Down syndrome
While some may need only a little support, while others may require a high level of support. However, most people growing up with Down syndrome today will be able to achieve and participate as one individual to another. People with Down syndrome generally need more support than most other people in order to achieve their potential – some will need very little support, while others may require a high level of support.

Everyone with Down syndrome will experience some delay in their development and some level of learning disability, but the extent and specific areas of delay vary from one individual to another. People with Down syndrome generally need more support than most other people in order to achieve their potential – some will need very little support, while others may require a high level of support. However, most people growing up with Down syndrome today will be able to achieve and participate as one individual to another.
valued members of their communities.

**Down syndrome and capabilities and potential**

Down syndrome affects, but does not determine, development and achievement in a person. People with Down syndrome demonstrate a wide range of capabilities. What happens after birth will be far more important in shaping the outlook for a person with Down syndrome than the occurrence of the extra chromosome at conception.

These days, children with Down syndrome attend childcare settings, pre-schools and primary and high schools alongside other children of their age. Adults with Down syndrome attend post-school training, participate in the workforce and lead full lives as productive and valued members of their communities. An increasing number are achieving independent living, with some level of support, within the community.

People with Down syndrome often have their abilities and potential underestimated. A person with this condition will achieve in many areas when offered the opportunity and encouraged to do so.

**Diagnosis of Down syndrome**

Down syndrome is usually recognisable at birth and confirmed by a blood test. A doctor can usually tell if a baby has Down syndrome from their physical appearance.

Prenatal tests that can help to detect Down syndrome in a fetus include:

- ultrasound scans
- maternal serum screening
- amniocentesis
- chorionic villi sampling.

**Genetic counselling and support regarding Down syndrome**

Genetic counselling is available to parents in all states of Australia and may be helpful if you are concerned that you may have a child with Down syndrome. Community support is also available for parents whose child has been diagnosed with Down syndrome.

**Where to get help**

- Your doctor
- Your local community health centre
- Maternal and child health nurse
- Down Syndrome Victoria Tel. 1300 658 873
- Genetic Support Network Victoria Tel. (03) 8341 6315
- Association for Children with a Disability Tel. (03) 9818 2000
- Better Start for Children with Disability Tel. 1800 242 636

**References**

- Down syndrome today, Down Syndrome Victoria. [More information here](#)
- Buckley, S, Living with Down syndrome, Down Syndrome Education International. [More information here](#)
- Trisomy 21 – Down Syndrome, Centre for Genetics Education. (pdf) [More information here](#)
- Changes to chromosome structure – translocations, Centre for Genetics Education. (pdf) [More information here](#)
- The Genetics File – Down syndrome, Murdoch Childrens Research Institute, Parkville Vic.
- Down syndrome, Mayo Clinic, Mayo Foundation for Medical Education and Research. [More information here](#)
Please note that we cannot answer personal medical queries. If you are looking for health or medical advice we recommend that you:

- talk to your doctor or pharmacist
- dial triple zero (000) in an emergency
- ring NURSE-ON-CALL Tel. 1300 60 60 24.

2/4 Your Comments

Tell us who you are  
Select an option

Enter your comments below (optional)

Next Submit Now Cancel

Send us your feedback

- Rate this website
- Your comments
- Questions
- Your details

3/4 Questions

What are you here to do? Looking for information on

Did you find what you were looking for?

- ☑ Yes
- ☐ No

Next Submit Now Cancel

Send us your feedback

Thank you. Your feedback has been successfully sent.

More information

Genes and genetics

The following content is displayed as Tabs. Once you have activated a link navigate to the end of the list to view its associated content. The activated link is defined as Active Tab

- A-Z of genetic conditions
- Genes and genetics explained
- Genetic testing

A-Z of genetic conditions

- Ambiguous genitalia
  The causes of ambiguous genitalia include genetic variations, hormonal imbalances and malformations of the fetal tissues that are supposed to evolve into genitals.
- Angelman syndrome
  The characteristic features of Angelman syndrome are not always obvious at birth, but develop during childhood.
• **Ankylosing spondylitis**
  Ankylosing spondylitis (AS) is a type of inflammatory arthritis that targets the joints of the spine.

