Case reports

Journal of Medical Genetics 1989, 26, 190–201

Unilateral absence of the hand in second cousins

MARGARET LAMONT* AND A J SALISBURY†
*Department of Child Health, Southampton General Hospital, Southampton SO9 4XY; and †Department of Paediatrics, Princess Margaret Hospital, Swindon SN1 4JU

SUMMARY This paper reports two second cousins with absence of the left hand.

Congenital absence of the hand, acheiria, is a rare defect occurring in 1:65 000 live births. It is always unilateral and almost always sporadic; reports of acheiria recurring in a family are sparse. They include those of Hecht and Scott, in which two sibs born to consanguineous parents were affected, and of Pilarski et al who recorded two affected first cousins. Familial occurrence of acheiria and forearm amputation has been recorded, in sibs, uncle and niece, uncle and nephew, and in a great grandmother and her great granddaughter. However, we know of no previous reports of acheiria occurring in children sharing great grandparents.

Case reports

Case 1 is the younger child of healthy, unrelated parents. His father was aged 33 and his mother 29 years at the time of his birth. Neither they, nor his older sib, have any hand abnormality. His mother had primary amenorrhoea and both pregnancies were induced with clomiphene. The pregnancy was...
uneventful with no illness or medication. Birth weight at term was 2890 g. He was noted at birth to have an absent left hand with rudimentary digits. Apart from this (fig 1) he has no physical defect and is developing normally. His right hand and both feet are of normal size with no digital anomalies. X ray at 17 months (fig 1) showed a normal radius and ulna with two ossification centres in the central region.

Case 2 was born 17 months after case 1 to a first cousin of his mother (fig 2). The two families live some 60 miles apart. She is the only child of a 28 year old father and a 29 year old mother. There was no history of menstrual problems. The pregnancy was uneventful with no medication apart from iron therapy and no history of illness. Her birth weight at 40 weeks' gestation was 2700 g. She was born with absence of the left hand (fig 3).

The defect is similar to that of case 1 but with greater reduction of digital development. She has no other physical defects and, like her cousin, is developing normally. An x ray (fig 3) done just after birth shows a normal left radius and ulna.

No other family members have any known anomalies of their hands or feet. The hands of the mother and maternal aunt of case 1 have been personally examined and show no clinical or radiological abnormality.

Discussion

The occurrence of this identical defect in two cousins suggests a genetic basis. The possibility that both children could have been affected by chance alone, when the incidence of acheiria is 1:65 000, is too remote to be considered. The parents of the two children live in different towns and environmental factors are unlikely to have any aetiological signifi-
Identification and characterisation of a small marker chromosome using non-isotopic in situ hybridisation with X and Y specific probes

JOHN A CROLLA*, MARY SMITH, AND ZOE DOCHERTY

From the Paediatric Research Unit, Division of Medical and Molecular Genetics, United Medical and Dental Schools of Guy's and St Thomas's Hospitals, Guy's Hospital, London SE1 9RT.

SUMMARY A 13 year old male with mild mental retardation, obesity, and poor secondary sexual differentiation was found to have a 46,X,+mar karyotype. In situ hybridisation with X and Y specific probes proved the marker to be composed of Y centromeric and short arm material.

*Present address: MRC Experimental Embryology and Teratology Unit, MRC Laboratories, Woodmansterne Road, Carshalton, Surrey SM5 4EF.

Received for publication 10 June 1988.
Accepted for publication 28 June 1988.

Case report

The proband, a 13 year old mentally retarded male, was referred because of poor secondary sexual development and gynaecomastia. He was thought to be a possible case of Prader-Willi syndrome. He was born at 37 weeks' gestation weighing 1956 g to healthy and unrelated parents. Two sibs, one younger and one older, are both healthy and normal. At two years, the patient's right testis was brought down surgically and the other descended spontaneously at seven years. His development has...