Birth Defects, Dysmorphology, Skeletal Dysplasia, Craniofacial Anomalies

AB107. Study of functional independence of patients with osteogenesis imperfecta

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Background: Osteogenesis imperfecta (OI) is a genetic bone disorder characterized by fragile bones that break easily. It is a complicated, variable and rare disorder. Its major feature is a fragile skeleton, but many other body systems are also affected. OI is caused by a mutation in a gene that affects bone formation, bone strength and the structure of other tissues. People with OI experience frequent broken bones from infancy through puberty. This study aims to evaluate functional independence for patients with osteogenesis imperfect.

Methods: Study included 32 patients with OI diagnosed and managed at Department of Endocrinology, Metabolism and Genetics, National Children's Hospital in 2017. This is a cross-sectional study using WeeFIM questionnaire (self-care, mobility and cognition) for the parents or caregivers.

Results: About 62% of OI patients were complete independence of self-care (grooming, eating, etc.), but less than half of OI patients (40%) need total assistance of self-care (bathing, toileting, etc.). Mobility ability (transfer to bath, toilet, etc.) without supervision were more than 50% of patients with OI, and 12% of OI patients need total assistance. Half of OI patients need total assistance of up and down stair. All OI patients had complete independence on communication.

Conclusions: Excepting communication, almost self-care and mobility of OI patients were affected and patients need assistance from their parents or caregivers.

Keywords: Osteogenesis imperfect (OI); functional independence

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