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Genomic Structure of Down Syndrome

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Down syndrome is caused by trisomy 21. The individual has three copies (trisomy) of chromosome 21, instead of two copies, in all cells. This trisomy condition is caused by the abnormal cell division during the development of the egg cell or sperm cell. Chromosome 21 is the smallest chromosome and it contains 200 to 300 genes in humans [1].

Down syndrome incidence are ranging from 1 in 650 to 1 in 1000 live births depends on the population. In an analysis of the chromosome it is revealed that there are 127 known genes, 98 genes are predicted genes and 59 genes are pseudogenes [2]. The genetic abnormality in the individual involves in the production of increased amounts of genes on chromosome 21 which overexpress in the cells and tissues of Down syndrome individuals, showing abnormalities of phenotipic. Because half of the individuals will have a normal heart; this suggests that genetic modifiers interact with the dosage of sensitive genes on chromosome 21 and which results in congenital heart disease.

In the cytogenetic investigation all the individuals are suspected with Down syndrome is very important to establish a diagnosis and it is mandatory to determine the recurrence risk of the syndrome in forthcoming generations [3].

Free trisomy 21 occurs as a sporadic event and recurrence is rare. When recurrence of the Down syndrome exists, the hypotheses are gonadal mosaicism which is a parental predisposition to nondisjunction, it effects on environmental factors and endogenous factors may also play a role.

For Mosaic trisomy 21, two different mechanisms were described for the formation of mosaicism: one is a mitotic error in a normal, euploid zygote resulting in a mosaic embryo having 46/47,+21 karyotype, the 45,-21 cell line being nonviable, and the other one is a nondisjunction in parental gametogenesis followed by an early postzygotic malsegregation of chromosome 21 ("tri - somy rescue"). A significant proportion of the mosaic parents have been considered as trisomics.

Down Syndrome and Congenital Heart Diseases

The first report on association between Down syndrome and malformation of heart was in 1894 and the first correlation between AVSD (Atrioventricular Septal Defects) and Down syndrome has been suggested 25 years later [4].

About half of the individuals with Down syndrome have congenital heart disease. One of the major causes of morbidity and mortality, and the spectrum of congenital heart disease pattern varies widely, encompassing any structural abnormality in the heart and great vessels. The most common defects are found in the Atrioventricular septal. About half of the Atrioventricular Septal Defects occur in patients with Down syndrome [5]. Even though trisomy 21 is a risk factor for congenital heart disease it is not a sufficient requirement (60% of the individuals with trisomy 21 do not have congenital heart disease), so it is important to identify the susceptible genes. Understanding the genes and their responsible steps of cardiac morphogenesis is necessary which may help to define a better framework of embryologica in all these aspects.

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