Downloaded from www.ajronline.org by Univ of Pittsburgh on 11/25/14 from IP address 198.184.147.58. Copyright ARRS. For personal use only; all rights reserved

Triad of Anorectal, Sacral, and Presacral Anomalies

Guido Currarino,[1] Dale Coln,[2]
and Theodore Votteler[2]

Three infants in whom a congenital anorectal stenosis was associated with a sacral defect and a presacral mass are described. The mass was a meningocele in the first case, a teratoma in the second, and an enteric cyst in the third. On the basis of these cases and similar material in the literature, we suggest that this association of anomalies may represent a syndrome complex with a common embryogenesis.

Case Reports

Case 1

A 5-month-old girl with congenital anal stenosis was admitted because of progressive constipation since birth. Physical examination showed a severe stenosis of the anus and distal rectum. Pelvic films showed a butterfly deformity of S1 and a large crescentic defect of the lower sacrum (fig. 1A). Barium enema examination confirmed a severe anorectal stenosis and suggested a presacral mass (fig. 1B). A vaginogram showed a septate vagina. Myelography outlined a presacral meningocele with an associated soft-tissue mass between the meningocele and the strictured anorectal canal (fig. 1C). Neurosurgical consultation elsewhere resulted in a laminectomy with exploration of the lower spinal canal. At surgery there was spinal dysraphism with an anterior meningocele and myelodysplasia with a tethered spinal cord. Abnormally located nerve roots were found on the surface of the area of myelodysplasia. Surgical excision could not be performed.

The patient was managed by rectal dilatation until age 1 year. At that time, a second neurosurgical procedure was carried out in Canada; a meningocele and a lipoma were excised. Subsequently, very mild bladder dysfunction was noted in addition to the persistent rectal stenosis. At age 17 months, she was admitted to Fort Worth Children's Hospital with *E. coli* meningitis and presacral abscess. A diverting colostomy was performed and the abscess was drained. Despite these maneuvers and intensive antibiotic chemotherapy, she died soon after.

Autopsy showed acute and chronic inflammation in the presacral area. A fistulous tract lined with granulation tissue extended from the mucocutaneous junction of the rectum to the sacrum where it was confluent with the spinal canal. Much fibrofatty connective tissue surrounded the fistulous tract with small cystic spaces, lined by partially stratified epithelium, containing an admixture of glial tissue and isolated neurons in a disorganized pattern.

Case 2

An 11-month-old boy with congenital anal stenosis was referred because of progressive constipation since birth. Physical examination detected a marked stricture of the anus and distal rectum. Radiographs of the abdomen showed a large crescentic defect in the lower sacrum (fig. 2A). A barium enema examination confirmed the anorectal stricture and suggested a presacral mass (fig. 2B). A voiding cystourethrogram revealed an increased space between the posterior urethra and the strictured anorectal canal. Dilatations under general anesthesia were carried out but severe constipation continued.

He was readmitted at age 17 months when a barium enema study again suggested a presacral mass which was confirmed by rectal examination. Myelography showed no meningocele. Through a retrorectal approach, a 2 × 2 cm cyst was dissected with difficulty from the posterior wall of the rectum to which it was firmly adherent. The cyst contained some creamy material. Histologically, its wall was composed of stratified squamous epithelium, fibrous connective tissue, skeletal muscle, and mature neural elements. A diagnosis of benign presacral teratoma was made. The patient was discharged in satisfactory condition to have further rectal dilatations but was lost to follow-up after 6 months.

Case 3

A 13-month-old boy was referred with possible closure of a transverse colostomy which was performed at age 2 days in another institution because of an "imperforate anus with a perineal fistula." The anus appeared normal externally, but neither the small finger nor a probe could be advanced more than 2 or 3 cm. Radiographs of the abdomen demonstrated a small defect in the lower part of the sacrum (fig. 3A). A simultaneous contrast study through the distal limb of the colostomy and from the anus showed a long stricture of the anorectal canal with a short segment of near obliteration about 3 cm from the anus (fig. 3B). The study also revealed a presacral mass. Myelography showed no presacral meningocele.

