CASE REPORTS AND TECHNICAL NOTES

AGENESIS OF THE SACROCOCCYGEAL REGION

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Congenital anomalies of the sacrococcygeal region associated with neurological abnormalities are not uncommon and sacral agenesis has been reported over 50 times since Hohl's case in 1852. The first clinical description was made by Litzmann in 1885, and Coleschi reported the radiographic appearance in 1918. Díaz Lira described a case in which surgical exploration was done. The present report deals with a case of this abnormality with surgical exploration and electrical stimulation of the roots of the cauda equina for study of the function of the urinary bladder.

CASE REPORT

NCBH #204210. A 6-year-old boy was admitted on May 12, 1955, for investigation of sacrococcygeal abnormality (Fig. 1). He was the third sibling and one of twins. It is not known whether or not the twins were identical. In the third trimester of the pregnancy the mother was found to have lymphosarcoma involving the left hip; however, the pregnancy terminated normally. At birth the patient weighed 4 pounds and there were no initial complications. The child was considered normal except for deformities of the lower extremities. Bilateral equinovarus deformities were corrected surgically and he learned to walk at 22/4 years. He did not achieve control of bladder or bowel. At the age of 4 a transurethral resection of the neck of the bladder was done with some improvement of bladder capacity but with no improvement of the sphincter. He was considered to be mentally normal at the age of 6 years.

Family History. The twin died at 4 weeks of age of an acute infection of the urinary tract. No roentgenograms were made and bilateral hydronephrosis and hydroureters were found at postmortem examination. No special studies of the spine were made.

Physical Examination. Temperature was 98.6°F., blood pressure 94/60, pulse rate 86, and respiratory rate 20. The abnormal findings were limited to the lower extremities, genitalia, and sacral region. The anterior muscles of the thigh were well developed and strong, while the buttocks were flat and atrophic. Below the knees the legs tapered down to the ankles with no visible muscles. The arches of the feet were high with the toes fixed in a flexion deformity. Scars of the previous orthopedic procedures were evident on the feet. The anal sphincter was relaxed. He dribbled urine intermittently.

Neurologic Examination. He was an alert, bright boy of 6 years. The cranial nerves were intact. There was good flexion and adduction at the hips but poor extension. Voluntary flexion and extension of the knee were normal but there was only a flicker of movement in the ankles and toes. The deep reflexes were brisk and equal in the upper extremities, but the knee jerks were barely present and the ankle jerks were absent. Cutaneous plantar stimulation on each side caused a quick voluntary withdrawal of the leg with slight flexion of the toe. All
sensation was normal including the cutaneous areas in the region of the motor deficit. There was no subjective sensation of bladder on filling the bladder. He occasionally experienced rectal sensation with defecation.

**Accessory Clinical Findings.** Blood counts and urine were normal; serological test of the blood for syphilis was negative. Roentgenograms revealed bilateral cervical ribs with fusion abnormalities of the C5, C6, C7, and possibly the T1 vertebrae (Fig. 2). There were five lumbar vertebrae and the sacrum was absent with the iliac bones meeting at the midline (Fig. 3). The hip joints were normal. Roentgenogram of the chest showed the lungs to be clear; the size of the heart was reported to be at the upper limit of normal, and there was slight prominence of the shadow of the pulmonary artery. Excretory urograms showed normal renal pelves and calyces. A cystogram revealed a small contracted bladder with reflux of dye into the right ureter. Cystometrograms done in the waking state were unsatisfactory. A closed system could not be maintained because of leakage around the catheter and a small contracted bladder. A lumbar puncture was attempted in the lower lumbar region but was not successful.

**Operation.** On July 11, 1955, a complete laminectomy was performed of L2 through L4. The dural cul-de-sac was found to terminate at the L2-L3 interspace. The nerve roots below this level were embedded in a loose fatty connective tissue and the manner of exit of the roots from the canal was normal (Fig. 4). Faradic stimulation (1–2 volts) of the nerve roots produced active motor responses in the hamstring muscles bilaterally. A cystometer was connected with the bladder through a urethral catheter but there were no contractions on electrical stimulation of the sacral nerve roots. The dural sac was opened and cerebrospinal ligation performed. There was no change in muscle function after this procedure.
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Fig. 2. Roentgenogram of cervical spine showing bilateral cervical ribs, with fusion abnormalities of C5, C6, C7, and possibly T1 vertebrae.

fluid flowed freely. The conus medullaris was not seen but a rubber catheter could be passed cephalad 10 cm, without meeting resistance in the subarachnoid space. The dura mater and wound were closed.

Postoperative Course. The wound healed *per primam* and there was no alteration in the neurologic status.

Three months after operation the parents reported that in the morning he could void 1 or 2 ounces of urine with a good stream. He had required no enemas, although occasionally he was incontinent of feces and continued to wear special waterproof pants.

**DISCUSSION**

The striking configuration of the sacral region and the lower extremities, which taper down to the feet with equinovarus deformities, identifies this abnormality. This appearance has been described as “mermaid-like,” or “sirenen” by Feller and Sternberg. The buttocks and sacrococcygeal region are flat with prominent iliac crests, shortened intergluteal fold, and iliac bones that touch one another in the midline. Freedman reviewed 45 cases and reported 8 cases (18 per cent) in which the hips were dislocated; club feet were present in over half of the cases.

