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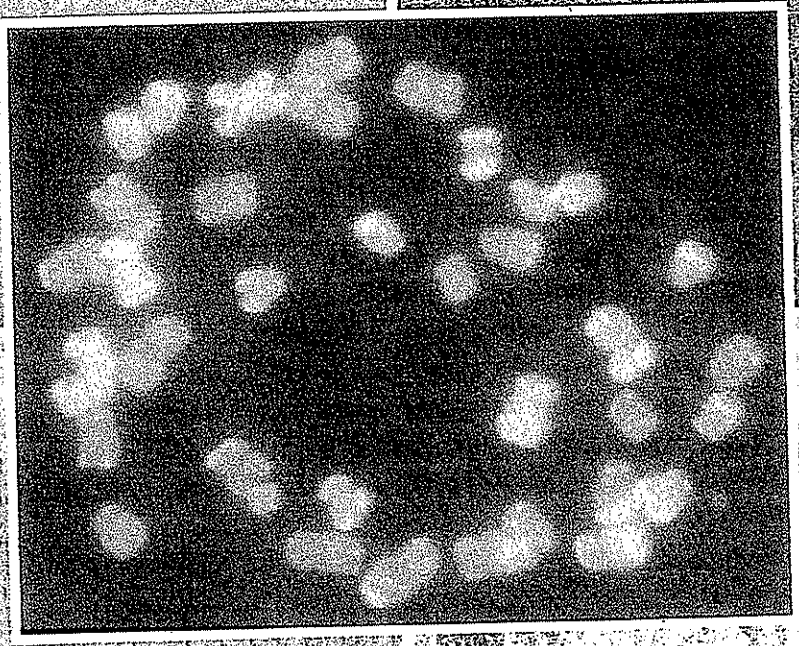
CHROMOSOME RESEARCH

Cytogenetics, Genomics, Chromatin and The Nucleus



SPECIAL ISSUE

The Cytogenetics and Genomics of Crop Plants

Guest Editors: Ingo Schubert and Andreas Houben

that this extra segment originated from chromosome 22. DGS probe (D22S75) was used to identify the breakpoints on chromosome 22. The signal for q11.2 was seen on the extra segment of chromosome 22 additionally to the normal signal. To our knowledge, 6 cases with partial or full phenotype of CES associated with an interstitial duplication of 22q11.2 have been reported. Our case is interesting because of major features of CES and the presence of a dicentric structure of the derivative chromosome 22, which has not been reported before.

1.91-P

Deletion of 8p: a case of a child with multiple congenital anomalies

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Deletions of the proximal part of 8p (8p11 to 8p21) are typically associated with congenital spherocytosis, which is most the common of hereditary hemolytic anemias. Genes for ankyrin and glutathione reductase (GSR) were localized to chromosome bands 8p11 and 8p21, respectively. Also associated with the deletion of the short arm of chromosome 8 are postnatal growth retardation, microcephaly, subnormal mentality, epicanthal folds, malformed ears, brevicollis, widely spread nipples, heart defects, and other abnormalities. Individals with 8p- are reported to share a distinctive pattern of clinical features. This has led several authors to propose a distinct 8p deletion syndrome phenotype.(Dobyns *et al.* 1985, Qstergaard and Tommerup 1989, Pecile *et al.* 1990, Claeys *et al.* 1997) We describe a 8 month-old boy with multiple congenital anomalies (microcephaly, cleft lip and palate, clinodactyly, micropenis etc.) with spherocytic anemia. Analysis of conventional G-banded metaphase chromosomes from short term lymphocyte cultures of the patient showed a small interstitial deletion on the short arm of chromosome 8: 46.XY,del(p11p21) Both parents had normal karyotypes. Prior reports of 8p deletions have shown mostly to be de novo mutations. In the future, Fluorescence in situ hybridisation will be performed with whole chromosome 8 painting probe for the chromosome mapping of break points.

1.92-P

Result of 4680 cytogenetic studies: a retrospective study in karyotype abnormalities

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Introduction: As a genetic center serving mostly for infertile couples, the results have significance for assisted reproduction. Here, we present our results of prenatal and postnatal samples.

Materials & Methods: Results of 4680 cytogenetic studies performed in a period of 7 years were included in this study. Karyotyping was performed by using conventional cytogenetic methods such as GTG and C-banding. Investgations on amniotic fluid, CVS and abortion material were carried out on long-term cultures. 3660 samples for peripheral blood (PB), 714 samples for prenatal diagnosis (PND) and 268 miscarriage materials were studied. Analysis of 714 cases for PND, 598 cases from amniotic fluid, 64 cases from CVS and 52 cases from cord blood were performed.

Results: Chromosomal aberrations were found in 421 of 3660 cases (11,5%) in PB studies. 238 patients (6,5%) have gonosomal and 183(5%) patients have autosomal abnormalities. The frequency of gonosomal abnormalities for Klinefelter's Syndrome, 46,XX males, 46,XY female and cases with mosaic sex chromosome abnormalities were 54.2%(129), 7.5%(18), 1 and 33.2%(79), respectively. The number of structural abnormalities for Reciprocal and Robertsonian translocations (RecT, RobT), double translocations, insertional translocations, and inversions were 75(41%), 31(17%), 6(32.8%), 3(1.6%), 54(29.5%), respectively. 44 chromosomal aberrations were found in 714 cases after PND. 26 out of 44(%3,6) have numerical and 18(%2,5) have structural abnormalities. Trisomy 21 was the most frequent numerical abnormality and RecTs were the most frequent structural abnormality. 92 out of 268(%34,3) miscarriage materials carried chromosomal abnormalities. The most frequent abnormality was trisomy 16(%15,2) and two cases had double trisomies.