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The Mermaid Malformation: Cloacal Exstrophy and Occult Spinal Dysraphism

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Five infants with cloacal exstrophy underwent neurological evaluation and radiographic examination of the caudal spine shortly after birth. Each was found to have occult spinal dysraphism. Four had terminal myelocystoceles, and one had a lipomyelomeningocele. Pathological anatomy was confirmed during surgery for the release of the tethered spinal cords. The striking association between cloacal exstrophy and occult spinal dysraphism suggests a common developmental defect in the caudal pole of the embryo. A hypothesis is offered to explain this association. Terminal myelocystocele and lipomyelomeningocele appear to be part of a continuum of lesions associated with skin-covered spina bifida. (*Neurosurgery* 28:834–843, 1991)

Key words: Cloacal exstrophy, Lipomyelomeningocele, Magnetic resonance imaging, Occult spinal dysraphism, Spina bifida, Spinal dysraphism, Terminal myelocystocele

INTRODUCTION

The concurrence of multiple congenital anomalies in a single child can provide clues about normal embryological processes. When multiple anomalies appear together systematically there is likely to be a common defect of embryological development. Recently, the author has had the opportunity to examine a group of infants born with cloacal exstrophy, a rare deforming developmental malformation. Each infant was found to have occult spinal dysraphism. In four of the five, the dysraphic abnormality was a terminal myelocystocele, an extremely uncommon variant of skin-covered spina bifida. One child had a lipomyelomeningocele. These disorders are similar and appear to represent a continuum of occult spinal dysraphism. The relationship between cloacal exstrophy and occult spinal dysraphism forms the subject of this report.

DEFINITIONS

Cloacal exstrophy is an extraordinarily rare congenital malformation characterized by evagination of the intestines between two bladder halves, imperforate anus, and omphalocele. Sometimes referred to as *vesicointestinal fissure* (14), this defect occurs sporadically and is the most severe form of the epispadias-exstrophy spectrum. Cloacal exstrophy occurs in fewer than 1 in 200,000 live births (23, 55, 59). The sex distribution is equal. Although cloacal exstrophy was first reported in 1709, the first successful surgical closure was not performed until 1960, when Rickham reported a single survivor among four children who underwent operation (11, 49). By 1976 there had been only 35 survivors of 157 reported cases of cloacal exstrophy (14). Cloacal exstrophy is now considered a rare but treatable problem, with excellent survival rates and even the potential for urinary continence (11, 22, 23, 38).

Spinal dysraphism has been reported to occur in association with exstrophy of the cloaca (25). Patients with anorectal malformations may have spinal deformities with varying degrees of neurological deficit (12, 13). Carey et al. (8) described the *OEIS complex* (omphalocele, exstrophy, imperforate anus, spinal defects) and reported its occurrence in six patients, four of whom had a skin-covered lumbar or sacral meningocele; the other two had hemivertebrae without cysts. Other spinal defects reported in association with cloacal exstrophy include myelomeningocele (22), lipomyelomeningocele, double discontinuous lipomyelomeningocele (17), and lipomeningomyelocystocele (57).

There is a paucity of information about the spinal abnormalities associated with cloacal exstrophy. Most descriptions have been concerned primarily with the grossly deforming bowel and bladder defects. Associated spinal cord lesions have not been discussed in detail. The terminology used to describe these associated spinal disorders is confusing, which is not surprising, given that the terminology describing spinal dysraphism in general is confusing.

The author believes that the spinal abnormalities seen in association with cloacal exstrophy represent a spectrum of occult spinal dysraphism, or *skin-covered spina bifida*. This condition is to be contrasted with *spina bifida cystica* (myelomeningocele), the dorsal protrusion of a malformed spinal cord through a bifid spine to form a membrane-covered midline mass on the back, frequently associated with hydrocephalus and the Chiari II hindbrain malformation (9). In occult spinal dysraphism, the skin overlying the back is closed, and there is a lesser degree of dysplasia of the spinal cord and its overlying tissues and no association with hydrocephalus or hindbrain malformations.

Terminal myelocystocele, the spinal abnormality most frequently encountered in this series, is a rare form of occult dysraphism. It consists of a localized cystic dilatation of the caudal central canal of the spinal cord (30, 47). The cord is low-lying and passes through an arachnoid-lined meningocele and expands into an ependyma-lined terminal cyst continuous with the central canal, as detailed by McLone and Naidich (36, 37). *Lipomyelomeningocele*, the spinal lesion encountered in one child, is the most common form of occult dysraphism seen in patients without exstrophy (51). The spinal cord is tethered by a lipoma of the conus medullaris, which usually passes posteriorly through defects in the dura, neural arch, and lumbodorsal fascia and ends subcutaneously (5, 6, 10, 20, 34, 35, 40). The author believes that terminal myelocystocele and lipomyelomeningocele are variants on the same theme.

SUMMARY OF CASES

Five infants born with cloacal exstrophy were referred to the Floating Hospital for Infants and Children–New England

Medical Center from 1985 to 1989. The median age of the mothers at time of delivery was 30 years (range, 20 to 34 years). No family had a history of cloacal exstrophy or spina bifida. No mother had diabetes mellitus. One father (Case 5) was an insulin-dependent diabetic. There was no history of consanguinity.

Each newborn underwent surgical reconstruction of the perineum and anterior abdominal wall on the first day of life. Neurological evaluation was performed within the first week of life, although neurosurgical intervention was deferred until recuperation from the cloacal exstrophy repair.

Cloacal exstrophy repair

The gastrointestinal and urological abnormalities observed in each patient were remarkably similar, with varying degrees of severity (Fig. 1). A large omphalocele was present, along with a centrally located eversion of the intestines surrounded by a hemibladder on either side. The symphysis pubis was widely diastatic. The anus was imperforate. The genitalia were ambiguous in every case. Four children were genetic males (46,XY karyotype) and one child, Patient 4, was a genetic female (46,XX karyotype). The genetic males were born with a bifid scrotum and duplex microphallus.

