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Short reports

Familial occurrence of omphalocele suggesting sex-linked inheritance

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SUMMARY A family is described in which 4 males in two generations had omphalocele. There was no case of omphalocele in any of the women. It is suggested that the mode of inheritance could be a sex-linked recessive trait.

The occurrence of omphalocele in siblings is rare (McKeown *et al.*, 1953; Irving, 1967; Rothemberg and Barnet, 1974; Rott and Truckenbrodt, 1974) and there is only one report of the presence of this malformation in two generations of a family (Osuna and Lindham, 1976). We report a family in which 4 cases of omphalocele occurred in two generations. Only males were affected, suggesting a sex-linked mode of inheritance.

Case reports

Case 1. (Figure). A male term infant was born to an 18-year-old primigravida, labour was induced because of hypertension. The infant, who weighed 3490 g, had an omphalocele of 4·5 cm in diameter and the sac contained liver and small intestines. A primary closure was done at age 8 hours. He needed to be digitalised for congestive cardiac failure due to a ventricular septal defect which has since closed spontaneously. He did not have macroglossia nor did he develop hypoglycaemia. He is now 36 months old and has developed normally.

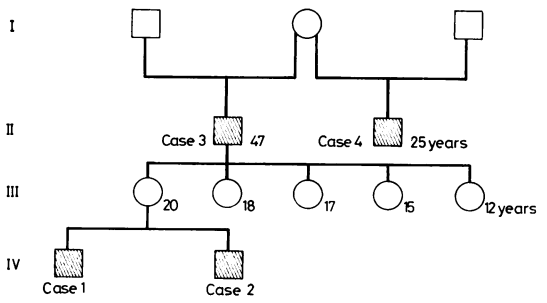

Figure *Family tree showing distribution of omphalocele in male siblings only.*

Case 2. A younger brother of Case 1, was born after a gestation of 40 weeks. The pregnancy was complicated by hydramnios, but labour and delivery were normal. The baby, who weighed 3870 g, had an omphalocele 4 cm in diameter. The sac contained liver and small intestines. Primary closure was performed at age 8 hours and the child had an uneventful postoperative course. There was no evidence of hypoglycaemia. He was discharged from the hospital on the 9th day. Follow-up to age 12 months shows normal development and there is no evidence of any other congenital abnormality.

Family cases. The maternal grandfather (Case 3) of Cases 1 and 2 had had an omphalocele at birth for which surgical closure had been needed. He had no other abnormality. His step brother (Case 4), who was born after his mother remarried, also had an omphalocele at birth for which surgical closure had been needed. There were no other siblings in that generation.

Discussion

Omphalocele is caused by a defect in the development of the abdominal wall. Physiological herniation of the intestines into the extra embryonic coelom of the cord occurs between the 6th and 11th week of embryonic life. Failure of the intestines to re-enter the abdominal cavity results in an omphalocele.

McKeown *et al.* (1953) reported the incidence of omphalocele as 1:3200 in Birmingham, and Soper and Green (1961) reported the incidence as 1:3800 in Ohio.

Omphalocele sometimes occurs in chromosomal aberration as part of the trisomy 13 and 18 syndromes, but its mode of inheritance as a single defect is not known.

Osuna and Lindham (1976) reported a family in which the condition was present in two generations (3 females and one male) suggesting the possibility

that omphalocele could be inherited. In the present family omphalocele occurred only in male children and both male siblings of the 2nd generation were affected. All the children in the 3rd generation were female and none had omphalocele. Both male siblings in the 4th generation had omphalocele.

The distribution of the defect in this family suggests an X-linked recessive inheritance with the males being affected and the females acting as carriers.

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Nonaccidental poisoning in childhood

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SUMMARY A boy aged 7 years 10 months was admitted to hospital on several occasions in an unconscious state with twitching and apnoeic episodes. Initial investigations failed to show a specific cause. During his time in hospital he had recurrent episodes of loss of consciousness and, on the last occasion, hypotension and ventricular tachycardia. A diagnosis of imipramine poisoning was established by the presence of imipramine in stomach washings and blood. The drug was being given to the child, both at home and in hospital, by his mother. The possibility of nonaccidental poisoning must be considered if there is no obvious cause for a child's illness. In this case the mother responded to psychiatric treatment.

The syndrome of child abuse incorporates a wide range of features including nutritional and emotional deprivation and violence. However, reports of nonaccidental poisoning are relatively rare. Meadow (1977) and Rogers *et al.* (1976) stated that occasionally parents may assault their children by administering a drug overdose. We describe the case of a child who was systematically and deliberately given toxic doses of a drug for a period of 2 months before the cause could be proved.

Case history

A boy aged 7 years 10 months was admitted to Scartho Road Hospital, Grimsby, in an unconscious state. He was twitching and had apnoeic episodes. Investigations at that time, including a full blood count, blood urea and electrolytes, and cerebrospinal fluid, were normal. He recovered consciousness over a period of 48 hours and was well on discharge from hospital 14 days later. At that time his parents, in response to specific questioning, denied that there was any possibility of the child having had an overdose of drugs. The inquiry was specific as the patient had been treated for enuresis in 1974, 1976 and, in the month before this illness, with amitriptyline. After discharge he vomited each day and was readmitted to hospital in a drowsy but rousable state one month later. He was treated with intravenous fluids for mild dehydration and his level of consciousness gradually improved. During the next week while in hospital, he had episodes of drowsiness and disorientation associated with bizarre jerking of eyes and limbs. These episodes occurred mainly in the evenings after his mother had visited him. He was referred to Dr J. Lorber at the Sheffield Children's Hospital for further assessment. At the time of transfer he was drowsy and had writhing movements of the limbs. There were no other neurological abnormalities. Again, his parents specifically denied that he had had any access to toxic drugs. He fully recovered within 12 hours of admission, and therefore no further investigations were carried out.

He was discharged but readmitted 12 days later because of gradual loss of consciousness during the previous 24 hours. He was unconscious, had myoclonic twitching of his limbs, nystagmus, hypotension,