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Neural tube defects and their involvement with mutation in stem cells

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Introduction: NTDs, one of the most severe problems of CNS. Epidemiological studies reveal that the incidence of NTDs is highly variable 0.2-3.5 per 1000 births in different geographical region. During embryogenesis the role of genetic/epigenetic factors and stem cell (Oct4, Nanog3 & Sox2) mutation in etiology of closer of neural tubehas not been clearly defined.

Objectives: The curiosity has been developed with the aim to evaluate stem cell gene mutation in term of their expression and their interaction with MTHFR and GST gene polymorphism using specific forward/reverse primers of three germ layers. RTPCR based DNA analysis were carried out to evaluate the gene expression and for mutation analysis DNA sequencing by Sanger's method.

Results: Interestingly, variable the complete disappearance of complete Sox2 gene (16.00%) and over expression Nanog (48.00%) & Oct4 (40.00%) and down regulation of Nanog3/Sox2 observed in Lumbosacral myelomeningocele (LMMC) cases of NTD. The odd ratio was also computed at 95% CI varies between 0.132-2.31 (0.005-1.29) & (0.424-13.812) between cases and controls. The variable frequency of stem cell gene expression is due to severity of disease between encephalocele, anencephaly and myelomeningocele, differences in the allele frequency of MTHFR between homozygous TT genotype (12.00%) and heterozygous CT (28.00%) conditions. Statistical analysis showing significant ($p<0.001$) differences and "T" allele observe as"risk factors" (CC vs. CT+TT) in NTDs.

Conclusion: Variations in stem cell gene mutation may be due to heterogeneous group of population varying in age, sex and different ethnicity, environmental genetic susceptibility, inappropriate dietary habit with low concentration of folate/vitamins due to poor socio-economic status and lead to decrease free radical scavenging capacity to increase cellular toxicity based on antioxidants.

Biography

Professor Ajit K. Saxena has received his Ph.D. (Cytogenetic & Molecular Genetics) from Institute of Medical Sciences, Banaras Hindu University,Varanasi, (U.P) in 1989/90. After receiving his Ph.D. degree he joined NIH funded Indo - US project at AIIMS New Delhi where he identified the role of novel antigen IL6 as a signal traducing agent in human glioblastoma. Dr Saxena, retains more then 24 years of academic (teaching / research) experiences in various renowned Institutions of India and abroad. Currently, he is working as a Professor of Clinical Genetics and Head of the Department of Pathology & Laboratory Medicine in AIIMS Patna. Of course, he has published more than 100 articles with high citations. Based on creditability in academics' he has received several awards and honours, including Gold Medal Award, Confer 'Vivek Ratan Award' and Millennium Award from USA and Excellence Award in 2014.

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