

# Down syndrome and Its Management (Review)

Dr. Sanjay Ganjoo

Associate professor and H.O.D. department of Munafe -ul - Aza. University college of unani Tonk, Rajasthan

---

## ABSTRACT

**A Genetical disorder of humans where medical science seems helpless . Down Syndrome or trisomy 21, caused by the presence of a third copy of chromosome 21, .It is named after John Langdon Down, a British doctor who fully described the syndrome in 1886 . child with Down syndrome has charsteristic clinical features such as moon like face, protuberant tongue, wide spaced nipples ,epicanthic fold, simian crease ,it (simian)may not be present in some of the cases.**

---

## INTRODUCTION

In recent years Down syndrome has become more common ,and children living with down syndrome are living longer, between 1979 and 2003 the number of babies born with down syndrome increased by 30% . The annual mortality rate per 1,00000 people from down syndrome in developing countries have decreased by 41.1% since 1990 and average of 1.8% year . The life expectancy of people with down syndrome increased dramatically between 1960 and 2007 .In 1960 on average, person with Down syndrome lived to be about 10 years old .In 2007 on average people with Down syndrome lived to be about 47 years old .Down syndrome or trisomy 21 is the most common known genetic cause of intellectual disabilities associated with physical growth delays, characteristics facial features .The average IQ of a young adult with down syndrome is 50 equivalent to the mental ability of a 8 to 9 years old child but it can vary widely .The parents of the affected individual are typically genetically normal the extra chromosome occurs by chance ,the possibility increases from less than 0.1% in 20 year old mothers to 3% in those age 45.

They have high risk of respiratory and blood cancer.Down syndrome age prematurely ,50% die before 5th year of their life in developing countries while life expectancy is high in developed nations.

Four life stages of Down syndrome were identified prenatal childhood ,and early adulthood ,adulthood and Senescence. 23% of deaths are caused by pneumonia in adulthood to 40% in senescence, 13% of children have congenital heart defects in childhood ,23% in adulthood, 10% have coronary artery disease in senescence and 9 % died due to Cardiac, renal ,and respiratory failure . Also some deaths are due to the childhood leukaemia immune and endocrine system dysfunction .

Misfortune of Down syndrome Is that they have an extra chromosome number 21 in every cell

### Types of Down Syndrome

Down syndrome can be divided into three types. We normally have 23 pairs of chromosomes each made of genes. During the formation of egg (or sperm) a woman's (or a man's) pair of chromosomes normally split so that only one chromosome is in each egg.(or sperm).

**First type :-** in trisomy 21,the 25th chromosome pair does not split and a double dose goes to the egg .It is estimated that 95 to 97% of the extra chromosome is of maternal origin .

**Second type :-** This Type of down syndrome is known as translocation . Itoccurs in about 2 to 4% of people with Down syndrome in this type an extra part of the twenty first chromosome gets stuck on to another chromosome .In about half of these situations one parent carries the extra twenty first chromosome , material in a balanced or hidden form .

**Third type :-** This is called Mosaicism, the person with Down syndrome has an extra twenty first chromosome in only some of the cells ,but not all of these .The Other cells have the usual pair of twenty first chromosome ,1 to 2% of the people with Down syndrome have this type.(Cooley and Graham 1991 )

## CHARACTERISTICS

Characteristics associated with Down syndrome including low muscle tone and enlarged tongue, a flat facial profile, and increased risk of related medical conditions. However, every person with Down syndrome is a unique individual and many possess these characteristics to different degrees or not at all. Down syndrome where genetic cause is well understood, but the entire medical science is so helpless that there is no medical treatment for this disorder. Not only this, we are equally helpless in finding out the factors responsible for abnormal division of chromosome number 21, thus resulting in trisomy of this chromosome. Until and unless the factor responsible for the misdivision of chromosome 21 are identified, birth of Down syndrome shall never be controlled.

