Revista Mexicana de Neurociencia

PERMANYER

Check for updates

EDITORIAL

Clinical insights on some rare genetic neurologic diseases

Miguel García-Grimshaw

Department of Neurology and Psychiatry, Instituto Nacional de Ciencias Médicas y Nutrición Salvador Zubirán, Mexico City, Mexico

Early detection of genetic neurologic diseases remains crucial for improving the quality of life (QoL) of patients affected by these diseases. The current issue of *Revista Mexicana de Neurociencia* includes articles on two rare genetic neurologic disorders. The first one is a retrospective, cross-sectional study by conducted Cervantes-Arriaga et al., where they studied the factors associated with delays in diagnosing Huntington's disease (HD); a study conducted at the National Institute of Neurology and Neurosurgery, Mexico City, Mexico, including 107 patients with genetically confirmed HD treated at their movement disorders clinic between 2003 and 2018, in their study, finding that neuropsychiatric manifestations and younger age at presentations were directly associated with a delayed diagnosis. Although, as of this date, there is no effective treatment for this devastating disease, there are some ongoing basic science studies on the role of glial differentiation defects in murine HD models, specifically deficits in the transcription factor 7 like 2 (Tcf7l2), a factor that induces developmental deficiencies in myelination, maintenance, and remyelination, defects which can be potentially rescued by overexpressing Tcf7l2 as proved by Benraiss et al., bringing the light to a potential therapeutic target[1].

The second study is an interesting review article by Briseño-Godínez et al. on transthyretin-related hereditary amyloidosis, also an autosomal dominant disease caused by a mutation in the transthyretin (TTR) gene, which causes the deposition of mis-folded TTR protein in several organs, among them the peripheral and autonomic nervous systems, being cardiomyopathy, symmetric length-dependent sensory-motor, and autonomic polyneuropathy the hallmark manifestations[2]. During the past decade, multiple disease-modifying molecules have been developed and proven to increase the survival and QoL of these patients[3,4]. Hence, this review provides essential information for the general neurologist that may help establish an early diagnosis that can positively impact the outcomes of these patients.

We hope that this issue will greatly interest our discerning readership, especially neurologists and non-neurologist interested in genetic neurologic diseases.

Miguel García-Grimshaw
Editor in Biostatistics, Revista Mexicana de Neurociencia

References

1. Benraiss A, Mariani JN, Tate A, Madsen PM, Clark KM, Welle KA, et al. A TCF7L2-responsive suppression of both homeostatic and compensatory remyelination in Huntington disease mice. Cell Rep. 2022;40:111291.
2. Gonzalez-Duarte A. Autonomic involvement in hereditary transthyretin amyloidosis (hATTR amyloidosis). Clin Auton Res. 2019;29:245-51.
3. Adams D, Gonzalez-Duarte A, O'Riordan WD, Yang CC, Ueda M, Kristen AV, et al. Patisiran, an RNAi Therapeutic, for hereditary transthyretin amyloidosis. N Engl J Med. 2018;379:11-21.
4. Adams D, Tournev IL, Taylor MS, Coelho T, Planté-Bordeneuve V, Berk JL, et al. Efficacy and safety of vutrisiran for patients with hereditary transthyretin-mediated amyloidosis with polyneuropathy: a randomized clinical trial. Amyloid. 2022 Jul 23:1-9. DOI: 10.1080/13506129.2022.2091985. Epub ahead of print.

Correspondence:
Miguel García-Grimshaw
E-mail: miguel.garciag@incmnsz.mx

Date of reception: 12-01-2023
Date of acceptance: 18-01-2023
DOI: 10.24875/RMN.M23000094

Available online: 01-03-2023
Rev Mex Neuroci. 2023;24(2):29
www.revmexneurociencia.com