

What is Down Syndrome (DS)?

Down Syndrome is a disorder that includes a combination of birth defects; among them, some degree of mental retardation, characteristic facial features, heart defects, increased infections, problems with vision and hearing are common. The severity of the problems varies greatly among affected individuals.

How common is this condition?

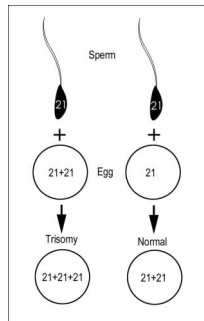
Down Syndrome is one of the most common genetic birth defects, affecting approximately one in 800 to 1,000 babies. It generally is caused by an extra 21st chromosome (Chromosomes are the structures in cells that contain the genetic information in the form of gene).

Is Down syndrome hereditary?

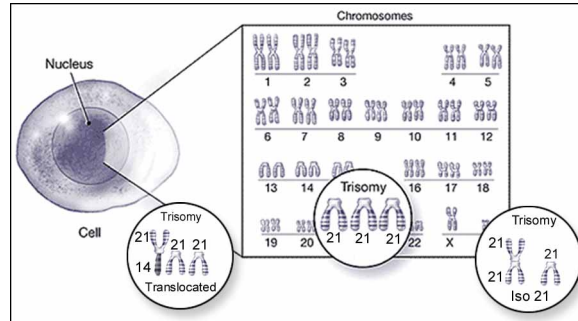
No, most often it is a sporadic event (a new event) very rarely familial chromosomal rearrangements causes DS.

What Causes Down Syndrome?

Every human cell contains 46 chromosomes (rod like structures with genetic material) which are present as 23 pairs. The only exception are the germ cells (egg and sperm) that contain 23 chromosomes. The union of egg (23) and sperm (23) creates 23 pairs or 46 chromosomes in total. DS is resulted when a germ cell (egg or sperm) with an extra 21st chromosome fuses with another normal germ cell resulting in a total of 47 chromosomes. All of the features and birth defects associated with DS result from the extra dose of genes present on this extra chromosome 21 in each of the body's cells (also is called trisomy 21).



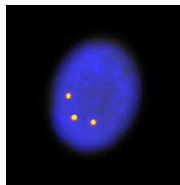
Trisomy 21 Conception



Karyotyping

What are the different forms of Down Syndrome?

- **Standard DS:** All cells of the affected individual have an extra 21st chromosome. This condition is usually present from the time of conception.
- **Mosaic DS:** Affected individuals have some cells with an extra chromosome 21 and others with normal chromosome number (46). This condition occurs as an accident in cell division after conception and during the course of embryonic development.
- **Translocation DS:** Occasionally, the extra chromosome 21 is attached to another chromosome and is called Translocation Down syndrome. If the chromosome that is attached to is a '21' it results in an "Iso 21 chromosome".



FISH Analysis

How is Down Syndrome diagnosed?

Down Syndrome can be diagnosed and confirmed by performing a chromosomal analysis (Karyotyping) on the blood sample of the patient. The test can be done immediately after birth if the doctor has a doubt although it is possible to do the test any time throughout the life. Fluorescence based analysis (FISH) can also be used to diagnose DS.

What Does a Child with Down Syndrome look like?

A child with Down Syndrome may have flat face, flat nasal bridge, small nose, eyes that slant upward and small ears that may fold over a little at the top. His/her mouth may be small, making the tongue appear large and protruding. Some babies with DS have short neck and small hands with short fingers and, due to less muscle tone, appear somewhat "floppy". The child or adult with DS is often short and has unusual looseness of the joints.

Most children with DS will have some, but not all of these features.

What health problems might a child with Down Syndrome have?

Almost half of babies with DS have heart defects. Some defects are minor and may be treated with medications, while others may require surgery. All babies with DS should be examined by a pediatric cardiologist, a doctor who specializes in heart diseases of children, and have an echocardiogram in the first 2 months of life so that any heart defects can be diagnosed and treated.

About 10 % of babies with DS are born with intestinal malformations that require surgery. More than 50 % have some visual impairment. A pediatric ophthalmologist should be consulted, usually within the first six months of life and some children may have hearing loss due to fluid in the middle ear, a nerve defect or both. Babies with DS should therefore be screened for hearing loss at birth or by 3 months of age. These children also tend to have frequent cold and ear infections, as well as bronchitis and pneumonia. They also are at increased risk of thyroid problems and leukemia. Children with the disorder should receive regular medical care including standard childhood immunizations.

What is the degree of mental retardation in Down Syndrome?