Abdominal and perineal reconstructions were performed as a combined procedure by a pediatric surgeon and pediatric urologist. The omphalocele was closed. The midgut and hindgut were dissected from the two hemibladders and an ileostomy or colostomy was fashioned. The hemibladders were closed primarily to form a sac. The bifid symphysis pubis was brought together. The genetic males underwent genitoplasty and gender reassignment to the female sex in the same sitting. Thus all children were reared as females.

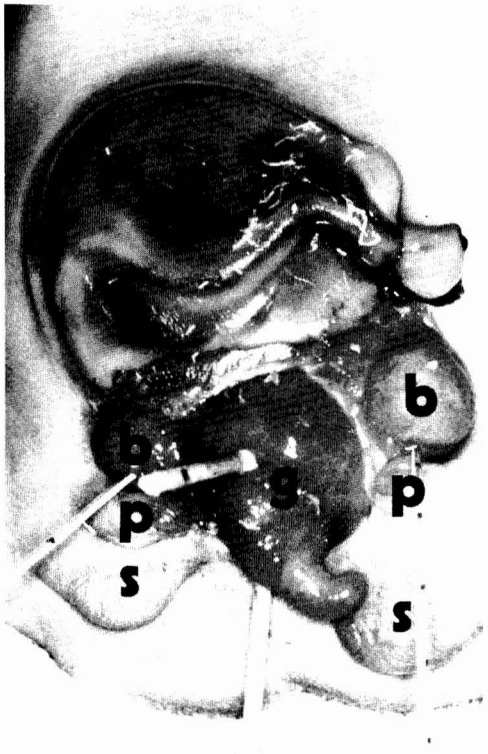

FIG. 1. Case 1. Exstrophy of the cloaca: *o*, omphalocele; *b*, hemibladders; *g*, hindgut; *p*, duplex phalli; *s*, scrotal folds.

Case 1

This child weighing 6 pounds 14 ounces at birth was born via normal spontaneous vaginal delivery to a healthy 20-year-old primigravida after 37 weeks' gestation. The pregnancy was complicated by hyperemesis in the 1st trimester, for which the mother took promethazine and metoclopramide at about the 6th week.

There was a large, midline, skin-covered lumbosacral mass measuring 10 × 8 × 7 cm on the back just above the intergluteal cleft (Fig. 2). Talipes cavus was present bilaterally. Hip flexion was normal, but knee extension was 1/5 on the left and 2/5 on the right, and knee flexion was 0/5 as was ankle dorsiflexion and plantar flexion. There was a sensory level to pinprick at L3. An ultrasonogram and computed tomographic (CT) scan of the head showed mild ventriculomegaly. A magnetic resonance imaging (MRI) scan of the spine showed terminal hydromyelia with a low-lying conus medullaris and a large anterior meningocele. There was a large distinct caudal cyst surrounded by fat, the terminal myelocystocele (Fig. 3).

The terminal myelocystocele was repaired when the infant was 2 months old. The patient was positioned prone, and a vertical midline incision was made and the soft tissue undermined to expose a large subcutaneous lipoma (Fig. 4*A*). The last intact neural arch above the dysraphic spine was removed to expose the normal dural tube. A thick, fibrous band was found tethering the meninges and cord just caudal to this, at the level of the most rostral bifid lamina. This band was divided. The dura was opened in the midline, exposing a low-lying hydromyelic conus with nerve roots directed rostrally. There was a large anterior meningocele. Just caudal to the conus, the dura was absent dorsally and blended into the subcutaneous lipoma laterally. The subcutaneous lipoma was debulked, and a large cyst filled with cerebrospinal fluid was entered. The low-lying cord expanded in a trumpet-like fashion as it approached this large terminal cyst. A probe inserted into this cyst passed freely into the hydromyelic central canal of the cord (Fig. 4*B*). The lipoma was detached from the terminal spinal cord using microdissection. The lipoma and terminal cyst were then further debulked, the edges of the terminal cyst were reapproximated, and the cord fell freely into the large meningocele. The dura could then be closed primarily, and the wound was closed in layers.

Histological examination of the wall of the terminal cyst showed that it was lined by ependyma and dysplastic glia and was surrounded by fat (Fig. 5). In a follow-up examination 3 years later, the patient was developmentally delayed but spoke in short phrases and sat and stood independently, but did not ambulate.

Case 2

This child weighing 4 pounds 8 ounces at birth was born via Caesarean section to a 31-year-old gravida 5, para 1, aborta 3 mother who experienced premature labor after a 36-week gestation. The pregnancy was complicated by vaginal bleeding during the first trimester and episodes of severe pain in the left flank experienced by the mother. She took no medication during the pregnancy. The pregnancy preceding

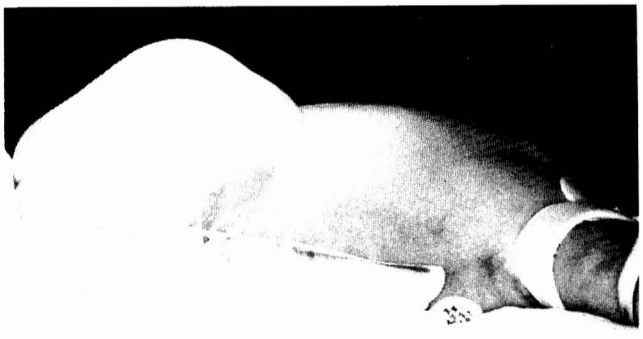

FIG. 2. Case 1. Terminal myelocystocele. Note the large, skin-covered, midline lumbosacral mass.

