SHORT COMMUNICATION

Toxicology and pharmacology congress: life with down's syndrome is no more challenging, rather beautiful

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ABSTRACT

When many people hear the words "Down syndrome" they picture a significantly disabled person with a poor quality of life. Advances in medicine mean that stereotype no longer applies. Advocates for people with Down syndrome are trying to educate the public on their abilities. The lives of the 250,000 Americans with Down syndrome today are radically different than a generation ago, says Brian Skotko, co-director of the Down syndrome program at Massachusetts General Hospital. Medical advances and educational supports have led to increased life expectancy and better quality of life for those with Down syndrome. A survey done by Skotko showed that 99% of those with Down syndrome were happy with their lives. Additionally, the survey found that 88% of siblings felt that having a person with Down syndrome as their brother or sister had made them better people. Down syndrome was first studied and described by a doctor named John Langdon Down. In every cell in the human body there is a nucleus, where genetic material is stored in genes. Genes carry the codes responsible for all of our inherited traits and are grouped along rod-like structures called chromosomes. Typically, the nucleus of each cell contains 23 pairs of chromosomes, half of which are inherited from each parent. Down syndrome occurs when an individual has a full or partial extra copy of chromosome 21. This additional genetic material alters the course of development and causes the characteristics associated with Down syndrome. A few of the common physical traits of Down syndrome are low muscle tone, small stature, an upward slant to the eyes and a single deep crease across the center of the palm – although each person with Down syndrome is a unique individual and may possess these characteristics to different degrees, or not at all. According to the Centers for Disease Control and Prevention, approximately one in every 700 babies in the United States is born with Down syndrome, making Down syndrome the most common chromosomal condition. About 6,000 babies with Down syndrome are born in the United States each year. It can be concluded that although those with Down syndrome still face significant medical and educational challenges, it is important that the public perception of these individuals accurately reflect their abilities.

INTRODUCTION

Down Syndrome (DS) could be an anomaly with huge medical and social costs, caused by a trisomy of the whole or a part of chromosome 21. It's the foremost prevalent congenital disease worldwide and also the common genetic reason behind intellectual disabilities appearing in about 1 in 400-1500 newborns. Although the syndrome had been described thousands of years before, it was named after John Langdon Down who described its clinical description in 1866. Scientists have identified candidate genes that are involved within the formation of specific DS features. These advances successively may help to develop targeted therapy for persons with congenital anomaly. Screening for DS is a crucial a part of routine prenatal care. Until recently, noninvasive screening for aneuploidy depends on the measurement of maternal serum analytes and ultrasonography. Newer progress has resulted within the development of Non Invasive Prenatal Screening (NIPS) test using cell-free fetal DNA sequences isolated from a maternal blood sample.

Down Syndrome (DS) is that the most often occurring chromosomal abnormality in humans and affecting between 1 in 400-1500 babies born in numerous populations, counting on maternal age, and prenatal screening schedules. DS is that the common genetic reason behind intellectual disabilities worldwide and huge numbers of patients throughout the globe encounters various additional health issues, including heart defects, hematopoietic disorders, and early-onset Alzheimer's disease. The syndrome is because of trisomy of the entire or a part of chromosome 21 altogether or some cells of the body and therefore the subsequent increase in expression thanks to gene dosage of the trisomic genes. It's let alone slowness, congenital heart defects, gastrointestinal anomalies, weak neuromuscular tone, dysmorphic features of the top, neck and airways, audiovestibular and disablement, characteristic facial and physical features, hematopoietic disorders and a better incidence of other medical disorders. The incidence of births of youngsters with DS increases with the age of the mother.

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However, thanks to higher fertility rates in young women, the probability of getting a toddler with DS increases with the age of the mother and quite 80% of youngsters with DS are born to women under 35 years old.