5. Bladder exstrophy and epispadias complex

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Definition

Bladder exstrophy is a urogenital malformation of the bladder and urethra in which the abdominal wall and the bladder are open below the umbilicus, exposing the insides of the bladder and urethra. The term “exstrophy” is derived from the Greek word “exstrophie”, that it means “the inside is everted”. Epispadias is a deficiency in part of the entire dorsal wall of the urethra.

Exstrophy of the bladder and epispadias complex include a wide spectrum of malformations ranging from continent epispadias to cloacal exstrophy. Specifically this entity include: The classic bladder exstrophy, the continent epispadias, the incontinent epispadias, the pubovesical cleft and the cloacal exstrophy. Associated anomalies: Undescended testes, bilateral inguinal hernia in males, duplication of the uterus and vagina in females. Also ureteral duplications, imperforate anus rectovaginal fistula, cleft palate, vertebral anomalies, spina bifida, congenital dislocation of the hip and clubfoot coexist 15.

History

Bladder exstrophy is an entity that is known circa 2000 B.C. as it is illustrated on Assyrian tablets that are exposed in the British Museum in London 15.
The first medical description was by Von Grafenberg in 1597 and the term extrophy was first used in 1780 by Chaussier. In 1849 Mowat completely described this entity. In 1894 Maydl described a more successful method of urinary diversion with transplantation of the trigone that contains the ureters into the rectum. Coffey, Nesbit and Leadbetter refined the technique in order to prevent the sigmoid -to ureter reflux 3,4,5.

Trendelenburg reported bilateral sacroiliac ostomies and application of a pelvic sling in 1906 in order to protect the anterior bladder and abdominal wall closures. In 1942 Young reported the first female with urinary continence after closure of bladder exstrophy. In 1948 Michon reported success in a male who has had full reconstruction. Lepor and Jeffs (1983), Mesrobian, Kelalis and Kramer (1988) and Ransley (1991) reported 75% to 80% continence rates after staged reconstruction with refinement of urethroplasty, epispidias repair and bladder augmentation 1,2,3,4,5,6,7,8,9,10,11,12,13,14,15.

Incidence

The incidence of the bladder extrophy has been estimated to be 1 in 10,000 to 1 in 50,000 live births. The male to female ratio derived from several series, is between 2.3 :1 and 4:1. Cloacal extrophy is rare, with an incidence 1 in 200,000 to 400,000 live births, an approximately equal incidence in both sexes 15, 16, 17.

Embryology

The extrophy results from a continuation of the cloacal cleft into the infraumbilical body wall. The rupture of the membrane has been attributed to the failure of the phallic primordium to form a mesodermal bar across the midline at the end of the cloacal groove. (Scandalakis –Gray 1972) 18. At the end of the fourth week, the cloacal plate bounds the whole ventromedial portion of the cloaca from the allantoic stalk to the tail gut (Frager 1935). Growth of the ventral cloaca (urogenital sinus) normally carries the insertion of the allantois away from the anterior end of the cloacal plate at about the time the phallus forms (5.0mm). If the anterior tip of the cloacal plate is carried upward by the elongation of the upper part of the urogenital sinus, instead of being left behind, the resulting extension of the cloacal plate and its eventual rupture would account for the open infraumbilical abdominal wall. These
defects are the failure of the mesodermal components of the ventral abdominal wall to develop normally. The infraumbilical wall is derived from mesoderm arising from the primitive streak surrounding the margin of the cloacal membrane and invading the inferior side of the body stalk. If this “secondary mesoderm” fails to form the endoderm of the urogenital sinus remains in contact with the skin ectoderm and eventually ruptures (Wyburn 1937). Paul and Kanagasuntheram (1956) argue that the mesoderm is not absent, but that its lateral halves have failed to fuse. Glenister (1958) reconciled these views by observing that the single layered secondary mesodermal sheet that forms the musculature of the ventral bladder wall is eventually reinforced by extension of the primary somatic mesoderm which forms the voluntary musculature of the remainder of the body wall. If the secondary mesoderm fails to unite at the midline, the somatic mesoderm will also be unable to unite, and the midline will remain a thin, double layer of ectoderm and endoderm. Thus there is no absence of the infraumbilical wall or of the anterior bladder wall in exstrophy but a failure of midline fusion only. Epispadias is the result of an unusual ventral prolongation of the cloacal membrane beyond the phallic primordium. Patients with variants of the exstrophy complex are born. Exstrophy of the cloaca arises from failure of secondary mesoderm from the primitive streak to cover the infraumbilical wall. Midline rupture has occurred earlier (about the fifth week) prior to the fusion of the genital tubercles and prior to the descent of the urorectal septum, which separates the cloaca into bladder and rectum. As the individual cloaca is exposed its central portion is the posterior wall of the gut, while its lateral portions receive the ureters and differentiate into bladder mucosa. Cloacal rupture early in the fifth week results in cloacal exstrophy while the same mishap in the seventh week results in vesicular exstrophy only. The origin of the blind caudal intestinal segment is not clear. Johnston (1913) believed that the blind caudal segment represents the persistent tailgut.