• **Barrett's oesophagus**
  Symptoms of Barrett's oesophagus are similar to regular heartburn, which means many people don't seek treatment until their condition is quite advanced.

• **Bipolar disorder**
  Bipolar disorder is a type of psychosis, which means the person's perception of reality is altered. It is characterised by extreme mood swings.

• **Central nervous system birth defects**
  Folic acid taken before conception, and during at least the first four weeks of pregnancy, can prevent around seven out of 10 cases of neural tube defects.

• **Charcot-Marie-Tooth disease (CMT)**
  Charcot-Marie-Tooth disease is the most common inherited disorder affecting the peripheral nervous system.

• **Cleft palate and cleft lip**
  Most cleft palates and cleft lips can be repaired so that appearance and speech develop normally.

• **Congenital adrenal hyperplasia (CAH)**
  CAH is a rare genetic disorder, but it is well understood and treatment is readily available.

• **Creutzfeldt-Jakob disease (CJD)**
  Creutzfeldt-Jakob disease is characterised by physical deterioration of the brain, dementia and walking difficulties.

• **Cystic fibrosis (CF)**
  When a person has cystic fibrosis, their mucus glands secrete very thick sticky mucus that blocks the tiny air passages in the lungs and traps bacteria.

• **Digestive tract birth defects**
  Too much amniotic fluid surrounding the baby during pregnancy (polyhydramnios) may indicate the presence of defects of the digestive tract.

• **Down syndrome**
  With the support and opportunities available to them today, most people with Down syndrome are able to achieve and participate as valued members of their community.

• **Dwarfism**
  Dwarfism refers to a group of conditions characterised by shorter than normal skeletal growth.

• **Eczema (atopic dermatitis)**
  Eczema can vary in severity, and symptoms may flare up or subside from day to day.

• **Essential tremor**
  Essential tremor causes involuntary shaking or trembling of particular parts of the body, usually the head and hands, but it is not Parkinson's disease.

• **Fragile X syndrome**
  The facts about fragile X syndrome are complicated, and parents and family members are invited to ask their doctor to refer them to a genetics clinic.

• **Friedreich's ataxia**
  To the casual observer, a person with Friedreich ataxia may seem to be drunk.

• **Genetic factors and cholesterol**
  Familial hypercholesterolaemia is an inherited condition characterised by higher than normal levels of blood cholesterol.

• **Haemochromatosis**
  Haemochromatosis (iron overload disorder) tends to be under-diagnosed, partly because its symptoms are similar to those caused by a range of other illnesses.

• **Haemophilia**
  All children with severe haemophilia are given preventative treatment with infusions of blood products before they have a bleed.

• **Hair**
  Human hair grows one centimetre every month.

• **Hearing problems in children**
The earlier that hearing loss is identified in children, the better for the child's language, learning and overall development.

- Huntington's disease
  The symptoms of Huntington's disease usually, but not always, first appear when the person is approaching middle age.

- Kabuki syndrome
  Kabuki syndrome affects males and females equally and there is no cure.

- Kennedy's disease
  Kennedy's disease is a rare inherited neuromuscular disorder that causes progressive weakening and wasting of the muscles, particularly the arms and legs.

- Kidney disease - medullary cystic kidney disease
  Medullary cystic kidney disease causes the growth of abnormal cysts in the kidneys.

- Kidney disease - polycystic kidney disease (PKD)
  Polycystic kidney disease is a common cause of kidney failure in Australia and equally affects men and women of different ethnic backgrounds.

- Klinefelter syndrome
  Klinefelter syndrome is often diagnosed at puberty, when the expected physical changes don't occur.

- Leukodystrophy
  Leukodystrophy refers to a group of inherited disorders that affect the white matter of the brain, which causes loss of normal brain functions.