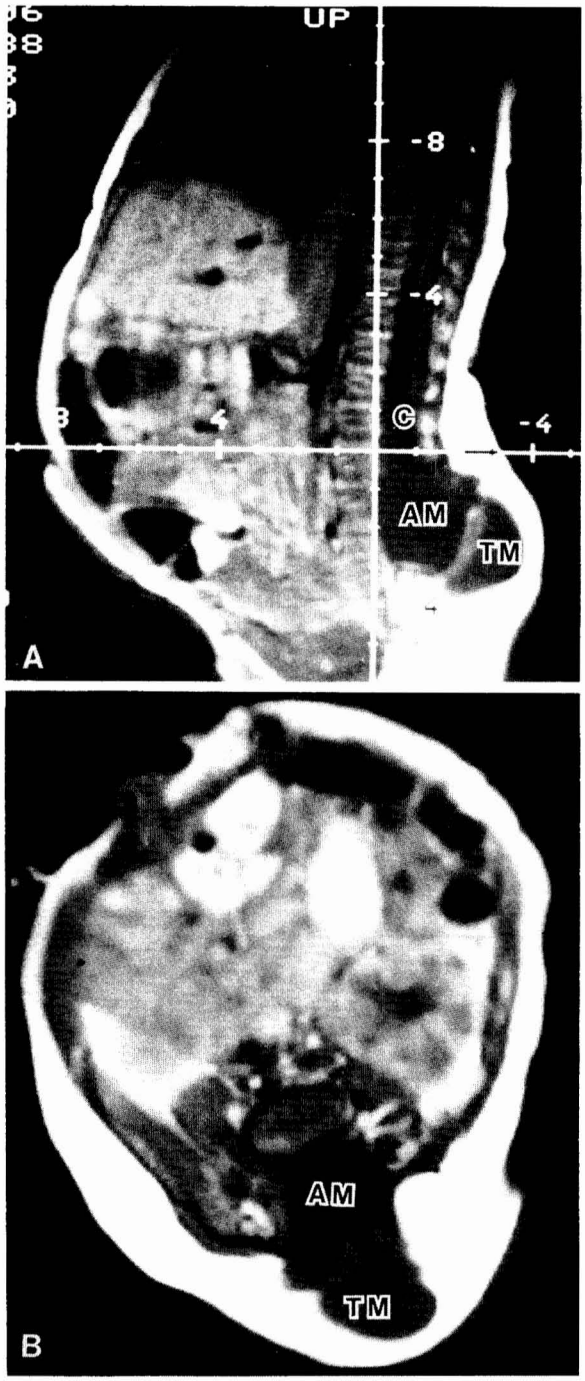

FIG. 3. Case 1. Sagittal (*A*) and axial (*B*) T1-weighted MRI scans demonstrating terminal myelocystocele. The conus (*C*) is low-lying, and there is an anterior meningocele (*AM*) as well as a terminal cyst (*TM*) surrounded by fat.

this one had ended in a miscarriage. A fetal ultrasonogram at 6 months' gestation revealed exstrophy and club feet.

Neurological examination revealed bilateral talipes cavus and analgesia from S3 to S5. There was a large dorsal subcutaneous lipoma in the lumbosacral midline. An MRI scan of the head found nothing abnormal. An MRI scan of the spine showed a low-lying hydromyelic cord with a small terminal cyst surrounded by fat.

Operation to repair the terminal myelocystocele and release the tethered cord was carried out when the infant was 3 months old. The findings were similar to those in Case 1, but less pronounced, and the defect was repaired in the same fashion. In a follow-up examination 2 years later, the child was ambulating independently without motor deficit, and speaking in short phrases.

Case 3

This child was born weighing 6 pounds 10 ounces via normal spontaneous vaginal delivery after 37 weeks' gestation. The mother was a 27-year-old gravida 6, para 2, aborta 3 recovering alcoholic. She took Alka Seltzer (Miles Inc., Consumer Healthcare Division, Elkhart, Indiana) during the 1st month of the pregnancy.

Examination revealed a large, dorsal, lumbosacral, subcutaneous lipoma. Talipes cavus was noted bilaterally, on the left more than the right, with mild bilateral weakness of ankle plantar flexion. Sensory examination and a cranial ultrasonogram disclosed nothing abnormal. An MRI scan of the spine showed a low conus adherent to a subcutaneous lipoma, with a hydromyelic central canal ending in a terminal cyst within the lipoma.

Surgery to release the tethered cord was performed when the infant was 3 months old. As with Case 2, the surgical findings were identical to those in Case 1, but less pronounced, and the repair was carried out the same way. At 27 months after surgery, the child spoke fluently and walked independently without weakness.

Case 4

This child weighing 5 pounds 2 ounces at birth was born via normal spontaneous vaginal delivery to a healthy, 30-year-old gravida 2, para 1 mother after 35 weeks' gestation. Examination revealed a large, subcutaneous, lumbosacral lipoma and talipes calcaneovalgus, worse on the right side than on the left. There was an L5 motor level and analgesia from S3 to S5. A cranial ultrasonogram and CT scan found nothing abnormal. A myelogram showed a tethered cord and lipoma. Surgical detethering was performed when the infant was 4 months old, and findings included a low conus stuck to a lipoma that surrounded a terminal cyst. Follow-up 4 years later found the child to be intellectually normal, ambulatory with ankle-foot orthoses and Canadian crutches, and without change in distal lower limb strength.

Case 5

This child was born weighing 5 pounds 11 ounces to a 34-year-old, healthy primigravida via Caesarean section after 38 weeks' gestation. There was a small subcutaneous sacral lipoma slightly to the left of midline. The child was normal neurologically except for mild bilateral talipes cavus. An ultrasonogram and MRI scan of the head found nothing abnormal. An MRI scan of the spine demonstrated a low conus with a lipomyelomeningocele from L3 to S1 (Fig. 6).

At 15 months of age the infant underwent surgical repair of the lipomyelomeningocele and release of the tethered cord. The findings differed from those in Cases 1 through 4. The conus was low-lying, with a dorsal intradural lipoma. This dorsal lipoma communicated with a subcutaneous lipoma via a narrow fatty stalk that traversed a small dural dysraphism at the site of a spina bifida at L5–S1 (Fig. 7*A*). A hemilamina was noted at L4. The intradural lipoma could be dissected from the dorsal surface of the conus, revealing a thickened, fatty filum that was divided to untether the cord (Fig. 7*B*). The dura was closed primarily. No terminal cyst was identified. At a follow-up examination 15 months later, the child remained ambulatory with no motor or sensory deficit.