Etiology

According to epidemiological data, a complex genetic as well as a multifactorial mode of inheritance could underlie BEEC. However, no single teratogenic agent or environmental factor has been identified, which could play a dominant role in the expression of the BEEC. A risk of recurrence of 0.5-3% has been described in families with one affected subject. These values correspond to an increased recurrence risk estimated to be as high as 200- to 800-fold when compared to the common population. Due to the paucity of affected sib pairs and suitable multiplex families, conventional linkage analysis to identify candidate genes causally related with BEEC appears to be unfeasible.
The inheritance of BEEC (Bladder Exstrophy Epispadias Complex) may be consistent with autosomal dominant inheritance with reduced penetrance (Reutter et al., 2003) or with autosomal recessive trait or X-linked transmission. Twin studies provide an efficient method to detangle the influences of genetic and environmental factors on a trait (Neale et al., 1994; Martin et al., 1997; Risch, 2001). Insufficient periconceptional folic acid intake and deficient folate metabolism in mothers and fetuses have been acknowledged as risk factors for several midline defects (Botto et al., 2002).

Methylenetetrahydrofolate is required for the conversion of homocysteine to methionine and of deoxyuridine monophosphate to deoxythymidine monophosphate in support of DNA synthesis, and it also serves as a major source of one carbon unit for purine biosynthesis. All of these pathways might be affected by the methylenetetrahydrofolate reductase (MTHFR) C677T polymorphism that causes an A222V substitution in the protein, thereby giving rise to a thermolabile variant with reduced activity (C/T, 35% reduction; T/T, 70% reduction) (Frosst et al., 1995). In that context, Mills et al. (2005) tested, apart from the MTHFR (A222V) variant, several polymorphisms in genes encoding proteins (MTHFD1: methylenetetrahydrofolate dehydrogenase [R653Q]; SLC19A1: folate transporter [R27H]; TCN2: transcobalamin II [P259R]) involved in folic acid metabolism for association with omphalocele in 25 cases. Although the latter three showed no significant association, the T-allele encoding the thermolabile MTHFR variant was associated with an increased risk ($p = 0.035$) for this malformation. Genetic factors have implicated in the formation of exstrophy bladder–epispadias complex.

