E-Poster

EP-586

Management of Motor Disorders in Turner Syndrome in Physical Medicine and Rehabilitation

Topic: Rehabilitation Across the Continuum of Care

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BACKGROUND

Turner syndrome is a rare genetic disorder caused by the partial or total absence of an X chromosome, affecting around one in 2,500 girls. It is characterized by short stature, gonadal abnormalities, skeletal malformations (such as scoliosis and flat feet), heart problems and neurocognitive deficits. It is also associated with an increased risk of vascular, renal, ENT and metabolic malformations. The management of this disease requires a multidisciplinary approach in order to treat the various complications and improve patients' quality of life.

CASE DESCRIPTION

We report the case of a 16-year-old girl with a history of bilateral varum knee for which she underwent surgery a year ago, and presenting with motor disorders associated with Turner syndrome. On clinical examination, lumbar scoliosis was noted, with accentuation of the waist crease and left gibbosity. There was no pain on palpation of the spinous processes, no paravertebral contractures, and no unevenness of the lower limbs. There was no imbalance of the pelvis or shoulder blades. Bilateral hollow feet were also observed.

Physical and Rehabilitation Medicine (PRM) management consisted of monitoring combined with regular motor rehabilitation, and the prescription of insoles for bilateral hollow feet.

DISCUSSION

Turner syndrome is a rare condition, occurring in approximately one in every 2,500 women born alive, it is a highly variable disorder with a wide range of clinical features including short stature, ovarian and cardiac abnormalities, as well as bone deformities and specific facial and physical features. Diagnosis is made following karyotyping or FISH analysis of the sex chromosomes, which reveals a monosomy 45,X in around 40-50% of cases. The initial assessment and management of the patient requires multidisciplinary cooperation between various health professionals (paediatric endocrinologist, cardiologist, ENT specialist, gynaecologist, clinical geneticist, PRM doctor, psychologist, etc.). Physical and Rehabilitation Medicine plays an essential role in managing the bone deformities, mobility problems and musculoskeletal complications of Turner syndrome. It also contributes to physical rehabilitation and improving patients' independence.

CONCLUSION

Turner syndrome, which is accompanied by motor disorders, requires multidisciplinary treatment including physical therapy, occupational therapy and specialised medical follow-up. Muscle strengthening exercises, postural re-education and walking exercises are recommended, as are functional treatments to optimize mobility and independence.

Statement of Disclosures

None

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Doi: 10.5606/archisprm.2026.EP-586