Correspondence


Sir,

I read with interest the case report entitled ‘Multiple congenital defects associated with an abnormal unclassifiable karyotype’ by Surana, Hunt, and Conen (Journal of Medical Genetics, 9, 247–249, 1972). The phenotypic abnormalities of that infant included a wide forehead, hypertelorism, a capillary haemangioma, outward and upward slanting of the palpebral fissures, opacities of both cornea, coloboma of the left iris, low set and poorly developed ears, depressed nasal bridge, receding chin, enlarged heart with grade I/VI systolic murmur, hydronephrosis, and abnormal dermatoglyphic patterns with arch fibular patterns on both hallucal regions, distally located palmar triradii (t'), reduced total finger ridge count of 73 and bilateral simian lines. The conventional karyotype analysis showed 46,XY, -G, +E; the extra E-group chromosome was metacentric and resembled a No. 16 chromosome. According to the authors, the fluorescent banding pattern of the chromosomes was not adequate when the Giemsa-stained slides were de-stained and then re-stained with quinacrine. The autoradiographic studies showed that the extra 16-like chromosome had a late replicating nature and there was one late replicating G and two early replicating G chromosomes. The labelling pattern of the three G-group chromosomes indicated the presence of one No. 21 and two No. 22 chromosomes.

Although many of the clinical features of the patient were rather nonspecific for any known chromosomal aberration, there were several features suggesting that this might possibly be a case of trisomy 13 syndrome. These features were capillary haemangioma, coloboma of the iris, low set and abnormal ears, slanting palpebral fissures, congenital heart disease, hydronephrosis, and abnormal dermatoglyphic patterns such as arch fibular patterns of both hallucal regions, simian lines, distally located palmar triradii (t') and a reduced total finger ridge count (Smith, 1970). Autoradiographic findings have further strengthened this possibility. The late replicating nature of the extra 16-like chromosome may represent a translocation chromosome involving a No. 21 chromosome and the long arm of a No. 13 chromosome, since both chromosomes Nos. 13 and 21 are late replicating in nature. It appears that the translocation site is between the short arm of No. 21 chromosome and the distal half of the long arm of an extra No. 13 chromosome. Thus the patient possibly had a partial trisomy 13 due to the translocation. This may also explain the modified clinical features for trisomy 13 syndrome in this patient. Although this possibility cannot be confirmed without fluorescent or trypsin banding staining, it must be considered.

Yours, etc,
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REFERENCE


Sir,

We reported in 1968, two sibships with familial microtia and mental atresia (Ellwood, Winter, and Dar, 1968). The parents of family A, Arab Moslem first cousins, who were related through their fathers had two children with bilateral anotia and mental atresia.

In 1972, Dr R. B. MacKay of the Nazareth EMMS Hospital referred a premature female infant (birthweight 1780 kg) with the identical malformation, to this hospital. She was the only child of the sister of the father in family A and the brother of the mother in family A.

![Diagram](http://jmg.bmj.com/fig.png)