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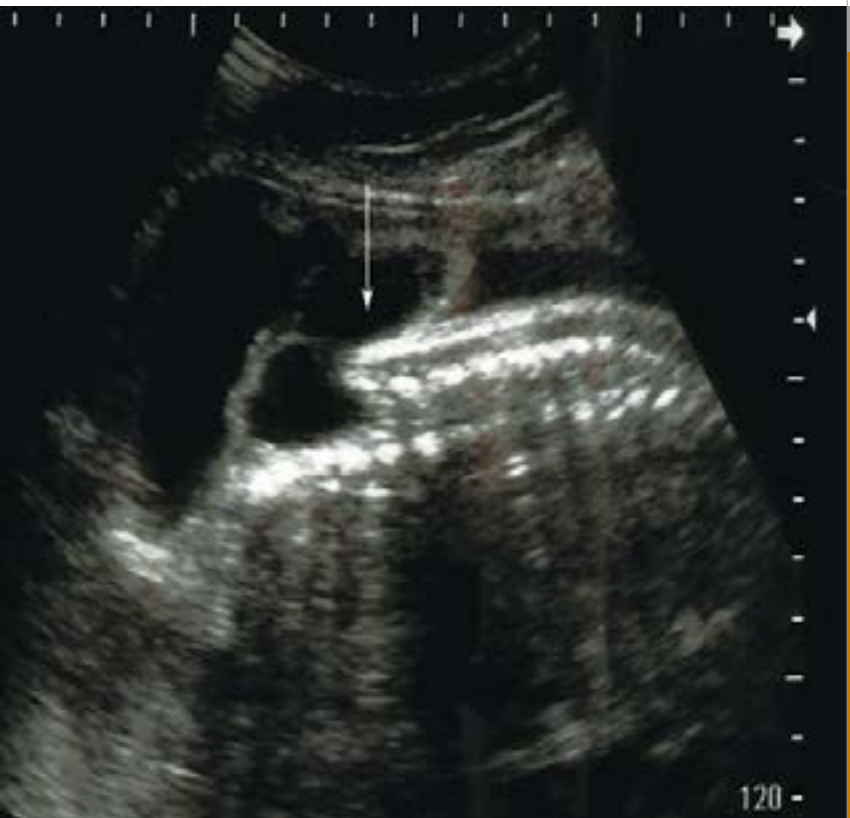
Exstrophy of the cloaca sequence



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A full term male baby was born to a nonconsanguinous couple by normal vaginal delivery. The mother had regular antenatal screening. Her rubella, HIV and hepatitis B screen were negative. Antenatal scan at 20 weeks showed sacrococcygeal meningomyelocele (Fig. 1) and a scan at 28 weeks revealed bladder exstrophy. At birth, the baby required only minimal resuscitation and the Apgar scores were 6 and 9 at 1 and 5 minutes, respectively.

His birth weight was 3 kg. He was breathing spontaneously in room air. There was no facial dysmorphism and on examination he had an omphalocele, cloacal exstrophy, imperforate anus and a large meningomyelocele (Fig. 2). His legs were flaccid with bilateral talipes equino-varus deformity. Scrotal folds were noted in the inguinal area on both sides and testes were absent. A penis was found just below the extrophied bladder. Both ureteric openings were well visualized in the bladder mucosa. Blind ending of the ascending colon was seen. His chest and upper limbs were normal.

Ultrasound showed a right pelvic kidney and left normal kidney. Echocardiography showed a normal heart. MRI of brain and spinal cord showed tonsillar herniation down to C 5-6 level, normal size ventricles, absence of lower lumbar and sacral segments with spina bifida aperta, tethering of the cord and a big lobulated meningomyelocele (Fig. 3-4). His karyotype was normal.

