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## **EFFECTS OF A CARDIORESPIRATORY REHABILITATION PROGRAMME IN EXERCISE TOLERANCE IN AN ADULT WITH PYCNODYSTOSIS: A CASE STUDY**

Cláudia AGUIAR<sup>1,2</sup>, Tiago TEÓFILO<sup>1</sup>, Énio PESTANA<sup>1</sup>, Pedro Diogo SILVA<sup>1</sup>, Inês ALVES<sup>2,3</sup>

<sup>1</sup> Physical Medicine and Rehabilitation Service, SESARAM, Funchal, Portugal; <sup>2</sup> ANDO Portugal, Évora, Portugal; <sup>3</sup> School of Health and Human Development, Évora University-CHRC, Évora, Portugal; [ines.alves@andoportugal.org](mailto:ines.alves@andoportugal.org)

### **1. Introduction and objectives**

Pycnodysostosis is an ultra-rare autosomal recessive skeletal dysplasia (1-5/1 000 000 births) caused by mutations in the *cathepsin K* gene<sup>1</sup>. Clinical features include disproportionate short stature, facial dysmorphism, bone fragility, and an increased risk of fractures<sup>2</sup>. Individuals with this condition often experience reduced exercise tolerance, impaired mobility, and a heightened risk of falls. The Timed Up and Go (TUG) test is a simple and reliable measure of functional mobility and balance, directly correlating with risk of falls. This study aimed to evaluate the impact of a structured cardiorespiratory rehabilitation program on balance and mobility in an adult with pycnodysostosis, with a particular focus on TUG performance.

### **2. Materials and methods**

A 42-year-old female with genetically confirmed pycnodysostosis (pathogenic variant c.436G C p. (Gly146Arg) in probable homozygosity in *CTSK* gene), with a body mass of 38kg and with 140cm height, was evaluated. The patient had a history of multiple fractures and was recovering from a surgically stabilized left tibial fracture. She underwent a 3-month outpatient cardiorespiratory rehabilitation program, from January to March 2024, consisting of twice-weekly sessions. Each session included a warm-up, strength training, aerobic exercise, balance and flexibility exercises, and monitoring of perceived exertion (RPE) using the modified Borg scale (rated from 0 to 10). The TUG test was administered over a standardized 3-meter distance at baseline, week 4, and week 8, to assess functional mobility and risk of falls.

### **3. Results**

The TUG performance improved considerably throughout the rehabilitation program: from 18 seconds at baseline to 11 seconds at week 4, and 9 seconds at week 8, representing a 50% improvement. In parallel with the TUG test results, the patient reported a reduction in exertional dyspnea, from 5 to 2 RPE, and expressed less fear of falling.

### **4. Conclusion and perspectives**

This case report demonstrates that the TUG test served as a simple, accessible and effective tool for monitoring mobility improvements in an adult with pycnodysostosis. Given the increased risk of fracture associated with this condition, interventions that enhance balance and reduce risk of falls are of particular value. Future research should investigate the long-term sustainability of these improvements and work towards the development of specific rehabilitation guidelines for patients with rare skeletal dysplasias. Multidisciplinary collaboration is essential to optimize care and create standardized rehabilitation protocols for this patient population.

### **BIBLIOGRAPHY**

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