Through a sacral approach, a 1.5 × 2 cm cyst behind the rectum, that had no apparent connection with the rectum or sacrum, was removed. Histologically, its wall was lined by squamous epithelium and contained fibrous stroma and epidermis; no neural elements were demonstrated. A diagnosis of enteric cyst was made. A probe was advanced through the narrow area and the rectum was then dilated. Repeated dilatations were performed for 4 months and then

Received October 21, 1980; accepted after revision February 13, 1981.

[1] Department of Radiology, University of Texas, Southwestern Medical School and Children's Medical Center, 1935 Amelia, Dallas, TX 75235. Address reprint requests to G. Currarino.

[2] Department of Surgery, University of Texas, Southwestern Medical School and Children's Medical Center, Dallas, TX 75235.

AJR 137:395–398, August 1981 0361–803X/81/1372–0395 $00.00 © American Roentgen Ray Society

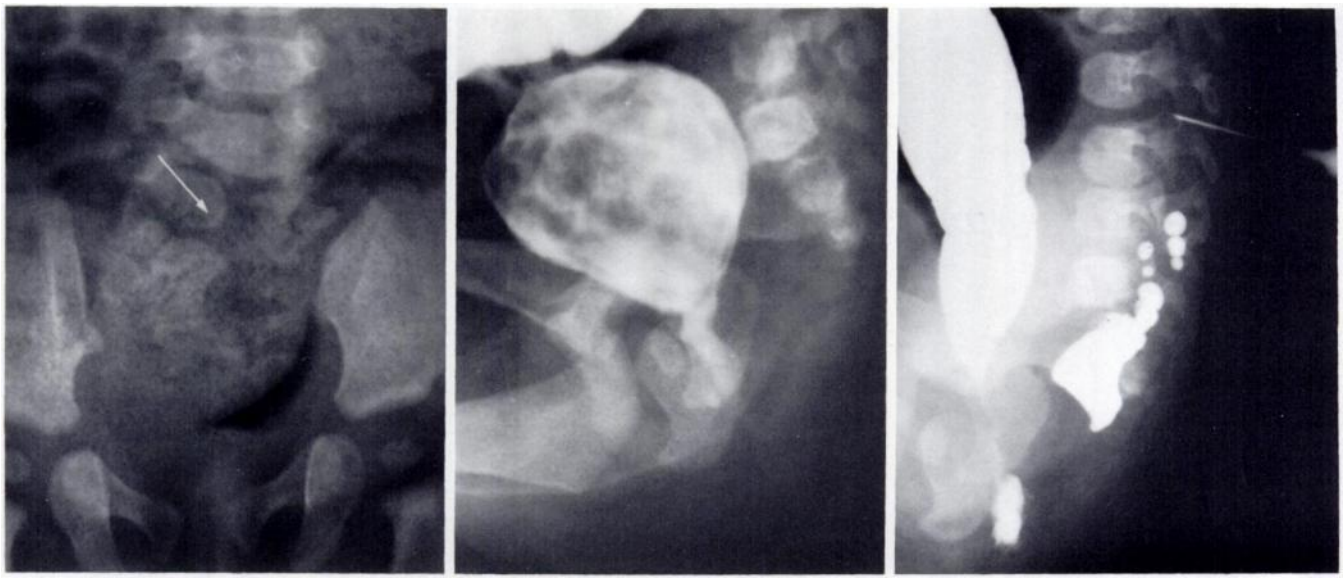

Fig. 1.—Case 1. **A**, Butterfly deformity of S1 (*arrow*) and large crescentic defect of lower sacrum. **B**, Barium enema study. Severe anorectal stenosis and suggestion of presacral mass. **C**, Myelogram. Presacral meningocele with soft-tissue mass between meningocele and strictured anorectal canal. Barium in colon from previous barium enema.

