Trisomy for 8p21→pter owing to a familial translocation

SUMMARY A girl with developmental delay and physical abnormalities was trisomic for the segment 8p21→pter owing to a familial translocation t(8;11). The child's father and paternal grandmother carry the same translocation.

Case report

In 1979, as part of a counselling work-up following three spontaneous abortions (at 10 weeks, 5 months, and 10 weeks), karyotypes using the Giemsa-trypsin banding method were obtained in III.2, a male aged 26 years, and in III.1, a female aged 24 years (fig 1). III.2 was found to be the carrier of an apparently balanced translocation t(8;11) (p21;q25), shown in fig 2 (a,b). The possibility of monitoring further pregnancies by amniocentesis was rejected by the couple. Subsequently, a pregnancy was carried to 34 weeks and resulted in the birth of a female (IV.1) (fig 3). Birth weight was 2060 g, length 43 cm, head circumference 32 cm, and Apgar scores 7 and 7. Features present at birth included hypotonia, hydrocephalus, ventricular septal defect, and coarctation of the aorta. She was operated on at 2 months of age for the cardiac defects. Other dysmorphic features seen included short up-turned nose, a long philtrum, carp mouth, frontal bossing, hypertelorism, micrognathia, and small low set ears with a prominent helix. Her palate was normal. Evaluation at 10 months showed her to weigh 7.3 kg with a head circumference of 47.5 cm. She was alert and responsive with no evidence of hearing or sight loss. Her developmental level was 12 to 16 weeks behind her chronological age. She still showed signs

FIG 2 (a) Diagram of t(8;11)(p21;q25). (b) Chromosomes 8 and 11 of II.2. (c) Chromosomes 8 and 11 of IV.1.

FIG 1 Pedigree of family.

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FIG 3 IV.1 with trisomy 8(p21→pter).

of hypotonia and weakness. Her karyotype by the Giesma-trypsin method was 46,XX,-11,+der(11)t(8;11)(p21;q25)pat (fig 2c).

The family history on the father's side included

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two other severely retarded subjects, a niece (IV.3) and half-brother (III.9) of III.2. The half-brother was reported to have had cleft lip repair surgery. These people were not available for study. Two of the nephews of III.2 died in infancy (IV.5, IV.8), one following heart surgery (IV.5). The mother (II.2) of III.2 was karyotyped and found to have the same translocation as her son.

Discussion

In reviewing reports of trisomy 8p, the only abnormal features our patient has in common with all other cases are mental and physical retardation.¹⁻⁷ Other features are shared with various cases, but no clearly defined trisomy 8p syndrome emerges. Features such as micrognathia, hypertelorism, cardiac defects, low set ears, epicanthal folds, and carp mouth, seen in our case and other cases of trisomy 8p, are often reported with other chromosome abnormalities. The large mouth and broad nose noted by Rethore *et al*⁶ were not seen in our patient.

Perhaps as more cases are examined, a cardinal feature or cluster of abnormalities may be found to be associated with trisomy 8p. The variable features may be the result of partial monosomy for genetic material on the other chromosomes involved in the translocations, in our case the q25 band of chromosome 11.

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Monosomy 22 with humoral immunodeficiency: is there an immunoglobulin chain deficit?

SUMMARY The cytogenetic analysis of a patient with selective deficit of IgA and decrease in IgM, IgE, and IgG is presented. Using trypsin-Giemsa banding the karyotype showed monosomy 22 (45,XX,-22). The interest of this case lies in the rarity of the illness and in the association of monosomy 22 with hypogammaglobulinaemia and selective deficit of IgA, particularly as this chromosome is known to contain genes coding for immunoglobulin chains.

Monosomy of a G group chromosome compatible with survival occurs rarely and there have been only 25 cases of partial or complete monosomy G reported.¹⁻³ Only in three cases^{1,4,5} was the monosomy identified as a chromosome 22. Our report presents the clinical and cytogenetic findings of a female infant aged 11 years with monosomy of chromosome 22, selective deficit of IgA, and decrease in IgM, IgE, and IgG.

Case report

The proband was a female of 11 years, with a weight of 37.7 kg (25th to 50th centile) and a height of 140 cm (10th centile). The head was dolichocephalic with a flat occiput and adenoid facies. She was mentally subnormal (IQ 60) and had genu valgus and splay foot. She had recurring severe respiratory infections.

Immunoglobulins were studied with the quantitative immunoglobulin test kit⁶ with the following results (mg/100 ml): IgG 500-620 (normal range 564-1565); IgM 48 (normal range 53-375); IgA undetectable on five occasions (normal range 85-385). The IgE was studied by radioimmunoassay

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