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 corneas, coloboma of the left iris, low set and poorly developed ears, depressed nasal bridge, receding chin, enlarged heart with grade I/VI systolic murmur, hydrenephrosis, 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, hydrenephrosis, 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,
Lillian Y. F. Hsu

Division of Medical Genetics,
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REFERENCE

Sir,

We reported in 1968, two sibships with familial microtia and meatal 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 meatal atresia.

In 1972, Dr R. B. MacKay of the Nazareth EMMS Hospital referred a premature female infant (birthweight 1-780 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.

Fig.

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This further case (IV.13 see Fig.) suggests autosomal recessive inheritance for the malformation.

Yours, etc,
H. Dar and S. T. Winter

Rothschild Hospital and
Aba Khousy School of Medicine,
Haifa, Israel

REFERENCE

Sir,

Your obituary on the late Lionel S. Penrose (Journal of Medical Genetics, 8, 253, 1972) has just come to my attention. Besides my own sense of personal loss, for the unstinting help he gave me during my limited research efforts years ago in the genetics of mental defect, there is the great indebtedness which American science owes him for direction and leadership during the early days of research in human and medical genetics. This was especially so in the uncharted field of mental defect, to which his contribution was truly unique in that it raised fundamental research in this branch of medicine to a level it had never known before; and his methods and techniques, as well as his results, are the foundations upon which present studies are based.

He was indeed most modest, and unselfishly devoted to science, and his kindness and warmth will always be remembered by those of us on this side of the Atlantic; although his humanity knew no boundaries. We will always miss him.

Yours, etc,
Sidney L. Halperin

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