DISCUSSION

Overview

The evolutionary significance of the human embryonic tail remains a mystery. The human tail has been better elucidated by mythologists than embryologists, and literature is rich with descriptions of beautiful mermaids and sirens. What remains

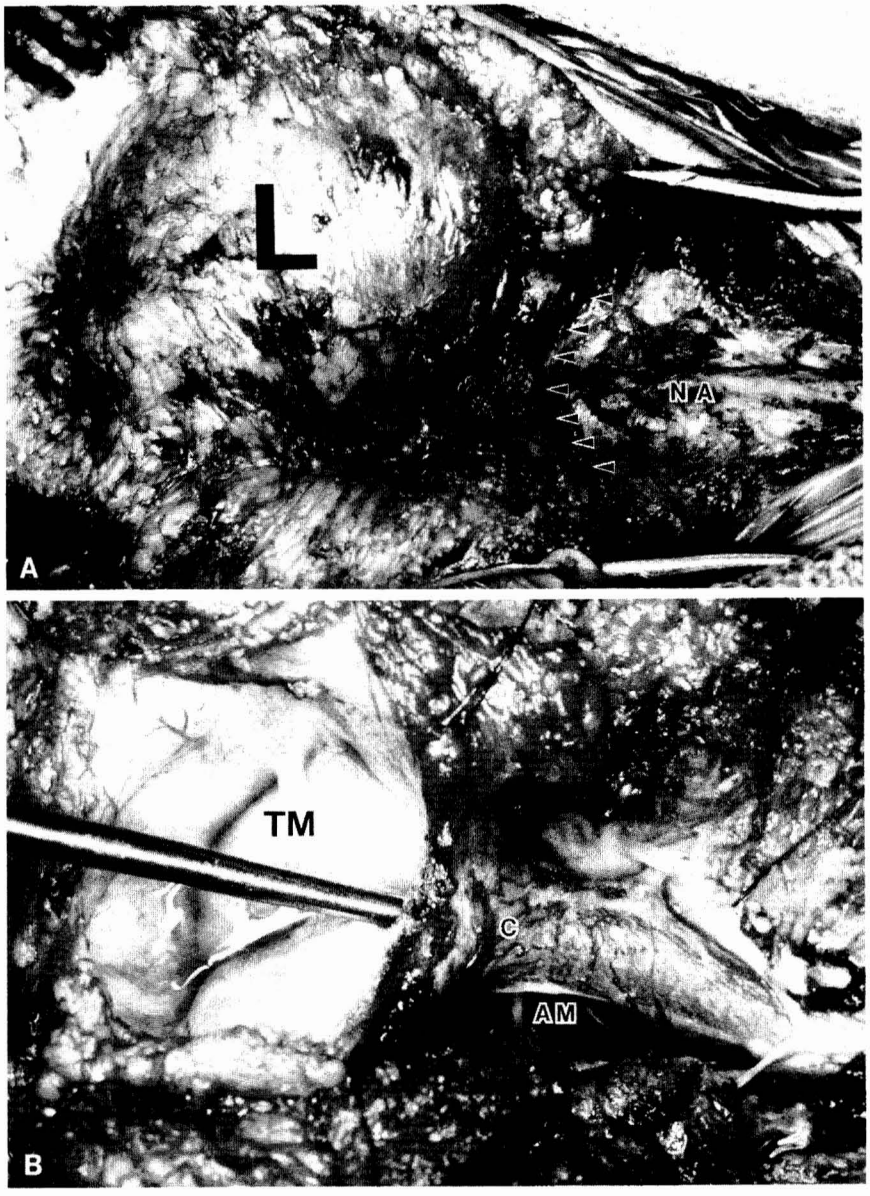

FIG. 4. Case 1. Operative exposure, direct posterior view, rostral to the viewer's right: *A*, dorsal dysraphism with a large subcutaneous lipoma (*L*) anchored by fibrous band (*arrowheads*) at the most rostral bifid laminae (*NA*, last intact neural arch above the dysraphic defect). *B*, view after debulking of the subcutaneous lipoma, division of the fibrous band, removal of the last neural arch, and opening of the dura. The conus (*C*) is low-lying and passes through an anterior meningocele (*AM*) and balloons into a large ependyma-lined terminal cyst (*TM*). A probe inserted into the terminal cyst passes freely into the hydromyelic central canal. Note the rostral course of the nerve roots.

unknown is why and how the human embryo elaborates a complex tail identical to a bluefish or striped bass, only to have this structure vanish completely before the child is born. To date, the development of the caudal neural tube and caudal embryo has received relatively little attention. The present observations suggest that the entire spectrum of abnormalities seen in both cloacal exstrophy and occult spinal dysraphism can be explained by a single defect in the developing embryonic tail.

The study of congenital abnormalities can provide clues about normal embryological development. Some abnormalities can be traced to specific gestational times. When two abnormalities occur together consistently, it is intriguing to search for a common embryological origin. The concurrence in this series of children of two extraordinarily rare congenital malformations traceable to the caudal pole of the embryo may help elucidate the normal development of this structure. Each of five patients with cloacal exstrophy was found also to have occult spinal dysraphism. In four of the five children, the dysraphic abnormality was a terminal myelocystocele. One child had a lipomyelomeningocele with a tight filum terminale and tethered cord. The strong association between cloacal exstrophy and occult spinal dysraphism, especially such an unusual dysraphic variant as terminal myelocystocele, permits one to speculate about the embryological development of the normal neural tube.

Pathogenesis of occult spinal dysraphism

Most of the central nervous system forms by the process of neurulation, in which the embryonic ectoderm thickens to form a plate and then folds upon itself to form a neural tube (15). Neurulation occurs early in embryological development, between the 18th and 27th days of gestation (Streeter horizons VIII–XII; crown–rump length, 1–4 mm). The notochord is contiguous with the ectoderm and is probably an inducer of neural plate and neural tube formation, as it lies just ventral to these structures (15, 21). The neural tube first closes at the level of the 3rd and 4th somites, paired mesodermal segments located at the site of the future occipital bone, and simulta-

FIG. 5. Microscopic section through the terminal myelocystocele demonstrating an ependyma-lined cyst (*C*) surrounded by dysplastic glia and fat (hematoxylin and eosin, ×100).