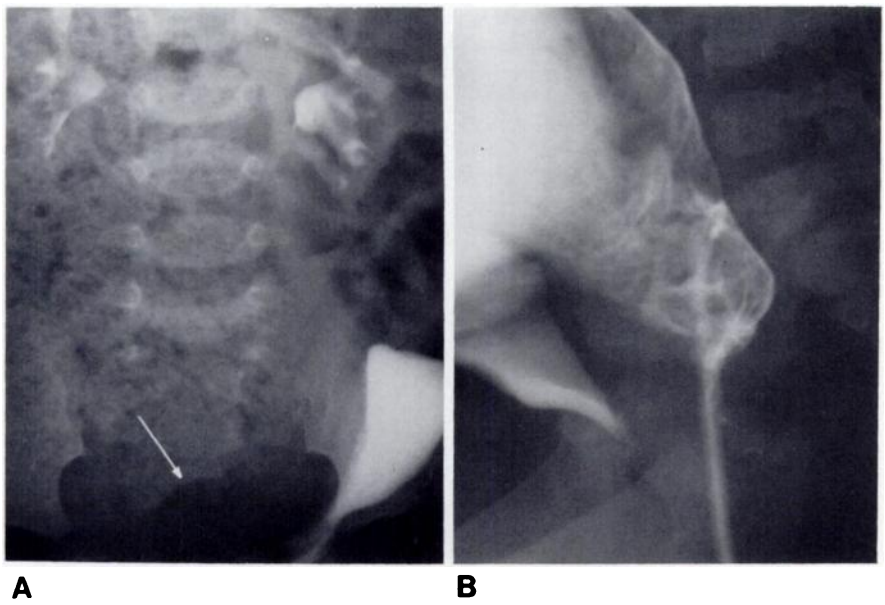

Fig. 2.—Case 2. **A**, Excretory urogram. Normal upper urinary tracts. Bladder displaced to left primarily by bulky fecal material in colon. Crescentic defect of sacrum (*arrow*). **B**, Barium enema study. Anorectal stricture and suggestion of presacral mass. Space between rectum and bladder is increased. Contrast material in bladder from previous cystogram.

the colostomy was closed. At age 2 years, the patient continues to do well.

Discussion

The major abnormalities in these three patients were congenital anorectal stenosis, a defect in the sacral bone, and a presacral mass. The presacral mass was a meningocele in the first patient, a teratoma in the second, and an enteric cyst in the third. A survey of similar and related material in the literature suggests that all these lesions may be developmentally related.

Thirteen unrelated patients with an anorectal anomaly, a presacral meningocele, and a sacral defect, similar to our case 1, have been reported [1–7]. Cohn and Bay-Nielson [8] described six members of three unrelated families having sacral defects. Two patients had anal stenosis and a presacral meningocele. Of the other four, two had a meningocele without anorectal malformation and two had an anorectal anomaly without a presacral mass. Aaronson [9] reported

Downloaded from www.ajronline.org by Univ of Pittsburgh on 11/25/14 from IP address 198.184.147.58. Copyright ARRS. For personal use only; all rights reserved

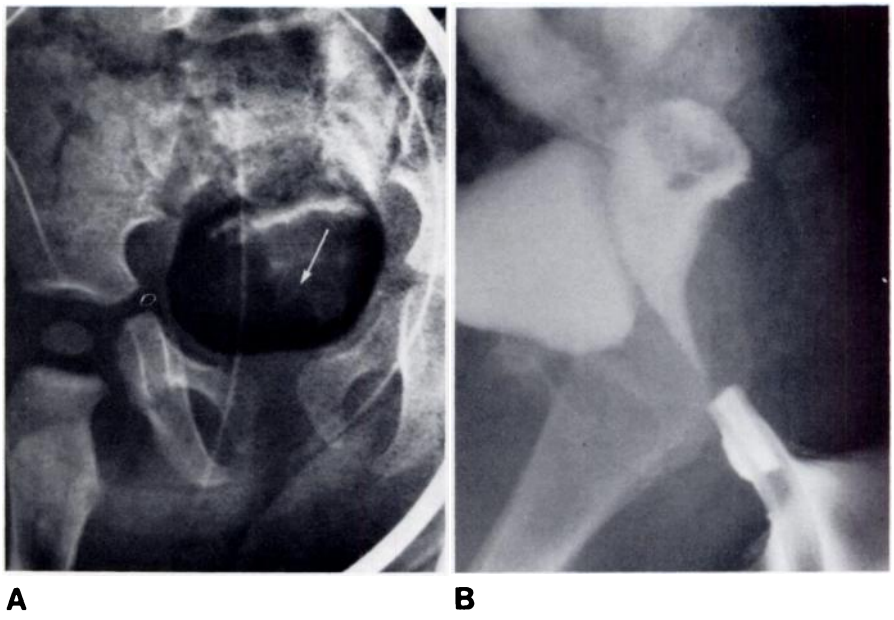

Fig. 3.—Case 3. **A**, Small defect in lower sacrum (*arrow*). **B**, Barium enema study. Marked narrowing of anorectal canal and presacral mass. Contrast material in bladder from previous cystogram.

three siblings with covered anus and sacral defects associated with a meningocele in one patient and presacral teratoma in another. Ashcraft and Holden [10, 11] and Hunt et al. [12] reported the association of presacral teratoma and sacral defects in 26 patients of six unrelated families. Thirteen patients had anorectal stenosis. An additional related patient had a sacral defect and anal stenosis without a presacral mass. All teratomas were benign except one and were firmly attached to the rectum in six patients and to the dura in three. Six patients presented with presacral abscess.

