Newborn Screening, Inborn Errors of Metabolism

AB112. Spectrum of mucopolysaccharidoses in Vietnam

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Background: Mucopolysaccharidoses (MPSs) are a group of lysosomal storage disorders (LSDs) caused by deficiency of a specific lysosomal enzyme, consisting of seven subtypes. In MPSs, the breakdown of the glycosaminoglycans (GAGs), chondroitin sulfate (CS), dermatan sulfate (DS), heparan sulfate (HS), keratan sulfate (KS), and/or hyaluronan is disrupted.

Methods: This report is to highlight the spectrum MPS patients diagnosed in Vietnam from 2013–2016.

Results: Seventy-one cases with MPSs were confirmed by enzyme assays and mutation analyses for MPS I, II, IV-A, and VI. Spectrum of subtype of MPS patients includes MPS II (38 cases, 53.5%), MPS IV-A (15 cases, 21.1%), MPS VI (8 cases, 11.3%), MPS I (7 cases, 9.9%), III-A (1 case, 1.4%), and III-B (2 cases, 2.8%). Seven patients (2 cases of MPS I and 5 cases of MPS II) are receiving enzyme replacement therapy.

Conclusions: Newborn screening, national registry, support group, and multidisciplinary care including ERT, genetic counseling were scheduled for MPS patients in Vietnam.

Keywords: Mucopolysaccharidoses (MPSs); enzyme replacement therapy; lysosome storage disorders

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