

Challenges and Future Perspectives of Down Syndrome and Chromosomal Abnormalities

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ABSTRACT

Down syndrome comes with birth defect and it is also comes with huge social and medical cost. This genetic disease is most prevalent and common genetic of disabilities in new born.

Albeit the syndrome had been described thousands of years afore, it was designated after John Langdon Down who described its clinical description in 1866.

More recent progress has resulted in the development of noninvasive prenatal screening (NIPS) test using cell-free fetal DNA sequences isolated from a maternal blood sample. A review on those achievements is discussed. These advances in turn may help to develop targeted therapy for persons with trisomy 21. Screening for DS is an important part of routine prenatal care. Until recently, noninvasive screening for aneuploidy depends on the measurement of maternal serum analytes and ultrasonography.

KEYWORDS: Down Syndrome, Trisomy 21, Chromosome Abnormality

INTRODUCTION

Down syndrome is customarily caused by an error in cell division denominated "nondisjunction" that leads to an embryo with three facsimiles of chromosome 21. This type of DS is called trisomy 21 and accepted to be the major cause of DS, accounting for about 95% of cases (20, 21). Since the tardy 1950s, scientists have additionally determined that a more minute number of DS cases (proximately 3-4%) are caused by chromosomal translocations. Because the translocations responsible for DS can be inherited, this form of the disease is sometimes designated as familial DS. In these cases, a segment of chromosome 21 is transferred to another chromosome, customarily chromosome 14 or 15. When the translocated chromosome with the extra piece of chromosome 21 is inherited together with two mundane facsimiles of chromosome 21, DS will occur. For couples who have had one child with DS due to translocation trisomy 21, there may be an incremented likelihood of DS in future pregnancies. This is because one of the parents may be a balanced carrier of the translocation. The chance of passing the translocation depends on the sex of the parent who carries the rearranged

chromosome 21. If the father is the carrier, the jeopardy is around 3 percent, while with the mother as the carrier, the jeopardy is about 12 percent. This difference is due to the fact that it seems to be a cull against chromosomal abnormalities in sperm engenderment which betokens men would engender fewer sperm with the erroneous quantity of DNA. Translocation and gonadal mosaicism are types of DS kenneed to have a hereditary component and one third of them (or 1% of all cases of DS) are hereditary (1, 22). The third form of disease denominated mosaicism, is a recherche form (less than 2% of cases) of DS. While kindred to simple trisomy 21, the difference is that the third facsimile of chromosome 21 is present in some, but not all cells. This type of DS is caused by eccentric cell division after fertilization. In cellular mosaicism, the cumulation can be optically discerned in different cells of the homogeneous type; while with mosaicism, one set of cells may have mundane chromosomes and another type may have trisomy 21 (1, 22).

CONCLUSION

DS is a birth defect with immensely colossal medical and convivial costs and at this time there is no medical remedy for DS. So, it is obligatory to screen all enceinte women for DS.

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