Brief Communication

J Mucopolysacch Rare Dis 2017;3(1):35
https://doi.org/10.19125/jmrd.2017.3.1.35
pISSN 2465-8936 · eISSN 2465-9452

JMRD
Journal of Mucopolysaccharidosis and Rare Diseases

New Born Screening Tests Status of Lysosomal Storage Diseases in Korea

Junghan Song

Department of Laboratory Medicine, Seoul National University Bundang Hospital and Seoul National University College of Medicine, Seoul Korea

Lysosomal storage disorders (LSDs) are caused by absent activity of one or more of these enzymes due to mutations of genes encoding lysosomal hydrolases or enzymes, which results in the accumulation of an intermediate metabolic product. Tandem mass spectrometry (MS/MS) screening for LSDs was first described by Li et al. in 2004. They performed direct multiplex assays for Fabry, Gaucher, Krabbe, Niemann-Pick A/B, and Pompe diseases by using dried blood spots. The findings of these studies demonstrated the usefulness of MS/MS in newborn screening for LSDs. MS/MS techniques have been also developed for the detection of some MPSs such as Hurler syndrome, Hunter, Maroteaux-Lamy, and Morquio syndrome type A. Recent technological developments enabled automated analysis as well as rapid high throughput screening with minimizing the number of separate incubations. In Korea, our laboratory revised multiplex enzyme reaction conditions and procedures for the assay using MS/MS for 6 LSDs including Niemann-Pick-A/B, Krabbe, Gaucher, Fabry, Pompe disease, and Hurler syndrome in 2010. Assay performance was of the acceptable standard. The assay also enabled unambiguous differentiation between samples obtained from healthy newborns and patients, suggesting that our method was also effective for newborn screening of LSDs in the Korean population. We also developed methods for measuring iduronate sulfatase activities in DBSs for the screening of Hunter disease which is relatively common in Korea. More detailed status on the NST of LSD in Korea will be presented.

References

1. Cho SE, Kwak JR, Lee H, Seo DH, Song J. Triplex tandem mass spectrometry assays for the screening of 3 lysosomal storage disorders in a Korean population. Clin Chim Acta 2016;454:20-7.
2. Han M, Jun SH, Song SH, Park KU, Kim JQ, Song J. Use of tandem mass spectrometry for newborn screening of 6 lysosomal storage disorders in a Korean population. Korean J Lab Med 2011;31:250-6.

Received April 28, 2017; **Revised** May 25, 2017; **Accepted** June 1, 2017
Correspondence to: Junghan Song
Department of Laboratory Medicine, Seoul National University Bundang Hospital and Seoul National University College of Medicine, 82 Gumi-ro 173beon-gil, Bundang-gu, Seongnam 13620, Korea
Tel: +82-31-787-7691, Fax: +82-31-787-4015, E-mail: songjhcp@gmail.com