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 aminocentesis 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 of hypotonia and weakness. Her karyotype by the Giemsa-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...
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 ab-
normal feature 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 chro-
some abnormalities. The large mouth and broad 
nose noted by Rethore et al8 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 chro-
mosome 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 hypogamma-
globinaemia 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 cases1 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 kit 6 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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