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Editorial

Down Syndrome: A Disease Insight

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ABSTRACT

One of the most common conditions with immense medical and social costs is Down syndrome (DS). Many phenotypes are associated with DS, including congenital heart defects, leukemia, Alzeihmer's disease, Hirschsprung's disease, etc. DS people are influenced to a different degree by these phenotypes, so it is a crucial challenge to grasp the origin of this difference. In this summary article, we highlight an overview of the diagnosis of DS, DS-associated phenotypes and disease control. In this article, the genes or miRNAs implicated with Alzheimer's disease along with Down syndrome, congenital heart defects (AVSD), leukemia like AMKL and Everything, hypertension and Hirschprung disease are discussed.

KEYWORDS: Down Syndrome, Trisomy 21, Chromosome Abnormality

INTRODUCTION

One of the major causes of intellectual disability is Down syndrome, and millions of these people face numerous health conditions, including learning and memory, congenital heart disease (CHD), Alzheimer's disease (AD), leukemia, cancer and Hirschprung disease (HD). Trisomy prevalence is impaired by maternal age and varies from population to population. DS has high genetic diversity and heterogeneity in phenotypes. Trisomic fetuses are at high risk of miscarriages and the likelihood of having multiple medical disorders in DS persons has risen. Recent improvement in socially funded medical care has improved the DS population's life expectancy. The estimated life expectancy for the DS population is 55 years in developing countries. The most common reason behind having a DS babies is presence additional copy body twenty one leading to chromosonal disorder. the opposite causes may be Robertsonian translocation and isochromosomal or ring body. Ischromosome may be a term wont to describe a condition during which 2 long arms of body separate along instead of the long and short arm separating along throughout egg sperm cell development. slowness (karyotype forty seven, XX, + twenty one for females and forty seven, XY, + twenty one for males) is caused by a failure of the body twenty one to separate throughout egg or sperm cell development. In Robertsonian translocation that happens solely in 2-4% of the cases, the long arm of the body twenty one is connected to a different body (generally body 14). whereas condition deals with the error or misdivision happens when fertilization at some purpose throughout organic process. because of this individuals with mosaic DS have 2 cell lineages that contribute to tissues and organs of people with Mosacism (one with the traditional range of chromosomes, and alternative one with an additional range 21).

Management of the Disease:

Severe complications can be avoided by prompt surgical treatment of heart abnormalities within the first 6 months of life. In around 3 percent of infants, congenital cataracts develop and must be removed shortly after birth to allow light to enter the eye. To maintain adequate weight, a healthy diet and daily exercise are required. Since heart surgery, eating disorders and inability to survive typically increase. Clinical geneticist - Referral to a genetics counseling program is highly desirable. These include: Developmental paediatrician, Cardiologist - Early cardiologic evaluation is crucial for diagnosing and treating congenital heart defects, which occur in as many as 60% of these patients, Pediatric pneumonologist -Recurrent respiratory tract infections are common in patients with DS, Ophthalmologist, Neurologist/Neurosurgeon – As many as 10% of patients with DS have epilepsy; therefore, neurologic evaluation may be needed, Orthopedic specialist, Child psychiatrist - A child psychiatrist should lead liaison interventions, family therapies, and psychometric evaluations, Physical and occupational therapist, Speech-language pathologist, Audiologist

CONCLUSION

The primary objective of this research is to unravel the typical genes, including APP, BACE2, PICALM, APOE, GATA 1, JAK 2, CRELD 1 and DSCAM, involved in DS-related phenotypes. MLPA and QF-PCR have been applied to the list of rapid aneuploid studies within a few years. The other approaches are: NGS scanning for maternal plasma for cell-free fetal DNA. Due to their operating expense, labour intensive protocol and complicated data processing, other approaches are not advertised for aneuploidy diagnosis except FISH, MLPA and QF-PCR. Since DS is correlated with multiple health conditions, the treatment of these patients thus involves a structured multidisciplinary approach and consistent supervision of these patients that has been addressed in this article..

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