E-Poster

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Functional Impact of Orthopedic Anomalies in Silver-Russell Syndrome or Syndromic Congenital Hemiatrophy in Adulthood: A Case Report

Topic: Disability Studies

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BACKGROUND

Silver-Russell Syndrom (SRS) is a rare genetic disorder that causes prenatal-onset growth restriction, metabolic disorders, and limb asymmetry. Its prevalence is estimated at 1:30,000 to 1:100,000 children. Diagnosis is primarily clinical and usually made in pediatric age, becoming more challenging in adulthood due to non-assessable clinical score criteria. Orthopedic issues associated with SRS mainly include hemibody asymmetry, resulting in leg length discrepancy (LLD). Long-term studies on SRS are scarce. We report the case of a patient referred to consultation for chronic lower back and right knee pain.

CASE DESCRIPTION

A 51-year-old woman (height: 1.35 m, weight: 44 kg, body mass index (BMI) : 24 kg/m²). She was single, childless, and postmenopausal. Physical examination revealed right hemibody atrophy with severe LLD. Examination of the spine showed exaggerated dorsal kyphosis, deviation of the spinous line, shoulder and pelvic girdle tilt, and a prominent dorsal gibbus. The lower limb assessment identified bilateral genu valgum with outward deviation of the leg and foot axis, as well as persistent left knee flexion despite LLD correction. The patient's gait required technical assistance and was characterized by an anteflexed trunk, compensatory right hip posture, taligrade foot strike, and shortened stance and swing phases on the right side. Podiatric examination revealed flat feet and calcaneal valgus. The patient's quality of life was severely impaired, as indicated by the SF-12 score, and moderate depressive symptoms were noted on the Hamilton scale. Radiographic imaging confirmed thoracolumbar scoliosis and femorotibial gonarthrosis.

DISCUSSION

Silver-Russell Syndrom is associated with short stature in adulthood, with studies reporting approximately 151-154 cm for men and 139-147 cm for women, in the absence of treatment. The LLD associated with SRS is a structural or anatomical inequality in length, the complications of LLD are very often mechanical: knee and hip pain (excessive stress on the hip or knee), muscle contractures, spinal pain, scoliosis, sciatica, stress fracture and plantar fasciitis. In the long term, there is a risk of osteoarthritis. Scoliosis has been reported in 9% to 36% of patients with SRS, probably partly due to muscle hypotonia. In addition, there is a risk of kyphosis in around 20% of cases.

CONCLUSION

The management of SRS must be multidisciplinary, initiated early, and maintained throughout adulthood. A preventive approach is crucial to detect and mitigate complications, ensuring better long-term functional outcomes. Particular attention should be given to orthopedic anomalies, as they are highly deforming and significantly impact mobility and quality of life.

Statement of Disclosures

None

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