Several instances of an incomplete or atypical form of this syndrome are described in the literature. Malangoni et al. [6] reported four unrelated patients with an anorectal anomaly associated with a teratoma in three and a sacral defect in one. Kennedy [13] described a case of presacral meningocele, a sacral anomaly, and a rectal "polyp" connected to the meningocele. Kenefick [14] reported seven patients in one family with sacral defects without anorectal anomaly. Four of the patients had a presacral mass: an enteric cyst in one, a meningocele in another, and a mass of unknown nature in the other two. The patient described by Vicky and Poccianti [15] had a sacral defect associated with a presacral meningocele and a presacral teratoma. The instructive patient described by Esterly and Baghdassarian [16] had anal stenosis, a presacral enteric cyst, and a presacral teratoma; a fistula connected the rectum to the two masses and the spinal canal.

This survey and our three cases suggest a specific malformation complex characterized by three main features: (1) a congenital anorectal stenosis or another type of low anorectal malformation; (2) an anterior sacral defect of the type seen in anterior sacral meningocele (scimitar, crescent, or sickle deformity of the sacrum); and (3) a presacral mass which may be a meningocele, a teratoma, occasionally an enteric cyst, or a combination of these. This triad may be familial. In members of the same family, one or two basic features may be lacking, suggesting an incomplete form of the syndrome.

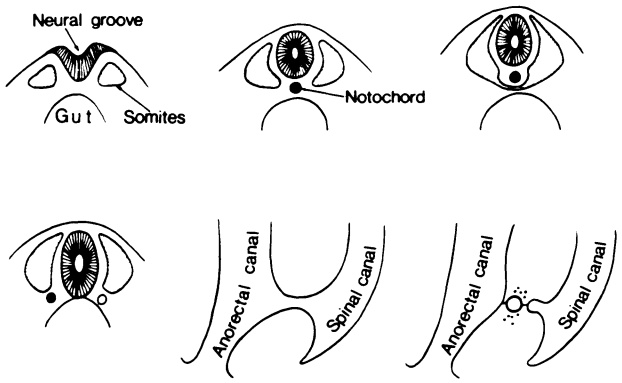

Fig. 4.—**Top**, Normal formation of neural tube. Appearance of notochord and formation of anterior arch of vertebra separates neural tube from gut. **Bottom**, Failure of closure of anterior part of vertebra. Notochord is split or displaced to one side, with formation of adhesions between gut and neural tube. Sagittal diagrams show consequent fistula between anorectal and spinal canals, and formation of meningocele, enteric cyst, and teratoma.

The embryogenesis of the malformation complex is not certain. The association of vertebral anomalies with neural and intestinal malformations at the same level suggests a similarity with the posterior neuroenteric cysts and duplications associated with thoracic vertebral anomalies. Abnormal endoectodermal adhesions and notochordal defects in very early fetal life have been postulated as the underlying embryologic cause for these lesions [17–21]. We suggest that a similar explanation may apply to the malformation complex we describe. (fig. 4).

Normally in early fetal life, the endodermal layer of the body stalk (the future gut) is in close approximation to the neural ectoderm. With the appearance of the notochord, the somites extend forward and medially, engulf the notochord

Downloaded from www.ajronline.org by Univ of Pittsburgh on 11/25/14 from IP address 198.184.147.58. Copyright ARRS. For personal use only; all rights reserved

and form the vertebral body, completely separating the gut from the neural tube. Persistent abnormal adhesions between the endoderm and the neural ectoderm, occurring before the appearance of the notochord, cause the notochord to split or be displaced laterally, preventing anterior fusion of vertebrae. Alternatively, a primary splitting of the notochord may bring the endoderm into more intimate contact with the neural tube causing adhesions between the two structures and a faulty vertebral development. In either situation a fistula may form between the gut and the spinal canal with enteric elements ventrally and neural elements dorsally. Partial resorption of the fistula may give rise to a meningocele or an enteric cyst. The anorectal anomaly is a consequence of these early abnormal adhesions between the hind gut and the neural tube. The presacral teratoma is formed by local enteric and neuroectodermal elements combined with mesodermal elements brought into the space by the developing somites and other local mesodermal structures.

