Collagen VI related myopathies. When to suspect, how to identify. The contribution of muscle magnetic resonance.

[Article in Spanish]
Suárez B1, Lozano-Arango A1, Araneda D2, Cortés F3, Hervias C1, Calcagno G1, Ortega X2, Castiglioni C1.

Abstract
Myopathies secondary to collagen VI mutations (COLVI-M) are the most frequent in the northern hemisphere, affecting the adult and pediatric population. There are no data on its prevalence in Latin America. They are characterized by a great clinical variability, from severe phenotypes, such as Ullrich congenital muscular dystrophy (UCMD), to intermediate and mild ones such as Bethlem myopathy (BM). Its onset is also variable and extends from the neonatal period to adulthood. Given the presence of joint hypermobility, the differential diagnosis should be made with various connective tissue diseases. The classical diagnostic algorithm in many patients has been insufficient to guide the genetic study in an adequate way, and from this the muscular magnetic resonance imaging has emerged as a very useful tool for a better diagnostic approach of this and other muscular pathologies. This objective of this review is to study the forms of presentation, clinical characteristics, specific diagnostic study, differential diagnosis and management of one of the most frequent hereditary muscular pathologies, with emphasis on the contribution of muscle magnetic resonance imaging.

PMID: 29999148 DOI: 10.4067/S0370-41062018005000305

Free full text