WHAT'S YOUR DIAGNOSIS? CLOACAL EXSTROPHY

What's Your Diagnosis [1] | September 30, 2005
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Female infant born to a gravida II, para I, 23-year-old mother at 38 weeks' gestation. Pregnancy complicated by oligohydramnios. Cesarean delivery performed because of prolonged time after rupture of membranes and fetal distress. Apgar scores, 3 and 6 at 1 minute and 5 minutes, respectively.

PHYSICAL EXAMINATION
Weight, 2258 g; length, 47 cm; head circumference, 33.5 cm. Defective lower abdominal wall. Bladder exposed and divided into halves by an interposed strip of bowel. The cloaca is the common chamber into which the urinary, genital, and intestinal tracts drain. The term ;exstrophy; is derived from the Greek words, exo, which means outside, and strophie, which means to twist or turn out. In cloacal exstrophy, the bladder is separated into halves by the interposed bowel. The condition was first described by Littr in 1709. Synonyms include vesicointestinal fissure, exstrophia splanchnica, ileovesical fissure, and ectopia cloacae.

WHAT'S YOUR DIAGNOSIS?

EPIDEMIOLOGY
Cloacal exstrophy is extremely rare and occurs in only about 1 of every 200,000 to 400,000 live births. There is no sex preponderance. The problem is usually sporadic. The condition is more common in premature infants.

EMBRYOGENESIS
The cloacal membrane is invaded by the medial migration of mesenchymal tissue at about the fourth week of gestation. Cloacal exstrophy is believed to result from either abnormal persistence of the caudal position of the body stalk on the embryo, or overdevelopment of the cloacal membrane, which produces a wedge effect and prevents the medial migration of the mesenchymal tissue between the inner endodermal layer and the outer ectodermal layer. About the fifth to eighth week of gestation, the urorectal septum divides the cloaca into the urogenital sinus anteriorly and the anorectal canal posteriorly. The cloacal membrane usually ruptures during the eighth week of gestation. Rupture before the cloaca is divided by the urorectal septum results in the classic presentation of cloacal exstrophy--a strip of bowel flanked by 2 hemiabladders.
PRENATAL DIAGNOSIS
A prenatal diagnosis can be established with ultrasonography. Such was the case with this patient. Major ultrasonographic criteria include a large midline infraumbilical anterior wall defect, a cystic anterior wall structure (persistent cloacal membrane), an omphalocele, nonvisualization of the bladder, and lumbosacral anomalies.\textsuperscript{4,9,11} Minor criteria include the presence of an elephant trunk-like structure that protrudes from the abdominal wall below the umbilicus (prolapsed ileum), lower extremity defects, renal anomalies, ascites, widened pubic arches, a narrow thorax, hydrocephalus, and a single umbilical artery.\textsuperscript{2,9,11,12}

CLINICAL MANIFESTATIONS
The classic presentation of cloacal exstrophy is a central exstrophic bowel field that contains 2 orifices.\textsuperscript{13,14} The proximal orifice leads to the ileocecal junction. An ileum that prolapses from the ileocecal junction gives the appearance of an elephant trunk.\textsuperscript{4,14} The distal orifice leads to the hindgut. The anus is usually absent.\textsuperscript{14-16} The exstrophic bowel field is flanked by 2 hemibladders

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The bladder is usually smaller than normal. An omphalocele is present in 90\% of cases (Figure 2).\textsuperscript{15} The rectus muscles are separated and the pubic symphysis is widened. In males, the penis is typically short, bifid, and epispadic; the scrotum is bifid; and the testicles are undescended.\textsuperscript{5,13,17} In females, the clitoris is usually bifid. The vagina may be duplex and the uterus bicornuate.