It should be appreciated that these presacral masses may be firmly adherent to the rectum or dura making surgical removal difficult. Postoperative abscesses or meningitis are not uncommon and may be due to injury to adjacent structures during surgery. These infections can occur preoperatively due to preexisting unrecognized rectal fistula, or to infection of enteric cysts.

REFERENCES

1. Eklöf O. Roentgenologic findings in sacrococcygeal teratoma. *Acta Radiol [Diagn]* (Stockh) **1965**;3:41–48
2. Kaufmann HJ. Anterior sacral meningocele. *Ann. Radiol* (Paris) **1967**;10:121–128
3. Werner JL, Tabyi H. Presacral masses in childhood. *AJR* **1970**;109:403–410
4. Shaker IJ, Lanier VC, Amoury RA. Congenital anal stenosis with anterior sacral meningocele. *J Pediatr Surg* **1971**;6:177
5. Anderson FM, Burke BL. Anterior sacral meningocele: a presentation of three cases. *JAMA* **1977**;237:39–42
6. Malangoni MA, Grosfeld JL, Ballantine TVN, Kleiman M. Congenital rectal stenosis: a sign of a presacral pathologic condition. *Pediatrics* **1978**;62:584–587
7. Brihaye J, Gerard A, Kiekens R, Retif J. Recto-meningeal fistulae in dysraphic states. *Surg Neurol* **1978**;10:93–95
8. Cohn J, Bay-Nielson E. Hereditary defect of the sacrum and coccyx with anterior sacral meningocele. *Acta Paediatr Scand* **1969**;58:268–274
9. Aaronson I. Anterior sacral meningocele, anal duplication cyst and covered anus occurring in one family. *J Pediatr Surg* **1970**;5:559–563
10. Ashcraft KW, Holder TM. Congenital anal stenosis with presacral teratoma. *Ann Surg* **1965**;162:1091–1095
11. Ashcraft KW, Holder TM. Hereditary presacral teratoma. *J Pediatr Surg* **1974**;9:691–697
12. Hunt PT, Davidson KC, Ashcraft KW, Holder TM. Radiography of hereditary presacral teratoma. *Radiology* **1977**;122:187–191
13. Kennedy RLJ. An unusual rectal polyp: anterior sacral meningocele. *Surg Gynecol Obstet* **1926**;43:803–804
14. Kenefick JS. Hereditary sacral agenesis associated with presacral tumours. *Br J Surg* **1973**;60:271–274
15. Vichi GF, Poccianti F. Su di un caso di meningocele sacrale anteriore associato a teratoma sacro-coccigeo in un lattante. *Minerva Chir* **1961**;16:504–511
16. Easterly JF, Baghadassarian OM. Presacral neurenteric cyst. *Johns Hopkins Med J* **1963**;113:202–210
17. Veeneklass GMH. Pathogenesis of intrathoracic gastrogenic cysts. *Am J Dis Child* **1952**;83:500–507
18. Fallon M, Gordon ARG, Lendrum AC. Mediastinal cysts of foregut origin associated with vertebral anomalies. *Br J Surg* **1954**;41:520–533
19. Bentley JFR, Smith JR. Developmental posterior enteric malformations: the split notochordal syndrome. *Arch Dis Child* **1960**;35:76–86
20. McLetchie NBG, Purves JK, Saunders RL de Ch. Genesis of gastric and certain intestinal diverticula and enterogenous cysts. *Surg Gynecol Obstet* **1954**;99:135–141
21. Beardmore HE, Wiggleworth FW. Vertebral anomalies and alimentary duplications: clinical and embryological aspects. *Pediatr Clin North Am* **1958**;5:457–474

Downloaded from www.ajronline.org by Univ of Pittsburgh on 11/25/14 from IP address 198.184.147.58. Copyright ARRS. For personal use only; all rights reserved