and are capable of engrafting themselves in the host's haemopoietic and lymphopoietic tissues; hence each chimera is immunologically tolerant of its twin. In this case this feature is substantiated by the non-reactivity of one twin to the other in the mixed lymphocyte culture system, and one would expect a skin or organ transplant between the two to survive as well as in the monozygotic twin situation. The difficulty in this particular case is the sexing of the twins. Physically they appear normal male and female children, apart from the undescended testes in the boy, and the sexes are confirmed by buccal smear examination. To date there is no evidence of the embryonic male or female hormones having any effect on the opposite sexed twin in the chimeric situation, and the majority of chimeric twins so far reported have proved to be fertile. However, in cattle, Lillie (1916) suggested that vascular anastomoses in unlike sexed bovine twins were the cause of the free-martin condition, caused by exchange of embryonal hormones affecting the normal sexual development of the female twin. If this were so in this case, and hormonal exchange does take place across the anastomoses, it would be difficult to explain why one twin had hypothyroidism and the other twin was euthyroidic. The answer to this query will have to wait until either PK's testes descend of their own accord, or with surgical assistance.

We wish to thank Dr Ruth Sanger for her help and encouragement on this case, and for confirming and extending our blood grouping results; also Mr M. Pepper for his excellent work on the HL-A typing. Thanks, too, to Dr Roberts for allowing us to study this interesting case.

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A case of ring chromosome G22

Summary. A girl with a G22 ring chromosome is described. There are few physical abnormalities, performance quotient is in the low normal range but verbal skills are much more retarded.

Many patients with ring G chromosomes have been reported since that of Lejeune et al (1964), but until chromosome banding techniques were available the specific identity of the ring was in doubt. Only four cases of definite ring G22, identified by Q- or G-banding, have been published to our knowledge—two by Crandall, et al (1972) and a set of twins by Lindenbaum, Bobrow, and Barber (1973). Three other cases are known, two by B. Noel (personal communication 1975) one of which is a mosaic 46,XY/46,XX,22r, and one by R. Nelson (cited by Lindenbaum et al, 1973).

We wish to report a further patient identified by G-banding who is less retarded than those previously described.

Case report

The patient, a girl, was born in June 1971 and was initially admitted to Alder Hey Children's Hospital at the age of 8½ months with an upper respiratory tract infection and bilateral otitis media. A week previously she had developed measles. The otitis media resolved with ampicillin and she was discharged from hospital after eleven days.

While she was in hospital it was noted that she had distinct epicanthic folds and a rounded face with prominent large ears. Her fontanelle was closed and her head circumference was below the third centile at 42.5 cm. Her weight was on the twentieth centile at 7100 g. The hands, including palmar creases, and feet were normal.

The patient's birth was normal and she was a vertex delivery. The pregnancy was uneventful and she weighed 3000 g at birth. Her mother was aged 25 years and was in good health. Her father was aged 29 years and was employed as a crane driver until six months
before her birth when she had a subarachnoid haemorrhage. A sister of the patient born in 1968 is alive and well.

Since her admission in February 1972 the patient has been followed up in the out-patient clinic. Her weight remains between the 10th and 20th centile, her height between the 40th and 50th centile, and her head circumference at the 3rd centile. She sat at 5 months and walked on her own at 11 months. By the age of 25 months she was toilet trained and was combining words.

At the age of 3 years and 4 months (Fig. 1) she was seen at the Assessment Clinic at Alder Hey Children’s Hospital. At that time on examination there were no neurological abnormalities and her vision and hearing were acute. Her gross and fine motor activities and also some items that represent non-verbal intelligence, were at no more than a 3-year level which was a low average for her age. Verbally however she was lower than this with an understanding of language below a 2½ year level and expressive capabilities at no more than a 2-year level.