- Long QT syndrome
  You should be investigated for long QT syndrome if you faint for no apparent reason, during or after exercise or emotional excitement.

- Marfan syndrome
  Some people may not realise they have Marfan syndrome, because their features are either mild or not obvious.

- McCune-Albright syndrome
  The severity of symptoms or how a child with McCune-Albright syndrome will be affected throughout life is difficult to predict.

- Muscular dystrophy
  People affected by muscular dystrophy have different degrees of independence, mobility and carer needs.

- Neurofibromatosis
  Neurofibromatosis is caused by faulty genes, which may be inherited or have spontaneously mutated at conception.

- Noonan syndrome
  Noonan syndrome is a genetic condition that usually includes heart abnormalities and characteristic facial features.

- Osteoporosis in children
  Osteoporosis in children is rare and usually caused by an underlying medical condition.

- Phenylketonuria (PKU)
  PKU is an inherited disorder that prevents the normal breakdown of a protein found in some foods.

- Prader-Willi syndrome
  A feature of Prader-Willi syndrome is the child's excessive appetite, which often leads to obesity.

- Premature and early menopause
  The symptoms of premature or early menopause are the same as for menopause at any age.

- Rett syndrome
  People with Rett syndrome have a keen desire to communicate.

- Spinal muscular atrophy (SMA)
  A child with spinal muscular atrophy type 1 rarely lives beyond three years of age.

- Tay-Sachs disease
  Tay-Sachs disease is a serious genetic disorder common in Ashkenazi Jews and French-Canadians.

- Thalassaemia

betterhealth.vic.gov.au
Thalassaemia is an inherited blood disorder that can cause anaemia or death if not treated.

- **Tourette syndrome**
  Milder forms of Tourette syndrome can be misdiagnosed, as it often occurs at the same time as attention deficit hyperactivity disorder (ADHD) and other disorders.

- **Treacher Collins syndrome**
  Treacher Collins syndrome is a genetic disorder that affects growth and development of the head, causing facial defects and hearing loss.

- **Tuberous sclerosis**
  Tuberous sclerosis is a genetic disorder that affects various parts of the body to varying degrees of severity.

- **Turner's syndrome**
  Turner's syndrome is a random genetic disorder that affects females, causing short stature and infertility.

- **Usher syndrome**
  Services aim to help a person with Usher syndrome prepare for the dual loss of sight and hearing.

- **Von Willebrand disease**
  A person with von Willebrand disease may have frequent nosebleeds, heavy menstruation or excessive bleeding from the mouth.

- **Williams syndrome**
  Williams syndrome often goes undiagnosed, which means that some people with the disorder fail to get the support and treatment they need until later in life.

- **Wilson disease**
  In Wilson's disease, a build-up of copper damages organs including the liver, nervous system, brain, kidneys and eyes.

### Genes and genetics explained

- **Genes and genetics explained**
  Children inherit physical characteristics such as eye colour from their parents through their genes.

- **Gene therapy**
  Gene therapy is an experimental form of treatment that targets the faulty genes that cause genetic diseases.

- **Genetic disorders**
  Genetic disorder is caused by an altered or faulty gene or set of genes.

- **Genetic services in Victoria**
  Genetic services can help people who are affected by, or who are at risk of, inherited conditions or birth defects, to make informed choices about their healthcare.

### Genetic testing

- **DNA profiling**
  DNA profiling is a way of establishing identity and is used in a variety of ways.

- **Egg freezing**
  You can freeze your eggs for medical reasons or for reasons that are more to do with your life circumstances.

- **Genetic services in Victoria**
  Genetic services can help people who are affected by, or who are at risk of, inherited conditions or birth defects, to make informed choices about their healthcare.

- **Genetic testing for inherited cancer**
  A predisposition to certain cancers can be inherited via altered genes.

- **Newborn bloodspot screening**
  Every newborn baby in Australia is offered a newborn bloodspot screening test to identify those at risk of rare, but serious, medical conditions.

