Hirschsprung’s Disease

A full-term male infant (3,400 gm) was delivered following an uncomplicated pregnancy of a gravida II, para II mother. Initial physical examination indicated a normal infant, but there was a delay in the passage of meconium and by the third day there was progressive abdominal distention accompanied by bile-stained vomitus. The infant was transferred to a special-care ward. A previous male sibling had died at 2 months of age of what was described as intestinal obstruction.

EXAMINATION

Rectal examination induced passage of meconium, followed by visible deflation of the distended abdomen; this proved temporary, however, and the distention recurred within 24 hours. Barium enema revealed a dilated sigmoid colon and a collapsed distal rectal segment. A film taken 24 hours later disclosed barium retention in the dilated colonic segment. Rectal biopsy confirmed the absence of ganglia, which were replaced by clusters of enlarged unmedullated nerve fibers in the submucosal (Meissner’s) and intermuscular myenteric (Auerbach’s) plexuses of the rectum.

DIAGNOSIS

Short-segment congenital aganglionic megacolon (Hirschsprung’s disease, hypoganglionosis, immature ganglia, segmental dilatation, congenital intestinal aganglionosis).
X-ray study showing barium retention in the dilated sigmoid colon, arrow, in Hirschsprung's disease.
Conservative treatment utilizing daily rectal washes with saline yielded unsatisfactory results, and a colostomy was performed on the normal segment colon just proximal to the collapsed aganglionic rectum. 

![Dilated distal colon](image)

**Fig. 41 Modified Duhamel operation:** A. The dilated distal colon is removed, and the rectum closed at the level of the pectinate line. C. Incision and re-established with lower rectum (From Kieswetter, W. B.: Arch. Surg. 79:111, 1963 Copyright 1963, with permission).

Two years the colostomy was closed, and a rectosigmoid modified Duhamel technique (Fig. 41) was performed by proximal ganglionated bowel to the rectum using a side-to-
h-outs of normal through the left dilated sigmoid segment. After the patient showed normal growth and development.

DISCUSSION

In 1888, Hirschsprung described the clinical and autopsy findings in two infants who had died of constipation associated with dilatation and hypertrophy of the colon. He was the first to recognize that a congenital malformation might be the cause of this disease, which occurs in 1 of 25,000 live births. Hirschsprung's disease may occur as a short- or long-segment type and 90 per cent of the short-segment involvement occurs in infants. By 1901 Tittel had reported cellular degeneration of Auerbach's and Meissner's plexuses distal to the dilated segment in the apparently normal colon. Swenson and Bills in 1948 performed the first curative operation for megacolon utilizing a "rectosigmoidoectomy".

Embryology

How do you explain embryologically the presence of a congenital megacolon? Those structures embryologically derived from the hindgut (distal transverse colon, descending colon, sigmoid colon, rectum, and part of the anal canal) are innervated predominantly by parasympathetic fibers via the pelvic splanchnic nerves, and it was suggested that failure in development of these pelvic splanchnic nerves might produce a congenital megacolon. Bodian conjectured that a genetically determined congenital megacolon resulted from a failure of the cells of the dorsal crest of the neuroectoderm to migrate in a normal craniocaudal direction; an early disturbance would cause a long aganglionic segment, a later disturbance would produce a short aganglionic segment. Wildegans showed that the ganglion cells within the wall of the intestine differentiate between the seventh and eighth week of gestation in a craniocaudal direction independent of any parasympathetic nerve supply. Studies by Okamoto and Ueda indicated that the ganglion cells enter the cranial part of the intestine during the sixth or seventh week of gestation, probably via the vagus nerve, and migrate caudally between the muscle layers of the intestinal tube, with the myenteric plexus maturing several weeks earlier than the submucosal plexus. From these theories, it appears that in Hirschsprung's disease, due to arrested development, the course of caudal migration of the ganglion cells is interrupted.

In the neonate, what intestinal malformations or syndromes may
mimic aganglionosis and must be considered in a differential diagnosis of congenital megacolon. Meconium plug syndrome, inspissated milk syndrome, immaturity of bowel functions, necrotizing enterocolitis, meconium ileus, intestinal malrotation, and lower ileal atresia must be considered in a differential diagnosis.

Appearance of Gut

Describe the histological appearance of the segments of gut involved in the present case of congenital megacolon:
1. Dilated proximal segment of megacolon (distal sigmoid colon). Normal ganglion cells are present.
2. Junctional zone (cone) of gut between sigmoid colon and rectum. The few ganglion cells present are abnormal in appearance.
3. Collapsed distal segment of megacolon (proximal rectum). Ganglion cells are absent and replaced by an increased number of clusters (whorls) of enlarged unmedullated nerve fibers.

