

Caudal Regression Syndrome

Dino Samartzis,^{1,2} DSc, MSc, Dip EBHC, Francis H Shen,^{2,3} MD



Fig. 1. Plain radiograph of the pelvic region and lower extremities illustrating the absence of the sacrum.

Quiz

What do you see in these images ?

- a. Down Syndrome
 - b. Jarcho-Levin Syndrome
 - c. Sirenomelia
 - d. Caudal Regression Syndrome
 - e. Klippel-Feil Syndrome
- (answer below)

Case

A white female with an uncomplicated history of birth delivery and a familial history of diabetes mellitus presented to the orthopaedic clinic at the age of 16 months old with bowel dysfunction, inability to walk, one kidney, and a gibbus noted at T12. A “frog-like” appearance was noted of the lower extremities with webbing of the popliteal space (Fig. 1). Although some strength of the hip flexor muscles was present, no muscle function was noted at the feet and ankles. Plain radiographic evaluation noted total absence of the sacrum, partial absence of the L5 vertebra, hemivertebrae, and left-sided congenital scoliosis (Figs. 1 & 2). The patient eventually underwent spinal fusion and bilateral knee disarticulations. At 19 years of age, the patient continues to wear cosmetic knee prosthesis with attached back support and is confined to a wheelchair.

Caudal regression syndrome (CSR) is a severe congenital condition characterised as premature termination of the spinal column with or without associated soft tissue, osseous,



Fig. 2. Anteroposterior plain radiograph of the spine and pelvis showing a congenital absence of the sacrum and the L5 vertebra, hemivertebrae, and congenital scoliosis.

or visceral anomalies.^{1,2} The precise aetiology of CSR remains speculative (i.e. genetic factors, vascular anomalies); however, evidence has indicated that maternal diabetes may play a role in the pathogenesis of this condition. Thorough clinical and radiographic evaluation is necessary³ to design proper therapeutic intervention to afford the patient optimal autonomy and function as well as psychological support. Early detection and evaluation of the extent of CRS is imperative to decrease the risk of incontinence, recurrent urinary tract infections, renal impairment, and the development of a neuropathic bladder.⁴ As such, prognosis relies on the stability of the spine, mobility of the lower extremities, and status of the urological function at time of presentation.

REFERENCES

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¹ Graduate Division, Harvard University, Cambridge, Massachusetts, USA

² Shriners Hospitals for Children, Chicago, Illinois, USA

³ Department of Orthopaedic Surgery, University of Virginia, Charlottesville, Virginia, USA

Address for Correspondence: Francis H Shen, Department of Orthopaedic Surgery, University of Virginia, PO Box 800159, Charlottesville, VA 22908-0159, USA. Email: fhs2g@virginia.edu

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