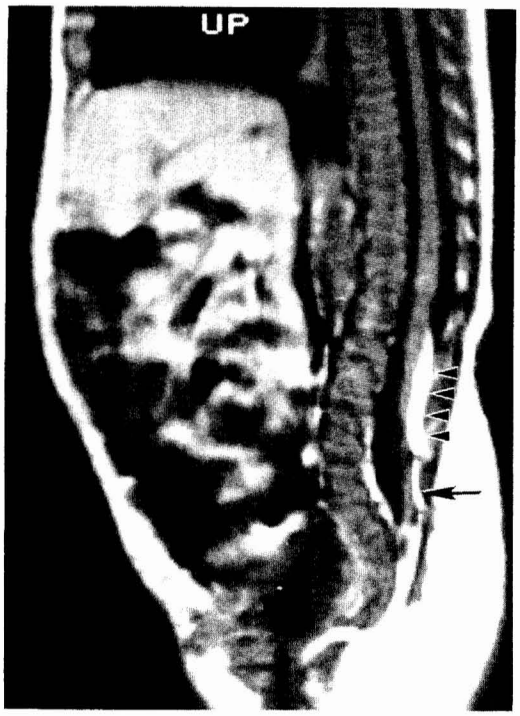

FIG. 6. Case 5. T1-weighted MRI scan, sagittal view. Note the low-lying conus stuck to dorsal lipoma (*arrowheads*). The bright signal caudal to this (*arrow*) proved to be a fatty filum.

neously proceeds in a rostral and caudal direction (31). The rostral neural tube closes at the anterior neuropore, the site of the future lamina terminalis, on Day 24 or 25 (43). Neurulation ends on Day 26 or 27 with closure of the caudal neural tube at the posterior neuropore. The posterior neuropore itself closes in a rostral-caudal fashion, with the site of final closure adjacent to the 2nd sacral vertebral level (42, 44).

The caudal neural tube forms in a different fashion via poorly understood processes of canalization and regression (3, 15, 29). Canalization occurs after closure of the posterior neuropore and occupies gestational Days 28 to 48 (Streeter horizons XII–XX; crown–rump length, 4–22 mm). Canalization begins when an undifferentiated conglomeration of cells, the caudal cell mass, transforms to develop a neural appearance and orient itself about small vacuoles. These vacuoles coalesce to form larger vacuoles, and ultimately become a large central canal that fuses with the more rostral tube formed by neurulation. At this point in development, the future spinal cord is a fluid-filled tubular structure extending caudally to the level of the coccyx (4). Since canalization is less precise than neurulation, it is not uncommon to see variations in development such as accessory lumens and canal forking (28). Toward the end of the period of canalization, the ventriculus terminalis becomes visible as a dilatation of the neural tube at the site of the future conus medullaris (26).

Retrogressive differentiation, the final stage of caudal neural tube formation, is characterized by a regression of structures formed during canalization and results in the complete disappearance of the embryonic tail (27, 31, 48, 54). Retrogressive differentiation begins near the end of canalization at about gestational Day 48 (Streeter horizon XX; crown–rump length, 22 mm) and continues throughout embryonic development and even postnatally. Atrophy of the caudal neural tube is responsible for formation of the filum terminale, a fibrous band anchoring the terminal spinal cord. The filum joins the ventriculus terminalis (future conus) with the coccygeal medullary vestige, a small ependymal cell rest stuck to the coccyx. Earlier in the development of the embryo, there is a level-to-level correspondence between the spinal column and spinal cord. Retrogressive differentiation and a disproportionate growth of the spine and spinal cord are responsible for the relative ascent of the conus to its adult level adjacent to the L1 vertebral body.

The embryological insult responsible for terminal myelocystocele probably occurs during the period of retrogressive differentiation. Since this occurs after neurulation has been completed, the defect is covered by skin. Failure of regression accounts for the abnormally low position of the conus medullaris. The neural tube remains open and becomes a terminal hydromyelia ending in an ependyma-lined cyst.

Pathogenesis of cloacal exstrophy

The striking coexistence of terminal myelocystocele with cloacal exstrophy can be explained by the proximity of the neural tube to the cloaca in the developing embryo (1, 16, 18, 39, 45, 46). An understanding of the defect responsible for cloacal exstrophy may further elucidate the mechanism re-

