Sacral Agenesis

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Summary

Sacral agenesis is a very rare congenital anomaly which presents in varying forms during childhood. The condition should always be kept in mind in unexplained cases of failure to achieve control over urinary and/or faecal continence during normal development. Such a case is presented here.

Introduction

Sacral agenesis is defined as an absence of all or part of two or more vertebral bodies at the lower end of the spinal column. Sometimes, the entire sacral and even some of the lumber vertebrae may be missing, and this has been labelled caudal regression syndrome[1] (Fig-1).

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The nerves most commonly affected are the motor efferents originating form S2 to S4 in the spinal cord. These roots are incorporated into the fibrous mass that develops in place of the absent vertebrae and they may be injured in the process. Sensation in the sacral area is usually intact and motor functions of the lower extremities are normal.

The true incidence of neurogenic bladder dysfunction has not been accurately determined because if the bladder or bowel is unaffected, there may be no other manifestations of this disease and the condition will go undetected. It is important to recognise the presence of sacral agenesis as early as possible because of its subsequent implications[1].

Case History

A five years old girl presented with failure to achieve continence of urine and faeces since birth. She used to be wet all the time. The prenatal period was uneventful and the mother never had diabetas mellitus. On examination she was found to have a waddling gait and small buttocks. She had an unusual softness over the sacrum and was constantly wet with urine. There was no muscle atrophy in the lower limbs and knee and ankle jerks were normal. Per rectal examination showed decreased anal sphincter tone. Sensations in the perianal area were normal. Intravenous pyelography and sonography of the abdomen and pelvis were normal. Cystoscopy showed some changes of neurogenic bladder. X-Ray of the spine showed absence of coccyx and sacrum below SI. (See Fig:2). A diagnosis of sacral agenesis (with secondry neurogenic bladder) was established.

Discussion

Sacral agenesis is an important congenital cause of micturition disorers[2].

Aplasia of the terminal spinal segments varies in extent[3]. There is absence of one or more lumbar vertebral segments, the iliac wings are not anchored to any bony structure and the femora and tibiae are short[4]. Severe cases with iliac bones approximation in the midline show flattened buttocks with dimples lateral to a short intergluteal fold[3]. Our case was a less severe form with aplasia of spine below SI. Associated congenital dislocation of hip, talipes, and neurological deficits affecting bladdar, bowel and lower limbs usually occur. Visceral anomalies occur in about one third

