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Etiopathology of Wilms' Tumor in India: Genetic heterogeneity and increasing risk factor involving novel gene mutations of *MTHFR* gene polymorphism

Wilms' Tumor (WT) is one of the rare tumors of pediatric age group. The etiopathology of WT is complex due to interactions between genetic and environmental factors. Genetic factors include deletion of the q13 region of chromosome-11. The functional aspect of loss of genetic material is unknown but might play a significant role in the process of malignant transformation. "Risk Factors" associated with the development of WT still remain elusive. The rationale behind this study was to identify new mutations and associated "Risk Factors" in the population of Eastern part of India, an area highly susceptible for development of cancer. We performed studies at cytogenetic and molecular level to identify the genetic abnormalities associated with WT cases in comparison with the respective controls. Interestingly, a variety of different karyotypes were observed in the cases of WT that includes the loss of Y-chromosome, presence of ring chromosomes, translocation and numerical (monosomy, trisomy) variations. Further, we evaluated the frequency of *WT1/WT2* gene mutation in cases and controls. PCR based analysis demonstrated that the frequency of WT1 (15%) mutation was three times higher than WT2 (5%), which may be due to interaction with environmental factors, or unknown reasons. Further, we performed Methylene Tetrahydrofolate Reductase (*MTHFR*) C677T gene polymorphism analysis to assess the genetic heterogeneity in WT cases using Amplification refractory mutation system (ARMS)-PCR, where allele "C" changes to "T" (C→T), increasing the "Risk" of the disease. Interestingly, DNA sequencing identified "novel mutations" in *MTHFR* gene, which has not been reported earlier in cases of WT. Since WT is an embryonic tumor, there may be an involvement of dysfunction of stem cells, therefore we further performed mutational studies to characterize early transcription factors (Sox-2, Oct-4, and Nanog) in WT cases. Such studies were found to be relevant to explore the transforming ability and activation of oncogenes during subtle changes observed during cytogenetics study (deletion, frameshift mutation, translocation and point mutation). The study showed differential expression of Oct-4, and mutation of Sox-2 in WT cases.

Biography

Ajit K Saxena has received his PhD (Cytogenetic & Molecular Genetics) from Institute of Medical Sciences, Banaras Hindu University, Varanasi, (U.P) in 1989/90. After receiving his PhD 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. He retains more than 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 honors, including Gold Medal Award, Confer 'Vivek Ratan Award' and Millennium Award from USA and Excellence Award in 2014.

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