

CONGENITAL ABSENCE OF THE LUMBAR VERTEBRAE AND SACRUM.
REPORT OF A CASE*

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Although the absence of a single vertebra is a not infrequent occurrence and may in fact be symptomless, the absence of a large number of vertebrae is very rare. Potter¹ recorded two cases in which a large number of vertebrae had failed to develop; in both these cases the lower dorsal and upper lumbar vertebrae were absent. Feller and Sternberg² describe in great detail the macroscopic and microscopic autopsy findings in cases of gross vertebral abnormalities but in only one of the six cases described by them was there any complete absence of vertebrae. Jorge³ describes two cases, in one of which there was also bilateral luxation of the hips; both these cases were alive at the time of the report. He also quotes a number of cases from the literature. Bauer⁴ reports a stillbirth with complete agenesis of the lumbar vertebrae but the sacrum was present. He quotes two similar cases from the literature; one of these reached the age of 32 (Walter)⁵ and the other was reported at the age of 5 years (Brailsford).⁶ Goldhamer⁷ reported a case of sacral agenesis and reviewed the literature of sacral and vertebral agenesis.

Case Report

B. S. (58/7086) was first seen by us at the age of eleven days. She was brought to the outpatient clinic because of "deformed legs" and the parents themselves had noticed that the baby had not moved her legs. The pregnancy and delivery had both been normal. The parents were not consanguineous and there was no family history of any congenital deformities.

On examination this was a female infant of normal size and weight for her age. In fact above the level of the umbilicus the infant appeared essentially normal. The abdominal muscles were flaccid and the abdomen protuberant. The spleen, liver and kidneys were not enlarged. The anal sphincter was patulous and on slight suprapubic pressure clear-looking urine could be expressed easily, suggesting a paralysis of the bladder sphincter. There was a marked hard protuberance on

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the back which coincided with the lower end of the vertebral column. The pelvis could be flexed and rotated easily in relation to the spine. The hips were externally rotated and abducted and the knees were moderately flexed. No passive movement could be elicited in these joints. A web-like fold of skin extended from the middle of the thigh to the upper calves on both sides. There was no active movement of the lower limbs, all reflexes were absent and the skin appeared to be completely anaesthetic.



FIGURE 1



FIGURE 2

Radiography

Antero-posterior and lateral radiographs (Figures 1 and 2) showed that below the level of the 12th dorsal vertebra all lumbar vertebrae had failed to develop. The sacrum and coccyx were absent and the bony pelvis, which itself appeared normal, was attached to the vertebral column by soft tissues only. The cervical and thoracic vertebrae were normal and there was no other bony abnormality. Excretion urography showed normal kidneys, ureters and bladder.

The child has been seen by us at intervals and was last seen at the age of four months. It was gaining weight and the upper half

of the baby was growing normally while below the level of the umbilicus (i. e. in the paralysed parts) there had been virtually no growth at all. There was definite incontinence of both urine and faeces although there was marked constipation. The blood N. P. N. was normal.

The prognosis as far as function of the lower extremities is concerned is of course hopeless in this case. She is likely to develop recurrent urinary infections and the resultant renal damage is the usual cause of death in this type of case. However some long-term survivals have been reported in the literature (see above).

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