.

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**What are autism spectrum disorders (ASDs)?**

The autism spectrum disorders (ASDs) represent a group of developmental conditions characterised by:

- Impairments in communication and social interactions.
- Restricted, repetitive and stereotyped behaviour.
- Anxiety and compulsions.

All of the conditions grouped under the ‘umbrella’ term of ASDs vary based on the symptom severity and development of language, cognition and social behaviour and include:

- ‘Classical’ autism.
- Asperger syndrome.
- Pervasive developmental delay not otherwise specified (PDD – N.O.S).

There are also a number of different genetic conditions where autistic-like features may be present. These include:

- Fragile X syndrome (see Genetics Fact Sheet 42).
- Rett Syndrome.

**What are the characteristics of autism?**

Autism is a lifelong, non-progressive neuro-developmental condition. In the ‘classical’ form of the condition:

- Symptoms are usually apparent before the age of thirty months.
- About 5-6 out of 10,000 Australians are affected. 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 autism rather than an increase in the number of people affected.
  - Males are 4 times more likely than females to be affected.
  - Early diagnosis and therapeutic treatment in childhood helps to improve outcomes.

The diagnosis of autism is based on criteria listed in the *Diagnostic and Statistical Manual of Mental Disorders (DSM IV)* and include, for example:

- A reluctance to engage with other people.
- Language delay or repetitive language.
- Repetitive movements (eg. hand flapping) and/or a need to regularly perform certain functions or rituals.
- Behavioural problems such as mood swings and tantrums.
- Sensory problems such as heightened sensitivity to light and sound.

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**What causes autism?**

The vast majority are of unknown cause.

There is no conclusive evidence to support any role in the development of autism of the following:

- The measles, mumps and rubella vaccine.
- Illness experienced by the mother during pregnancy.
- The child’s diet early in life.

There are a number of indications that the individual’s genetic make-up plays a major role in the development of autism. For example, it is more likely for the condition to affect both twins of an identical twin pair. When one twin is affected by ‘classical’ autism, there is about six chances in ten (60%) that the other identical twin will also have the condition.

In addition, as shown in Table 43.1 when one child in the family is affected, the risk for having further children with autism is, overall, about 4-5 chances in 100 (4.5%).

- If the first affected child is a girl, the risk is about 7 chances in 100 (7%).
- If two children are affected, more than 1 chance in 4 (25%).

**Genes and autism**

The cells of the body contain the genes or set of instructions for the cell to make all the necessary proteins (chemicals) for our bodies to grow and work normally (see Genetics Fact Sheet 1).

A gene that contains a variation in the information that stops it working properly is described as faulty. The variation that makes the gene faulty is called a *mutation*. The information contained in the faulty gene, and its product, is impaired (see Genetics Fact Sheets 4 & 5).

When, however, one twin of an identical twin pair has autism, the other twin does not always develop the condition, which suggests that there are likely to be ‘environmental factors’ that trigger the onset of symptoms. Without these triggers, the condition will not develop. It may be that there is a genetic (inherited) predisposition to develop autism, triggered by other unknown factors which may be the information in a number of different genes or external factors. See Genetics Fact Sheet 11 for more information about conditions due to environmental and genetic interactions.

The fact that autism affects males four times more often than females suggests that there may be genes on the X chromosome that can predispose to autism.
Intellectual features can be characterised by a pattern of unusual physical, behavioural and/or communicative skills. The term is also used when stereotyped like features? are at the more ‘functional’ end of the autism spectrum with average or above average intelligence and fewer language difficulties.

Other autism spectrum disorders (ASDs)

Two other developmental conditions thought to be on the same continuum (spectrum) as autism are Asperger syndrome and pervasive developmental delay – not otherwise specified (PDD – N.O.S.).

- Individuals with Asperger syndrome are at the more ‘functional’ end of the autism spectrum with average or above average intelligence and fewer language difficulties
- PDD – N.O.S (otherwise known as Autism Spectrum Disorder) is the term used to describe those children that do not meet the diagnostic criteria for conditions such as autism or Asperger syndrome but still have significant impairment in the development of social interaction or verbal and nonverbal communication skills. The term is also used when stereotyped behaviour, interests, and activities are present

It is thought that these conditions may be determined by the same set of genetic susceptibility factors as autism.

Genetic testing is still in the research phase.

Genetic conditions where autistic-like features may be present

Around 5% of all cases of ASDs are associated with specific genetic conditions.

Some of these conditions are caused by a change that has made just one of the 20,000 or so genes in the human genome, faulty. These conditions are described as syndromes because they are characterised by a pattern of unusual physical, behavioural and/or intellectual features.

**Fragile X syndrome**

Fragile X syndrome, its genetic basis and inheritance pattern in families are described in Genetics Fact Sheet 42.

The condition is characterised by intellectual problems which can vary from mild learning difficulties through to severe intellectual disability, emotional and behavioural problems. It usually affects males although females may show varying degrees of the condition.

The genetic basis of fragile X syndrome is complex but is due to having a faulty copy of *FMR-1* gene, which is located on the X chromosome.

- Males who have the faulty *FMR-1* gene will usually be affected as they do not have a partner back-up working copy available: boys only have one copy of the X chromosome and a Y chromosome
- Females have two copies of the X chromosome and so can be carriers of the faulty *FMR-1* gene: ie. they have a faulty copy of the gene on one X chromosome and a working gene copy on the other partner X chromosome. They may be unaffected or affected less severely
- Genetic testing is available

**Rett syndrome**

Rett syndrome is a rare neuro-developmental condition that usually affects females and rarely males and may include the following features:

- Normal development of infants and children until about 6 to 18 months of age, when they may begin to lose previously acquired skills such as purposeful hand movements and the ability to communicate
- Development of distinctive, uncontrolled hand movements, such as hand clapping, rubbing, or ‘wringing’
- Impaired control of voluntary movements required for coordination of walking
- Other autistic-like behaviours

The condition usually results from having a faulty copy of the gene called *MECP2* that is located on the X chromosome. The *MECP2* gene is thought to normally play an essential role in brain development.

- Genetic testing is available

**What can be done if a child has autism or autistic-like features?**

It is important to exclude a possible genetic syndromic cause in an individual showing signs and symptoms of autism. A genetic counselling consultation may be able to assist in diagnosis, provide current information, discuss genetic testing options and provide risk assessment for the condition affecting future children (see Genetics Fact Sheet 3).

Chromosomal microarray or molecular karyotype (see Genetics Fact Sheet 21) to determine very small chromosomal abnormalities has become the first-line genetic investigation for children with suspected or diagnosed ASD.

Currently there is no cure for autism but early diagnosis and therapeutic treatment in childhood helps to improve outcomes. Treatment is primarily educational with an emphasis on preventing the onset of difficult behaviours.

**Other Genetics Fact Sheets referred to in this Fact Sheet:** 1, 3, 4, 5, 9, 10, 11, 21, 42
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