NeoLSD™ MSMS kit

The first commercial IVD kit for screening of Pompe, MPS-I, Fabry, Gaucher, Niemann-Pick A/B and Krabbe disorders from a single DBS sample.

PerkinElmer®
For the Better

A PIONEER IN EXPANDING NEWBORN SCREENING

FIRST COMMERCIAL IVD KIT FOR NEWBORN SCREENING OF SIX LYSOSOMAL STORAGE DISORDERS FROM A SINGLE DRIED BLOOD SPOT

PerkinElmer, a pioneer and the global leader in mass spectrometry based newborn screening since 2002, currently offers the NeoBase™ Non-derivatized MSMS Kit, the commonly used reagent in mass spectrometry based testing for amino acid, organic acid, and fatty acid oxidation disorders, used to screen for millions of babies annually.

Due to advancements in treatments of lysosomal storage disorders (LSD) and the subsequently increasing demand for newborn screening of these disorders, we are proud to release the NeoLSD™ MSMS Kit, the first commercial mass spectrometry based IVD kit for newborn screening of up to six LSD disorders from a single blood spot punch. The NeoLSD MSMS kit enables running up to 500 tests per instrument per day and easily switching between NeoBase2™ (in development) and NeoLSD™ kits on a single MSMS instrument. In addition, the NeoLSD™ kit provides you a wide analytical range, leading to potentially lower false positive rates, less costs associated with repeat enzyme activity tests and unnecessary molecular confirmatory testing.

- **Pompe**
- **MPS-1**
- **Niemann-PICK A/B**
- **Krabbe**
- **Gaucher**
- **Fabry**

BENEFITS OF THE ASSAY

- **Screen for up to six LSDs from a single DBS punch and single incubation**
  You can screen for the most commonly occured LSDs from a single DBS punch, saving time, human resources, and consumables
- **Wide dynamic range**
  MSMS provides you a wide dynamic range, enabling better separation of affected and non-affected individuals, potentially leading to less false positives, less costs and subsequent parent anxiety
- **High sample sample volume per day**
  With a run time of two minutes per sample, you can achieve a throughput that meets the demand of newborn screening

KIT INCLUDES ALL NECESSARY FOR EFFECTIVE LSD NEWBORN SCREENING

KIT INCLUDES:

Package P1		Package P2	Package P3
1/1 NeoLSD Kit Controls C1 C2 C3 (3 filter paper cassettes each containing 2 sets of dried blood spots)	**2/2** NeoLSD Assay Buffer (1 Bottle, 35 mL)	NeoLSD Extraction Solution (1 bottle, 700 mL)	Microplate, U-bottomed (20 plates)
NeoLSD Substrates and Internal Standards (5 vials, dried)		Neo MSMS Flow Solvent (1 bottle, 800 mL)	Microplate, Deep Well (10 plates)
Barcode labels for the plate (30 pcs)			Adhesive aluminum foil microplate covers (20 sheets)
Lot-specific quality control certificate (1pc)			Adhesive microplate covers (10 sheets)

ASSAY PRINCIPLE

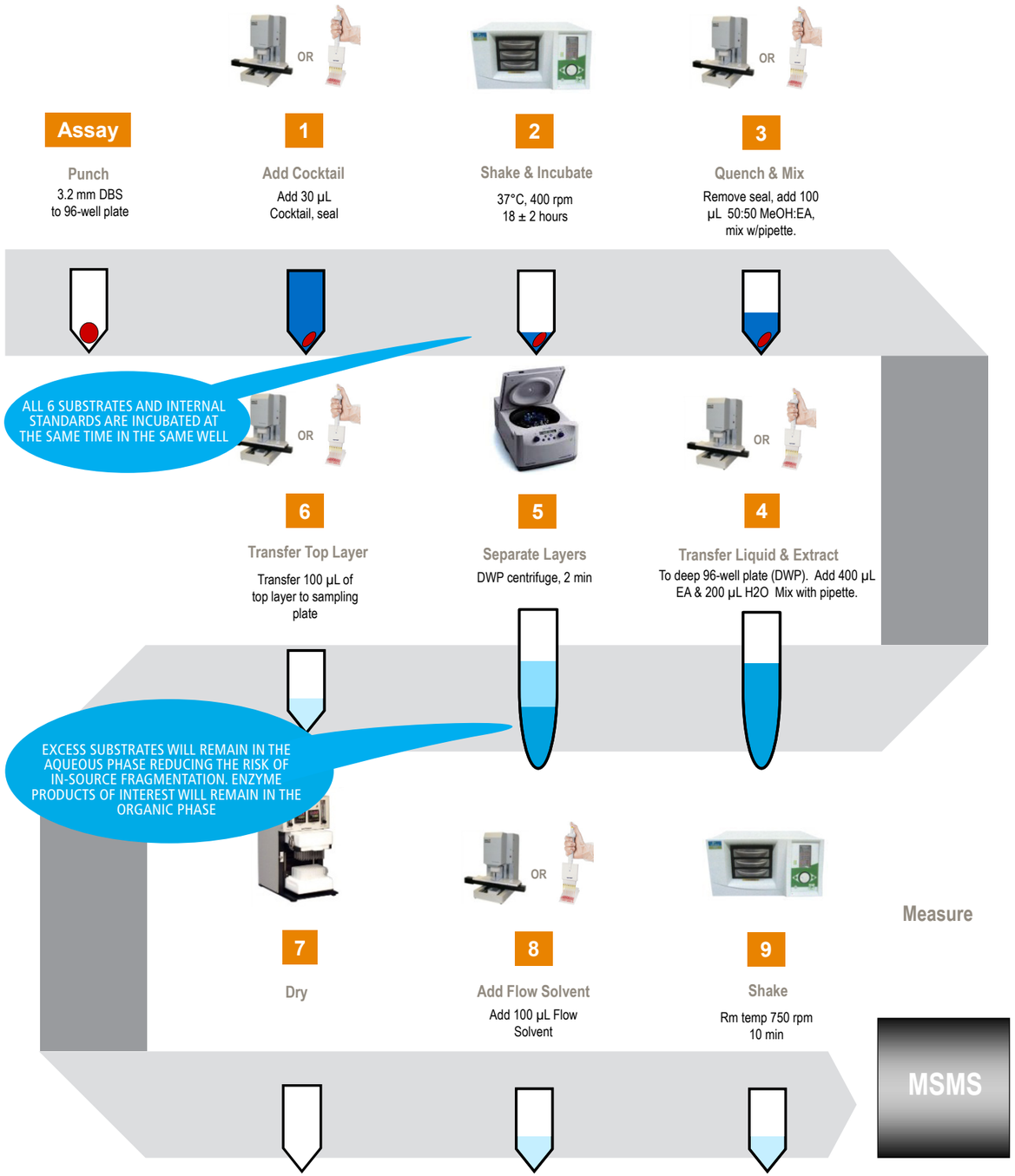
Enzyme activities are assessed by measuring the product generated when an enzyme reacts with a synthesized specific substrate to create a specific product as shown in the equation below:

