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Non- classical Down's syndrome: prevalence and importance

Priyanka Pandey
Hind institute of medical sciences, sitapur, india

Statement of the problem: Down syndrome constitutes the most common chromosomal abnormality among live births (1 in 730 live births) and most frequent form of intellectual disability. Genetic cause for this syndrome is trisomy of chromosome 21. The cytogenetic profile of Down syndrome includes free trisomy 21, Robertsonian translocations, mosaicism. Though the most common variant of Down's Syndrome is free trisomy which arises due to meiotic non- disjunction, Robersonian Translocation and Mosaicism also constitute important variant of this chromosomal aberrations. Robertsonian Translocation occurs between two acrocentric chromosomes most common being translocation 14,21 followed by translocation 21,21. This type of translocation occurs when the breakage of chromosome occurs at the centromere and the long arm of two chromosome fuse together while the fused short contain negligible genetic information, so gets degenerated. DS due to translocation can be de novo or inherited from a balanced carrier parent. In general, Robertsonian translocations carriers are phenotypically normal. In 50% of these cases of the rearrangements occur de novo and 95% of the de novo cases originate at the time of maternal meiosis. Around 3-5% of DS cases occur due to mosaicism which involves a non-disjunction postzygotic event. Indivduals with mosaic Down syndrome have two distinct cell lines with different karyotype. Some cells have a total of 46 chromosomes with normal karyotype, while others have trisomy in chromosome 21.There is no positive correlation between the maternal age and incidence of non- classical variant of Trisomy 21.

Biography

Dr. Priyanka Pandey completed her MD in human anatomy from King George's Medical University, India. Presently she is involved in teaching the medical undergraduates and research work at Hind Institute of Medical Sciences, India. She has 11 publications to her name.