

NBS07

Newborn Blood Spot Screening for Pompe Disease by Lysosomal Acid α -Glucosidase Activity Assays

This report discusses the detection of Pompe disease (PD) by population-based newborn screening using dried blood spot specimens to measure acid α -glucosidase enzyme activity. Classic infantile-onset PD is a lethal disorder that is not evident at birth, and therapy effectiveness is improved by presymptomatic detection.

A CLSI report for global application.