ASSOCIATED ANOMALIES
Associated anomalies are inevitable and are often multiple.\textsuperscript{14} GI anomalies. Omphalocele and imperforate anus are so commonly associated that most investigators consider these anomalies an integral part of the syndrome.\textsuperscript{4} Other GI anomalies include inguinal hernia, malrotation, small-bowel atresia, Meckel diverticulum, anorectal agenesis, duplication of bowel, and absent or duplicated appendix (Figure 3).\textsuperscript{6,12,16} CNS and skeletal anomalies. Anomalies of the CNS and spine include spina bifida, meningomyelocele, tethered cord, hemivertebrae, abnormal lumbosacral segmentation, scoliosis, and kyphosis.\textsuperscript{14,18} The OEIS complex refers to the presence of omphalocele, exstrophy, imperforate anus, and anomalies of the spine.\textsuperscript{12} Other skeletal anomalies include congenital hip dislocation, tibial torsion, and talipes equinovarus.\textsuperscript{12} Urinary tract anomalies. The most common anomalies are pelvis kidney, horseshoe kidney, renal agenesis, and vesicoureteral reflux.\textsuperscript{2} Other anomalies include ureteral atresia, ureteral duplication, and multicystic dysplastic kidney.
COMPLICATIONS
Urinary tract infection, bladder stone, hydronephrosis, and carcinoma of the bladder are more common in persons with cloacal exstrophy. Infrertility is the rule for both males and females. Fecal incontinence is common because of poor perineal sensation and weak anal sphincter muscles. Many affected children require permanent intestinal diversion. Psychological problems are inevitable and can be profound. The patient in this case had recurrent urinary tract infections and fecal incontinence.

INVESTIGATIONS
Renal ultrasonography is necessary to identify hydronephrosis or other renal anomalies. A voiding cystourethrogram should be obtained after surgical correction of the bladder exstrophy to look for vesicoureteral reflux. A DTPA radionuclide renal scan should be considered if hydronephrosis is present but is not thought to result from vesico-ureteral reflux. Periodic urinalysis is recommended (to search for urinary tract infection), and a urine culture should be ordered as needed. MRI scans of the spine and pelvis are necessary because the spinal abnormalities might be occult and poorly delineated on a plain film. A chromosomal study should be performed to clarify the genetic sex.

MANAGEMENT
The spectrum of possible defects requires an individualized treatment plan; a team approach is important. Parenteral nutrition should be started promptly in the neonatal period. Intestinal diversion with either an ileostomy or colostomy is necessary. Optimally, a pull-through procedure should be performed when the child is older to re-establish as near-normal bowel anatomy as possible. However, a pull-through procedure might not be possible if the child is not able to form solid stools, has only minimal colonic tissue available, or has severe spinal dysraphism or poor pelvic musculature.

SEX ASSIGNMENT
The decision regarding sex assignment is controversial. Formerly, the decision was made based on the status of the phallus and often without any consideration of the chromosomal identity. However, a Johns Hopkins follow-up study of 14 genetic males with cloacal exstrophy who were reassigned surgically and socially to the female sex in the neonatal period revealed that most had declared themselves to be male. All had moderate to marked interests and attitudes that were considered typical of males. This information led to a reevaluation of the advisability of considering sex reassignment in male patients with cloacal exstrophy. The decision to reassign sex should be made by the family in consultation with pediatric specialists familiar with the urologic, surgical, and psychiatric problems associated with sex reassignment. In genetically male patients with a satisfactory phallus, male sex assignment is appropriate. Such patients require orchidopexy and phallic reconstruction. Phallic lengthening might be achieved by detaching the upper portions of the corpora from the pubic bone, thereby allowing the corpora to be advanced into the shaft. Epispadias should be repaired. In genetically female patients--like the one in our case history--a vaginoplasty is usually carried out at about the time of puberty. Hormonal therapy is usually offered to stimulate the development of secondary sexual characteristics. The outcome of genital reconstruction in females is usually satisfactory from the sexual point of view. Fertility, however, is often impossible. Lifetime psychological support is necessary.

References: REFERENCES:

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