Expression of the p63 transcript at gestational days (GD) 9.5-12.5, the equivalent of human gestational weeks 4-6 (postulated time of BEEC organogenesis in humans) was studied. In addition, p63 expression analysis was performed in human blood and bladder derived samples of 15 BEEC newborns accompanied by sequencing analysis of their genomic DNA. In mouse embryos, p63 expression was detected at days 9.5-12.5 in the cloacal membrane and urethral epithelium, supporting its role in the morphogenesis of the external genitalia and the bladder. Tissue-specific expression of a novel and already-known mRNA isoforms were established and a reproducible dysregulation of variable p63 isoforms was observed in 11 of 15 patients indicating altered gene expression. However, no obvious p63 gene mutations were identified in any of the patients. These findings strongly suggest that p63 is not only involved in embryonic formation of the urogenital and ventrocaudal anatomy but is also highly dysregulated in human BEEC bladder tissue. Since p63 has been shown to self-regulate its expression through a balance of its isoforms, the dysregulation observed may contribute to the formation of BEEC. Cytogenetic and molecular analyses have revealed chromosomal
anomalies in 20 patients with BEEC although none of these anomalies appear to be causative. Numerical chromosomal aberrations (47,XXX; 47,XXY; 47,XYY; 47, [no sex reported], +18; 45,X0/46XX [mosaic]) were observed in six patients and, interestingly, were found in association with Down syndrome in an additional four CBE males, one CBE female, and one girl with CE. Aneuploidy of sex chromosomes in five of these cases might point to a gonosomal loci, or locus, involved in the formation of BEEC. The transcriptional activity of *Cyr61* during the development of the external genitalia was performed with in situ hybridization in mouse embryos at GDs 9.5, 10.5, and 11.5, with particular emphasis on the region of the ventrolateral trunk. Another gene which its expression was detected at GDs 10.5 and 11.5 and was found predominantly in the major vessels of the ventrocaudal trunk and the umbilical cord was the gene *Cyr61*. A strong and specific signal was detected in the left and right umbilical arteries, which are positioned between the hind limb bud and the genital tubercle and in the arteries of the umbilical stalk. The course of these vessels runs adjacent to the mesenchyme of the cloaca, the genital tubercle and the urogenital sinus—structures that are involved in the morphogenesis of the external genitalia and the bladder. *Cyr61* was also transcribed in a subpopulation of somite cells. To analyze *Cyr61* expression histologically, vibratome sections of the caudal trunk were taken from the embryos. These sections clearly demonstrate that the ventrocaudal expression of *Cyr61* was confined to the endothelium of the vessel systems. The gene encoding *CYR61* is located on human chromosome 1p22.3, a region that is not known to be rearranged in association with EEC. However, it was found to be the most downregulated gene in a study of the response to single-dose prenatal administration of FLU (Foster and Harris, 2005). The observation that FLU exposure on GD 16 induced the development of epispadias in approximately 12% of rat offspring (30% of the litters), led us to investigate *CYR61* in humans. It is possible that FLU generates several metabolites (activated by P450s), which have a variety of effects (Kang et al., 2008). This gene functions as a ligand of integrin receptors and promotes the proliferation, migration, and adhesion of endothelial cells and fibroblasts (Kireeva et al., 1996). It is a direct target of STAT3, an important regulator that inhibits the differentiation of embryonic cells into mesodermal and endodermal lineages (Bourillot et al., 2009). *CYR61* activity might affect the mesenchyme of the urogenital system as a secreted factor that promotes cell proliferation, chemotaxis, angiogenesis, and cell adhesion. This mode of action could be achieved from a distance, with *CYR61* acting as a soluble factor traveling throughout the infrabulbar abdominal wall or by other factors regulated by *CYR61*. Spatiotemporal analysis of gene activity in the mouse embryo provides important information regarding the possible function of a gene in any given
developmental process. The detailed expression analysis, which focused on the ventrocaudal trunk region during the developmental stages that are critical for the development of the malformations of the EEC, showed that Cyr61 is strongly and specifically expressed in the endothelial cells of the ventrocaudal embryonic and extraembryonic vessels—that is, the left and right umbilical arteries and the vessels of the umbilical stalk. These vessels surround the cloaca and the genital tubercle. It is therefore possible that the Cyr61 gene product is involved in the development of the external genitalia. This would be compatible with the role of Cyr61 in promoting proliferation and differentiation of mesenchymal cells (Wong et al., 1997). Cyr61 is a secreted molecule, and the whole mount in situ hybridization analysis suggests that the endothelial cells of the umbilical vessel system display the source for Cyr61 signaling. However, secreted Cyr61 remains associated with the cell surface and extracellular matrix. This observation makes its diffusion over long distances, and an activity as a wide range signal, unlikely (O’Brien and Lau, 1992). Signaling by Cyr61 should affect mainly those mesenchymal cells that are in close proximity to the umbilical vessel system. It remains to be shown whether these mesenchymal cells could function as driving force for the regular establishment of the external genitalia and the infraumbilical abdominal wall (e.g. by a higher proliferation rate or increased migratory behavior). In addition, it cannot be ruled out that the mesenchymal cell itself expresses Cyr61 at a level that is not detectable by whole mount in situ hybridization, but could be sufficient to enhance proliferation and migration. It is possible that FLU generates several metabolites (activated by P450s), which have a variety of effects (Kang et al., 2008).