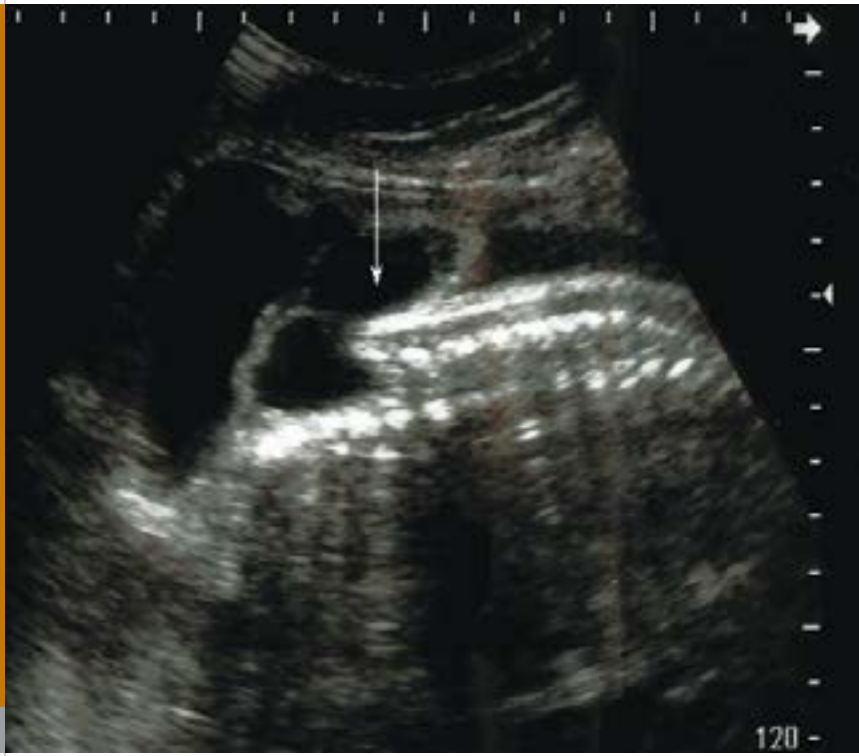

Fig. 1

Antenatal ultrasound scan at 20 weeks showing meningomyelocele (white arrow).

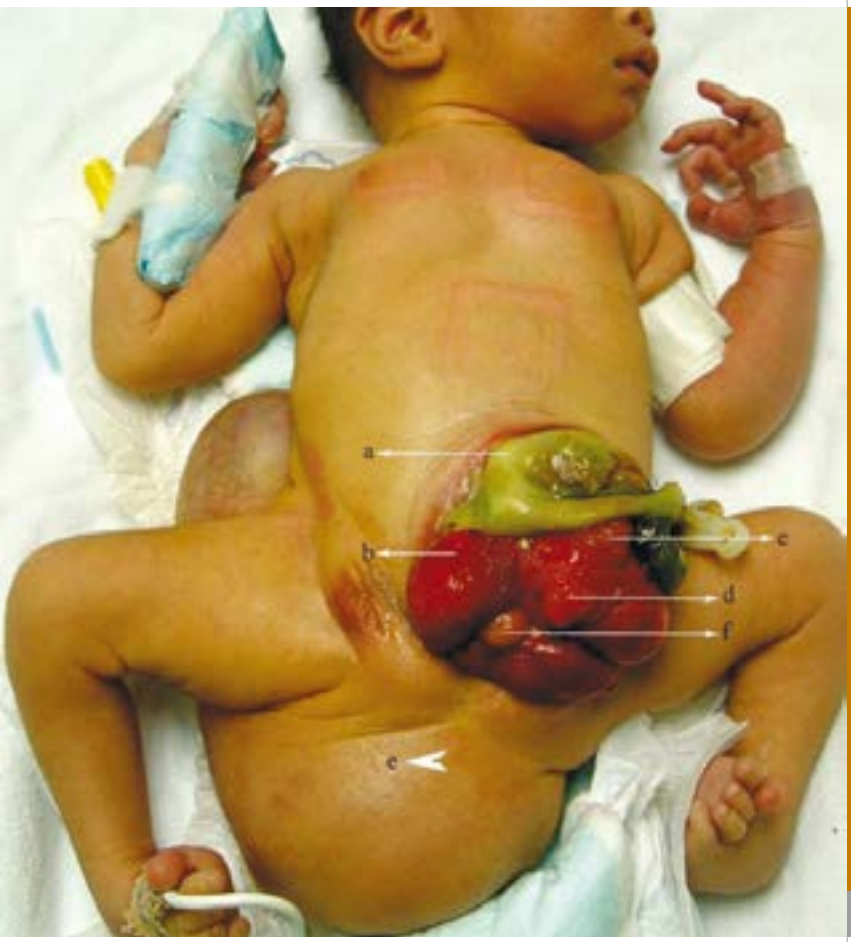

Fig. 2

a) omphalocele; b) bladder mucosa; c) and d) blind endings of gut; e) meningomyelocele; f) penis.

