

Down Syndrome

Version: April 2009

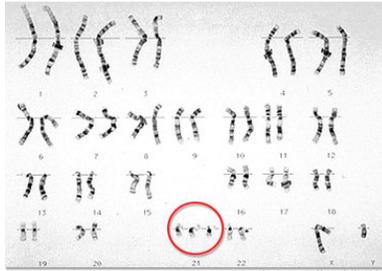


Image credit: University of Utah Medical Genetics

Condition & Description

Down syndrome, named after John Langdon Down who identified the syndrome¹, is one of the more common chromosomal disorders and includes some degree of mental defect³. It is caused by the presence of an additional chromosome 21, causing the affected person to have three and not the normal 2 and hence is referred to as Trisomy 21. It occurs in approximately 1 in 800 live births¹. Estimates indicate there are over 400,000 people in the United States living with Down syndrome². In California, the rate of Down syndrome is 13.66 per 10,000 births.

Associated Conditions

Approximately 50% of children with Down syndrome also have congenital heart disease which can be associated with high blood pressure in the lungs¹. Vision problems such as near-sightedness, cataracts and near or far sightedness affect approximately 60% of children with Down syndrome³. Intestinal malformations that require surgery are found in approximately 10% of children⁴.

In newborns, complications include poor muscle tone and malformation of the upper part of the spine. Young children are 10-15 times more likely to develop leukemia and also suffer from higher rates of infectious diseases. Adults with Down syndrome tend to age prematurely and exhibit characteristics similar to Alzheimer patients such as dementia, memory loss and impaired judgment¹.

Causes & Risk Factors

The causes of chromosome abnormalities such as Down syndrome are unknown.

The most common risk factor for giving birth to a child with Down syndrome is age. The risk for women 35 years of age is 1 out of 350 births which increase to 1 out of 25 births for women 45 and older¹.

Prevention

There are a number of steps a woman can take to reduce her risk of having a baby with a birth defect. Please see CBDMP's fact sheet on [Reducing the Risk of Birth Defects](#).

Preconception & Prenatal Care

Although prenatal care cannot prevent Down syndrome, it is possible to screen for and/or diagnose Down syndrome during pregnancy. Screening involves a blood test and ultrasound, done during the first trimester, in order to examine the back of the baby's neck. There is another blood test that is performed during the second trimester. If these show an elevated risk, then a diagnostic test such as chorionic villus sampling or amniocentesis can be performed³. For more information, please visit the [California Prenatal Screening Program](#) website.

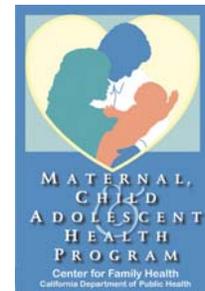
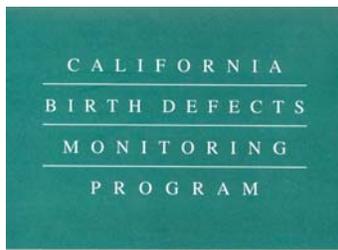
Medical Care

A syndrome is a pattern or combination of physical, mental, and health characteristics that are usually seen together. There is no standard treatment and therefore any course of treatment is decided on an individual basis.

Children with Down syndrome need hearing screening and regular dental care, although tooth eruption may be delayed. Early on, they may learn faster with sign language than with speech. Yearly thyroid function testing is recommended.

Long Term Outcomes

People with Down syndrome are living much longer lives. The average life expectancy in 1929 was nine years old. Today it is common to live to fifty and sometimes even longer⁴. Additionally, with assistance, people with Down syndrome can develop skills that may help them maintain employment and allow them to live semi-independently. Females are capable of becoming pregnant and giving birth¹, though there is a 50% chance of conceiving a child who also has Down syndrome⁴.



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Resources For Families

Regional Centers

Regional centers are nonprofit private corporations that contract with the Department of Developmental Services to provide or coordinate services and supports for individuals with developmental disabilities. They have offices throughout California to provide a local resource to help find and access the many services available to individuals and their families.

- Services Provided By Regional Centers
<http://www.dds.cahwnet.gov/RC/RCSvs.cfm>
- Who is Eligible for Services?
<http://www.dds.cahwnet.gov/General/Eligibility.cfm>
- Early Start
<http://www.dds.ca.gov/EarlyStart/Home.cfm>

California Healthy Families Program

Healthy Families is low cost insurance for children and teens. It provides health, dental and vision coverage to children who do not have insurance and do not qualify for free Medi-Cal.

- Phone: 1-800-880-5305 if you do not already have a child enrolled in Healthy Families.
- E-mail: HealthyFamilies@MAXIMUS.com
- Be sure to include your name and phone number.
- In person: Many community organizations have people who are trained to help you apply. Call to find a *Certified Application Assistants*, or CAAs in your area: 1-888-747-1222. The call is free.
- Website: www.healthyfamilies.ca.gov

California Children's Services (CCS)

California Children's Services (CCS) is a state program for children with certain diseases or health problems. Through this program, children up to 21 years old can get the health care and services they need. CCS will connect you with doctors and trained health care people who know how to care for your child with special health care needs.

(Service eligibility is income-based, but you may be eligible for services if the health care costs exceed 20% of your income.)

Find your local CCS Office –

- In the phone book under *California Children's Services* or *County Health Department*
- Or online at: www.dhs.ca.gov/ccs

Birth Defects Research for Children, Inc.

Birth Defect Research for Children is a resource for free birth defect information, parent networking and birth defect research through the National Birth Defect Registry.

- Birth Defect Research for Children, Inc.
800 Celebration Avenue, Suite 225
Celebration, FL 34747
- Phone: 407-566-8304
- Fax: 407-566-8341
- Website: <http://www.birthdefects.org/>

Other Information & Support Groups

- National Down Syndrome Society: <http://www.ndss.org/>
- National Down Syndrome Conference: <http://www.ndsccenter.org/>
- Down Syndrome Treatment and Research Foundation: <http://www.dsrtf.org/>
- Down Syndrome: www.downsyndrome.com/
- Down Syndrome Information Alliance: <http://www.downsyndromeinfo.org/>
- California Prenatal Screening Program: <http://www.cdph.ca.gov/programs/pns/Pages/default.aspx>
- Family Resource Center: <http://www.frcnca.org/>
- Down Syndrome: <http://emedicine.medscape.com/article/943216-overview>

References

1. National Institutes of Health. (2008). Facts about Down Syndrome. Retrieved on October 17, 2008, from <http://www.nichd.nih.gov/publications/pubs/downsyndrome.cfm>.
2. National Down Syndrome Society. (2009). Down Syndrome Fact Sheet. Retrieved on October 17, 2008, from http://www.ndss.org/index.php?option=com_content&view=article&id=54&Itemid=74.
3. March of Dimes (2007). Down Syndrome – Quick References and Fact Sheets. Retrieved on October 17, 2008, from http://www.marchofdimes.com/professionals/14332_1214.asp.
4. Children's Hospital of Philadelphia (2009). Down Syndrome. Retrieved on October 17, 2008, from http://www.chop.edu/consumer/your_child/wellness_index.jsp?id=8545.