Clinical presentation and associated problems

Patients with variants of the extrophy complex are born full term. The incidence of prematurity increases in extreme cases of bladder extrophy in particular, in cloacal extrophy. In patients with superior vesical fissure or cloacal extrophy, skeletal defects, myelomeningocele and cardiovascular anomalies are common. At birth the typical appearance of bladder extrophy – an everted posterior bladder wall of varying size associated with separation of the symphysis pubis – is obvious. The umbilicus is located at the immediate superior border of the bladder plate and a small umbilical hernia is seen. The mucosa of the exposed bladder may look normal or a few inflammatory polyps there are on its surface. With exposure and chronic inflammation, the mucosal surface becomes thickened and polypoid. As well the bladder wall musculature becomes fibrotic, rigid and unyielding. The symphysis pubis presents some
degree of widening caused by outward rotation of the innominate bones in relation to the sacroiliac joints. This event causes an outward rotation and eversion of the pubis rami at their junction with the ischial and iliac bones. Inguinal hernias are commonly seen in children with bladder extrophy. In addition the testes may appear undescended. Also the distance between the umbilicus and the anus is foreshortened, making the perineum appear short and wide. The anal canal and the anus itself are usually normal but may also be stenotic and require dilatation. In rare instances the anus is ectopic, presenting as a rectoperineal or rectovaginal fistula. The anal sphincter mechanism is also displaced anteriorly and the levator ani complex is divergent. The latter factor leads to weakness of the perianal floor and rectal prolapse in 10% to 20% of patients.  

**Genital defects in the males**

The penis appears foreshortened because of the wide separation of the symphysis pubis and the attachments of the corpora cavernosa. A prominent dorsal chordee is usual present and the urethral groove is short. In cases with severe dorsal chordee, the penis is short and the glans is noted to be adjacent to the verumontanum. If the penis is dystrophic or severely hypoplastic however, sex conversion may be appropriate. Therefore, careful evaluation by an experienced physician at the time of birth is critical if such a decision for sex conversion is to be considered. On inspection, the glans is flattened and the halves are everted, and the prepuce is noted on the ventral surface. The base of the penis and the scrotum are widely separated. The corporal bodies and the autonomic cavernous nerves are displaced laterally. The ejaculatory ducts and vas deferens are normal and their entrance into the posterior urethra is evident. The urethra is an open strip that extends from the bladder to the tip of the penis. This urethral strip is only about one third to one quarter the length of the normal male urethra. The transition between the bladder and urethra is indistinct. The verumontanum is exposed and may be normal or eccentric. The openings of the ejaculatory ducts may be obvious. The scrotum is flat and wide. The testes are usual descended but are often widely separated within the scrotum. The urogenital diaphragm is distorted and is not perforated by the urethra. Therefore the sphincter urethrae muscle that surrounds the urethra is not formed. In continent epispadias, the epispadiac urethral orifice is either glandular or penile and a groove extends from the normal site of the urethral meatus on the dorsum of the glans as far as the epispadiac orifice. In incontinent epispadias, associated with urinary incontinence, the urethral meatal orifice tends to be more proximal, the
urethral lumen wider, and the pubic bone separated at the symphysis. In case of pubovesical cleft, there is a complete cleft of the urethra to the bladder neck, but the bladder is otherwise intact 15.

Genital defects in the females

The clitoris is bifid with divergence of the labia, mons, pubis and the clitoral halves. The urethra and vagina are short and the vaginal orifice is often stenotic. The uterus, fallopian tubes and ovaries are normal. There are occasional cases of duplicated vagina and uterus, as well as uterine prolapse. The sphincter vaginae, the sphincter urethrae and the transversus perineii are represented as a relatively undifferentiated transverse skeletal muscle that runs behind the vagina and fuses in the midline posteriorly with the perineal body. Therefore, the urinary sphincters are abnormal. There is no external sphincter near the urethra. Rectum and anus appear to be anteriorly displaced. The external and internal anal sphincter appears to be normal, but the elevator ani and the puborectalis sling are abnormal. The pubic attachments are widely separated and the puborectalis becomes a widely open C shape. This results in a less efficient sphincter and an increased tendency for rectal prolapse 15, 38.

