Empowerment of genetic information by women at-risk of being carriers of Duchenne and Becker muscular dystrophies

HOEFEL, Alice M.L.[1]; ROSA, Bruna F.[2]; WESCHENFELDER,Cesar A.[2]; DONIS, Karina C.[3,4]; SAUTE, Jonas A.M.[1,2,3,4].

1. Graduate Program in Medicine: Medical Sciences, School of Medicine, Universidade Federal do Rio Grande do Sul, Porto Alegre, Brazil
2. School of Medicine, Universidade Federal do Rio Grande do Sul, Porto Alegre, Brazil
3. Medical Genetics Service, HCPA, Porto Alegre, Brazil
4. Internal Medicine Department, School of Medicine, Universidade Federal do Rio Grande do Sul, Porto Alegre, Brazil

E-mail para contato: jsaute@hcpa.edu.br

Introduction: The emergence of therapies acting on specific molecular targets for Duchenne and Becker muscular dystrophies (DBMD) led to expanded access of diagnostic DMD analysis. However, it is unclear how much of these advances have also improved healthcare and access to genetic testing for women at-risk of being carriers. Objectives: to evaluate the process of genetic counseling and empowerment of genetic information by women from DBMD families. **Methods:** We carried out a cross-sectional study between February and June 2022 in Brazil. The online survey with items regarding sociodemographic data; family history; access to health services; reproductive decisions; and the Genomic Outcome Scale was answered by 123 women recruited from a rare diseases reference service and a nationwide patient advocacy group. **Results:** Genetic counseling was reported by 77/123 (62.6%) of women and 53.7% reported having performed genetic analysis of DMD. Although the majority knew about the risks for carriers of developing heart disease and muscle weakness, only 35% of potential carriers have had cardiac studies performed at least once in their lives. Country region, type of kinship, number of affected males in the family, age, notion of genetic risk, education level and participation in advocacy groups were the main factors associated with adequate healthcare access to women and empowerment of genetic information. **Conclusion:** Education to health professionals and policies to expand access to carrier genetic testing, whether public policies or regulation of pharmaceutical companies´ diagnostic programs, are paramount to improve the care of families with DBMD in Brazil.

Key-Words: Genetic counseling; Duchenne Muscular Dystrophy; Becker Muscular Dystrophy; Genetic testing; Genomic outcome scale