Bladder Exstrophy with Several Other Associated Malformations

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ABSTRACT

A case of a newborn male infant with exstrophic bladder and several other malformations was reported and the theories as to the genesis of this tragic deformity were briefly reviewed.

Key Words: *malformation*; *omphalocele*; *meningocele*

INTRODUCTION

The exstrophy of the urinary bladder is a tragic malformation which is characterized by the extroversion of the posterior bladder wall with continuous leakage of urine. It has beed well known that other anomalies frequently occur in association with the bladder exstrophy[1].

With respect to its etiology, there is no evidence of familial or racial predisposition and no definite relationship can be found between this anomaly and some specific illness of the mother during pregnancy. Mechanical, pathologic and embryological theories that have been advanced failed to explain its genesis satisfactorily.

In this paper we report an autopsy case of newborn infant with exstrophy of the bladder associated with omphalocele, meningocele, atresia ani, vesico-intestinal fistula, retentio testis and agenesis of colon.

CASE REPORT

A male newborn infant was admitted to the Department of Surgery of Yamaguchi University Hospital on February 5, 1976, because of multiple malformations noted at birth. He was born at full-term: the delivery was spontaneous from a breech presentation. His birth weight was 2360 gm. There was no family history of anomalies or of any other significant illness.

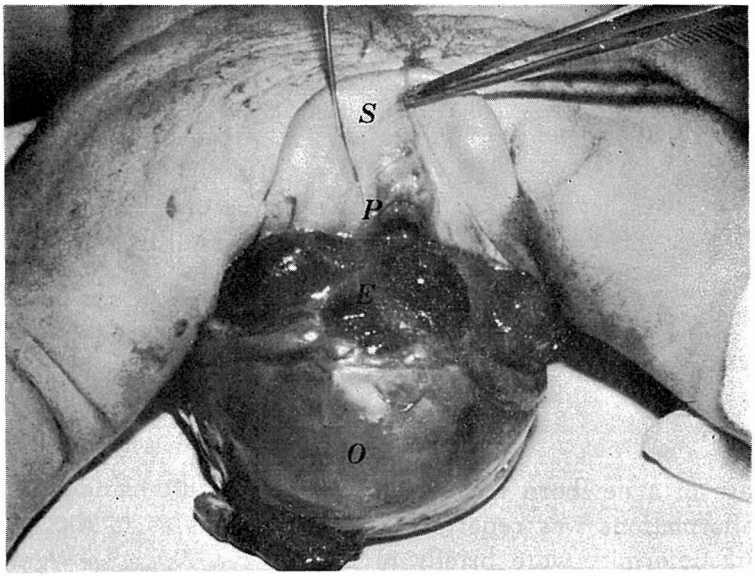

Fig. 1. Dorsal aspect of the abdominal mass and perineal region. It shows omphalocele (O), exstrophied bladder (E), rudimentary penis (P) and edematous scrotum (S).

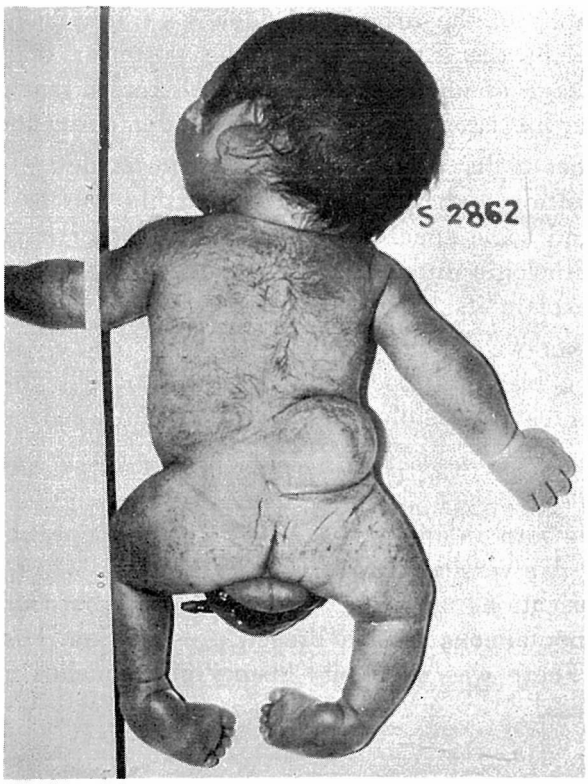

Fig. 2. Dorsal aspect of the baby shows large meningocele in the lumbosacral region.

