BASELINE FREQUENCIES OF SPONTANEOUS CHROMOSOME ABERRATIONS, SISTER CHROMATID EXCHANGES AND MICRONUCLEI IN HUMAN NEWBORNS

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A cytogenetic monitoring programme on human newborns was carried out from 1983-1987, with a view to find out the incidence of constitutional chromosome anomalies. During this period, studies were also undertaken to obtain baseline data on spontaneous chromosomal aberrations (CA) sister chromatid exchanges (SCE) and micronuclei (MN) in limited number of human neonates. We report here the data on mean frequencies of spontaneous chromosome aberrations in the lymphocytes of hundred and twenty newborns, sister chromatid exchanges in ten newborns and micronuclei in the lymphocytes of six newborns studied. The results are compared with data obtained in our laboratory on spontaneous chromosome aberrations and sister chromatid exchanges from adult blood lymphocytes. The micronuclei data is compared with the data from adult lymphocytes by the cytokinesis blocked lymphocyte assay by cytochalasin B.

The low incidence of spontaneous chromosome aberrations, SCEs and micronuclei reported in the present study may possibly reflect the influence of high folate levels in cord bloods on spontaneous chromosomal damage.

The present study will provide baseline data and accurate baseline data on spontaneous chromosomal aberrations, sister chromatid exchanges and micronuclei, which are sensitive cytogenetic end points in the assessment of genetic damage, are important in monitoring human population exposures to low levels of occupational and environmental genotoxicants.

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MOSAICISM INVOLVING CHROMOSOME NUMBER 17 p ARM DELETION IN A PREMATURE CHILD

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During a cytogenetic monitoring programme on human neonates, for constitutional chromosome anomalies, cord blood studies revealed abnormal karyotypic findings in a premature child. Two different cell populations were observed, one having normal chromosome 17 and the other having a chromosome 17 with a p arm deletion. 7% of the cells analysed showed the p arm deletion. Proband was the product of a para II pregnancy of a 20 year old mother. Para I was also a premature child with cerebral palsy. The family is being currently investigated in detail.

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AN EVALUATION OF SPONTANEOUS AND GAMMA RAY INDUCED CHROMOSOME DAMAGE IN BETA THALASSEMIA TRAITS BY CYTOKINESIS BLOCKED LYMPHOCYTE MICRONUCLEUS ASSAY.

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An earlier report on the increased chromosomal anomalies in lymphocyte cultures from beta thalassemia homozygotes and heterozygotes as well as the fact that beta thalassemia heterozygote frequency encountered in some communities in the Indian population is as high as 17.5% prompted us to undertake this study.

Spontaneous micronucleus frequency was determined in normal and beta thalassemia traits (heterozygotes) in cytokinesis blocked (CB) binucleate lymphoblasts accumulated by the addition of cytochalasin B at 44 hrs in 72 hr cultures. In addition comparison is made of yields of radiation induced micronuclei in CB lymphoblasts from normal individuals and thalassemia traits. The frequency of induced micronuclei in peripheral blood lymphocytes irradiated **in vitro** in whole blood with Cobalt 60 gamma rays at 2 gy was studied.

The **in vivo** micronucleus frequency in beta thalassemia traits was found to be almost two times higher than in normal individuals. Significantly higher number of miconuclei were induced in irradiated lymphocytes from thalassemia traits compared to normal individuals. The values were still found to be significant, even after the baseline number of micronuclei is substracted from the number expressed after radiation exposure.

However there was not necessarily a strong correlation between the number of lymphoblasts with micronuclei and the degree of chromosomal damage as evidenced from studies in our laboratory on spontaneous as well as radiation induced chromosome aberrations in beta thalassemia traits. The implications are discussed.

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MONOSOMY OF CHROMOSOME 16q 24 - A CASE REPORT

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A new case of chromosome 16 long arm deletion, 46, XX del (16) (q24) is reported , which seems to be inherited from a phenotypically normal father. Referral was at 1 year and 3 months of age with parental concern about delayed development. The physical examination of the propositus revealed widely open anterior fontonalle, marked fronto-parietal bossing, low set ears, tortuous vein all over the skull with very sparse hair on scalp, marked ichlhyosis on abdomen and lower chest and a small umblical hernia. Skeletal radiographs were unremarkable. There was no hypotonia. The analysis of parental karyotypes revealed a normal karyotype in the mother and a deleted long arm of chromosome 16 in the father. The deletion in the phenotypically normal father could be attributed to different breakpoints on the 16 q24 region or it could be a translocation, the identification of which might be difficult at the current detection level. The proband is phenotypically different from the previously reported cases of typical 16q deletion syndrome and differ from them in the location of the band detection.

Erythrocyte APRT activity as well as hybridisation studies using APRT gene may provide a more precise cytogenetic assignment of the long arm break point in the proband and her father, since the gene for adenosine phosphorybosyl transferase has been assigned to 16q 24 by Southern analysis and in situ hybridisation studies.

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UNCERTAIN BREAKPOINT ASSIGNMENT FOR A BALANCED TRANSLOCATION INVOLVING CHROMOSOMES 19 AND 20 IN A NEWBORN

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During a cytogenetic monitoring programme on human neonates for constitutional chromosome anomalies, we observed a balanced translocation involving chromosomes 19 and 20 in cord blood lymphocytes. The probands karyotype was designated as 46, XX t (19:20). She was the product of a G2P2 uneventful pregnancy of a 22 yr. old mother with no history of genetic problems. Follow up of the case revealed physical and mental milestones to be normal. Considering the limitations in the banding pattern the breakpoints were interpreted as 46, XX t (19:20) (p 13.1 q 13.3.). A more precise breakpoint assignment of this balanced translocation can be carried out by molecular techniques using quantitative hybridisation, dosage studies and in situ hybridisation and further chromosome analysis. Subsequent family studies can reveal whether the translocation is a de novo one.

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