A Griffiths Mental Development Test was performed on the patient at the age of 4 years 2 months. Results were as follows: locomotor scale, 4 years; persono-social, 3 years 2 months; hearing and speech, 1 year 10 months; eye/hand coordination, 3 years; performance scale, 2 years 10 months; practical reasoning scale, 2 years 3 months. The resulting quotients were approximately 71 for performance items and 47 for verbal skills. Her speech was noted to be unclear and appeared immature. It was thought that at least part of the patient’s poor verbal performance may have resulted from poor parental stimulation.

Cytogenetic studies

From peripheral blood cultures in 1972 chromosome counts were made on 30 cells. All cells contained 46 chromosomes including three normal G chromosomes, the fourth being represented by a ring chromosome. Investigation of both parents showed they had normal karyotypes.

Cultures from a blood specimen of the patient in 1975 confirmed the original finding, a further 30 cells being scored. Trypsin banding (modification of Seabright, 1971) showed the ring chromosomes to be that of a G22 (Fig. 2) and all other chromosomes to be normal.

Other studies

Blood group studies for groups ABO, CDE, MN, S, P, K, and Fy showed heterozygosity only for the blood group Kell indicating that the locus for this blood group could not be on the deleted portion of G22.

The finger print patterns of the right hand showed four ulnar loops, the index finger being the exception with a swirl. The left hand exhibits two ulnar loops on the index and middle fingers, the other three fingers showing whirls. Both palms have the axial triradii in the thumb position and normal C triradii.

Discussion

Attempts to classify G-deletion phenotypes into two categories have been made both before banding of chromosomes (Weleber, Hecht, and Gibblett, 1968; Warren and Rimoin, 1970) and after, specifically for G ring phenotypes (Crandell et al, 1972; Lindenbaum et al, 1973). The published cases which have been identified as a G22 ring show few common physical features other than microcephaly, retardation of growth, and epicanthus. Other features seen are high arched palate and large prominent ears (see Table). None of them show the syndactyly of toes or hypotonia which were described as specific features of ‘Syndrome II’ by Warren and Rimoin (1970) and which were interpreted by them to be associated with the partial deletion of a G22. Syndactyly of the third and fourth toes was seen in the mosaic case of Noel (1975, personal communication).

The present case agrees with other known G22
ring cases in the microcephaly, with a head circumference initially below the 3rd centile but now at the 3rd centile, and epicanthus which was obvious when first born but has become less apparent as the patient grew older. The ears are large and prominent, but there is no high arched palate. Muscle tone is normal and there is no evidence of strabismus of the toes.

The main interest in this case is the level of intelligence, for all previous G22 ring cases have indicated severe mental retardation. On the Griffiths Mental Development Scale our patient has a performance quotient in the low normal range (71) but her quotient for verbal skills was much lower (47). Though in this case the latter may be depressed by home environment it is in agreement with the pronounced slowness in speech development in other recognized ring G22 cases. The two patients of Crandall et al (1972) had IQ’s of 10 and 22 on the Binet scale, one showing no speech and the other very slow speech development. The twins described by Lindenbaum et al (1973) had IQ’s of 35 to 40 by the Merrill-Palmer test, they were hyperkinetic and often resorted to ‘low-pitched cries than to recognizable words’. The case of Nelson, cited by Lindenbaum et al (1973), and the two cases of Noel (personal communication 1975) also had severe mental retardation. Neither of the two latter cases had any speech.

The intelligence of the girl reported here is within the subnormal range with an overall IQ of 59 and above those cases of ring G22 previously described. As with all chromosomal deletions the possible variable amount of chromatin loss in different cases could account for the variability in expression.

We are grateful to Dr R. J. Derham for access to his patient, to Dr L. Rosenbloom and Miss E. J. Horn for the developmental assessments, to Dr D. W. Fielding for the palm prints, and to Mr W. T. A. Donohoe and Miss P. Ball for technical assistance.

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Familial Ebstein’s anomaly

Summary. A family is described in which both a father and son are affected with Ebstein’s anomaly, while several other family members manifest different cardiac malformations. Five additional instances of familial Ebstein’s anomaly