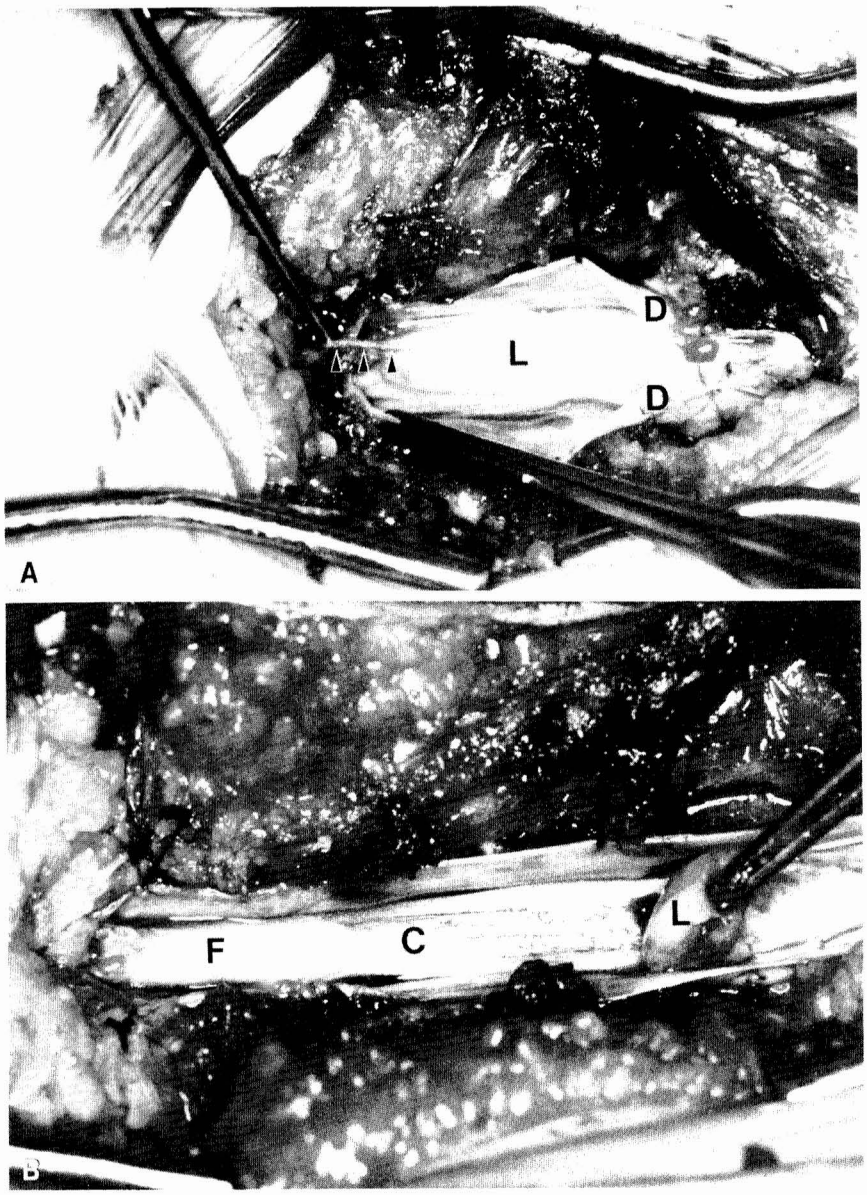

FIG. 7. Case 5. Operative exposure, direct posterior view, rostral to the viewer's right: *A*, the dura (*D*) has been opened, revealing a dorsal lipoma (*L*) stuck to the low-lying conus. A narrow fatty stalk (*arrowheads*) traverses a dorsal dural defect and communicates with a subcutaneous lipoma. *B*, the dorsal lipoma (*L*) has been dissected from the conus (*C*), exposing a thickened, fatty filum terminale (*F*), which was subsequently divided.

sponsible for spinal dysraphism. *Cloaca* is Latin for *sewer*. The cloaca is an endoderm-lined chamber in early embryos into which the hindgut (future bowel) and allantois (future bladder) both empty. A transitory bilaminar veil, the cloacal membrane, sits at the ventral wall of the cloaca and consists of only ectoderm and endoderm. In normal embryogenesis, a urorectal septum grows to divide the single-chambered cloaca into a posterior alimentary system and an anterior urogenital system (53). Along with the craniocaudal march of the urorectal septum, the cloacal membrane atrophies at about 6 weeks' gestation as the primitive streak migrates to form the anterior abdominal wall (24).

The most widely accepted explanation of cloacal exstrophy was offered by Marshall and Muecke (33) and has been called the *wedge effect* [see also Saltzman et al. (50)]. They proposed that overdevelopment of the cloacal membrane creates a mechanical barrier preventing medial migration of the lateral mesoderm to form the anterior abdominal wall. The ultimate rupture of the cloacal membrane creates exstrophy. Rupture of the cloacal membrane after formation of the urorectal septum accounts for the classic syndrome of exstrophy of the bladder. Rupture of the cloacal membrane before formation of the urorectal septum leads to exstrophy of the cloaca.

Association of occult spinal dysraphism and cloacal exstrophy

Although cloacal exstrophy and terminal myelocystocele are two extraordinarily rare and complex entities, both may be explained by a single defect at a specific time in embryonic development. Several observations suggest that inability of the abdominal wall to close at the site of the cloacal membrane may be the cause of this defect. Experimentally, cloacal exstrophy has been induced in chick embryos simply by manipulating the cloacal membrane (41, 56). This implicates the cloacal membrane in the pathogenesis of cloacal exstrophy, but no comment can be made about spinal dysraphism, because the pathology of the spinal cord was not systematically studied. Recently, Alles and Sulik (2) have reported spina bifida cystica induced by retinoic acid in a mouse model. They

used supravital staining and scanning electron microscopy to demonstrate increased cell death in the tail bud, primarily the gastrulating mesoderm. The increased cell death occurred in isolated areas of the primitive streak and appeared to be an exaggeration of physiological cell death in these regions seen in control animals. Thus, a teratogenic insult to the primitive streak can result in spinal dysraphism. In fact, multiple malformations, including both terminal myelocystocele and omphalocele, have been demonstrated in the offspring of pregnant hamsters treated with retinoic acid (52). Thus, failure of the primitive streak to form the anterior abdominal wall is sufficient to explain the morphogenesis of both cloacal extrophy and terminal myelocystocele.

In the clinical setting, cloacal exstrophy is usually associated with terminal myelocystocele or some form of occult spinal dysraphism, but the converse is not true. That is, occult spinal dysraphism frequently occurs unassociated with cloacal exstrophy. This holds for common dysraphic lesions, such as lipomyelomeningocele, and even uncommon ones, such as terminal myelocystocele, which may occur sporadically.

