

Down Syndrome; The Trisomy of Chromosome 21

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Down syndrome, the trisomic effect of the 21st chromosome, is seen more frequently and continually increasing. DS is becoming more prevalent in society creates many troubles for individuals born with the trisomic effect, along with families of such individuals. DS patients demonstrate many physical characteristics that coincide with the many medical illnesses that each individual suffers. Maternal age is the only factor known to contribute to children developing DS. And as one would guess, as maternal age increases, so does the probability of conceiving a child with DS. The purpose of this review is to examine DS as a whole. To provide the reader with some insight and to recent advances in medicine taking place to benefit individuals diagnosed with the trisomic effect. Throughout the article we will discuss recent advances and look into the steps being taken to develop a better understanding of the trisomy that occurs at the gene Hsa21.

Introduction

Down syndrome, DS, is considered the most common genetic contribution to developing a learning disability. Cases of DS relate back to well over a century. Down syndrome is a result of what is called a trisomy of chromosome number 2; or also known as Hsa21, in the patient. DS contributes to a number of different disabilities, which can include mental retardation. Most often individuals suffer from heart defects, as well as early onset of Alzheimer's disease, and an increased risk of childhood leukemia. Most individuals with DS show an onset of intellectual disease consisting of language deficiencies, and memorization disabilities. "Neurological phenotypes associated with DS include increased incident of seizure disorder in relation to the general population, motor and oculomotor dysfunction and a virtually universal incidence of a neuropathology indistinguishable from Alzheimer's Disease"(Boda et. al 2012). This is just a couple of the characteristics associated with the disease. Many physical characteristics are also caused by the trisomy of the 21st chromosome, which are seen in almost every patient. Studies show that the leading cause for the

trisomic event occurring on chromosome 21 relates back to the maternal age of the mother.

The enigma of trisomic events relating to the case of Down syndrome show that maternal age is the only known factor determining whether a trisomy of chromosome 21 has the probability of occurring. "The incidence of trisomy is influenced by maternal age and differs between population (between 1 and 319 and 1 in 1000 live births are trisomic for Hsa 21)" (Wiseman et. al 2009). DS has been recognized for over a century but advanced maternal age was established as an occurrence for over 50 years. (Gaulden 1992)

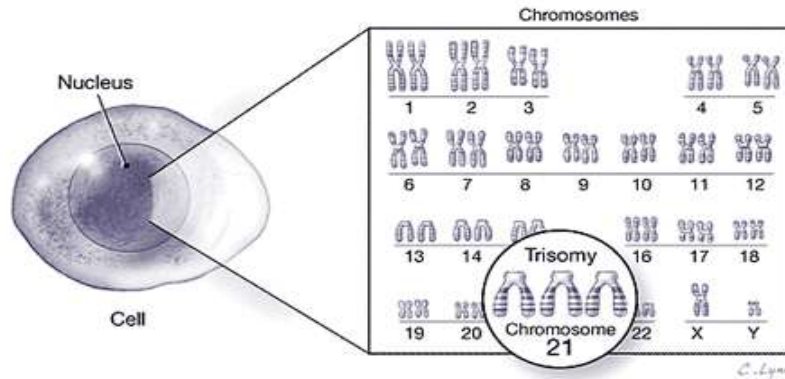


Figure 1. (a). Picture showing what the trisomic effect to the Hsa 21 genes looks like when viewed on a gene map. (*Trisomy of 21st Chromosome*. N.d. Photograph. *Gulf MD*. Web. 7 Oct. 2012. <http://www.gulfmd.com/pregnancy/downsyndrome.asp>)

Recent Progress

Many advances are constantly being made in science to develop a better understanding of DS and the effects it causes in many infants each year. The central goal of the research dealing with DS patients relates to understanding the gene Hsa21. Depending on the expression of Hsa21 determines the phenotypes of the DS individual. Two distinct approaches are being taken to address the issues. Genomic association studies along with animal model of the trisomy of Hsa21 have been established. Utilizing mice as strains of the Hsa21 gene both dosage sensitive genes can be mapped out and develop understandings relating to the pathological mechanisms (Wiseman et. al 2009). Learning and memory problems are common characteristic of all DS patients. “Over expression of a number of Hsa21 genes, including DYRK1A, synaptotagmin 1, and single-minded homologue 2 (SIM 2), result in learning and memory defects in the mouse models, suggest that trisomy of the genes may contribute in learning disability in people with DS” (Wiseman et. al 2009).

Leukemia in children is another major issue that is seen in patients with DS. The onset of learning disabilities is another major issue that is constantly being reviewed to developing a better understanding as to why this trisomy of the 21 gene causes so many irregularities to the newborn.

Discussion

Even though research is being performed each day to develop a better understanding as to why DS occurs there is still is no definite understanding of DS and the effects it causes for many people in today’s society. Down syndrome has an ongoing history as we know, but the only understanding of this defect is that it is a trisomic

event that occurs on the 21 chromosome producing three copies of the chromosomes instead of the normal two. When researching the causes of Down syndrome the only evidence shown to affect this trisomy is maternal age. Research shows that paternal age plays no factor in the occurrence of this disease; it solely relies on maternal age. As the maternal age increases the likely-hood of producing offspring with DS greatly increases.

This brings about one question amongst many. How does an individual try and control the outcome of producing an offspring with DS? The only way to decrease the opportunity of producing such offspring is to decrease maternal age. In today’s society many are having offspring later in life because by this time they are usually more financially stable and have established a base to start a home.

As each day continues research to possibly develop a cure for this disease is being sought out. At this point the only understanding we have of the disease is that maternal age plays a role in the trisomy of the 21st chromosome. Therefore scientist only suggestion is to try and conceive children early in life because as life progresses so do your chances of possibly producing a child with DS.

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