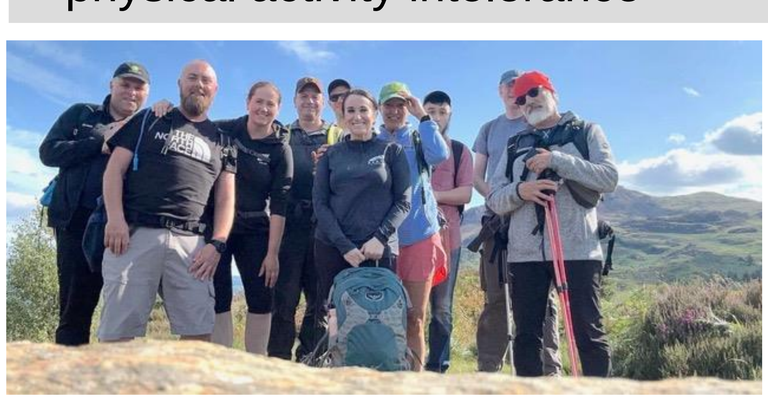
Toward an understanding of GSD5 (McArdle disease): How do patients learn to live with the metabolic defect in daily life

Karazi W[1], Coppers J[1], Maas D[1], Cup E[1], Bloemen B[1], Voet N[1], Groothuis J[1], Pinós T[2], Marti R[2], Quinlivan R[3], Løkken N[3], Vissing J[4], Bhai S[5], Wakelin A[6], Reason S[6], Voermans N[1]

[1]Departments of Neurology, Rehabilitation and Health Evidence, Radboud University Medical Center, Nijmegen, the Netherlands; [2]Biomedical Network Research Centre on Rare Diseases (CIBERER), Vall d'Hebron Research Institute, Barcelona, Spain; [3]MRC Centre for Neuromuscular Diseases, UCL Institute of Neurology, National Hospital, London, UK; [4]Copenhagen Neuromuscular center, Rigshospitalet, Copenhagen, Denmark; [5]Department of Neurology at the University of Texas Southwestern Medical Center, USA; [6]International Association for Muscle Glycogen Storage Disease, California, USA

Background

- Glycogen storage disease type 5 (GSD5; McArdle disease) is an AR metabolic myopathy caused by a deficiency of muscle glycogen phosphorylase
- Symptoms include myalgia, fatigue, cramping, contractures and rhabdomyolysis
- Individuals with GSD5 experience physical activity intolerance
- We aimed to capture the daily life experiences of individuals with GSD5
- The focus was on adapting to and coping with their physical activity intolerance



The patient organization IamGSD has organized annual walking courses to offer peer support and share recommendations for adapting and coping (www.iamgsd.org). This photograph shows the participants of the Future Leaders course in 2023.

Methods

- An online survey composed with IamGSD
- Customized and validated questionnaires on demographics, general health and comorbidities, physical activity, psychosocial well-being and functioning, pain, fatigue and adapting to and coping with GSD5

Results

- 162 Participants from 16 countries
- 60% female; mean current age 49.7; mean age at diagnosis 31.5; mean age at onset 7.7 years
- The majority, n = 86 (69%) was from NL, USA or UK
- We observed a high rate of misdiagnosis prior to a definitive GSD5 diagnosis (49%)
- Being diagnosed had a strong impact on emotional status, daily life activities and important life choices
- A large proportion had not received rehabilitation (41%) nor medical treatment (57%) before diagnosis
- Engagement in vigorous and moderate physical activity was reduced compared with the general population
- Health related quality of life was low, most likely related to low physical health.
- Median score on Fatigue Severity Scale was 4.3, indicating moderate to severe fatigue
- Patients reported limited receipt of information upon diagnosis
- Patients had found various ways to adapt to and cope with their disability (in school, work, sports, household and social activities) themselves
- Coping consisted of using assistive devices, using help from others, avoiding certain activities, and using second wind
- Patients had useful recommendations for newly diagnosed patients:

Ten most frequent recommendations	# (of 162)
Accept your abilities and maintain an active lifestyle	75
Use information from patient organization IamGSD	56
Find support from family members	51
Visit a specialized neurologist	40
Find psychological support	28
Seek support from colleagues	27
Start physical therapy	26
Find support at school	21
Try the ketogenic diet	20
Start a rehabilitation program (multidisciplinary)	19

Conclusion

- Participants recommendations included:
  - *Accept one's limited abilities*
  - *Maintain an active lifestyle*
  - *Educate yourself (IamGSD website, publications, YouTube channels, Facebook group and courses)*
- Adequate counseling on ways of adapting and coping is expected to increase both health related quality of life and physical activity

More info: nicol.voermans@radboudumc.nl

References: *Karazi W et al. EUROMAC Consortium. Data from the European registry for patients with McArdle disease (EUROMAC): functional status and social participation. Orphanet J Rare Dis. 2023 Jul 25;18(1):210.*
Reason SL et al. Development of Continuum of Care for McArdle disease: A practical tool for clinicians and patients. Neuromuscul Disord. 2023 Jul;33(7):575-579.
Karazi et al. Toward an understanding of GSD5 (McArdle Disease): How do individuals learn to live with the metabolic defect in daily life Journal of Neuromuscular Diseases. In press.