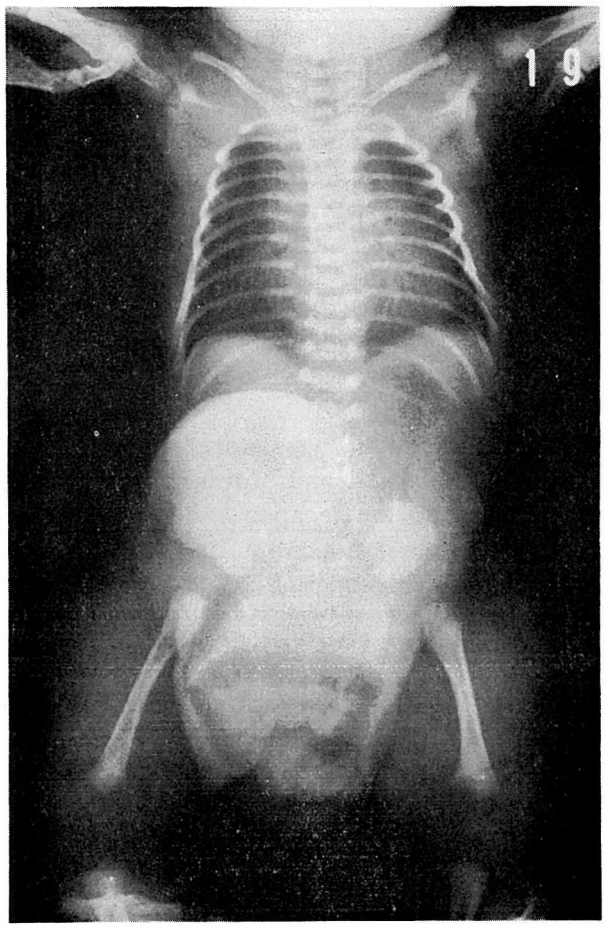

Fig. 3. Roentgenogram of the abdomen shows translocation of the intestinal loop into the omphalocele. Note the wide separation of the pubic arch.

Physical examination revealed the following abnormalities: In the lower abdominal region, there were two large bulging areas devoid of covering skin. The upper one was larger measuring 4.5 cm in diameter and was an omphalocele. The other bulging which lied lower than the omphalocele was exstrophied bladder measuring 2.5×2 cm (Fig. 1). An intervening band of normal skin separated the omphalocele and exstrophied bladder. The penis was of small size, measuring 0.8 cm in length, and was flat with complete epispadias. The scrotum was edematous. In the perineal region there was a shallow dimple, but no anal orifice could be found. A soft projection of meningocele, 5×4.5×2 cm in dimension, was also present in the lumbosacral area (Fig. 2). The additional clinical finding was club feet.

A plain roentgenogram of the abdomen revealed the displacement of the intestinal loop into the hernia sac and separation of the pubic symphysis (Fig. 3). The discharge of meconium was noted on the central part of exstrophied bladder mucosa three days after birth. The patient died on the third post-natal day (February 6). Autopsy was performed from two hours after death.

POSTMORTEM EXAMINATION

The autopsy material was that of a small male infant, 39 cm in stature and weighing 2370 gm. As noted clinically many anomalies were in the lower half of the body.

Omphalocele: The intestinal loops were displaced into hernia sac devoid of skin. It was composed of fibrovascular connective tissue. The umbilical cord was attached to the apex and umbilical vesseles coursed on the wall and entered beneath surrounding skin. Histological examination revealed suppurative inflammation on the outer surface of the hernia sac.

Exstrophy of the urinary bladder: The vesical mucosa was reddish violet due to submucosal hemorrhage. The central area of evaded bladder mucosa was stained with small amount of feces and a small vesico-intestinal fistula was suspected. Histologically the surface of exstrophied bladder was eroded and in most part it was devoid of normal transitional epithelium and severe submucosal hemorrhage was also found.

Gastro-intestinal tract: The intestinal tract was short. The duodenum was located in normal position but throughout its entire length it was free from the retroperitoneum. No macroscopic distinction could be made between the duodenum, jejunum, ileum and colon. The bowel ended at the right lower portion of the abdominal wall. Approximately 5 cm proximal to the bowel end there was a small opening of vesico-intestinal fistula. The mucosa at the end of the bowel showed microscopic appearance indistinguishable from normal colon with numerous goblet cells. The appendix could not be found.