Classic bladder exstrophy

Classic bladder exstrophy has a completely open urethral strip. In addition the bladder is open on the surface of the lower abdomen. More severe is the association of the classic bladder exstrophy with imperforate anus. The pelvic girdle is an open C-shaped structure. The hips are externally rotated, and the thighs are widely separated by a broad perineum. There is no pubic symphysis. The pudendal vessels and nerves are also widely separated. The abdominal wall in suprapubic region is replaced by the exstrophied bladder. In the upper abdomen, the recti are usually contiguous, but in the lower abdomen the recti diverge and attach to the superior aspect of the widely separated pubic bones. The umbilical cord is usually in this area, although it is sometimes situated further superiorly. There are three vessels, the single umbilical vein and the two umbilical arteries. The urachus is sometimes a short, thick cord running from the bladder to umbilicus. Even a small omphalocele may be present. The external oblique, internal oblique and transversus abdominal muscles are foreshortened in the lower abdomen because of the lateral displacement of the recti. The inguinal canal is laterally displaced and its contents are normal although the processus vaginalis is frequently patent. The bladder area is relatively small. The trigone tends to be retracted and smooth, whereas the
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The remainder of the mucosa is usually irregular with polypoid appearance. The ureteric orifices may be obvious and widely gaping 15.

**Cloacal exstrophy**

The various forms of cloacal exstrophy are thought to be related to different levels of descent of the urorectal septum. The hemibladders are situated on either site of the centrally placed exstrophied strip of intestine (usually cecum). However, the hemibladders may be confluent either superior or inferior to the intestinal patch. The bladder may be covered in whole or only on one side. The genitalia also may have a varying appearance. The penis or clitoris may be widely split or united. The uterus or vagina may be duplicated in 2/3 of patients and vaginal agenesis may be seen in 1/3 of patients. Also there are inguinal hernias and undescended testes in males. Skeletal anomalies including congenital dislocation of the hip, talipes equinovarus and various limb deficiencies are common in up to 50% of patients. The typical appearance includes split bladder halves and a centrally placed island of exstrophied intestine, usually cecum, with two prolapsed lumens, one for the appendix and the other the terminal ileum. There may be only a single opening with multiple apertures or multiple apertures may be evident with a proximal orifice superiorly and a distal bowel inferiorly. Either one or two appendiceral orifices with duplication of the appendix are present half the time. The small intestine may be foreshortened or with normal length. Associated intestinal anomalies include malrotation, intestinal atresia and Meckel’s diverticulum. Various neural, cardiovascular and pulmonary anomalies including myelomeningoceles, various cyanotic and acyanotic forms of congenital heart disease, double aortic arch and pulmonary anomalies have been reported 15, 65.

**Rare variants of bladder exstrophy**

Rare variants of bladder exstrophy include: covered exstrophy, duplicate exstrophy, superior vesical fissure and pseudoxstrophy. In covered exstrophy patients present with diastasis of the symphyses pubis and rectus abdominis muscles. The urethra is tubular and covered but the bladder may be covered only by skin. There may be a small vesicocutaneous fistula. In duplicate exstrophy patients present with diastasis of the symphyses pubis and a patch of exstrophic bladder just below the umbilicus with either a normal or a smaller than normal complete bladder underneath. In superior vesical fissure the diastasis of pubis and recti are present but the only bladder event ration is below the umbilicus. A small opening into the bladder often permits some degrees of bladder prolapse on the anterior abdominal wall. In pseudoxstrophy patients present with a low
umbilicus and some degree of separation of the rectus muscle attaching to modestly separated pubic bones. When full, the bladder may bulge outward between the separated lower abdominal wall structures but the urinary tract is completely normal 15, 30, 34.

Diagnostic studies

The prenatal ultrasonographic findings include the presence of normal kidneys and amniotic fluid, the absence of the bladder and diastasis of the pubic bones. Color Doppler has been proved to aid the diagnosis of bladder exstrophy by depicting the urine flow in direct communication with the abdominal cavity and has been useful in showing the course of the perivesical umbilical arteries. Prenatal 3D ultrasound with tomographic ultrasound imaging (TUI) and antenatal MR imaging might be useful adjuncts to conventional 2D scan in aiding the prenatal diagnosis of such malformation. After birth renal ultrasonography is performed to determine whether two kidneys are present, whether one or both are dysgenetic or whether evidence indicates hydrourteronephrosis. Physical examination permits estimation of the size of the bladder plate, how many ureteral orifices are present, the opening of ejaculatory ducts and the status of the vagina and the phallic components. In patients who undergo staged reconstruction of the bladder and lower urinary tract over several years, additional imaging and functional studies, including renal ultrasonography, voiding cystogram and radionuclide and urodynamic studies are performed 29, 15.

