Ring chromosome 1 associated with radial ray defect

The patient was born at 38 weeks' gestation and was very small, weight 1.48 kg (approximately 1 kg below the 3rd centile), length 42 cm (5 cm below the 3rd centile), and head circumference 28 cm (4 cm below the 3rd centile). She had an abnormal face with a sloping forehead, prominent nose, thin lips, and micrognathia. The hands were radially deviated, with absent first metacarpals, and hypoplastic thumbs (fig 1). The right thumb was attached by only a narrow pedicle and the left had no tendinous attachment. Both were biphalangal. Her subsequent development has been retarded and at a recent assessment at 2 years 3 months she was functioning at a 9 month level. She remained very small, with a weight of 6 kg, length 68 cm, and head circumference 38.5 cm, being respectively 4 kg, 12 cm, and 7 cm below the 3rd centile.

G and C banding was done on 72 hour cultured lymphocytes. Most cells had a ring chromosome 1, in which no loss or gain of chromosomal material could be detected (fig 2). In a few cells, variant rings were seen. Some showed a duplication of bands 1q24→44, some had a very long dicentric chromosome, which appeared to be a duplicated and broken ring, and in some cells

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FIG 1 The baby's hands.

FIG 2 The ring 1 and the normal chromosome 1.

a much smaller ring was seen. The parents' chromosomes were normal and C banding was not informative as to the parental origin of the ring.

A ring chromosome 1 is very rare, only four cases having been previously recorded.¹ The constant phenotypic features have been profound growth retardation and mild mental retardation. In none has a consistent loss of chromosomal material been detected. Thus, these cases may be examples of the 'general ring syndrome', as proposed by Coté *et al*,² in which telomere-to-telomere fusion occurs, with mechanical disruption of mitosis being the process which determines phenotypic abnormality.

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