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Spectrum of Lysosomal Storage Disease Targeted for Newborn Screening in Vietnam

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Vietnam is the easternmost country on the Indochina Peninsula in Southeast Asia. With an estimated 94 million inhabitants as of 2016, it is the world's 13th-most-populous country, and the eighth-most-populous Asian country. Congenital anomalies accounted about 22% of causes of deaths in children under-5 years old[1)]. The aim of this report is to highlight the spectrum and future direction of newborn screening for Vietnamese patients with some lysosomal storage diseases (LSDs) in Vietnam. The first service for LSDs was set up at the Northern referral center of Pediatrics–National Children's Hospital, Hanoi (NCH) in 2004 officially. There was no specific biochemical & molecular laboratory for LSDs in Vietnam so far. 178 cases from 10 years with suspected mucopolysaccharidosis (MPS) were diagnosed clinically and using urinary total glycosaminoglycans (GAGs) analysis and specific GAGs using tandem mass spectrometry (LC-MS/MS). 67/178 cases were confirmed using enzyme assay and mutation analysis for MPS I, II, IVA and VI. Spectrum of subtype of MPS patients is MPS II (35 cases, 52.2%), MPS IVA (15 cases, 22.4%), MPS VI (8 cases, 12.0%), MPS I (6 cases, 8.9%), IIIA (1 case, 1.5%) and IIIB (2 case, 3.0%). 19 cases had I-cell disease; one case had multiple sulfatases deficiency; and 21 cases had Pompe disease and 9 cases had Gaucher disease. 2 patients with MPS (1 case-MPS I and 1 case-MPS II) are receiving ERT with support from Genzyme from 2.5 years. Five patients with Pompe disease was received ERT from 20 to 2 months. Five patients with Gaucher disease are under ERT from 14 to 3 years. A specific LSDs laboratory in Vietnam, training for medical staffs, newborn screening, national registry, support group and multidisciplinary care including ERT, genetic counseling were scheduled for LSDs patients in Vietnam[2)].

References

1. Lee HY, Van Do D, Choi S, Trinh OT, To KG. Trends and determinants of infant and under-five childhood mortality in Vietnam, 1986-2011. Glob Health Action 2016;9:29312.
2. Kubaski F, Suzuki Y, Orii K, Giugliani R, Church HJ, Mason RW, et al. Glycosaminoglycan levels in dried blood spots of patients with mucopolysaccharidoses and mucolipidoses. Mol Genet Metab 2017;120:247-54.

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