Meningocele: Meninges bulged through the defect on the dorsal aspect of lumbar vertebrae and sacrum to form a subcutaneous soft mass. Spinal cord and its nerves were in normal position.

Other organs: The right kidney was situated just below the liver and lobulated with a few urinary cysts. The right ureter ended at about 1.5 cm above the bladder. The other parts of the urinary tract including renal pelvis were dilated remarkably. The testes were in the retroperitoneum about 3 cm below the level of the kidney. All lobes of the lung

showed marked congestion with scattered spots of hemorrhage. There was no abnormalities in the cardiovascular system. The central nervous system was not autopsied.

The pathological diagnosis can be summarized as follows:

1) Large omphalocele with membranous covering
2) Exstrophy of the urinary bladder with a small vesicointestinal fistula
3) Spina bifida with meningocele in the lumbosacral region
4) Separation of the pubic arch
5) Rudimentary penis with complete epispadias
6) Agenesis of colon
7) Retentio testis
8) Club feet
9) Right hydroureteronephrosis
10) Pulmonary congestion

COMMENT

Exstrophy of the urinary bladder is said to occur once in 50,000 births[2] and more frequently in males than in females[1-4]. Although the cause of different sex incidence is unknown, Marshall and Muecke[3] speculated that male preponderance is due to hormonal stimulus during embryonic development to sustain cloacal membrane.

The bladder exstrophy was usually classified according to the severity of involvement into two main groups; 1) incomplete exstrophy, in which there is a defect in the upper or lower part of the bladder with normal pubic arch and normal genitalia, 2) complete exstrophy, which is common and is characterized by the protrusion of the posterior bladder wall with wide separation of the pubes and epispadias or cleft clitoris[5]. On the other hand, Uson et al.[1] showed these abnormalities even in incomplete exstrophy. According to Marshall and Muecke[3], a complete exstrophy of the bladder is commonest in a broad spectrum of exstrophic lesions which ranges from epispadias to ectopia viscera abdominalis. The case reported here belongs to the complete exstrophy of the bladder.

It is known that other anomalies occur frequently in association with the exstrophied bladder and defomities are sometimes so extensive as to involve the lower gastrointestinal tract, genital and musculoskeletal systems. Uson et al.[1] found such association in 32 out of 49 males (65%) and 17 out of 23 females (74%). The incidence of concomitant anomalies of intestinal or musculoskeletal system was 22% and 4% in his series respectively.

Several theories have been advanced to explain the genesis of bladder exstrophy and allied anomaly. Mechanical theory attributes its genesis to intra-uterine rupture of the bladder from over-distension. However, Abeshouse[5] considered it unlikely because the union of the pubes is normally complete before the kidney begins to function and hence the theory fails to explain its non-union. Pathological theory presumes that ulceration of the abdominal wall is the cause but there is no scar or any other lesions around the opening of the bladder or cloaca to the abdominal wall. Thus Abeshouse was led to the theory of developmental arrest and stressed the importance of mesodermal deficiencies in midline region. Patten and Barry[6], noticing coexistence of epispadias, considered that genital tubercle primodia is formed too far caudally and this abnormal position would entail the absence of the ingrowth of mesenchyme. However, because no exstrophic stage can be observed in normal embryo of man and animal, a mere developmental arrest is by no means a satisfactory explanation. Marshall and Muecke[3] propounded the opposite theory that the exstrophic lesion is the result of over-development and non-regression of the cloacal membrane during the first six weeks of gestation. An abnormally large and persisting cloacal membrane would act as a mechanical barrier to mesodermal ingrowth and hold apart the developing structures of the lower abdominal wall. Experimentally, Muecke[7] induced cloacal exstrophy in the chick embryo by putting a minute plastic graft into the tail bud region during early development which could interfere with normal cloacal membrane regression.

Another interesting aspect of exstrophied bladder is the frequent association of various pathological conditions such as 1) acute or chronic inflammation, 2) squamous metaplasia of the bladder epithelium, 3) glandular formation and 4) malignant change[8,9]. In our case, erosion and hemorrhage were found in the exstroverted vesical mucosa.

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