

Huntington's disease-like disorders in Latin America and the Caribbean.

Walker RH¹, Gatto EM², Bustamante ML³, Bernal-Pacheco O⁴, Cardoso F⁵, Castilhos RM⁶, Chana-Cuevas P⁷, Cornejo-Olivas M⁸, Estrada-Bellmann J⁹, Jardim LB¹⁰, López-Castellanos R¹¹, López-Contreras R¹², Maia DP⁵, Mazzetti P⁸, Miranda M¹³, Rodríguez-Violante M¹⁴, Teive H¹⁵, Tumas V¹⁶.

Author information

Abstract

Diseases with a choreic phenotype can be due to a variety of genetic etiologies. As testing for Huntington's disease (HD) becomes more available in previously resource-limited regions, it is becoming apparent that there are patients in these areas with other rare genetic conditions which cause an HD-like phenotype. Documentation of the presence of these conditions is important in order to provide appropriate diagnostic and clinical care for these populations. Information for this article was gathered in two ways; the literature was surveyed for publications reporting a variety of genetic choreic disorders, and movement disorders specialists from countries in Latin America and the Caribbean were contacted regarding their experiences with chorea of genetic etiology. Here we discuss the availability of molecular diagnostics for HD and for other choreic disorders, along with a summary of the published reports of affected subjects, and authors' personal experiences from the regions. While rare, patients affected by non-HD genetic choreas are evidently present in Latin America and the Caribbean. HD-like 2 is particularly prevalent in countries where the population has African ancestry. The incidence of other conditions is likely determined by other variations in ethnic background and settlement patterns. As genetic resources and awareness of these disorders improve, more patients are likely to be identified, and have the potential to benefit from education, support, and ultimately molecular therapies.

Copyright © 2018. Published by Elsevier Ltd.

KEYWORDS:

Chorea-acanthocytosis; Genetics; HDL2; Huntington disease; Spinocerebellar ataxia

PMID: 29853295