Birth Defects, Dysmorphology, Skeletal Dysplasia, Craniofacial Anomalies

AB106. Study of functional independence of patients with Hunter syndrome (mucopolysaccharidosis type II)

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Background: Hunter syndrome or mucopolysaccharidosis type II (MPS II) is caused by deficiency of lysosomal enzyme iduronate-2-sulfatase and characterized by neurologic and movement functions. Aims: To evaluate functional independence for patients with Hunter syndrome.

Methods: Study included 17 patients with Hunter syndrome 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: The percentage of patients needed total assistance of self-care was 35.9%, and only 4.69% of patients were complete independence of self-care. Mobility ability with supervision was 41.25% of patients, and patients with complete independence on mobility account for 11.25%. Only 12.5% patients had complete independence on communication.

Conclusions: All self-care, mobility and communication of patients with Hunter syndrome were affected, and patients need assistance from their parents or caregivers.

Keywords: Mucopolysaccharidosis type II (MPS II); Hunter syndrome; functional independence

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