Enzyme (E) + Substrate (S) → [ES complex] → Enzyme + Product (P)

In the NeoLSD Kit the internal standards and the enzyme-generated products are measured by Flow Injection Analysis – Tandem Mass Spectrometry (FIA-MSMS) using Multiple Reaction Monitoring (MRM). Enzymatic activities (Ae) are expressed as micromoles (µmol) per hour (h) per Liter (L) calculated from the peak ratio of each measured product and its associated internal standard and the known internal standard concentrations using the formula listed below:

$$Ae = \{(P/IS) * [IS] * V\} / (3.1 * ti * RRF)$$

EASY ASSAY WORKFLOW



EFFICIENT NEWBORN SCREENING

DESCRIPTIVE STATISTICS OF NEWBORN SCREENING SPECIMENS IN A ROUTINE SCREENING LABORATORY

Enzyme	n	Enzyme activity (µmol/L/h)				
		Range	Mean	Median	Lower percentiles	
					0.5th	1.0th
ABG	1981	2.48-52.0	11.19	10.31	3.52	3.89
ASM	1981	1.30-35.1	7.12	6.63	2.66	2.95
GALC	1981	0.30-42.2	4.68	3.91	0.79	0.97
IDUA	1981	2.06-22.1	7.06	6.75	2.52	2.86
GLA	1981	3.31-85.1	11.83	10.16	4.08	4.19
GAA	1981	1.98-37.2	9.77	9.07	3.45	3.77

WIDE MEASURING RANGE

Enzyme	Xevo
	Measuring Range µmol/L/h)
ABG	0.67-19.1
ASM	0.88-20.0
GALC	0.32-6.1
IDUA	0.47-18.8
GLA	0.92-20.3
GAA	0.63-26.2

MSMS WORKSTATION SOFTWARE

Completely new software for managing, reviewing and reporting results

KEY FEATURES:

- **Import data from instrument software**
  - Concentrations, IS intensties and TIC raw data
- **Automatically perform LSD specific calculations**
- **Visualize results**
  - Plate map with colored wells
  - Flagging of wells violating cutoffs or other QC criteria
  - View TIC & spectra
  - View results by disorder
  - Split grid view for easy review of results
- **SQL server database to store and consolidate data from one or more MSMS instruments**
- **Perform QC trend analysis and reporting**
- **Sample and assay audit tracking**
- **Automatic export to R4S for analysis**
- **Export results to LIMS**

COMPLETE MASS SPECTROMETRY SOLUTIONS FOR NEWBORN SCREENING. A COMMUNITY OF SUPPORT

From dried blood spot cards, punchers, instruments, reagents to informatics, PerkinElmer's mass spectrometry solutions empower newborn screening laboratories across the globe to meet their demands.

By joining the PerkinElmer newborn screening community, you become part of a movement that includes the majority of the newborn screening laboratories and leaders worldwide. In addition to all necesasry equipment, reagents and informatics, we also provide on-site installation and training, ongoing support, phone consults and various training courses. We are here to help you improve your newborn screening program.

WHY SCREEN FOR LYSOSYMAL STORAGE DISORDERS?

LSDs comprise a heterogeneous group of nearly 50 disorders that are caused by genetic defects resulting in the dysfunction, deficiency or absence of a lysosomal enzyme. Although each disorder is rare, LSDs as a group have a frequency of 1 in 7,000-8,000 live births. Affected individuals are unable to metabolize the disease specific substrate of the deficient lysosomal enzyme, which leads to progressive accumulation of the substrate in the lysosomes of tissues. Mutations that result in an LSD are highly heterogeneous. Affected individuals exhibit a broad variety of disease severities with widespread symptoms and ages of onset. Because symptoms are absent at birth, early diagnosis of these diseases by clinical observation can be difficult. Enzyme replacement therapy or hematopoietic stem cell transplantation are available to treat some LSDs, however, if an available therapy is delayed, irreversible tissue damage may result.

Infantile onset forms of some LSDs (Pompe and Krabbe disease) may be fatal within the first year of life when left untreated. Early diagnosis and treatment is essential to the wellbeing of individuals with these diseases.

Products may not be licensed in accordance with their laws in all countries, such as Canada. Please check with your local representative for availability.

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