A family study of bladder extrophy

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SUMMARY The families of 102 index patients with bladder extrophy treated at The Hospital for Sick Children, Great Ormond Street were studied in an attempt to arrive at an empirical risk of recurrence for sibs. Of the 102 patients, 89 had complete extrophy, eight had partial extrophy (variant), and five had extrophy of the cloaca.

In all they had 162 sibs, none of whom had bladder extrophy. The risk to sibs, in this study and from reports by surgeons of sib pairs in their consecutive series, is low and probably less than 1%. There is a suggestion of an increase in central nervous system malformations in sibs as well as in the index patients.

Bladder extrophy is a defect of the infraumbilical abdominal wall. The minimal degree of such a defect is epispadias alone and this is not included in this study. The relatively common intermediate degree of the defect is complete bladder extrophy, with epispadias or bifid clitoris, anteriorly ectopic and often stenotic anus, and wide separation of the pubic symphysis. Frequently associated anomalies include duplication of the uterus and vagina. In a minority of cases only part of the bladder is exposed. This may be at either the upper or lower end, or a portion of extrophic bladder may be separated from the remainder of the bladder which is covered. These forms are usually labelled 'variants' of bladder extrophy. The rare and most severe degree is extrophy of the cloaca, in which the lateral portion of the exposed mucosa represents the two halves of the posterior wall of the bladder, but the central portion is intestinal epithelium. The anus is imperforate. Above the extrophy there is a broad exomphalos. The female genital tract is usually duplicated.

The basic defect is probably failure of fusion of the secondary mesoderm (from the primitive streak) in the midline of the anterior abdominal wall, with subsequent rupture of the thin wall consisting only of ectoderm and endoderm. Early rupture (5th week) results in extrophy of the cloaca, later rupture (7th week) in extrophy of the bladder.

The earliest known report must surely be the description on an Assyrian tablet of about 2000 BC, now in the British Museum, London.

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for parents having already had one child with this severe and troublesome condition, a systematic family study was undertaken.

Material and methods

The index patients were 102 children (71 boys and 31 girls) with extrophy of the bladder attending The Hospital for Sick Children, Great Ormond Street between 1966 and 1973. Complete bladder extrophy was present in 89 (67 boys and 22 girls), partial bladder extrophy in eight (one boy and seven girls), and extrophy of the cloaca in five (three boys and two girls). Details of the three groups together with associated malformations present are shown in the table.

We set out to trace and, with the permission of the family doctors, to interview the parents of each index patient. It was not possible to include seven families because one family was not willing to participate, it was considered unwise to visit another family with special problems, and five families had gone overseas. Family data were collected during home visits for the remaining 95 (65 boys and 30 girls) index cases. The family pedigree taken included sibs, parents, grandparents, aunts, uncles, and cousins, as well as the history of the pregnancy of the index patient.

Results

The average birthweight of the 82 patients, where the information was available, was 3.2 kg and no different from that of all live births. Similarly, measurements obtained wherever possible on current height and weight gave no indication of any interference with growth in childhood. Two patients were mentally retarded but intelligence in the remainder was within the normal range. Only three index patients were dead and two of these had extrophy of the cloaca.

There is no indication of any effect of maternal age, birth order, or season of birth. Questioning all the mothers of index patients about possible prenatal factors, maternal illness, febrile episodes, or medications gave no indication of any contributory environmental factors.

The sex ratio is 2:3 and similar to that found in other series.

TWINS

There was one set of discordant twins, both boys, who were thought to be dizygotic; they are now separated and adopted.

SIBS

The index patients had, in all, 162 sibs (90 brothers and 72 sisters), none of whom had bladder extrophy or associated abnormality. The patients with cloacal extrophy had ten brothers and three sisters, those with the variants had seven brothers and five sisters, and those with complete extrophy had 73 brothers and 64 sisters. Parents reported that no other children had evidence of a possible minor degree of the extrophy malformation complex, other genital anomalies, or difficulties with urinary control other than two sibs with mild nocturnal enuresis.

PARENTS

No parents were consanguineous and none had bladder extrophy. The mother of the twins had right duplex kidney and double ureters.

Cousins

The index patients had 424 male and 395 female cousins none of whom had bladder extrophy.

Sibs with other malformations included one with anencephaly, one with spinal dysraphism, one with congenital heart disease (VSD and ASD), and one with unilateral cleft lip and palate. A further pregnancy was terminated at 5 months because of anencephaly.

Discussion

The recurrence risk after bladder extrophy is clearly low. In this systematic study, of 162 sibs none was affected. The reports by urological surgeons of consecutive series of patients quoted above also suggest that the proportion of sibs affected is low. These in sum include three sib pairs of 421 patients, but the total of the normal sibs is not stated. For genetic counselling, at present, a recurrence risk of about 1% would appear appropriate.
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It is well known that there is an association of bladder extrophy and spina bifida. In this series, two of 102 index patients had sacrococcygeal meningoceles. Of the 162 sibs, one had anencephaly and one spinal dysraphism, and a further pregnancy was terminated because of anencephaly. However, large studies of neural tube malformations report no instances of bladder extrophy among index patients or their sibs.

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References


Requests for reprints to Professor C O Carter, MRC Clinical Genetics Unit, Institute of Child Health, 30 Guilford Street, London WC1N 1EH.
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