Brief Communication

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Newborn Screening Status of Lysosomal Storage Diseases in Japan

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Patients with lysosomal storage diseases (LSDs) during newborn period are typically asymptomatic and have no specific symptom. Therefore, to diagnose patients with LSD at early stage is often difficult and delayed. It is well known that early initiation of treatment significantly improve the therapeutic outcome. National Center for Child Health and Development (NCCHD) started newborn screening for Pompe disease in 2011 by measuring acid α-glucosidase activity in dried blood spots using the classic 4-methylumbelliferone method. So far over 8000 infants have been screened and no cases of Pompe disease have been found. Now we have been expanding newborn screening around Tokyo area with a population of about 12 million. In 2015, two hospitals (St. Marianna University Hospital and Kawasaki Municipal Tama Hospital) joined the newborn screening for infantile Pompe disease. In addition, two major children's hospitals (Kanagawa Children's Medical Center and Chiba Children's Hospital) also joined screening program in 2016. Another two flagship hospitals (Saitama Children's Medical Center and Saitama Medical University hospital) are also going to participate in the newborn screening program. At this opportunity, we are planning to change and improve our newborn screening system for LSDs in several points. First new LSDs (Mucopolysaccharidosis Type I, Fabry disease and adrenoleukodytrophy (ALD)) will be added to the list of target diseases. Second, tandem mass spectrometry (LC-MS/MS) assay will replace the current fluorescent assay as measurement method for lysosomal enzyme or accumulated substrate[1]. Kazusa DNA Research Institute will engage in this screening system project and perform the LC-MS/MS assays. This new screening system will start in 2017.

Reference

1. Mashima R, Sakai E, Kosuga M, Okuyama T. Levels of enzyme activities in six lysosomal storage diseases in Japanese neonates determined by liquid chromatography-tandem mass spectrometry. Mol Genet Metab Rep 2016;9:6-11.

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