ANTERIOR SACRAL MENINGOCELE OCCURRING IN A FAMILY

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Anterior sacral meningocele is an uncommon congenital anomaly. The earliest case report that we have found appeared in the *Lancet* in 1837 from the Medical Society of London. Cohn and Bay-Nielsen (1969) reported sixty-nine patients and Thierry, Archimbaud, Fischer, Freidel and Mansuy (1969) reported sixty-seven: about fifty of these patients were common to both reviews. The condition arises in association with a congenital defect of the sacrum and coccyx through which the caudal part of the meninges may herniate to form a cyst-like structure. A familial incidence has only been recorded twice.

Without operation the condition carries little or no morbidity provided a patient is not pregnant. However, if an abdominal operation is done in ignorance of the correct diagnosis the mortality and morbidity are high. The following case report is presented to remind orthopaedic surgeons of the difficulties which may be encountered in the diagnosis, and because of the occurrence of the anomaly in a father, his brother and his daughter.

A woman aged nineteen years attended the gynaecological department with a history of oligomenorrhoea. At laparoscopy a large smooth, rounded mass was seen lying behind the rectum. The uterus was noted to be double with a normal ovary and tube on each side. The cervix and vagina were single. Intravenous pyelography showed normal functioning kidneys in the normal situation, but the sacrum was found to be deformed and hypoplastic (Fig. 1). On further questioning the patient said that she developed headaches when in a hunched position or bent down. There had been difficulty with bowel movement throughout childhood. On examination the tone of the anal sphincter was found to be poor and there was hypoaesthesia on the left side of the saddle area in the distribution of the third, fourth and fifth sacral segments.

CASE REPORT

A woman aged nineteen years attended the gynaecological department with a history of oligomenorrhoea. At laparoscopy a large smooth, rounded mass was seen lying behind the rectum. The uterus was noted to be double with a normal ovary and tube on each side. The cervix and vagina were single. Intravenous pyelography showed normal functioning kidneys in the normal situation, but the sacrum was found to be deformed and hypoplastic (Fig. 1). On further questioning the patient said that she developed headaches when in a hunched position or bent down. There had been difficulty with bowel movement throughout childhood. On examination the tone of the anal sphincter was found to be poor and there was hypoaesthesia on the left side of the saddle area in the distribution of the third, fourth and fifth sacral segments.
The diagnosis lay between a pre-sacral dermoid cyst, a chordoma or an anterior meningocele.

Myelography showed a large anterior sacral meningocele (Fig. 2). Neurosurgical advice was sought and the lesion was excised completely through a posterior approach (Mr L. Symon). There was no change in the neurological findings, but menstruation became normal. **Affected relatives**—The patient's general practitioner pointed out that the father had been found to have a deformed sacrum during intravenous pyelography (Fig. 4). He did not have any symptoms referable to the sacral anomaly except that his bowels acted only twice weekly.

Because of the finding of similar anomalies in father and daughter as many of the relatives were seen as was possible. The family (Fig. 3) showed the defect in an uncle (Figs. 5 and 6) as well as in the father of the patient.

![Family tree diagram](#)

**DISCUSSION**

Anterior sacral meningocele is characterised by a defect of the anterior aspect of the sacrum involving one or more segments. The defect is smooth, with well demarcated edges, and usually oval in shape. At its lower margin the tip of the sacrum and coccyx hook under the meningocele and produce a typical scimitar or sickle appearance which is diagnostic. As in the present patient, this is sometimes not well seen because of the acute angulation of the sacrum, which was almost at right angles to the x-ray beam in the conventional antero-posterior view of the pelvis. The sacrum appears to have developed around a mass and the deformity varies with the position of the sac (Sherman, Caylor and Long 1950).

The condition is of importance for a number of reasons. It may give rise to difficulties during pregnancy, although normal deliveries have been recorded (Sherman et al. 1950). Constipation is commonly a problem (Thierry et al. 1969). Urinary retention is less troublesome but may also occur. Puncture of the sac, or operation in ignorance of the true diagnosis, may lead to meningitis and death (Llywelyn Jones and Evans 1959). Communication with the subarachnoid space may be very narrow and it may prove difficult to fill the cyst on myelography. It is, however, the type with a narrow neck which is most easily excised.

The presenting symptoms may be constipation, urinary difficulties, or gynaecological problems such as dysmenorrhoea. Neurological symptoms may include disturbance of sphincter control, muscle weakness and sensory disturbance in the lower limbs. Changes in
intra-abdominal pressure may be reflected in changes of intracranial pressure producing intermittent headache and nausea.

In the reported patients there is a striking preponderance of females. Sixty out of the sixty-nine patients in Cohn and Bay-Nielsen's series were female as were fifty-eight of the sixty-five patients in Thierry's series in whom the sex was stated. Thierry said that out of

![Image](64x30 to 548x762)

**FIG. 4**
Radiograph of the sacrum of the father of the patient.

![Image](186x180)

**FIG. 5**
Antero-posterior and lateral view of the sacrum of the patient's uncle.

![Image](180x714)

**FIG. 6**

eleven children aged under fifteen years the sexes were almost equal with five boys and six girls. The family described by Cohn and Bay-Nielsen consisted of two affected female cousins with three other female relatives showing sacral anomalies including meningoceles. Aaronson's (1970) family had one patient with an anterior meningocele and other siblings with anal atresia and sacral anomalies.
This is the first report of sacral meningocele occurring in members of the opposite sex in the same family. The details available are insufficient to allow a definite statement about the nature of the genetic defect, but it is compatible with an autosomal dominant with variable penetrance.

The co-existence of uterine abnormalities is common. Thierry mentioned ten cases of reduplication of the genital tract. Other abnormalities are also common: six of Thierry's patients had spinal anomalies and three of them had meningoceles in a second site. There is also an increased incidence of teratomatous tumours, dermoids and lipomata. The condition may be slightly more common than is realised in view of the fact that it tends to cause few symptoms, especially in males.

SUMMARY
1. A case of anterior sacral meningocele is described in members of a family consisting of a woman, her father and his brother.
2. This is the first recorded case of this anomaly in members of the same family of different sex, and only the second recorded case of occurrence in the same family.

We would like to thank Mr A. M. Fisher for referring this patient, Dr B. Slater for his help in the investigation of the affected family and Mr Lindsay Symon of the National Hospital for Nervous Diseases, London, for his advice and help.

REFERENCES