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Down Syndrome

What is Down syndrome?

Down syndrome is a birth defect that occurs in approximately one in 900 births. It is characterized by varying degrees of mental retardation and changes in appearance. Less than half of all babies born with Down syndrome have congenital heart defects, and many have visual and hearing problems.

Currently, there are over 250,000 people with Down syndrome living in the U.S. Down syndrome is not curable. The life expectancy of someone with Down syndrome is about 55 years.

What causes Down syndrome?

Down syndrome is a chromosomal birth defect. A normal baby develops with 23 chromosomes contributed by both the mother's and the father's cells. When one cell contributes 24 chromosomes instead of 23, the extra chromosome (number 21) results in Down syndrome. The birth defect is also known as Trisomy 21, because of the presence of three number 21 chromosomes.

How does Down syndrome make a person different?

- **Appearance:** A person with Down syndrome may have eyes that slant upward. Their ears are smaller and often fold inward at the top and their nose is usually smaller and flatter, too. Their mouth is smaller, making their tongues look large. Babies with Down syndrome tend to have shorter necks and smaller hands with smaller fingers. Finally, a person with Down syndrome tends to be short in height and very limber in the joints.
- **Mentally:** The degree of retardation varies greatly. Most retardation is moderate. Advances in training have shown that fewer than 10% will be considered to have severe retardation.
- **Developmentally:** Most persons with Down syndrome develop into happy, healthy individuals that can do most everything a normal person can do, it just takes them a little longer to learn and develop skills. Almost all attend school. It has been found that the earlier a child with Down syndrome is involved in special learning programs, the more likely they are to read and write later down the road.
- **Health:** Less than half will have heart defects, which will need to be treated. It is recommended that all babies born with Down syndrome see a pediatric cardiologist. Also, about 10-12% will be born with intestinal disorders. More than 50% will have sight problems that are easily treatable with glasses and/or simple surgery. Often, hearing is impaired. Both hearing and sight impairments should be assessed as early as possible so that development is not delayed. Other health concerns for children with Down syndrome are frequent colds, bronchitis, pneumonia, thyroid problems and leukemia.

Other important facts:

- Parents of a child with Down syndrome are only slightly more likely to have another child with Down syndrome (1% higher) than parents of the same age who do not have a child with Down syndrome. One exception is when the first child is born with translocation Down syndrome. This greatly increases the parent's chance of having another child with it. Translocation Down syndrome is very rare.

- Older maternal age, (greater than 35) and mothers and fathers who have chromosomal rearrangements involving chromosome 21 (which results in translocation Down syndrome) are the greatest risk factors for having a child with Down syndrome.
- 70 % of all children born with Down syndrome are to mothers younger than 35.
- Down syndrome often can be determined prenatally (before birth) through the use of tests like Quad Screen (which is a blood test performed in the first trimester) and through amniocentesis (a test which takes amniotic fluid from the uterus so that cells can be examined).

There are many support groups that you can contact for more information. Here are just a few:

National Down Syndrome Society
666 Broadway
New York, NY 10012
1-800-221-4602
212-460-9330

National Down Syndrome Congress
1605 Chantilly Drive, Suite 250
Atlanta, GA 30324
1-800-232-NDSC
404-633-1555

International Resource Center for Down Syndrome
CWRU-School of Medicine
Cleveland, OH 44106-4921
1-800-288-8804
216-368-8806