

## Down Syndrome

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### What is Down syndrome?

Persons with Down syndrome are first and foremost human beings who have recognizable physical characteristics and limited intellectual endowment that are due to the presence of an extra chromosome 21.

The estimated incidence of Down syndrome is between 1 in 800 to 1 in 1,100 live births. Each year approximately 3,000 to 5,000 children are born with this chromosome disorder. It is believed there are about 250,000 families in the United States who are affected by Down syndrome.

### How do children with Down syndrome develop?

Children with Down syndrome are usually smaller, and their physical and mental developments are slower, than those who do not have Down syndrome. The majority of children with Down syndrome function in the mild to moderate range of mental retardation. However, some children are not mentally retarded; they may function in the borderline to low average range; and only a few children may be severely mentally retarded. There is a wide variation in mental abilities and developmental progress in children with Down syndrome. Also, their motor development is slow; e.g. instead of walking by 11 to 14 months as other children do, children with Down syndrome usually learn to walk between 15 to 36 months. Their language development is also markedly delayed.

It is important to note that a caring and enriching home environment, early intervention, and integrated educational efforts will have a positive influence on the child's development.



### What are the physical features of a child with Down syndrome?

Although individuals with Down syndrome usually have distinct physical characteristics, generally, they are

more similar to the average person in the community than they are different. The physical features are important to the physician in making the clinical diagnosis, but no emphasis should be put on those characteristics otherwise. Not every child with Down syndrome has all the characteristics; some may only have a few, and others may show most of the signs of Down syndrome. Some of the physical features of children with Down syndrome include flattening of the back of the head, slanting of the eyelids, small skin folds at the inner corner of the eyes, depressed nasal bridge, slightly smaller ears, small mouth with narrow palate, decreased muscle tone, loose ligaments, and small hands and feet. About fifty percent of children with Down syndrome have one line across the palm, and there is often a wide gap between the first and second toes. The physical features observed in children with Down syndrome (and there are many more than described above) usually do not cause any significant disability in the child.

### How many chromosome subtypes are observed in Down syndrome?

There are four main types of chromosome abnormalities in Down syndrome:

The vast majority of children with Down syndrome (approximately 95 percent) have an extra 21 chromosome. Instead of the normal number of 46 chromosomes in each cell, the individual with Down syndrome has 47 chromosomes. This condition is called trisomy 21.

The second type is called translocation. Here, the extra 21 chromosome is attached or translocated on to another chromosome, usually on chromosome 14, 21 or 22.

If translocation is found in a child with Down syndrome, it is important to examine the parents' chromosomes, since in at least one-third of the cases, a parent may be a carrier of the translocation. This form of chromosome error is found in 3 to 4 percent of individuals with Down syndrome.

Another chromosome problem, called mosaicism, is noted in about 1 percent of persons with Down syndrome. In this case, some cells have 47 chromosomes and others have 46 chromosomes. Mosaicism is thought to be the result of an error in one of the cell divisions after conception. Children with mosaicism Down syndrome usually have fewer physical characteristics and function better intellectually than those with trisomy 21 and translocation Down syndrome.

A very rare chromosome abnormality in persons with Down syndrome is called partial trisomy 21. Here, part of an extra chromosome 21 is usually attached to another chromosome.

### **What is the cause of Down syndrome?**

Although many theories have been proposed, at the present time it is not known what actually causes Down syndrome. Some professionals believe that hormonal abnormalities, X-rays, viral infections, immunologic problems, or genetic predisposition may be the cause of the improper cell division resulting in Down syndrome, but there is no evidence that any of these conditions causes Down syndrome.

It has been known for a long time that the risk of having a child with Down syndrome increases with advancing age of the mother; i.e., the older the mother, the greater the possibility that she may give birth to a child with Down syndrome. However, most babies with Down syndrome (more than 85 percent) are born to mothers younger than 35 years of age. Recent studies revealed that if the mother has only one ovary, she also has an increased risk of having a child with Down syndrome.

The extra chromosome in trisomy 21 could either originate in the mother or the father. Most often, however, the extra chromosome comes from the mother.

### **What kind of information can be provided through genetic counseling?**

Parents who have a child with Down syndrome have an increased risk of having another child with Down syndrome in future pregnancies. It is estimated that the risk of having another child with Down syndrome is about one in 100 in trisomy 21 and probably also in mosaicism. If, however, the child has translocation Down syndrome and if one of the parents is found to be a translocation carrier, then the risk of recurrence increases markedly. The actual risk depends on the type of translocation and whether the translocation is carried by the father or the mother.

### **What health concerns are often observed in people with Down syndrome?**

Generally, the child with Down syndrome is in need of the same kind of optimal medical care as any other child. The pediatrician or family physician should offer support and counseling to the family, provide general health maintenance, immunizations, and attend to medical problems. There are, however, situations when children with Down syndrome need special attention.

Sixty to eighty percent of children with Down syndrome have hearing deficits. Therefore, audiologic assessments at an early age and follow-up hearing tests are indicated. If there is a significant hearing loss, the child should be seen by an ear, nose and throat specialist.