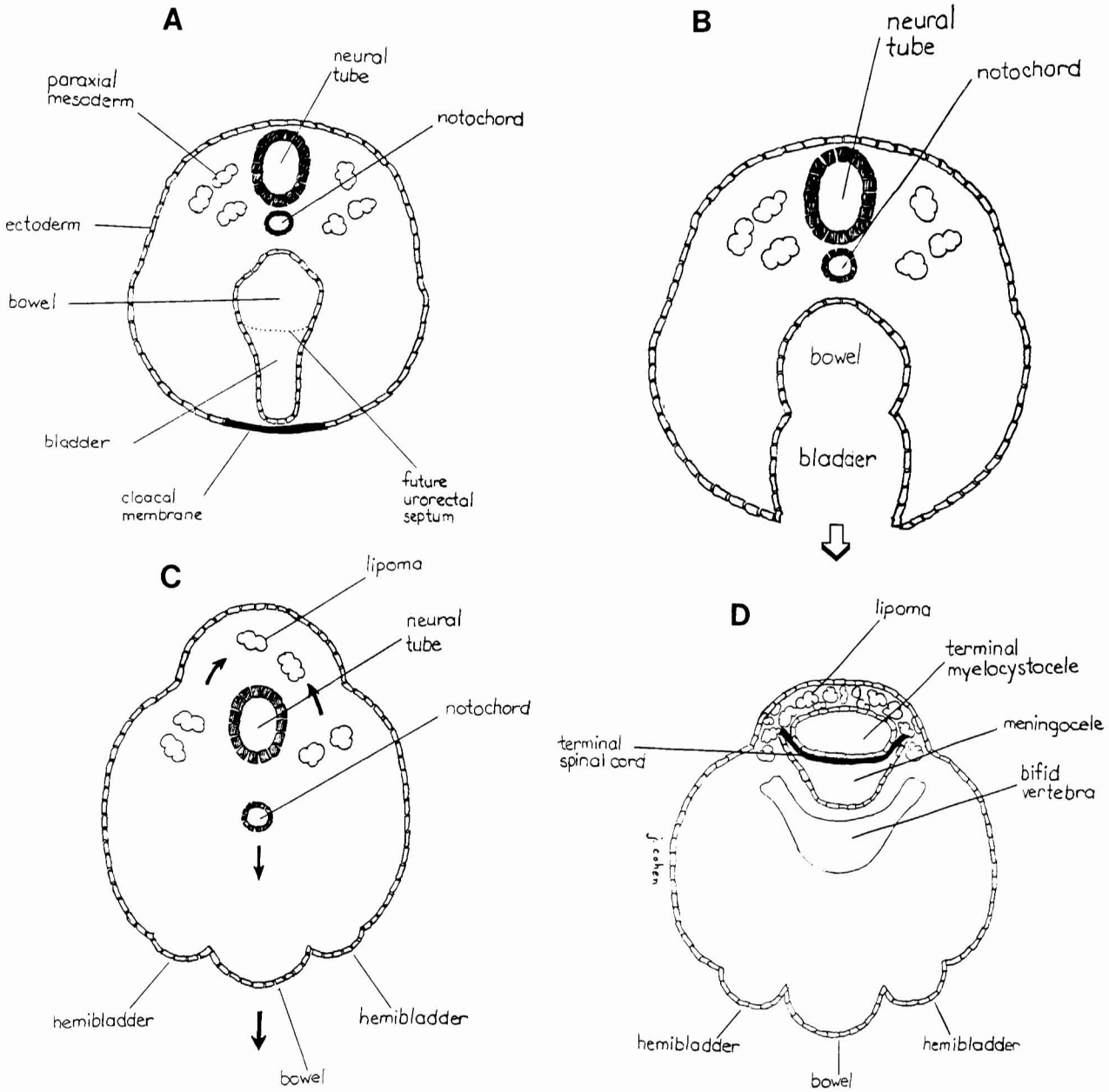

FIG. 8. Diagrammatic explanation of the embryology of cloacal exstrophy and terminal myelocystocele: *A*, horizontal section through the caudal pole of a normal human embryo at about 6 weeks. *B*, dehiscence of the cloacal membrane before formation of the urorectal septum permits prolapse of the future bowel and bladder fields. *C*, prolapse creates a central, exstrophied bowel surrounded by two hemibladders. This prolapse pulls the notochord away from the neural tube and interrupts the signals for retrogressive differentiation. Relative displacement of the paraxial mesoderm explains the dorsal lipoma. *D*, the final result, with exstrophy of the bowel and bladder. The cord is low-lying and tethered, and expands into a large, ependyma-lined terminal cyst surrounded by fat. The spine is bifid dorsally. Forward displacement of the notochord creates an anterior meningocele.

Therefore, it is unlikely that dysgenesis of the neural elements plays a primary role in the development of the bowel, bladder, and genital abnormalities. Also, simple exstrophy of the bladder, a far more common occurrence than cloacal exstrophy, is not usually associated with occult spinal dysraphism. Although a pathological process involving the cloacal membrane is responsible for exstrophy of the bladder, the problem here probably occurs later in embryonic development, after formation of the urorectal septum. The urorectal septum permits formation of a relatively normal hindgut and helps to isolate the neural tube from pathological processes arising from the anterior abdominal wall.

The spinal cord of the early embryo is directly adjacent to the future bowel and bladder. Mechanical stresses associated with flexion of the tail bud could interfere with the normal development of these contiguous structures. Copp et al. (7) studied spinal dysraphism in the curly tail mouse and suggested that an imbalance in the growth of neural and nonneural structures in the tail bud could account for the development of caudal neural tube defects. Consider what would happen to all neighboring structures if there were a single defect in the vicinity of the cloacal membrane. Figure 8*A* is a horizontal drawing through the caudal pole of an embryo at about 6 weeks' gestation. The neural tube is a hollow cylinder and the notochord lies just ventral to it and adjacent to the cloaca, the site of the future bowel and bladder. The cloacal membrane lies at the anterior aspect of the abdominal wall. An insult to the primitive streak leading to increased cell death would prevent the mesoderm of the primitive streak from invading the cloacal membrane to form the anterior abdominal wall. Subsequent rupture of the cloacal membrane before completion of the urorectal septum would create a large abdominal wall dehiscence (Fig. 8*B*). This would then allow the cloaca, with its future bladder field and future bowel field, to eventrate at the anterior abdominal wall, turning itself inside out and opening like a book. The result would be a central bowel field surrounded by two hemibladders (Fig. 8*C*).

Such a major eventration could pull the notochord forward and away from the neural tube. The notochord is the major inducer of neurulation and may be an inducer of retrogressive differentiation. Van Straaten et al. (58) studied the effects of notochord fragment implantation on neural tube development in chick embryos. They found effects on the neural tube only up to 80 μm from the implant, suggesting that contiguity of notochord and neural tube are important. Thus, if the notochord were no longer contiguous with the neural tube, retrogressive differentiation might not occur. The caudal spinal cord would remain a fluid-filled tube and the conus would be low-lying. Mechanical movement of the neural tube anteriorly could allow posterior migration of the paraxial mesoderm and explain the dorsal, skin-covered lipoma. The embryo would have both cloacal exstrophy and terminal myelocystocele (Fig. 8*D*). Dorsally, one sees skin-covered spina bifida and a large subcutaneous lipoma. The hydromyelic spinal cord is tethered and ends in an ependyma-lined cyst, the terminal myelocystocele, which is surrounded by fat. A meningocele is created by the separation of the notochord from the neural tube. Figure 9 demonstrates the multiple deformities in a child born with cloacal exstrophy and terminal myelocystocele.