At present, there is no treatment for Down syndrome; therefore, management or rehabilitation of this syndrome is the only answer. The first part of the management of Down syndrome is to get the chromosomal study done so as to establish the nature of the chromosomal abnormality. **In a Down syndrome child, milestones like social smile, stability of head, rolling over, sitting, walking and talking are all delayed.** Once through chromosome study a child is confirmed as a Down syndrome, the rehabilitation process should start when parents are told that their child is bearing Down Syndrome, their first reaction is of shock and disbelief.

Rehabilitation of Down Syndrome is the combined effort of parents and experts, if it is done with compassion and truthfulness, the child can be made as a normal part of the society. In our country, classes are overcrowded and individual attention is a rarity in schools. Doing so, these children may learn to read and write to a certain extent. Some of the specific points in the rehabilitation of the Down Syndrome are:

## PHYSIOTHERAPY

A newborn Down Syndrome child has low muscle tone than the normal child. Physiotherapy forms an essential part of training right from the birth if properly given, it will help them turn, sit, and walk, and the delay in these milestones may be reduced.

### Speech Therapy :

Most of the Down Syndrome children speak the first word only after 2 years and sentences between 3 and 5 years. Parents should learn from the speech therapist what is to be done and train the child accordingly. Language development may be learnt by the parents and then taught to the child. Group counselling is absolutely necessary in Down Syndrome.

### Special Medical Care :

Down Syndrome is associated with many other congenital anomalies like, birth defects. Prominent among these defects are **heart defects, anomalies of the gastrointestinal tract, malformed teeth, eye defect, decreased intelligence etc.** **The most common associated condition is recurrent respiratory tract infections due to reduced immunity. They are also prone to blood cancer and Alzheimer disease.** If all these defects are monitored in early infancy, the quality of life of individuals with Down Syndrome can be improved significantly and their contribution to society substantial. This can be achieved through special medical attention only.



Puberty development in both sexes is there, but males remain infertile while as females are fertile. As adults, the girl child should be taught to do house work or simple repetitive jobs. Boys also could be taught simple repetitive jobs.

Rehabilitation of Down Syndrome involves a lot of time and financial burden on family, society, and government.

#### **How Can The Birth Of A Down Syndrome Be Prevented:**

It can be done by being aware of the disease risk factors and also utilizing available screening test. It is seen that older the mother >30 years of age, the higher the chances of chromosomal errors and Down Syndrome. Also if there is history of a Down Syndrome child before, the greater chances of having another affected child. Although a woman of any age can have a baby with Down Syndrome.

Down Syndrome cannot be prevented, it is a Genetic abnormality that happens spontaneously. Screening tests are now standard in most prenatal cases and can give parents an idea of the likelihood of chromosomal abnormalities.

#### **CONCLUSION**

Down syndrome is a disorder which cannot be cured, but it is the responsibility of society to give love affection and care to them, also it is the duty of government to provide schools for them so that they can live a better life. Down syndrome are achieving some of the same milestones as other young people. In foreign countries people with Down syndrome get jobs, move into group homes or individual housing. Some time young people with down syndrome undergo major mood changes in these cases additional School assistants and evaluation for hypothyroidism or depression should be ruled out. Teenagers with Down syndrome undergo hormonal changes like any other teen, parents should encourage their teenagers with Down syndrome to develop independent Skill in hygiene and self care, to be aware of privacy issues. Teenagers with Down syndrome should be educated about puberty, sexual activity and consequences of such activities. In India, "Down syndrome federation of India" started by Dr. Surekha Ramachandran.

#### **REFERENCES**

- [1] <https://www.miss.off>>down syndrome
- [2] <https://www.medicinenet.com>>article
- [3] <https://in.m.Wikipedia.off.wiki> down
- [4] what causes Down syndrome 2014-01-17;05 Jan2016
- [5] Morris JK Mutton, DE, Alderman, E (2002). "Revised estimates of the maternal age specific live birth prevalence of down syndrome" journal of medical screening 9(1):2-6.
- [6] Pathophysiology of disease an introduction to clinical medicine 6th edition Stephen J. NEPheW, DayS (2010)