



Review on Down Syndrome

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Received: March 28, 2017; **Accepted:** April 23, 2017; **Published:** April 28, 2017

Abstract

Down syndrome is a naturally occurring chromosomal disarrangement that has always been a part of the human condition, common across all races, genders or socio-economic lines, and affects approximately one in every 800 births worldwide, resulting in intellectual and physical disability and associated medical issues. Around 50% of children with Down syndrome also have congenital heart disease which may be associated with pulmonary hypertension. Visual disorders like cataracts and near or far sightedness is seen in 60% of children with Down syndrome. Intestinal malformations requiring surgical interventions are seen in approximately 10% of children. Typical characters of Down syndrome are slightly small head which is flattened in the back, extra skin folds at the inner corners of the eyes, up-slanted eyes, small ears, nose and mouth, short stature, small hands and feet and some degree of intellectual disability.

Keywords: Down syndrome; Chromosomal abnormality; Trisomy

Introduction

John Langdon Down, an English physician, who is considered as father of Down syndrome published an accurate description of a person with Down syndrome in 1866. Although many other people had previously recognized the characteristics of the syndrome, it was Down who first described the condition as a distinct and separate entity. Later in 1959, a French physician Jerome Lejeune identified Down syndrome as a chromosomal anomaly. Lejeune observed 47 chromosomes instead of the usual 46 in the cells of individuals with Down syndrome. It was further identified that an extra partial or complete chromosome 21 results in the characteristics associated with Down syndrome.

Complications of Down syndrome include poor muscle tone and malformation of the upper part of the spinal cord. Young children are 10-15 times more prone to develop leukemia and high rates of infectious diseases. Adults tend to age prematurely and exhibit characteristics such as dementia, memory loss and impaired judgment. Though recent advances in medical treatment with social support has raised the life expectancy of people with Down syndrome averaging upto 55 years in developed nations It is necessary that the parents and caretakers to have ample knowledge about Down syndrome so that they can identify and take proper care of the children with Down syndrome. Open access journals provide a platform for the

practitioners as well as researchers to reach out to the parents and the caretakers. They provide a valuable support to the parents and caretakers to improve the quality of life and rise the life expectancy of people effected with Down syndrome.

Many researchers, physicians, geneticists, and NGOs unite to form associations and/or societies to support the caretakers and parents of the patients suffering with down syndrome and to bring awareness among them by regularly publishing parent's guide, handbooks and other literary materials. Some societies like Down syndrome association offeres developmental programme to children with Down syndrome, and training to the parents and others. These societies help in people understand about down syndrome by creating global awareness and help to provide better service to the patients and work together to improve their quality of life.

Many international peer-reviewed scholarly journals like Journal of Genetic Disorders and Genetic Reports, publishes valuable scientific papers on various themes of Down syndrome. Kavak SB et al. had published an interesting article entitled 'Fetuin A Concentration in the Amniotic Fluid of Fetuses with Down Syndrome' in which they found amnion fluid Fetuin-A significantly lower in the presence of Down syndrome. Reduced Fetuin-A levels may be attributed to antenatal and postnatal developments of the Down syndrome foetus. Journal of Alzheimer's Disease & Parkinsonism frequently publishes 'Special Issues' on Down Syndrome under the supervision of Elizabeth Head, Associate Professor, Department of Molecular and Biomedical Pharmacology, University of Kentucky, USA who acts as the Special issue editor. Journal of Down Syndrome & Chromosome Abnormalities publishes articles on Downs syndrome in open access mode making the research easily available to the desired.

Conferences like [Euro Nursing & Medicare Summit](#) provides platform for students and researchers to present their research and ideas to the delegates attending the conference and can receive suggestions of the experts, which will motivate and help them to excel in their respective fields. Susette Brynard presented a poster entitled 'The role of parents in educating learners with Down syndrome successfully: A narrative journey' in the 15th Euro Nursing & Medicare Summit held in Italy [1-20].

Etiology and Risk factors

The etiologies of chromosomal abnormalities such as Down syndrome are unknown. Age of the carrying mother is the most common risk factor for giving birth to a child with Down syndrome. The risk for 35 years old women is 1 out of 350 births which raises to 1 out of 25 births for 45 year and above aged women 45 [21-36].

Comorbidities

Around half of children with Down syndrome found to have congenital heart disease which can be associated with high blood pressure in the lungs. Visual defects like cataracts and near or far sightedness is observed in about 60% of children with Down syndrome. Complications in newly born include poor muscle tone and malformation of the upper part of the spine. Young children more prone to develop leukaemia at a rate of 10-15 times and also suffer from higher rates of infectious diseases. Adults with Down syndrome tend to age prematurely and exhibit Alzheimer's like characteristics such as memory loss, dementia and impaired judgment [37-62].

Diagnosis and Detection

Down syndrome can be screened and/or diagnosed Down syndrome. It involves a blood test and ultrasound, in the first trimester, to examine the back of the baby's neck. Other blood test is performed during the second trimester, if these show an elevated risk, then diagnostic test such as amniocentesis or chorionic villus sampling is performed [63-86].

Medical attention

Down syndrome doesn't have any standard treatment and therefore any course of treatment is decided on an individual basis. Children with Down syndrome need screening of listening abilities and regular dental care, although tooth eruption is slightly delayed. Annual thyroid function testing is recommended. In the initial stages they may learn faster with sign language than with speech [87-98].

Conclusion

People with Down syndrome needs special attention from the initial stages, addressing them properly had proved to be successful in improving their condition. Huge amount of information is shared in open access journals which can be accessed freely by various professionals like physicians, geneticists and caretakers and parents for improving the quality of life of the patients with Down syndrome [99,100].

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