RECOGNIZING EARLY SIGNS OF FIBRODYSPLASIA OSSIFICANS PROGRESSIVA IN INFANCY: A CASE REPORT

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Background

Fibrodysplasia ossificans progressiva (FOP) is an ultrarare, autosomal dominant disease characterized by congenital malformations of the great toes and progressive heterotopic ossification of the connective tissues. It has a prevalence of approximately 1 in 2 million individuals. As the disease progresses, patients experience increasing limitations in mobility, which in turn compromises thoracic cavity expansion, affecting both pulmonary and cardiac function. To date, around 800 cases have been reported worldwide, however, nearly 90% of patients are initially misdiagnosed. Clinical examination, radiological imaging, and genetic testing for mutations in the ACVR1 gene are critical for early and accurate diagnosis.

Case report

We present a case of a one-month-old infact referred to a Physical and Rehabilitation Medicine consultation due to congenital bilateral hallux valgus associated with microdactyly and bilateral thumb adduction. Clinical examination revealed bilateral symmetrical bilateral hallux valgus with microdactyly of the great toes, adducted thumbs and clinodactyly of both fifth fingers. Foot radiographs demonstrated malformation of the first metatarsal bilaterally. Physical rehabilitation was initiated with the goals of functional improvement and correction of hand deformities. At four-months-old, the appearance of multiple hard bony swellings over the cranial vault raised suspicion of FOP, which was subsequently confirmed by genetic testing revealing a mutation of the ACVR1 gene.

Conclusion

FOP is a rare and highly disabling musculoskeletal condition for which no curative or progression-halting treatment currently exists. Misdiagnosis can lead to unnecessary and potentially harmful interventions. The presence of congenital hallux valgus with microdactyly and malformation of great toe in a child, particularly when associated with other skeletal anomalies, should prompt early consideration of FOP, even before the developmental of soft tissue lesions. Prevention of injury by all means, medical management of acute painful flare-ups and rehabilitation efforts should be the goal in the care strategy of individuals with FOP.

Keywords: Fibrodysplasia Ossificans Progressiva, Rehabilitation, Children