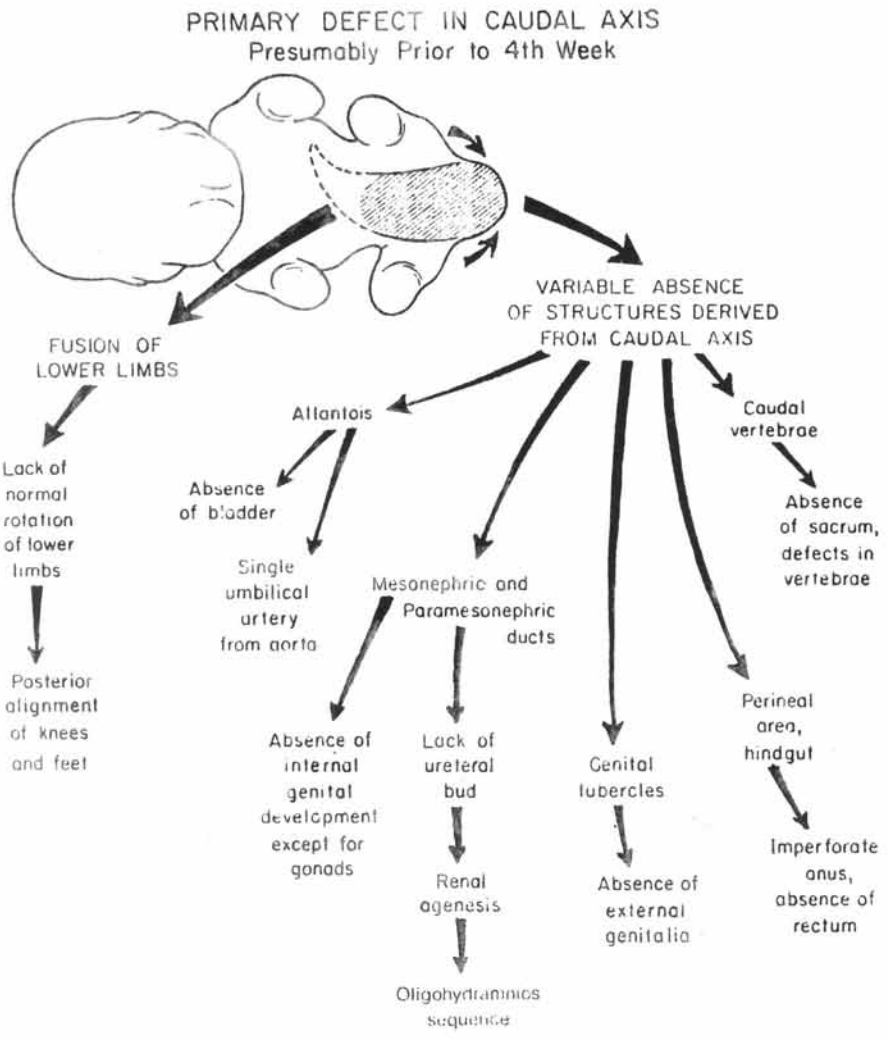

Fig 1. Adapted from Smith D.W Recognizable patterns of human malformation. :3rd Ed. W.B.Saunders company. 1982.

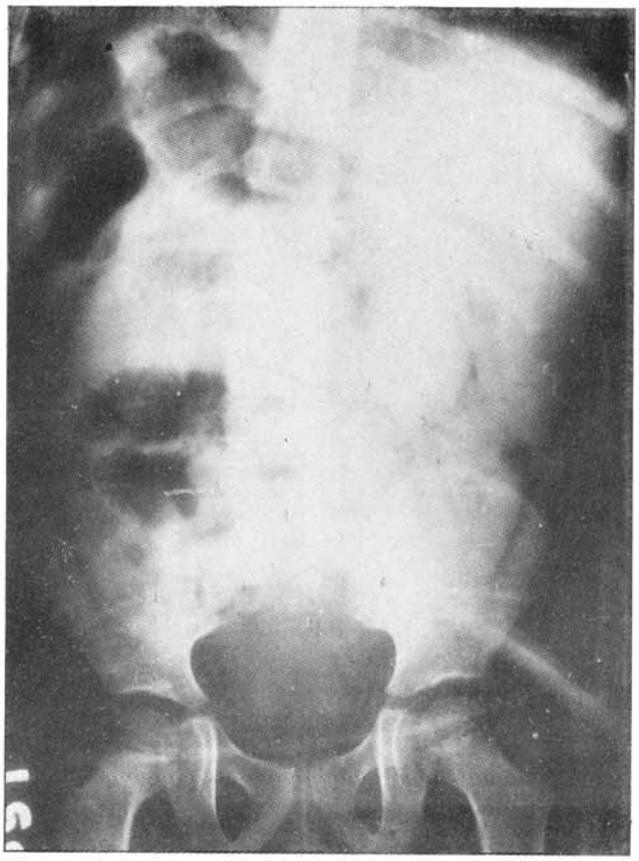

Fig 2. X-Ray showing absence of the spine below S1.

of cases. This defect is rare but occurs with increased frequency in infants of diabetic mothers[3]. Our patient did not have any of these associated anomalies.

Management of such children is of great concern for the pediatrician specially in a patient like ours with otherwise normal physical, mental and intellectual development. Psychosocial repercussions of being incontinent are innumerable and may severely impede the development of such children. The current management is mainly supportive including physiotherapy, orthopaedic surgery and prosthesis as necessary, and management of the neurological complications[4].

References

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