



Lysosomal Disorders Screening in DBS LC-MS/MS Analysis Kit

Lysosomes serve enzymes that metabolize excess sugars and lipids into substances which cells can utilize. As lysosomes do not function properly, these sugars and fats accumulate in the cell rather than being used or disposed of. Lysosomal storage diseases are rare but can cause death if left untreated. There are more than 50 types of lysosomal storage disease. The most common types in infants and children include:

- Gaucher
- Niemann-Pick
- Fabry
- Krabbe
- Pompe
- Mucopolysaccharidoses (MPS)

Highlights of the Analysis Kit



Simultaneous screening of six lysosomal storage diseases (Pompe, Fabry, Krabbe, Gaucher, Niemann–Pick A/B, and MPS I)



Total run time is 7.5 min.



Lyophilised cocktail of the substrates includes stable isotope standards of corresponding products

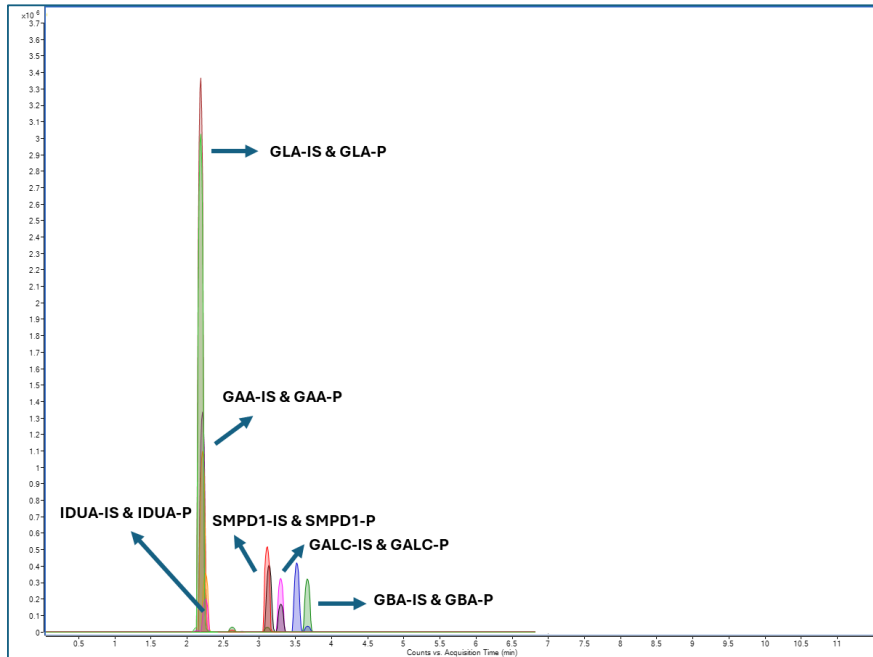


Simple and single sample preparation protocol for all parameters

Parameters			
GAA (α -Glucosidase) GLA (α -Galactosidase)	GALC (Galactosylceramidase) GBA (α -Glucocerebrosidase)	SMPD1 (Acid Sphingomyelinase)	IDUA (α -L-Iduronidase)
Sample Type			
Dried Blood Spot (DBS)			

Sample Preparation

1	Place a 3 mm DBS punch in microcentrifuge tube
2	Incubate at 37 °C for 16 h in an orbital shaker at 250 rpm
3	Quench the reaction by the addition of 100 μ L of Reagent-1 and vortex for 5 sec.
4	Add 400 μ L Reagent-2 and Reagent-3 and vortext for 10 sec.
5	Centrifuge the microtube at 4000 rpm for 5 min. at ambient temperature
6	Transfer 100 μ L of the upper layer to a new microtube and dry it under a stream of nitrogen at room temperature
7	Add 200 μ L of Reagent-4 to the microtube, pipette up and down 3 times
8	Transfer the final solution into a inserted-HPLC vial prior to injection



Total ion chromatogram of the analysis



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