COVID-19 is an emerging, rapidly evolving situation.
Get the latest public health information from CDC: https://www.coronavirus.gov.
Get the latest research from NIH: https://www.nih.gov/coronavirus.

Case Reports
Epub 2019 Oct 3.

Two middle-aged women with the Finnish variant of muscle-eye-brain disease (MEB)

Maria Arvio ¹ ² ³ ⁴, Laura Määttänen ⁵, Maria Haanpää ², Jaana Lähdetie ⁶

Affiliations
PMID: 31580529 DOI: 10.1002/ajmg.a.61369

Abstract

Muscle-eye-brain disease (MEB) is a recessively inherited rare disease. Sixteen different gene mutations are known, with the most common mutations in the POMGNT1 gene. The disease is now called congenital muscular dystrophy-dystroglycanopathy type A3 (MDDGA3). It manifests itself as muscular dystrophy with eye and brain anomalies and intellectual disability. Previous clinical reports describe young patients. We have been able to follow two patients for almost 40 years. Their clinical picture has remained quite stable since adolescence, appearing as severe intellectual and motor disability, extremely limited communication skills, visual impairment, epilepsy, joint contractures, repeated bowel obstructions, teeth abrasion due to bruxism, an irregular sleep pattern and as a previously unreported feature hypothermic periods manifesting as excessive sleepiness.

Keywords: POMGNT1 gene; MEB; dystroglycanopathia; muscle-eye-brain disease.

© 2019 Wiley Periodicals, Inc.

Related information
MedGen

LinkOut – more resources
Full Text Sources
Ovid Technologies, Inc.
Wiley