On the 3rd day of life he was examined under general anesthesia. The omphalocele was excised and the bladder was separated from the bowel. The blind end of the ascending colon was identified and a colostomy was done. The exposed bladder was closed leaving a vesicostomy over the lower anterior abdominal wall. Bilaterally atrophied testes were identified in the lumbar area.

A small phallus noted at birth was buried in the pelvis. Closure of vesicostomy and reconstruction of the open phallus was done at a later stage.

Because of parental concern, surgery for the meningomyelocele was postponed. With increasing size of the meningomyelocele on day 78 of life a ventriculo-peritoneal shunt with repair of the meningomyelocele was done. During the repair 1600 ml of CSF was drained and an adequate closure was achieved.

The patient is paraplegic but growing well with near normal mentation at 3 years of age (Fig. 5).

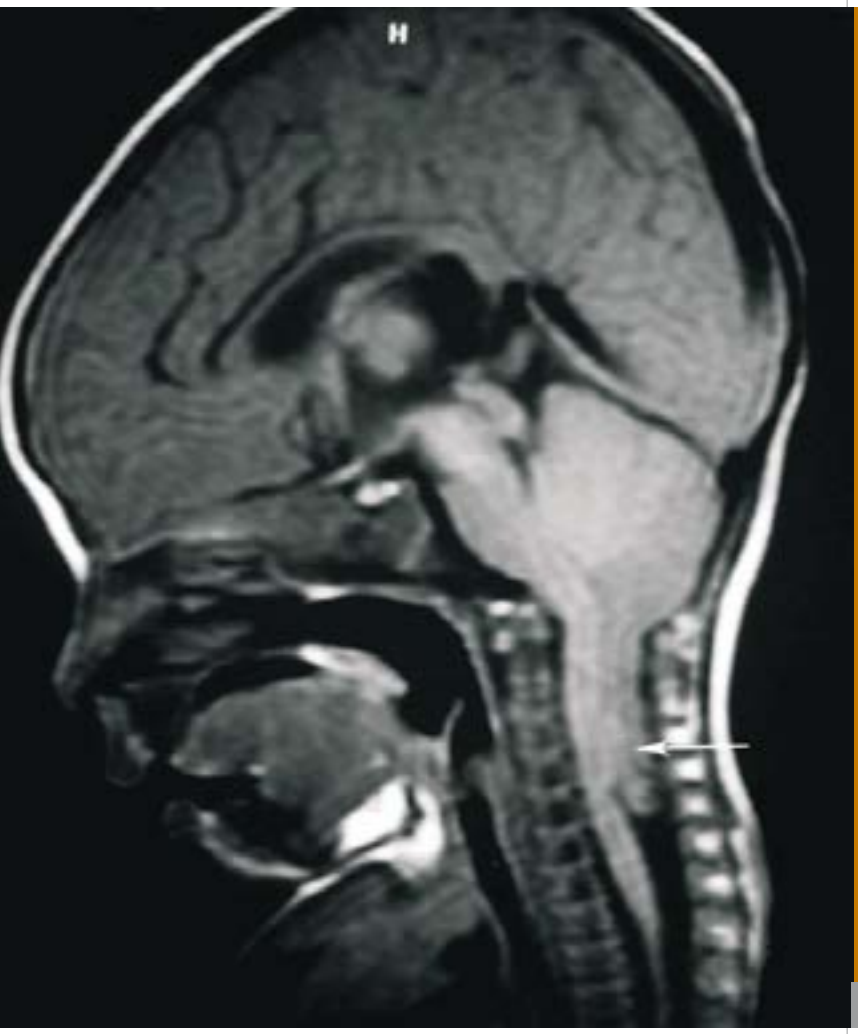

Fig. 3

MRI: Tonsillar herniation.