Forty to fifty percent of children with Down syndrome have congenital heart disease. Many of these children will have to undergo cardiac surgery and often will need long term care by a pediatric cardiologist. Persons with Down syndrome beyond adolescence often have mitral valve prolapse which is thought to be a benign condition.

Intestinal abnormalities also occur at a higher frequency in children with Down syndrome. For example, a blockage of the food pipe (esophagus), small bowel (duodenum), and at the anus (anal atresia) are not uncommon in infants with Down syndrome. These may need to be surgically corrected at once in order to have a normal functioning gastro-intestinal tract.

Celiac disease which is due to an intolerance to a particular part of protein in flour (gluten) is also more often observed in persons with Down syndrome.

Children with Down syndrome have more eye problems than other children who do not have this chromosome disorder. For example, 3 percent of newborn infants with Down syndrome have cataracts that need to be removed surgically. Other eye problems such as cross-eyedness (strabismus), near-sightedness, far-sightedness and other eye conditions are frequently observed in children with Down syndrome. Therefore, regular ophthalmological examinations are recommended.

Another concern relates to nutritional aspects. Some children with Down syndrome, in particular those with severe congenital heart disease, often fail to thrive in infancy. On the other hand, obesity is frequently noted during adolescence and early adulthood. These conditions can be prevented by providing appropriate nutritional counseling and dietary guidance.

Thyroid dysfunctions are more common in children with Down syndrome than in normal children. Between 15 and 20 per cent of children with Down syndrome have hypothyroidism. A few children may have hyperthyroidism. It is important to identify individuals with Down syndrome who have thyroid disorders and institute appropriate treatment, since it may compromise central nervous system functioning.

Skeletal problems have also been noted at a higher frequency in children with Down syndrome, including kneecap subluxation, hip dislocation, and atlantoaxial instability. The latter condition occurs when the first two neck bones are not well aligned because of the presence of loose ligaments.

Approximately 15 percent of people with Down syndrome have atlantoaxial instability. Most of these individuals, however, do not have any symptoms, and only

1 to 2 percent of individuals with Down syndrome have a serious neck problem (symptomatic atlantoaxial instability) that requires surgical intervention.

Other important medical aspects in Down syndrome, including immunologic concerns, leukemia, seizure disorders, sleep apnea, skin disorders, zinc deficiency, and Alzheimer disease may require the attention of specialists in their respective fields.

### **Can Down syndrome be medically treated?**

Although many medications and various therapies including nutritional supplements have been touted as treatment for people with Down syndrome, there is no effective medical treatment available at the present time. However, recent advances in molecular biology make it feasible now to examine the genetic basis for Down syndrome. In the spring of 2000 nearly all genes on chromosome 21 were identified and their DNA was sequenced. However, at the present time we do not know how the triple genetic dose interferes with normal developmental sequences. Once we have more knowledge of the genes' function and how to counteract these genes, a rational approach to medical therapy could emerge.

### **What educational services and vocational opportunities are available for people with Down syndrome?**

Today early intervention programs, pre-school nurseries, and integrated/inclusive special education strategies have demonstrated that youngsters with Down syndrome can participate in many learning experiences that will positively influence their overall functioning. Research has shown that early intervention, environmental enrichment, and assistance to the families will result in marked progress that is usually not achieved by those infants who have not had such educational and stimulating experiences.

Children with Down syndrome, like other children, can benefit from sensory and cognitive stimulation, specific exercises involving gross and fine motor activities, and speech therapy. Also, preschool nurseries play an important role in the young child's life since exploring the environment beyond the home enables the child to participate in a broader world.

Later, the school can give the child a foundation for life through the development of academic skills and physical as well as social abilities. School should provide an opportunity for the child to engage in sharing relationships with others and help to prepare the child to become a productive citizen. Contrary to some views, all

children can learn, and they will benefit from placement in a normalized setting with support as needed.

During adolescence, youngsters with Down syndrome should be exposed to prevocational training in order to learn good work habits and to engage in proper relationships with co-workers. Appropriate vocational counseling and job training will result in meaningful employment, and this, in turn, should lead to a feeling of self-worth and of making a contribution to society. In addition, post-secondary school educational opportunities should be made available for young people with Down syndrome.

### **What attitude should society have?**

It is important that society develop attitudes that will permit people with Down syndrome to participate in community life and to be accepted. They should be offered a status that observes their rights and privileges as citizens, and in a real sense preserves their human dignity. When accorded their rights and treated with dignity, people with Down syndrome will, in turn, provide society with a most valuable humanizing influence.

### **Selected Resources**

#### **Organizations:**

National Down Syndrome Congress  
1370 Center Drive, Suite 102  
Atlanta, Georgia 30338  
Toll-free: 800-232-6372  
Local: 770-604-9500  
E-mail: [info@ndscenter.org](mailto:info@ndscenter.org)  
<http://www.ndscenter.org>

National Down Syndrome Society  
666 Broadway, 8<sup>th</sup> Floor  
New York, New York 10012-2317  
Toll-free: 800-221-4602  
Local: 212-460-9330  
E-mail: [info@ndss.org](mailto:info@ndss.org)  
<http://www.ndss.org>

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