Treatment

Treatment in classic bladder exstrophy

The first step is staged functional reconstruction, the second step is urinary diversion (when reconstructive options are not available or when repeated efforts at functional reconstruction fail). Sex conversion becomes a consideration in males who have inadequate phallic tissue for male orientation and the decision must be the result of a medical team. The first reconstructive stage is performed in the immediate neonatal period. This is the most favourable time from the standpoint of pliability of the pelvic ring. Furthermore, reconstruction in the newborn period prevents the damage and scarring of the bladder plate. The bladder is completely mobilized with preservation of its blood supply. The corpora cavernosa are dissected off the inferior pubic ramus as far as permitted to preserve the neurovascular bundles. The corpora are then approximated in the midline to promote penile
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elongation. The paraexstrophy flaps are mobilized and are then approximated in the midline to the base of the bladder. Then the bladder and neourethra are tubularized after exteriorization of the ureteral stents and placement of a Malecot suprapubic tube in a small silastic urethral stent. After closure, this permits the pubis to be approximated anteriorly to protect the bladder closure and urethra reconstruction from tension and possible disruption. Marshall, Muecke and Csontai et al approximate the pubis but not use osteotomies. Others use anterior innominate or superior pubic ramus osteotomy. Other authors such as Mollard, favour posterior iliac osteotomy. Patient’s follow up include determination of residual urine and renal ultrasonography. Antimicrobial drugs should be continued because vesicoureteral reflux is present in almost all cases and urinary tract infection can damage the upper tracts. In the male the second stage of repair includes reconstruction of the phallus. This surgery is performed between 6 and 12 months of age. Gearhart and Jetts have shown that this stage also enhances bladder capacity as a result of increased outflow resistance. In the repair of epispadias the goals are to provide adequate length of the phallus with appropriate dangle and release of dorsal chordee. The final stage of reconstruction concerns the reconstruction of a continence mechanism. This is generally undertaken around 4 years of age. Before this the patient has been periodically evaluated with renal ultrasonography. It is important to avoid urinary infection and to ensure adequate urinary drainage without hydronephrosis. The bladder capacity is also measured and this must be greater than 60 ml in order to have enough functional capacity after bladder neck reconstruction. The Young-Dees-Leadbetter technique remains the most common procedure for reconstruction of the bladder neck. In this procedure, the ureters are mobilized and reimplanted in a cephalad position in the bladder either by a cross-trigonal or cephalotrigonal procedure. In patients who remain incontinent after bladder neck reconstruction, will require augmentation of the bladder in order to decrease intravesical pressure to an acceptable level for continence. All segments of intestine have been used for augmentation. The ileum is the most preferred in most cases. Patients with augmentation require clean intermittent catheterization and the use of Mitrofanoff procedure has markedly changed their management. In this procedure, the appendix is anastomosed to the bladder in an antirefluxing fashion and brought through the abdominal wall as a continent catheterizable stoma. Other options include augmentation of outlet resistance with collagen or Teflon injections, repeated Young-Dees-Leadbetter surgery with or without augmentation cystoplasty, artificial urinary sphincter, closure of the bladder neck with a continent catheterizable stoma and urinary diversion. Urinary diversion is preserved for patients who have inadequate bladder plates or those in whom all attempts at reconstruction fail. The first
approach to diversion was that of ureterosigmoidostomy. However, problems of intermittent pyelonephritis, hyperchloremic acidosis, ureteral obstruction, rectal incontinence and adenocarcinoma at the ureterocolic anastomosis have been reported 33, 35, 15, 40, 44, 45, 46, 47, 48, 49, 50, 51, 52, 53, 54, 55, 57, 58, 59, 60, 62, 63, 31, 37.