Methods of Treatment

What are the major methods of treating congenital megacolon? Surgery has been the most successful treatment since 1948 when Swenson and Bill introduced “rectosigmoidectomy” (pull-through), a technique most suitably used in children past infancy. A modification of the Swenson and Bill technique by Duhamel is more frequently employed on infants, as in the present case. Recently, Soave introduced an abdomino- perineal approach which avoids pelvic dissection. The normal segment of colon is carried through a portion of aganglionic rectum and united to the anal sphincter, forming an anocutaneous anastomosis.

Associated Malformations

What malformations might be associated with congenital aganglionic megacolon? Megalobradder and megaloureter occasionally accompany a congenital megacolon. Additional abnormalities include mongolism, cardiac malformations, malrotation or nonrotation of the midgut, Meckel’s diverticulum, and congenital short bowel.

REFERENCES

Imperforate Anus

Initial inspection of the perineal area of a full-term newborn male infant revealed no anal opening. Pigmented skin surrounded an anal dimple. Upon “thumping” the buttock, the area surrounding the dimple contracted, indicating an intact and functional external anal sphincter.

EXAMINATION

Squamous cells and small amounts of meconium were identified in a urinalysis, signifying a fistular connection between the cul-de-sac of the gut and the urinary system. Further examination was postponed for 24 hours, allowing gas to enter the lowest part of the bowel prior to x-ray evaluation. The infant was placed in an inverted position for three minutes, after which the perineal area was painted with barium paste, the knees flexed, and two lateral pictures made. The x-rays showed the rectum ending in a blind pouch above the pubococcygeal line of Stephens (line from the center of the pubis to the sacrococcygeal junction). Dye injection via the urethra confirmed a fistular connection between the blind rectal pouch and the prostatic urethra.

DIAGNOSIS

Imperforate anus (type III, high lesion) with rectourethral fistula

TREATMENT

No additional malformations were noted and surgery utilizing an abdominoperineal approach was immediate. Mobilization of the bowel
and division of the fistular connection was made through an incision in the anal dimple allowed the mobilization pulled through this newly formed opening. Care was taken through the puborectalis muscle, which acts to preserve sensory and motor nerves to the levator ani and genitourinary systems. Four days after surgery a moment occurred, and by the tenth day regular dilatation of the formed anus were begun and continued at home by the infant was 1 year old, fecal and urinary continence and bowel movements were regular. The complete pair cannot be evaluated until toilet training is begun.

**DISCUSSION**

Imperforate anus, which occurs in 1/5,000 births, was described by early Greek, Roman, and Arabic physicians. In A.D. 634, a Byzantine physician, Paulus Aegineta, described the first mention of surgical intervention, while Littre described the use of colostomy as a palliative procedure. In 1936, one of the most significant contributions to this field was the method of perineal dissection to locate the rectal pouch abdominoperineal approach for repair of an imperforate anus was initiated in 1948 by Rhodes, Pipes, and Randal.

**Embryology**

Explain the formation of the rectum. In the how do you account for the imperforate anus?

Embryonic changes from the third to ninth week of life transform a common chamber (cloaca) into a digestive tract and a digestive portion.

By the end of the third week (Fig. 42), the primitive cloaca receives the paired mesonephric ducts, and the hindgut forms a primitive cloaca. The cloaca itself is separated from the hindgut by a cloacal membrane composed of entoderm and mesoderm.

At approximately the sixth week (Fig. 47B), the cloaca is a mesodermal mass covered with peritoneum, becoming anteriorly and ventrally, resulting in the formation of the sinus and a dorsal primitive rectum; moreover, the posterior the bladder and the hindgut is narrowed to:

By the end of the seventh week (Fig. 50D), see
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genital passage from the rectum to the urorectal septum with the cloacal membrane; the latter is then temporarily divided into a urogenital and an anal membrane. Rapid proliferation of mesenchyme beneath the ectoderm surrounding the cloacal membrane forms the genital swellings. Thus, the cloacal membrane comes to lie at the bottom of a depression (external cloaca) surrounded by the genital swellings. Investigators suggest that the urorectal septum divides the cloaca into a urogenital portion and a rectal portion only up to the point of entrance of the paramesonephric (müllerian) ducts into the posterior wall of the urogenital sinus. The remainder of the urorectal septum (from the entrance of the paramesonephric ducts to the cloacal membrane) is formed by lateral invaginations (folds of Rathke) from the wall of the definitive urogenital sinus.