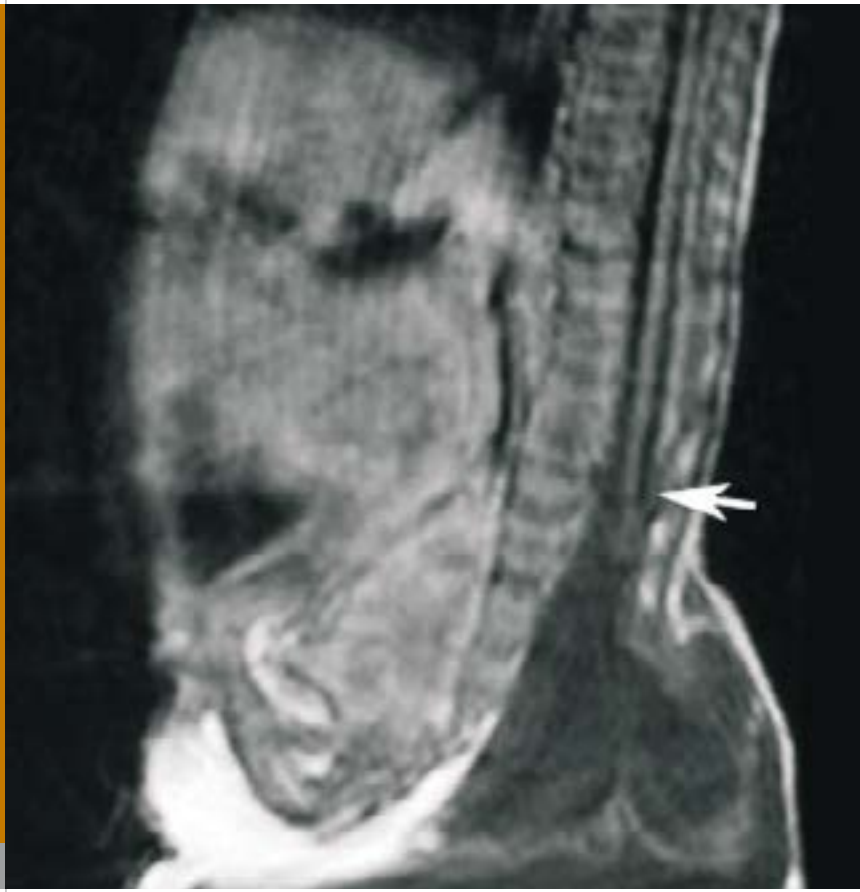

Fig. 4

MRI: Absence of lower lumbar and sacral segments with tethering of the spinal cord (white arrow).

Fig. 5

Patient at tree years of age.

DISCUSSION

Exstrophy of the cloaca sequence occurs approximately in 1 of 200'000 to 400'000 births (1). Both sexes are affected with a male to female ratio of 2.5:1. Embryogenesis of exstrophy of the cloaca sequence is still unclear. The primary defect is thought to be in the development of early mesoderm which will later contribute to the infraumbilical mesenchyme, cloacal septum and caudal vertebra. This results in (a) failure of cloacal septation, with persistence of a common cloaca; (b) complete breakdown of the cloacal membrane with the exstrophy of the cloaca, failure of fusion of genital tubercles and pubic rami, and often in the formation of an omphalocele; and (c) incomplete development of lumbosacral vertebrae with herniation of a grossly dilated central canal of the spinal cord (2). Cryptorchidism is a common finding in the male. Urinary tract anomalies like pelvic kidneys are common. The exstrophied bowel is usually the ileocecal region with little or no large bowel distally as in our case.

Excellent survival with surgical repair is possible. Genetic females do not raise a problem as they will be raised as females. Gender assignment is one of the difficult tasks in the management of newborns with cloacal exstrophy. In genotypic males with exstrophy of cloaca sequence, the phallic size is usually small and reconstruction of an adequate penis is nearly impossible. Testes are usually undescended and abnormal. Males with unilateral or bilateral phallic structures should be raised as males. There is general consensus

that in genetic males with insufficient phallus reassignment as phenotypic females with early orchidectomy to minimize testosterone imprinting on central nervous system should be considered (3, 4).

Surgical management of babies born with cloacal extrophy sequence has progressed over the years and reasonably good outcome is being reported in the literature. Interestingly the reported child is doing well even though he has marked tonsillar herniation.

REFERENCES

1. Ziegler MM, Duckett JW, Howell CG. Cloacal extrophy. In Welch KJ, Randolph JG, Ravitch MM, et al., editors. Pediatric Surgery. Chicago IL: Year Book, 1986:764-71

2. Exstrophy of Cloaca Sequence. In: Jones KL, ed.Smith's Recognizable Patterns of Human Malformations. Fifth edition. Philadelphia; WB Saunders Company, 1997:627-628

3. Lund DP, Hendren WH. Cloacal extrophy: experience with 20 cases. J Pediatr Surg 1993;28:1360-9

4. Matta H, Hemes A, Nawaz A, Jacobz A, Shawis R N, Salem AHA. An unusual variant of Cloacal extrophy. Annals of Saudi Medicine 2002;22:204-205

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