April 2, 2025
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Announcement of Publication of a Paper on Secondary Screening for Mucopolysaccharidoses Using Dried Blood Spots

We are pleased to announce that Anges Clinical Research Laboratories (ACRL), a clinical laboratory primarily providing screening tests for rare inherited disorders, has developed a novel secondary screening method for mucopolysaccharidoses (MPS), one of the diseases covered by its screening services. A paper describing this secondary screening method was published in Biochemistry and Biophysics Reports on March 31, 2026.

In newborn screening for mucopolysaccharidoses based on enzyme activity measurements, it has long been a major challenge that among screen-positive newborns, the overwhelming majority are false positives caused by carriers or pseudodeficiency. In Japan, screen-positive cases for MPS have traditionally undergone follow-up testing such as urinary glycosaminoglycan analysis, which requires 50 mL of urine, or genetic testing. These approaches impose a substantial burden, both in collecting sufficient urine samples from newborns and on parents who must wait anxiously for definitive test results.

In this study, we evaluated a secondary screening method for mucopolysaccharidoses using dried blood spot samples obtained during expanded newborn screening. This method quantifies disaccharides derived from dermatan sulfate, heparan sulfate, and keratan sulfate in blood using LC–MS/MS.

As a result, for MPS type I, the method achieved 100% sensitivity and 100% specificity, completely eliminating false-positive cases. For MPS type II, by applying a strict criterion requiring concurrent elevation of both dermatan sulfate and heparan sulfate, specificity was improved to 100% while maintaining sensitivity, demonstrating the practical value of this method.

Since last year, ACRL has been the first in Japan to implement this approach as a secondary screening test for mucopolysaccharidoses, contributing to the reduction of burdens on infants, their families, and medical institutions caused by false-positive results.

For further details, please visit the following website:
https://www.sciencedirect.com/science/article/pii/S2405580826001299?via%3Dihub#

〈Paper Title〉
Quantification of glycosaminoglycans in dried blood spots, and evaluation of its usefulness as a secondary newborn screening test for mucopolysaccharidoses

Biochemistry and Biophysics Reports
https://www.sciencedirect.com/journal/biochemistry-and-biophysics-reports