Thus, a single insult to the primitive streak after closure of the posterior neuropore can explain the pathogenesis of both cloacal exstrophy and terminal myelocystocele. Terminal myelocystocele is a rare form of occult spinal dysraphism accounting for about 5% of all skin-covered lumbosacral masses (36), but it is extremely common in patients with cloacal exstrophy, and was found in four of the five patients in this series. One would predict that, if patients with cloacal exstrophy were screened routinely with a sagittal MRI scan, terminal myelocystocele would be identified even more frequently. The only patient in our series in whom we did not find a terminal myelocystocele had the least severe abnormality of the ventral abdominal wall. She did have spinal dysraphism in the form of a lipomyelomeningocele and tight filum terminale. Lipomyelomeningoceles occur in the general population with much greater frequency than terminal myelocystoceles. The skin-covered dorsal dysraphism in lipomyelomeningocele is very similar to that seen with terminal myelocystocele, but there is no large terminal cyst; however, we have frequently encountered small cysts within the lipoma during surgery for lipomyelomeningocele. Ependymal tracts and rests have been identified in pathological specimens of lipomyelomeningoceles, and even within the normal filum (28, 29, 32). Therefore, lipomyelomeningocele and terminal myelocystocele appear closely related and represent variants of skin-covered dorsal dysraphism.

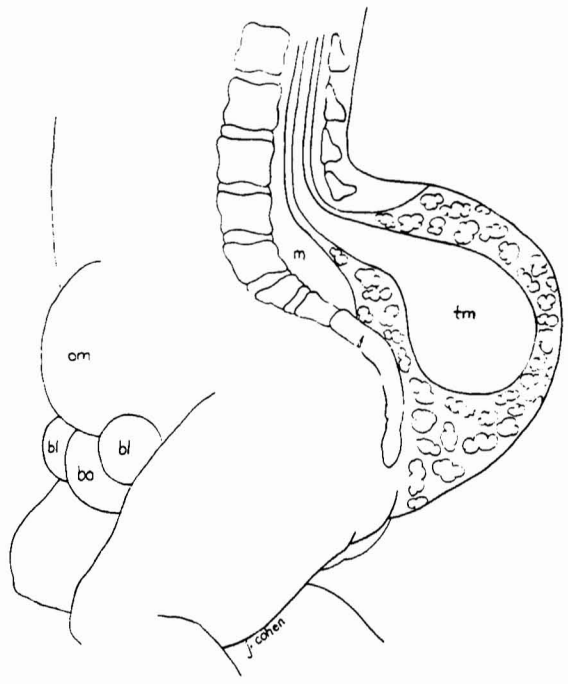

FIG. 9. Sagittal illustration of cloacal exstrophy and terminal myelocystocele. The hydromyelic central canal expands into a terminal cyst lined by ependyma, the terminal myelocystocele (*tm*). Key: *m*, meningocele; *bl*, hemibladders, *bo*, bowel; *om*, omphalocele.

Children with cloacal exstrophy and occult spinal dysraphism present the surgeon with serious ethical and social issues. The children are born with extensive congenital deformities and must undergo multiple gastrointestinal, genitourinary, neurosurgical, and orthopedic procedures. They usually require an ileostomy or colostomy. The genitalia are duplex and ambiguous, and gender reassignment has been recommended to convert genetic males to females, rather than attempt to maintain a male phenotype with inadequate genitalia (19, 22, 55). The moral problems are exceedingly complex. It should be remembered, though, that neurologically, these children usually do quite well. They have neither hydrocephalus nor hindbrain malformations, may have normal intelligence, and often become ambulatory.

CONCLUSIONS

Cloacal exstrophy is strongly associated with occult spinal dysraphism. The association may be due to a single insult to the embryo during its 2nd month of development. A single abnormality at the site of the cloacal membrane could explain all of the findings encountered in cloacal exstrophy and occult spinal dysraphism. The spinal dysraphic abnormality most commonly associated with cloacal exstrophy is terminal myelocystocele. Terminal myelocystocele and lipomyelomeningocele are closely related variants of occult spinal dysraphism. All patients born with cloacal exstrophy or anorectal malformations should undergo evaluation of the lumbosacral spine to search for lesions of the caudal spinal cord.

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COMMENT

Until recently, our understanding of the surgically relevant anatomy of complex caudal dysraphic lesions has been distressingly incomplete. This had been due largely to a lack of detailed studies of pathological material that might guide the surgeon who attempts to unravel the anatomy of lesions similar to those addressed in the present paper. Now, because of better imaging techniques and renewed interest in the careful documentation of what we see at operations, our understanding of occult dysraphic lesions has become much clearer and more systematic. The present paper emphasizes the association between cloacal exstrophy and myelocystocele in particular. The descriptions and the illustrative material contained in this paper should be helpful to anyone confronted with the same problem. Although the author has relied on magnetic resonance imaging alone, I have found computed tomographic myelography to be a useful preoperative adjunct. This allows one to identify the precise relationship between the subarachnoid space and the hydromyelic cavity and to exclude the possibility that the dorsal cerebrospinal fluid space is merely an extension of the subarachnoid space. That information can be quite helpful intraoperatively in maintaining orientation.

I cannot critically review the author's theory of embryogenesis. As an interested reader, however, I remain unclear as to how the proposed etiology relates to cases of lipoma and lesser degrees of occult dysraphism in which there is no associated anomaly of the ventral structures. Secondly, theories of the embryogenesis of myelocystocele should probably take into account the occasional occurrence of a choristoma in the large subcutaneous mass. One of our patients, for example, had a testis and epididymis in addition to a well-formed stomach with a chronic gastric ulcer (1).

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