

# Down Syndrome

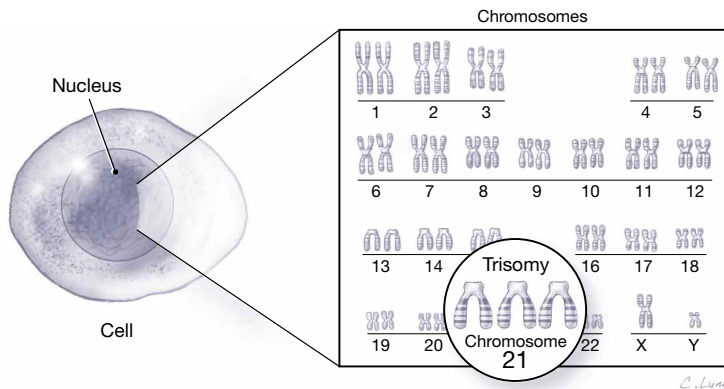
**D**own syndrome, a **chromosomal** disorder, is the most frequent genetic cause of mild to moderate mental retardation in the United States. An article in the February 28, 2001, issue of JAMA discusses whether an ultrasound examination of the fetus during pregnancy is an accurate screening test for Down syndrome. The authors conclude that ultrasound examinations are not practical to use for this purpose.

## WHAT IS A CHROMOSOME?

A **chromosome** is a package of genetic material found in the center (**nucleus**) of every cell. Human cells normally contain 23 pairs of chromosomes, half of which are inherited from each parent. Each chromosome pair is designated by a number, except for the sex chromosomes, which are designated by X and Y.

## WHAT CAUSES DOWN SYNDROME?

Down syndrome occurs when one of the reproductive cells from the parents that combine at fertilization contains extra material from chromosome 21. Individuals with Down syndrome usually have 3 copies of chromosome 21 (**Trisomy 21**) instead of 2 copies.



The risk of having a baby with Down syndrome increases with increasing age of the mother at conception. You can discuss with your doctor the possibility of receiving **genetic counseling** and prenatal screening for Down syndrome. Genetic counseling helps you and your partner learn about the benefits and risks of prenatal screening.

## DEVELOPMENTAL NEEDS OF INDIVIDUALS WITH DOWN SYNDROME

Most individuals who have Down syndrome have intelligence quotient (IQ) scores in the mild to moderate range of retardation. Children with Down syndrome need to be in special educational programs that give them a good environment for learning and that keep pace with their intellectual capabilities. Early intervention with the help of special educators, speech therapists, occupational therapists, and social workers can provide the stimulation and encouragement that can make a difference in the life of a person with Down syndrome.

Sources: National Institute of Child Health and Human Development, American Academy of Family Physicians, March of Dimes, The AMA Family Medical Guide, The AMA Complete Guide to Children's Health, The AMA Home Medical Library, The AMA Encyclopedia of Medicine

## MEDICAL PROBLEMS ASSOCIATED WITH DOWN SYNDROME

Children who have Down syndrome have the same health care needs as other children, but they also need extra care. Some of the conditions that may be associated with Down syndrome include

- Heart defects
- Hearing problems
- Eye problems
- Seizure disorders
- Stomach or intestinal problems
- Trouble swallowing
- Thyroid problems
- Bone and joint problems
- Leukemia

## FOR MORE INFORMATION

- American Academy of Family Physicians  
800/274-2237 or [familydoctor.org](http://familydoctor.org)
- National Institute of Child Health and Human Development Clearinghouse  
PO Box 3006  
Rockville, MD 20847  
800/370-2943 or [www.nichd.nih.gov](http://www.nichd.nih.gov)
- National Down Syndrome Congress  
800/232-6372 or [www.ndscenter.org](http://www.ndscenter.org)
- National Down Syndrome Society  
800/221-4602 or [www.ndss.org](http://www.ndss.org)

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