Prior to loss of the anal membrane (Fig. 45A), two small eminences that are connected by a medial band appear externally and

![Diagram](image)

Fig. 45 Division of external cloaca: A, 7-week-old embryo; B, 8-week-old embryo; C, 9-week-old fetus

dorsally on either side of the future anus and ventral to the tail of the embryo (Fig. 45B). These eminences elongate in a ventral and then a medial direction to converge at the midline. The genital swellings fuse
with these anal eminences at approximately the same time as the urorectal septum fuses with the cloacal membrane. The anal eminence genital swellings, and urorectal septum fuse to form the perineal body (Fig 45C). Once the perineal body is formed, the external cloaca divided into a urogenital and a rectal portion. Continued growth the anal eminences produces a depression externally; this is the procident and is surrounded by the anal mound. The anal mound is composed of external sphincter fibers.

In the present case study, the imperforate anus is the result of incomplete urorectal septum. This incomplete septum yields a fistula connection between the rectal cul-de-sac and a portion of the definitive urogenital sinus (prostatic urethra of the male).

Classification

Congenital malformations of the rectum and anus are frequently correctly listed under the term “imperforate anus.” This group of

![Diagram of anal and rectal malformations]

Fig 46 Classification of anal and rectal malformations: A, Stenosis of the anus; B, Imperforate anus with separation of rectum from anus; C, Membranous obstruction the anus; D, Separation of rectum from anal canal and anus.
verse anomalies includes the "commonest" malformations that are "incompatible" with life. How are these malformations classified? The classification introduced by Ladd and Gross in 1934 is most widely used (Fig. 46), although a number of anomalies do not conveniently fit into this taxonomy. In the present case study, the Ladd and Gross classification with modifications is utilized:

Types of Imperforate Anus

Type I Congenital anal stenosis
Type II Persistent membrane covering the anus
Type III Imperforate anus and rectum in which the rectal pouch terminates blindly some distance above the anus
   A. High lesions—The colon terminates above the levator ani
      Surgical approach is abdominoperineal
      1. Anorectal agenesis
      2. Persistent cloaca
      3. Fistulas
   B. Low lesions—The colon terminates below the levator ani
      Surgical approach is perineal
      1. Thick anal covering
      2. Anal stenosis or agenesis
      3. Persistent anal membrane

Type IV Anus and proctodeum are normal, but rectal pouch terminates blindly in the sacrum.

Complications

What postoperative complications may occur after repair of a high lesion as in the present case study? Possible postoperative complications are: mucosal prolapse at junction of rectum and new anal opening; stenosis at surgical junction; separation of the anastomosis; urethral stricture; urethral diverticulum; recurrent rectourinary fistula; urinary or rectal retention or incontinence; and inadequate blood supply to surgical junction.

Associated Malformations

What malformations are most commonly associated with an imperforate anus? The highest percentage of malformations accompanying an imperforate anus are of the urinary and genital systems and remaining portions of the digestive system.
Hypospadias

An 18-year-old primigravid mother delivered a full-term male infant. Routine examination of the infant revealed that the urethral meatal opening terminated ventral and proximal to its normal location at the apex of the glans penis. Redundant dorsal preputial tissue gave the penis a hooded appearance, while the prepuce itself was deficient on the lateral sides and absent on the ventral side of the penis. The prostatic urethra was not involved; thus, normal function of the sphincters and urinary control existed. The right testis was palpated in the superficial inguinal pouch and could be manipulated into the scrotal sac.

DIAGNOSIS

Subcoronal hypospadias with chordee and meatal stenosis and right unilateral undescended testis

TREATMENT

In repair of hypospadias the prime objectives are (1) to correct the ventral deformity (chordee); (2) to construct a penile urethra at or near the apex of the glans—the construction must be free of hair or scar and capable of normal sexual function; and (3) to utilize or remove the hooded prepuce. In the present case, repair was completed in two stages. When the boy was 2 years of age, the chordee and meatal stenosis were corrected. Due to the straightening of the penis, the urethral opening was now further from the glans than originally. After a lapse of one year to allow the penile skin to regain its normal elasticity and loose-
Catheter in the urethra of a penile-type hypospadiac
Fig. 47 Male urethral formation: A, 5-week-old embryo — ventral view; B, 6- to 7-week-old embryo — sagittal section; C, 7-week-old embryo — ventral view; D, 9-week-old embryo — ventral view; E, 12-week-old embryo — ventral view.
ness, urethroplasty (stage two) was performed using the Browne technique, in which the urethral tube is lengthened by tubular formation of a buried strip of the prepuce skin.

