

Down syndrome

Chapter 2

Awareness and Current Knowledge of Down Syndrome

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Abstract

Down's syndrome is an inherited disorder which has higher rate with increasing age. This is high identifiable disease which affects physical and mental health. Babies are unable to walk, talk, think or maintaining body posture. 1 baby out of 700 children is affected by Down's syndrome. It is a chromosomal disorder. Each human cell contains 23 pairs of chromosomes with equal ratio of transfer from parents to babies. There is an extra chromosome inherit from parents in Down's syndrome in the form of 47 chromosomes. This extra chromosome may be completely or partially present at 21st chromosome known as trisomy 21. Downs syndrome is categories into three main types according to chromosome localization. The data about Down's syndrome was taken from different site such as NHS, Down's syndrome society and web med etc. this article reviewed about latest research on the causes, pathophysiology and management of Down's syndrome.

1. Introduction

Down syndrome, also called Trisomy 21, is an inherited ailment which characteristically grounds some level of erudition debility and some specific type of somatic properties [1]. 700-900 children are born with Down's syndrome in the world. If we see the pathophysiology of Down's syndrome normally 46 Chromosomes are present in each human cell but in down's syndrome babies born with 47 Chromosomes. Human body is made of billions of cells. In these cells 46 Chromosomes are present which produce DNA which controls all the activity of a Cell there is one extra chromosome in Down's syndrome [2]. This disease is known from centuries. In late 19th Century an English physician whose name was John's down gave the first concept of Down's syndrome [1]. Due to his discovery he was called father of this disease. Down's syndrome is derived from his name Jerome who was a French physician by his profession gave the concept of down's syndrome as chromosomal disorder in 1959 [3]. Jerome observed 47 chromosomes in patient suffering from Down's syndrome instead of 46 chromosomes (4). Later it was observed that complete or incomplete copy of 21st chromosome is present which is related with down's syndrome [5]. In 2000, scientists discovered that in 21 Chromosome there are 329 Genes which cause Down's syndrome [2].

2. Types

2.1. Mosaic Down's Syndrome

In this syndrome there is mixture of 47 or 46 chromosomes. An extra chromosome is present in 47 containing group. It is less communal form of Down's syndrome and present only in 1% people that suffer from Down's syndrome. It is also called mosaicism [6].

2.2. Non disjunction Down's syndrome

It is the most common type of downs syndrome. 95% people suffer from this type who engaged Down's syndrome. Normally 21st copy of Chromosomes has two copies while in this type 21st copy contain 3 copies at embryonic level. Before commencement Egg or Sperm is unable to separate. When embryo grows it is duplicated to every cell. Due to this reason it is also called trisomy21 [7].

2.3. Translocation

In this type normal 46 Chromosomes are present. 21 copy of Chromosome is partial or complete and this partial or complete copy attaches usually on Chromosome 14. This interchange shows partial or complete symptoms of Down's syndrome [8].

2.4. Pathophysiology

In human 46 numbers of chromosomes are present. In reproduction 23 chromosomes come from father and other 23 come from mother. These 46 chromosomes control all the activities of a human. In Down's syndrome 23 pairs of chromosomes are not dispersed especially 21st pair of chromosome. Consequently baby is born with defective chromosomes in which there is one extra chromosome. Extra chromosome may be partial or complete which cause physical and mental disabilities. In USA 1 child is susceptible of Down's syndrome out of 700 children. It has been observed that Down's syndrome occurrence ratio greatly increases in above 35 years of age mother and father above age 40 has twice the risk of Down's syndrome to his children. Down's syndrome also occurs in masses that have positive family history of Down's syndrome and Gene Translocation [9].

2.5. Clinical feature

Clinical features vary from person to person. Down's syndrome child may have features of bottomless groove between second and first toes, diminished or poor muscle tendency, a solitary, profound, crease transversely palm of hands, short neck, with extra covering at the spinal of the neck, wide, short hands with short fingers, mounting oblique eyes, frequently with a skin crinkle that comes out from the higher eyelid and concealments innermost crook of the eye, trodden facial shape and nose, white acnes on the colored portion of eye (called brush field acnes) and small head, mouth or ears. Babies born with Down's syndrome have low muscle tone as compared to babies without Down's syndrome. Cognitive deficiency, learning and thinking disability is common in child with Down syndrome short attention span, cognitive disability may include impulsive conduct, overdue linguistic and communication growth, sluggish learning and deprived techniques of judgment [10]. Child with Down's syndrome is at greater risk of epilepsy and Alzheimer's disease [11].