The degree of mental retardation varies widely, from mild to moderate to severe. Most fall within the mild to moderate range, and studies suggest that, with proper intervention, only 10 % will have severe mental retardation. There is no way to predict the degree of mental development in a child with DS based upon physical features.

What can a child with Down Syndrome do?

Children with DS usually can do most things that any young child can do, such as walking, talking, dressing and being toilet-trained. However, they generally start learning these things later than other children. The exact age that these developmental milestones will be achieved cannot be predicted. However, early intervention programs beginning in infancy can help these children achieve their developmental milestones sooner.

What is the life expectancy?

The survival rate for infants born with DS is found to be 88% at 1 year and 82% at 10 years. The major cause of death in the first year of life is due to heart defects and/or their complications. Adults with good self-help skills could be expected to live up to 50 years.

Can Down Syndrome be cured or prevented?

There is no cure for DS, nor is there any prevention for the chromosomal abnormality that causes DS. However, recent studies suggest that some women who have had a baby with DS had an abnormality in how their bodies metabolize (process) the B vitamin folic acid. If confirmed, this finding may provide yet another reason why all women who might become pregnant should take a daily multivitamin containing 400 micrograms of folic acid which has been shown to reduce the risk of certain birth defects of the brain and spinal cord. Several therapies have been beneficial in reducing the severity of DS which include antioxidant, nutritional and hormonal therapy.

Is there any special care that will help Down Syndrome individuals?

Individuals with DS benefit from loving homes, early intervention, special education, appropriate medical care and positive public attitudes. In adulthood many individuals with DS hold jobs, live independently and enjoy recreational opportunities in their communities.

Can people with Down Syndrome marry?

Some people with Down Syndrome marry. With rare exceptions, men with DS cannot father a child. In any pregnancy, a woman with Down Syndrome has a 50% chance of conceiving a child with DS, but many affected fetuses are miscarried. A substantial number of adults with DS (15 to 20%) develop Alzheimer's disease in middle age.

Who has the greatest risk of having a baby with Down syndrome?

Parents who have already had a baby with Down syndrome, mothers or fathers who have a rearrangement involving chromosome 21, and mothers over 35 years old are at greatest risk. The risk of DS increases with maternal age, from about 1 in 1,250 for a woman at age 25, to 1 in 1,000 at age 30, 10 in 100 at age 40.

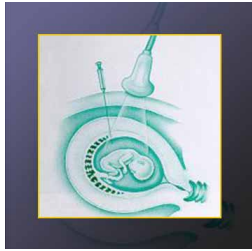
What is the risk that parents of Down Syndrome child will have another affected child?

In general, in each subsequent pregnancy, the chance of having another baby with Down Syndrome is 1% plus whatever additional risk a mother has, based upon her age. If, however, the first child has translocation DS, the chance of having another child with DS may be greatly increased (up

to 15%). It is therefore important to confirm the type of DS through chromosomal analysis (Karyotyping). The doctor may refer parents to a genetic counselor who can explain the results of chromosomal tests in detail, including what the recurrence risk may be in another pregnancy.

Can Down syndrome be diagnosed before the child is born?

Yes. Prenatal testing using amniocentesis or chorionic villus biopsy can diagnose DS during early pregnancy. Amniocentesis is an invasive procedure during which a small amount of amniotic fluid is tapped by an experienced radiologist or gynecologist under ultrasound guidance. Amniocentesis is a safe and simple procedure usually performed around 13-16 weeks of gestation.



Amniocentesis

Prenatal diagnosis to rule out Down Syndrome is strongly recommended for pregnant women with any of the following indications:

✦ History of child with Down Syndrome ✦ Parents with balanced translocation of chromosome 21 ✦ Low AFP levels or abnormal triple results screen test ✦ Increased maternal age (35 years and above) ✦ Abnormal findings in ultrasound which indicate the possibility of DS ✦ In addition to the above, any family with a mentally retarded child or a child with other birth defects can discuss these tests with their doctor or genetic counselor.

What can genetic research offer to Down Syndrome patients and their families in future?

Scientists are investigating why errors in chromosome division occur, in the hope of preventing DS and other birth defects caused by abnormalities in the number or structure of chromosomes. Research in investigating the role of a gene in causing the brain abnormalities associated with DS, with the goal of treating the mental retardation associated with the disorder is also under progress. An international team of scientists has mapped all the genes of chromosome 21, which eventually could pave the way for treatment of many features of this disorder.

How does genetic counseling help?

Genetic counseling provides information on DS, its inheritance and latest development in genetics research. This information along with chromosomal analysis of the affected individual or child provides an immediate benefit to families. Recurrence risk in future pregnancies can be calculated based on genetic studies. Prenatal tests and options available will be discussed

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Patient Information Material



Down Syndrome

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