- **Pregnancy tests – chorionic villus sampling**
  Chorionic villus sampling (CVS) is a pregnancy test that checks the baby for some abnormalities.
Pregnancy tests - maternal serum screening

Maternal serum screening can indicate increased risk of abnormalities in the unborn child, but is not a diagnosis.

Related Information

- **Tay-Sachs disease**
  Tay-Sachs disease is a serious genetic disorder common in Ashkenazi Jews and French-Canadians.

- **Dwarfism**
  Dwarfism refers to a group of conditions characterised by shorter than normal skeletal growth.

- **Kabuki syndrome**
  Kabuki syndrome affects males and females equally and there is no cure.

- **Neurofibromatosis**
  Neurofibromatosis is caused by faulty genes, which may be inherited or have spontaneously mutated at conception.

- **Prader-Willi syndrome**
  A feature of Prader-Willi syndrome is the child's excessive appetite, which often leads to obesity.

Related information on other websites

- Centre for Genetics Education
- Child and Youth Health SA
- Down Syndrome Victoria
- Genetic Support Network of Victoria

Support Groups

- Down Syndrome Association of Victoria Inc.

Content Partner

This page has been produced in consultation with and approved by: Down Syndrome Association of Victoria

Last updated: April 2016

Content on this website is provided for information purposes only. Information about a therapy, service, product or treatment does not in any way endorse or support such therapy, service, product or treatment and is not intended to replace advice from your doctor or other registered health professional. The information and materials contained on this website are not intended to constitute a comprehensive guide concerning all aspects of the therapy, product or treatment described on the website. All users are urged to always seek advice from a registered health care professional for diagnosis and answers to their medical questions and to ascertain whether the particular therapy, service, product or treatment described on the website is suitable in their circumstances. The State of Victoria and the Department of Health & Human Services shall not bear any liability for reliance by any user on the materials contained on this website.
My Health Life helps you manage your health

With tools, information and recommendations tailored to you, it’s your personal and secure health dashboard.

Learn more

Medical Dictionary

Enter a search term

Search

Search for your topic using the Merriam Webster medical dictionary

Service Search

Service Search

Find services near you

Service: Select a service

Location:

Type a minimum of three characters then press UP or DOWN on the keyboard to navigate the autocompleted search results

Melbourne or 3000

Use my location

Find a service

Find a doctor

Need to find a doctor in your local area? Take a look at the general practitioners entry in our health service profiles.

see general practitioner

Recent Activity

45 people have watched a video today
Polls polled today are 1.

• Health topics
  • Conditions and treatments
Healthy living
Services and support

• Explore
  - Recipes
  - Healthy pantry
  - Videos
  - Consumer medicine information
  - Multilingual health information - Health Translations Directory

• About
  - About us
  - Accessibility
  - Content partners
  - Privacy
  - Terms of use
  - Contact us

• Connect with us
  - Facebook
  - Twitter
  - YouTube

Page last reviewed: 29 Apr 2014

This web site is managed and authorised by the Department of Health & Human Services, State Government of Victoria, Australia

© Copyright State of Victoria 2018.

Back to Top

This page has been produced in consultation with and approved by:

Down Syndrome Association of Victoria

Content on this website is provided for information purposes only. Information about a therapy, service, product or treatment does not in any way endorse or support such therapy, service, product or treatment and is not intended to replace advice from your doctor or other registered health professional. The information and materials contained on this website are not intended to constitute a comprehensive guide concerning all aspects of the therapy, product or treatment described on the website. All users are urged to always seek advice from a registered health care professional for diagnosis and answers to their medical questions and to ascertain whether the particular therapy, service, product or treatment described on the website is suitable in their circumstances. The State of Victoria and the Department of Health & Human Services shall not bear any liability for reliance by any user on the materials contained on this website.

For the latest updates and more information, visit www.betterhealth.vic.gov.au