**Treatment in cloacal exstrophy**

The spectrum of anomalies of cloacal exstrophy includes:

1. Omphalocele and abdominal wall deficiency
2. Bladder exstrophy (two exstrofied hemibladders and interposed midline colonic segment)
3. Ileocecal exstrophy (superior – orifice prolapsed ileum, single or duplicate inferior – orifice short blind – ending hindgut and duplicate appendiceal stumps in intermediate position)
4. Imperforate anus
5. Split symphysis anomaly
6. Bifid external genitalia (diminutive penis, bifid clitoris and labia and cryptorchidism)
7. Duplex mullerian structures (exstrophied vagina and atretic vagina)
8. Myelomeningocele
10. Renal anomalies (agenesis or multicystic kidneys, megaureter, hydronephrosis, ectopia and fusion anomalies).

Extensive repair is undertaken within 48 hours of birth to take advantage of the pliability of the pelvis in the neonatal period. The approach to management of the present omphalocele is closure with complete repair of the abdominal wall. Primary omphalocele closure is possible in at least 80% of cases. On gastrointestinal reconstruction, regardless of whether or not the small-bowel length is normal, every effort should be made to preserve the entire intestine, because of the additional need for use of segments of the intestine track for continent urinary track reconstruction. At the time of repair, the mucosal junction between the bladder wall on each side and the exsrophied intestine should be incised, preserving every bit of intestinal mucosa. The ileocecal junction is tubularized and brought out as an end colostomy well laterally in order to avoid contamination of the abdominal wall- closure. Some patients have intact colons with an anteriorly placed anus, in which case anoplasty may be required. The pubis rami are approximated either with or without iliac osteotomy and closure of the bladder is performed. In males with
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Cloacal exstrophy reconstruction of genitalia is rarely possible because of the diminutive nature of the male external genitalia. Therefore, most of the authors recommend that the males with this anomaly should be converted to the female sex at the time of the initial operation in the newborn period. Final reconstruction of internal and external genitalia is best deferred until later, when the child is of adequate size. Corporeal tissue is preserved for construction of a clitoris, and split scrotal remnants are used to construct labia and lower portion of vagina. Orchietomy is best performed at the time of birth. Up to 50% of children with cloacal exstrophy have either myelomeningocele or a lipomeningocele. In case of leakage of spinal fluid a priority closure of myelomeningocele defect is usually undertaken. Otherwise, closure of the posterior defect can be deferred for 4 to 6 weeks 15, 64.

Complications

Wound infections (>42%), wound breakdown (>25%), abdominal distention, penile ischemia, penile urethral fistula, urethral stictures, urethral stenosis, urethral calculi, bladder prolapse bladder calculi, infection and dehiscence and renal damage have been noted. Also, loss of urethral and suprabubic catheters, urinary incontinence, urethrocutaneous fistula, adenocarcinoma of the bladder (due to chronic inflammation, infection and metaplasia), adenocarcinoma of the ureter colic anastomosis (due to long term contact of the intestinal mucosa with urine) have been reported. Squamous cell carcinoma and rhabdomyosarcoma have been described in patients with bladder exstrophy. Diminished fertility and retrograde ejaculation in males is related to injury to the verumontanum during initial closure. Uterine prolapse has been reported in women 15, 32.

Discussion

Bladder exstrophy –epispadias complex is a severe congenital anomaly with a complicated nature and many attendant problems. The survival for 1960s was in the range of 20% and for the 1970s was 50%. In the 1980s survival has increased to 90% where it stands today. Despite improvements in modern surgical reconstructive techniques, the major problem of urinary incontinence remains. Continent diversion is performed to achieve continence and in order to improve quality of life. In a study of the quality of life in adults with bladder exstrophy –epispadias complex (Wittmeyer at al), the quality of life scores in their patients were less than the norm based scores on 2 of the 8 health concepts including limitations in physical activity due to health problems and general health perception. Results were statistically different
among patients depending on dryness, voiding and urinary reconstruction. Patient’s scores did not differ in regard to gender, number of interventions, sexual life, cosmesis or renal function. Despite a high degree of social integration and adult adaptation, children and adolescents with extrophy bladder suffer from psychosocial and psychosexual dysfunction. Anxiety about genital appearance and sexual activity is a common phenomenon among them, even when they present with near normal genitalia 36, 37, 15, 39, 41, 42, 43, 56.

References