Several days after surgery, a fistula occurred between the old and new urethral tubes and was promptly repaired. At 5 years of age the boy voided from the tip of the glans with a good urinary stream, an repair was completed prior to entrance into school.

The undescended testis was not treated at this time.

**DISCUSSION**

Dieffenbach in 1837 was the first to attempt correction of hypospadias, a condition that occurs in 1/300 live male births. In 1874, Ang and Mettauer accomplished the first satisfactory correction of both chordee and an abnormally located meatal opening. The successful techniques in hypospadias repair are the result of the early works of Duplay, Ombredanne, and Nové-Josserand. More than 123 surgical techniques and modifications have been utilized for the repair of chordee and hypospadias.

**Embryology**

Discuss the development of the male urethra. In the four-week-old embryo, the primitive urogenital sinus and the hindgut empty into the cloaca; the latter is separated from the amniotic cavity by the cloacal membrane (Fig. 42). By the end of the seventh week (Fig. 47B), the cloacal membrane fuses with the growing urorectal septum and then is divided into a urogenital portion and an anal portion. The urogenital portion is separated into: (1) the vesicourethral canals from which arise the urinary bladder and the upper portion of the prostatic urethra, and (2) the definitive urogenital sinus, which is further divided into pelvic and phallic portions. In the male, the pelvis portion of the sinus becomes the lower part of the prostatic urethra and the membranous urethra. The phallic portion forms the external genitalia, with the exception of the urethral portion of the glans penis.

By the end of the seventh week (Fig. 47A, C, 48), a urethral plate develops from entodermal cloacal epithelium related to the base of the genital tubercle. This plate is located immediately above and internal to the cloacal membrane. The urethral plate grows forward into the mesenchyme of the genital tubercle such that the lower margin of this entodermal plate remains against the ectodermal surface epithelium covering the genital tubercle. While the urethral plate is growing for
ward to reach the tip of the genital tubercle, the mesenchyme on either side of the plate proliferates. Thus, on the ventral aspect of the genital tubercle, the ectodermal surface epithelium covering the mesenchyme is raised into two ridges, called the primitive urethral folds, between which lies the primary urethral groove. Simultaneously, the urorectal septum contacts and divides the cloacal membrane into the urogenital and anal membranes. The urogenital membrane then disintegrates, resulting in the external opening of the phallic part of the urogenital sinus. Presently, the entodermal urethral plate, which is in contact with the ectoderm of the primary urethral groove, thickens and disintegrates along with the related ectoderm. This results in the formation of a deep secondary urethral groove, called the definitive urethral groove, which is continuous proximally with the open phallic portion of the urogenital sinus.

Fig 48 Male urethral formation X-rays through the developing phallic to show formation of primitive and definitive penile urethra. (From Hamilton, W. J., Boyd, J. D., & Mossman, H. W.: Human Embryology, ed. 3. W. Heffer & Sons, Cambridge England, 1964, with permission.)

At nine weeks (Figs 47D; 48) the genital tubercle becomes elongated and transforms into the penis, while the genital (scrotal) swellings become more definite. The definitive urethral groove co
continues forward on the ventral aspect of the penis, but not to the tip. The urethral folds unite and fuse such that only entodermal epithelium from the urogenital sinus (secondary urethral groove) comprises the lining of the penile urethra. The urethra of the glans penis is formed in a manner similar to that described for the penile urethra, but in addition, surface ectoderm grows into the substance of the glans to meet the urethral plate, thus giving an ectodermal and entodermal lining to the glans urethra.

In the male, development of the external genitalia is completed by the eleventh week (Fig. 47E).

Explain the development of hypospadias Failure of fusion of the
urethral folds results in maintenance of the secondary urethral groove. This condition is hypospadias. Concurrently, epispadias is occasionally present. In epispadias, which is due to failure in the development of the infraumbilical mesoderm, the dorsal wall of the urethra is partially or completely absent. In extensive cases, the bladder mucosa is exposed on the anterior abdominal wall (ectopia vesicae) together with cleft pelvis.

Classification

How are the types of hypospadias classified? Denis Browne describes five forms of this condition (Fig. 49). In descending order of frequency, they are subcoronal (balanitic), penile, penoscrotal, scrotal, and perineal.

Complications

What are the most common complications in treatment of hypospadias? Most failures are the result of formation of fistulas, sloughing of skin, meatal retraction, recurrence of chordee, and infection.

Associated Malformations

What malformations are commonly associated with hypospadias: Chordee, cryptorchidism, urethral meatal stenosis, and the glans penis hooded by a redundant prepuce are the most frequently associated defects. Inguinal hernia, anomalous upper urinary tract, and cardiovascular defects are sometimes encountered.