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Short note on down syndrome

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DESCRIPTION

Down's syndrome is a series of physical, mental, and functional abnormalities resulting from the presence of chromosome 21 that results in a collection of clinical features commonly known as Down Syndrome (DS). DS is one of the most genetically complex disorders compatible with human postnatal survival and is the most common surviving autosomal aneuploidy. A mouse model of DS containing all or part of human chromosome 21 in a trisomy or orthologous mouse genomic region provides valuable insights into the contribution of triple genes or gene groups too many clinical manifestations of DS. This is difficult because chromosome 21 has more than 200 protein-encoding genes that can directly and indirectly affect the homeostasis of cells, tissues, organs, and systems. This complexity presents difficult challenges in understanding the molecular basis underlying each of the many clinical features of DS, but underlies the development and function of many cell types, tissues, organs, and systems. It also provides an opportunity to better understand the genetic mechanisms. Since Trisomy 21 was first described, we have learned a lot about the intellectual disability and genetic risk factors of congenital heart disease. The low incidence of solid tumours in DS patients supports the identification of genes on chromosome 21 that prevent cancer when overexpressed. The universal occurrence of histopathology of Alzheimer's disease and the high prevalence of dementia in DS provide insights into the pathology and treatment of Alzheimer's disease. Clinical trials to improve the intellectual disability of DS represent a new era in which therapeutic interventions based on knowledge of the molecular pathophysiology of DS can be explored. These efforts give us a legitimate hope for the future.

There are three types of Down syndrome. Because their physical characteristics and behaviors are similar, it is often difficult to tell the difference between each type without looking at the chromosomes. Trisomy 21 approximately 95% of people with Down syndrome has trisomy 21. In this type of Down's syndrome, each cell in the body has three separate copies of chromosome 21 instead of the usual two copies. Translocation Down Syndrome this type accounts for a small proportion (about 3%) of people with Down syndrome. This occurs when some or the entire extra chromosome 21 is present, but attached to another chromosome or instead "trans-located". That it is another chromosome 21. Mosaic Down Syndrome this type affects about 2% of people with Down syndrome. 2 Mosaic means a mixture or combination. In children with Mosaic Down Syndrome, some cells have three copies of chromosome 21, while others have two typical copies of chromosome 21. A child with Down syndrome can have the same characteristics as any other child with Down syndrome. However, the disease may be less characteristic due to the presence of a small number (or many) of cells with a typical number of chromosomes.

Regardless of the type of Down syndrome capable of having a person, all people in the Down syndrome may have an additional important part of the chromosome 21 present in all or part of those cells. This additional genetic material changes the development sport and causes characteristics associated with Down syndrome. The cause of additional full chromosomes or partial chromosomes is not yet known. Mother's age is connected with the only factor associated with elevated opportunities, Down syndrome caused by the lower junction or mosaicism. As the birth rate of recent women is high, 80% of the Down syndrome is born in women under 35 years old. There is no scientific re-

search that revealed Down syndrome during pregnancy or during pregnancy causing environmental factors and parents' activities. Additional partial or all copies of the 21st chromosome that causes Down syndrome can come from fathers and mothers. About 5% of cases were persecuted by his father.

Diagnostic tests are usually performed after a positive screen inspection to check the diagnosis of Down syndrome. The type of diagnostic test is as follows. Chorionic Villus Sampling (CVS) examines material from the placenta. Amniocentesis examines the amniotic fluid (the fluid from the sac surrounding the baby). Percutaneous Umbilical Blood Sampling (PUBS) examines blood from the umbilical cord. These exams are looking for chromosomal changes that specify Down syndrome diagnosis.

Treatment of patients with Down syndrome is interdisciplinary. Karyotype analysis should be performed on newborns with suspected Down syndrome to confirm the diagnosis. The family must be referred to a clinical geneticist for genetic testing and counseling of their parents. Parental education is one of the most important aspects of treating Down's syndrome, as parents need to be aware of a variety of related conditions so that they can be properly diagnosed and treated. Treatment is basically symptomatic and complete recovery is not possible. These patients need to have their hearing and eyesight tested and are prone to cataracts, so timely surgery is needed. Thyroid function tests are performed annually and should be treated appropriately if interrupted. Although nutritional problems improve after heart surgery, optimal growth and weight gain require a balanced diet, regular exercise, and physiotherapy. A cardiac referral should be sent to all patients, regardless of the clinical signs of congenital heart disease. If congenital heart disease is present, it should be corrected within the first 6 months of life to ensure optimal growth and development of the child. Other disciplines involved include developmental pediatricians, pediatric respirators, gastroenterologists, neurologists, neurosurgeons, orthopedic surgeons, pediatric psychologists, physiotherapists, occupational therapists, speech therapists, Includes auditors. Down's syndrome is caused by the presence of extra chromosome 21. This is the most common pattern of human malformations. People with Down syndrome need the same screening and care that is available to everyone. Children with Down syndrome are at increased risk of developing certain birth defects, and adults with Down syndrome are at increased risk of developing certain health problems. Links to online man pages are provided for the most common issues. The daily health care recommendations for adults are listed above.