3. Diagnosis

3.1. Chorionic villus Sampling

In this test cells are taken from mother placenta to check abnormality in chromosome. There is also a risk of abortion. This test should be done by discussion and cooperation of doctors and parents [12].

3.2. Amniocentesis

In this test a needle is positioned by using USG into the mother uterus to get a sample of the amniotic fluid which is present into fetus. Chromosomes of fetus are observed through amniotic fluid analysis. This process is called amniocentesis. Baby can also be diagnosed after birth by physical outlook [13].

3.3. Cordocentesis or percutaneous umbilical blood sampling

This is a technique which is least used for diagnosis of Down's syndrome. In this procedure doctor take a sample from mother umbilical cord. This procedure is done after eighteen weeks of pregnancy and there is higher menace of abortion. This technique is done when Down's syndrome is not diagnosed from all other tests. So this is the last option of diagnosing Down's syndrome [14].

3.4. Screening test

Screening test is done when pregnant mother is at the age of thirty five while in case of father age should be forty. In first trimester USG and complete blood count is done to evaluate pseudo positive testify test does not confirm then a physician should follow amniocentesis test after fifteen weeks of pregnancy. In second trimester weeks between fifteen and twenty ultrasonography and Quadruple Marker Screen (qms) is done to valuate spinal cords disorders, brain abnormality and Down's syndrome [15].

4. Common Problems Related to Down's syndrome

Alzheimer's disease which grounds dementia and thinking disability, Lower IQ level, autism spectrum disorder with communal assistances, monotonous actions and communication, Cardiac disorder commonly existing at delivery, hypothyroidism, GIT problems like gastroesophageal reflux disease celiac disease and constipation, visual or hearing loss are the common diseases which may occur due to down's syndrome [16,17].

4.1. Treatment

Different methods of treatment are used in Down's syndrome. It depends upon the severity of disease. It is treated as surgically, medically or different types of psychotherapies are done. Following method of treatment can be done to avoid complication in adulthood.

4.2. Early Intervention

Early Intervention consists of group of therapist and teachers which provide services to child up to three years old to improve their mental and physical activity. This group help the child to improve expertise in talking, understanding, listening, solving problems, thinking, meetings with people ,playing, feeding, crawling, dressing themselves, roll over and walking etc. Individuals with disabilities education act orders that entirely baby's birth with Down's syndrome should get initial intrusion facilities as soon as possible after natal. Early intervention, according to the national diabetes services scheme, is a "Systematic package of Exercise, treatment and deeds intended to discourse late growth that might be knowledgeable by infants with Down's syndrome or other debilities. Early intervention includes three psychotherapies.

4.3. Occupational psychotherapy

This is basically an independent therapy for child. This therapy is done for children so that they may do their routine work independently. Activities include picking up something, dressing, assertive Buttons to self-Feeding and spinning a Knob.

4.4. Physical psychotherapy

Most children born with Down's syndrome have low muscle activity. If they are not managed at early stages irregular body tone occur in later stages. To avoid from later complication physical therapy of muscle is done. Physical therapy guide parents how to move the poster of baby. It also helps to develop accurate muscle position, working and daily activities by these muscles [18].

4.5. Speech psychotherapy

Babies affected by Down's syndrome have distended tongue and small Mouth which cause problem while speaking. If baby has low muscle tone along with little mouth and distended tongue then it became worse for child to speak. To meet this problem Speech Therapy is performed which help the baby to speak in adulthood. Hearing Loss devices also aid in speaking. Speech therapy enable child to communicate with other person. Some adults also use clue sign to understand.

4.6. Medicine

Some child born with some diseases like hypothyroidism, developmental delay or Alzheimer's disease and dementia as well. Medication is given to improve medical condition. Thyroxin is given orally in hypothyroidism [19]. Multivitamins and growth enhancer are given in developmental delay and antioxidant is given in dementia [20].

5. Conclusion

From above article we reviewed about the causes, pathophysiology, diagnosis and treatment of down's syndrome .we concluded that the risk of down's syndrome can be minimize by early diagnosis especially in positive family history .we also concluded that down's syndrome cannot managed like other medical disease but it can conservatively be managed.

6. References

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