Bony abnormalities of the spine can occur from the cervical to the sacral region. These may consist of hemivertebra, abnormal fusions, failure of development of the neural arches, sacralized and wedged 5th lumbar vertebra and congenital aplasia. Congenital synostosis of several ribs, genu recurvatum, and congenital subluxation of the knees also have been described. The relation of the dural sac to the vertebral
canal may be altered, as in our case, with the cul-de-sac ending at the L2–L3 level. This would explain the failure of the attempted lumbar puncture at the lower level. Lausecker reported a sacral deformity in a 24-year-old man who had bowel and urinary incontinence. The patient died of acute infection of the urinary tract. There was striking deformity of the L4 and L5 vertebrae with a rudimentary sacrum. The lower lumbar canal was narrowed and deformed, but he stated that the spinal cord ended at the L1 level and the nerves issued normally from the canal. From observation of the photograph and drawing in his case it is conceivable that the bony deformities compressed the cauda equina (Figs. 5 and 6).

A multiplicity of visceral abnormalities may be associated with this condition. In one review of 45 cases, there were 16 (35 per cent) instances of associated geni-

![Fig. 3. Anteroposterior and lateral roentgenograms of lumbar spine and pelvis showing absence of sacrum and coccyx.](image-url)
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tourinary or bowel involvement, usually rectal, and there was 1 case of transposition of the viscera. White described the postmortem findings in a case of sacral agenesis with an accessory spleen, an umbilical hernia filled with bowel, undescended right testicle, and median kidney with two suprarenals. The right ureter was normal; the left was a fine impervious cord. In our case there were no visceral abnormalities evident, but the dead twin was known to have had bilateral hydronephrosis and hydroureters. It is not known whether or not the twin had agenesis of the sacrum and coccyx.

Fecal and urinary incontinence of varying degrees are the rule. In our case cystometrogram done at the time of surgery under general anesthesia revealed a small contracted bladder and electrical stimulation of the nerve roots produced no contraction. The reason for the apparent improvement in bladder function after laminectomy is not known, although Díaz Lira has made a similar observation.

Motor defects predominate over sensory with associated atrophy of the muscles below the knees and in the sacral region. Walking may be possible in some patients if the associated anomalies of the bones and joints are not severe. In our case the child walked as if on stilts, using his thigh muscles; however, he had a remarkable degree of agility in spite of his deformities.

Sensory findings have been listed as "confused," "chaotic," or "incomplete." The sensory defect when it exists is much less than one might anticipate in view of the profound motor deficit; careful testing of our patient revealed normal sensation in a cooperative child. The discrepancy between the obvious large motor deficit and the normal sensory function remains unexplained.

Reflexes were usually absent below the knees and mass reflexes from cutaneous stimulation have not been reported.

Surgical exploration of this type of abnormality was first reported by Díaz Lira in a 9-year-old boy with sacrococcygeal agenesis. He expressed the opinion that the cauda equina was compressed. The spinal canal was exposed from L3 to S1. The spinous process of the 4th lumbar vertebra was formed by two bony masses separated by a tight fibrous band and beneath this the nerve roots were entangled in a fibro-
fatty mass. The nerve roots could not be separated from the mass. Postoperatively the child retained some urine and had the desire to urinate, voiding approximately 20 cc. At the end of the third postoperative day a cathartic was given and the patient expressed the desire to defecate. From that time he had no fecal incontinence,

regained urinary control and retained 40 cc. for several hours. A later examination revealed normal anal reflexes with good function of the sphincters. Páez\(^\text{10}\) reported a case of subtotal sacrococcygeal agenesis and mentioned two others in which there was improvement after transurethral resection of the neck of the bladder. He also referred to a case of Heras in which the superior hypogastric plexus was resected and another case in which the cauda equina was decompressed.

In view of the experiences cited above, surgical exploration is indicated to obtain further information regarding function of the bladder in the individual patient. Some improvement has been reported and there may be variation in innervation of the bladder from patient to patient as in those with lumbosacral myelomeningocele. The possibility of improving function of the bladder in this type of abnormality

Fig. 5. Photograph from Lausecker's case, showing multiple deformities of the lumbar spine. (Reproduced by courtesy of Dr. H. Lausecker.)
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by altering the motor supply of the bladder awaits further trial in selected cases.¹

A wide range of variation exists in the congenital abnormalities of the sacrococcygeal region, from a minimal defect such as spina bifida occulta to the more severe

deformities associated with sacrococcygeal agenesis. We believe that these deformities are more prevalent than indicated in the literature and know of 5 unpublished cases reported to us by Martinat.⁹

SUMMARY

A case of sacrococcygeal abnormality is reported with surgical exploration and electrical stimulation of the lumbosacral nerve roots. This abnormality consists of minor bony defects of the cervical and lumbar spine with absent sacrum and coccyx. There was a severe loss of motor function below the knees but normal sensation. Rectal and bladder incontinence improved subjectively after laminectomy.

REFERENCES

9. Martinat, E. Personal communication.