Down syndrom was first described in 1866. The cause of Down syndrome (trisomy 21) was discovered in 1959. Down syndrome is relatively well known, with distinctive characteristics such as mental retardation, distinguishing facial features, and other traits. In the United States, Down syndrome occurs in 1 in 800 live births, and approximately 6000 children are born with Down syndrome each year. About 85% of infants with Down syndrome survive 1 year, and 50% of people with Down syndrome live longer than 50 years.

The Genetics of Down Syndrome – Trisomy 21

Down syndrome is a genetic disorder caused by extra genetic material (DNA) (ie, presence of an extra 21st chromosome). Chromosomes, which are microscopic thread-like structures that are present in every cell of the body except red blood cells, carry genes. Genes are necessary for development. Human cells normally have 46 chromosomes that can be arranged in 23 pairs. One set of 23 chromosomes comes from the mother (egg cell or ovum) and the other half of the 23 pairs comes from the father (sperm cell).

Trisomy 21

In Down syndrome, 95% of all cases are caused by either the sperm or the egg cell having two 21st chromosomes instead of one, so the resulting fertilized egg has three 21st chromosomes. Hence the scientific name, trisomy 21. Recent research has shown that in these cases, approximately 90% of the time the abnormal cells are the eggs. The cause of the extra chromosome isn't known, but there is definitely connection with the mother's age.

The Effects of Trisomy 21 – Symptoms of Down Syndrome

In trisomy 21, there is extra genetic material from chromosome 21. This extra material means that there are more genes expressed than normal. For most genes, this overexpression has little effect because the body regulates genes and their products. But the genes that cause Down syndrome appear to be exceptions.

Scientists have been trying to determine exactly which genes are involved in Down syndrome ever since the third 21st chromosome (trisomy 21) was found. Current research has led to a theory that only certain areas of chromosome 21 need to be tripled to get the effect of Down syndrome. These regions are called the Down syndrome critical region. Exactly which genes cause Down syndrome when tripled is not known, but some genes are suspected.

Down Syndrome Treatment

There is no specific treatment for Down syndrome. Early intervention programs, such as physical therapy, occupational therapy, and speech therapy, are helpful. Special education and training is offered in most communities for mentally handicapped children. Because the risk of vision problems, hearing loss, infection, and hypothyroidism (low thyroid hormone) is increased, screening and treatment may be necessary.

Timely surgeries for cardiac and gastrointestinal anomalies are necessary to prevent serious complications. Digitalis and diuretics are usually needed for the medical management of cardiac anomalies along with prophylaxis for subacute bacterial endocarditis.

People with Down syndrome should have plenty of opportunities to participate in community life. Children with Down syndrome should participate in social activities, sports, and other aspects of society during the growing years.

Characteristic Features of Down Syndrome

Despite the variability in Down syndrome, children with Down syndrome have a widely recognized characteristic appearance. The head may be smaller than normal (microcephaly) and abnormally shaped. Typical facial features include a flattened nose, protruding tongue, and upward slanting eyes. The inner corner of the eyes may have a rounded fold of skin (epicanthal fold) rather than coming to a point. The hands are short and broad with short fingers, and they often have a single crease in the palm. Normal growth and development is usually retarded, and most affected children never reach average adult height.

Heart defects present from birth are often present in people with Down syndrome. Early death is often caused by cardiac abnormalities. Gastrointestinal abnormalities (such as obstruction of the esophagus, called esophageal atresia, and obstruction of the duodenum, called duodenal atresia) are also relatively common. Obstruction of the gastrointestinal tract may require major surgery shortly after birth. Acute lymphocytic leukemia is also more common in children with Down syndrome.