E-Poster

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Total Sacral Agenesis: A Case Report on Bladder, Sphincter, and Motor Dysfunction

Topic: Miscellaneous

Daouda Diouf, Santana Ireke, Douae El Ouazzani, Rime El Beloui, Hasnae Boutalja, Nada Kyal, Fatima Lmidmani, Abdellatif El Fatimi

Department of Physical Medicine and Rehabilitation, CHU Ibn RODHD, Casablanca, Morocco

BACKGROUND

Sacral agenesis is a rare malformation, due to an in-utero developmental defect of the caudal part of the spine, which may be partial or total, isolated or associated with a poly-malformation syndrome. The etiopathogenesis remains unknown, but its relationship with maternal diabetes is well established. Symptoms vary according to the degree and severity of spinal damage. Diagnosis is often delayed. We report the case of a patient presenting with bladder and bowel dysfunction (BBD)

CASE DESCRIPTION

A 15-year-old patient with no notion of consanguinity or similar cases in the lineage, with maternal type 2 diabetes, was referred to Physical and Rehabilitation Medicine department at CHU Ibn ROCHD Casablanca for BBD with urinary incontinence by urgency, primary enuresis, dysuria, recurrent urinary infections, and anal incontinence. Questioning revealed delayed psychomotor development and BBD without genital malformation. Physical examination revealed normal muscular strength and objective sensitivity, neuropathic pain, and abnormal spinal curvatures affecting plantar seating with a hollow foot (according to the descending postural chain of lesions) with bilateral hallux valgus, clawed toes, and widened forefeet. On walking, there is a plantigrade step attack, internal rotation of the feet, an oscillating phase, and instability of the pelvis. Squatting without support is impossible, and making use of the Turkish toilet is difficult. Perineal examination reveals a normal sensitivity, perineal reflexes, and anal tone. Muscle testing of the levator muscles was 3/5.

Magnetic resonance imaging shows total sacrococcygeal agenesis from S2 with sacralization of L5, trochoid aspect of the medulla (D12). A bladder ultrasound revealed a small-capacity, diverticular bladder with significant post-void residual volume, minimal hydronephrosis on the right, and moderate hydronephrosis on the left. Urodynamic assessment (UDA) was favorable on flowmetry: dysuria with vesical-sphincter dyssynergia and on cystomanometry: small cystomanometric bladder capacity, sensitive, hypo-compliant, without detrusor hyperactivity or leakage on exertion. Anal manometry reveals sphincter hypotonia at rest, lack of voluntary anal contraction, and microrectia.

DISCUSSION

Sacral agenesis, like all caudal malformations, affects the ponytail to varying degrees. Bladder and bowel dysfunction and the resulting motor disorders impair quality of life, and must be diagnosed exhaustively. The therapeutic arsenal is vast and gives good results.

CONCLUSION

This is a rare congenital malformation with no identified cause. Imaging confirms the diagnosis and assesses the degree of spinal involvement. Urodynamic assessment and anal manometry allow better assessment of BBD and adaptation of therapy to improve patients' quality of life.

Statement of Disclosures

None

Corresponding Author:
Daouda Diouf
Department of Physical Medicine and Rehabilitation, CHU Ibn RODHD, Casablanca, Morocco
Email: dioufdaouda92@gmail.com

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