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Human Genetics

Cases Observed

De Novo Duplication 13q (46,XX,dup(13)(q21→q333))

A female born 10.12.1977, birth weight 3250 g. Ascertained because of trigonocephaly. Low hair implantation, bilateral epicanthus, hypertelorism, malformed ears, high arched palate, bilateral clinodactily of the fifth toes. Severe psychomotor retardation. Diagnosis by G- and Q-bands. Q- and NOR-bands were not informative for the parental derivation of the abnormal chromosome. Dermatoglyphics in progress.

We would like to receive from or to send to colleagues full information for eventual joint publication on similar cases.

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Three Cases of Familial Pericentric Inversion 2

Case 1: A 25-year-old mentally retarded female with congenital heart disease (VSD) showed a pericentric inversion inv(2)(p13q21). This was inherited from her healthy mother.

Cases 2 and 3: Two 37-year-old healthy women were referred to amniocentesis because of advanced age. Fetal karyotypes were respectively 46,XY,inv(2)(p11q13)mat and 46,XX,inv(2)(p11 q13)pat.

We would propose a collaborative study on the clinical significance and the genetic risk of pericentric inversions of chromosome 2. Information on further observations on this apparently frequent chromosome rearrangement for joint publication would be appreciated.

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Familial Translocation t(1;17)

A reciprocal translocation t(1;17)(q12;p11.3) was detected in a 23-year-old male who had coarctation of the aorta subclavia. The proband's sister and her daughter have also been shown to be a carrier of the balanced translocation. The father of the proband was deceased but he must have been a carrier of the translocation. A first cousin of the proband had a daughter who was reported to be statomotorically retarded while her brother had an abnormal configuration of head. Further analysis of the family is planned.

Our data are available for a study on the frequency and the segregation ratios of chromosomal rearrangements involving such heterochromatic regions.

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Partial Trisomy 13 (q14→qter) Due to a Familial Translocation t(13;18)(q14;q23)

A newborn female with multiple malformations suggestive of Pätau's syndrome had the karyotype 46,XX,der(18),t(13;18)(q14;q23)pat. She died of bacterial sepsis at the age of one month. Autopsy was not allowed. The paternal aunt was also carrier of the translocation which must have been inherited from the grandmother. Prenatal diagnosis was performed during the next pregnancy of the patient's mother. The karyotype of the fetus was 46,XY,der(18),t(13;18)(q14;q23)pat. He had multiple external and internal malformations including brachycephaly, bilateral epicanthus, malformed ears, hexadactyly, vitium cordis, and cystic kidneys.

We would like to send to or receive from colleagues complete information on this chromosomal disorder for a collaborative study to further delineate the associated clinical syndrome. Fibroblasts are also available for biochemical studies of gene dosis effects.

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