Clinical Genetics

AB114. Study of functional independence of patients with Duchene muscular dystrophy

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Background: Duchene muscular dystrophy (DMD) is an X-linked inherited neuromuscular disorder due to mutations in the dystrophin gene. It is characterized by progressive muscle weakness and wasting due to the absence of dystrophin protein that causes degeneration of skeletal and cardiac muscle. Aims of this study were to evaluate functional independence of DMD patients.

Methods: The study included 30 patients with DMD diagnosed and managed at Department of Endocrinology, Metabolism and Genetics, National Children's Hospital in 2016. This is a cross-sectional study using WeeFIM questionnaire (self-care, mobility and cognition) for the parents or caregivers.

Results: We found that only 3.3–6.6% of DMD patients needed total assistance of self-care (eating, grooming, bathing, and bowel and bladder management). More than 30% of DMD patients were completely independent on their self-care. Half of the DMD patients did not require assistance (transfer to wheelchair, to toilet, etc.) but only 3.3% of the patients reported total independence on going up or down stairs. The majority of DMD patients had normal ability in communication, while 33% of the patients had difficulty in social communication and making friends.

Conclusions: DMD which is a genetic disease with no current effective specific treatment leads to substantial negative impact on self-care and mobility. Patients often need assistance from their caregiver or parents.

Keywords: Duchene muscular dystrophy (DMD); functional independence

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