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Case report

Familial omphalocele

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An omphalocele is an extrusion of the intestine and other abdominal contents through the umbilical ring. The prevalence rate varies with the methods of ascertainment. McKeown et al,[1] in Birmingham in 1941-1951, found a prevalence rate of 1 in 3,200 live- and stillbirths, and in 1982, in Northern Ireland, the figure was 1 in 2,000 live- and stillbirths.[2] It is usually sporadic, and familial occurrence is rare. DiLiberti,[3] who reported a family with five affected individuals in four generations, could identify only eight previous reports of familial occurrence. We report a family with seven affected persons in four generations. Although there was no male-to-male transmission, the congenital abnormality in this family probably has an autosomal dominant mode of inheritance.

FAMILY HISTORY

The family, from Ghana, now living in Northern Ireland, was referred for advice on the management of the omphalocele (Figure). In addition to the three affected children of the proposita, there were three other affected relatives. The proposita (III.7) had an omphalocele about 10 cm in diameter covered with skin. She had no symptoms of obstruction or other congenital abnormalities; in particular, there was no macroglossia or visceromegaly. Surgical intervention was unnecessary. She and her husband were not consanguineous. Her eldest daughter (IV.1) aged 8 years had an omphalocele measuring 1.5 cm in diameter. This abnormality was present at birth and, according to her mother, in the past few years had become smaller. The proposita's other two children, dizygotic female twins (IV.2 & IV.3), each had an asymptomatic omphalocele 2 cm in diameter. None of the three children

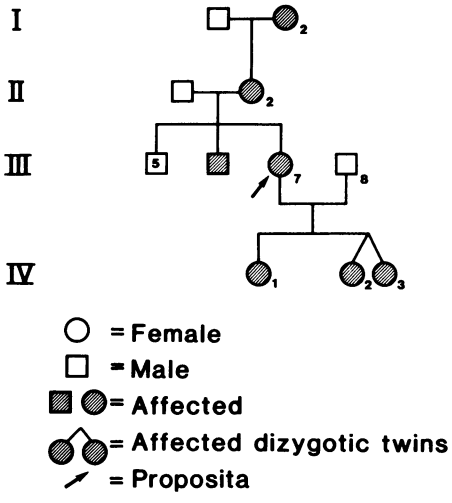

Figure. Pedigree of the family.

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(IV.1-3) had any clinical symptoms or other congenital abnormality. The proposita's brother (III.6) and mother (II.2) were not available for examination, but each was said to have an omphalocele. The maternal grandmother (I.2) who was deceased also had had an omphalocele.

DISCUSSION

Omphalocele is usually a sporadic congenital abnormality affecting approximately 1 in 3,000 to 1 in 2,000 total births.[1,2] Familial occurrence is unusual. Only nine families have been documented with two or more affected relatives.[3] DiLiberti[3] described a family in which two sisters had an omphalocele, and their father, paternal grandfather, and paternal great-grandfather, had an umbilical hernia. Of the eight other families reported in the literature[3] two also showed the abnormality in several generations.[4,5] Havalad et al[5] described a family with four affected males in a pedigree pattern suggestive of X-linked inheritance. In the six remaining families,[3] the abnormality affected only sibs or half-sibs. Our family is similar to that of DiLiberti[3] with seven affected individuals, one male and six females, in four generations. It has been postulated that in familial cases of omphalocele there may be an inherited defect in muscle or connective tissue.[3] Inheritance in our family is probably as an autosomal dominant trait with variable penetrance although X-linked dominant inheritance cannot be excluded. In counselling parents of a child with an isolated omphalocele, although the overall recurrence risk in sibs is small (under one per cent),[6] it is worth remembering that, occasionally, the abnormality may have a genic origin and be associated with a high risk of recurrence.

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