EXPANDED NEWBORN SCREENING FOR METABOLIC DISORDERS

USING THE NEW NEOBASE<sup>™</sup> NON-DERIVATIZED METHOD



ENVIRONMENTAL HEALTH





# With the new NeoBase<sup>®</sup> non-derivatized kit you can measure amino acids, acylcarnitines and succinylacetone simultaneously

For screening programs that are implementing tandem mass spectrometry, derivatized assay allows for the measurement of 30-40 markers to

support screening of more than 30 genetic metabolic disorders simultaneously. As impressive as the derivatized assay is, two questions still remain:

## Can the assay be simplified? Can it accept new analytes?

PerkinElmer's NeoBase non-derivatized MSMS assay provides a positive answer to both questions. While retaining equivalent analytical performance, it requires only 4 steps compared to 12 steps in the derivatized assay. In addition, it supports two new analytes: Succinylacetone and Proline.



# Now it's easy to detect succinylacetone together with amino acids and acylcarnitines

By ordering the Succinylacetone Assay Solution and adding this to samples at the same time as you add internal standard, you can extract succinylacetone along with amino acids

and acylcarnitines. This allows the simultaneous detection of succinylacetone alongside other key metabolic disease markers.

The amino acid and acylcarnitine recoveries are not affected by the addition of the NeoBase Succinylacetone Solution. Also the total assay imprecision is equally good whether or not Succinylacetone Solution is used.



### Succinylacetone provides earlier and more specific detection of Tyrosinemia type I

Up to now, the commonly used marker for Tyrosinemia type I has been tyrosine. This is not an ideal primary marker because increased tyrosine levels are not always visible when samples are taken at the usual sampling time, which ranges from 24 h to 7 days.

Succinylacetone (SA) accumulates at an early stage in Tyrosinemia type I cases making it a better choice as the primary marker.

### Tyrosinemia type I

Tyrosinemia type I, the most common of the tyrosinemias, is an inherited metabolic disorder attributable to deficiency of fumarylacetoacetate hydrolase, the terminal enzyme in the degradation pathway of tyrosine.

Signs and symptoms of the disorder include failure to thrive, fever, vomiting, diarrhea, hepatomegaly, ascites, jaundice, and renal Fanconi syndrome. If the disorder remains untreated the child may die of acute liver failure before the second year of life, or from chronic liver failure or hepatocellular carcinoma before the end of the second decade of life.



### By measuring SA rather than tyrosine, you can detect Tyrosinemia Type I at a much earlier age







**NeoBase Non-derivatized MSMS kit** 

The NeoBase SA assay provides good linearity and good precision



## **ORDERING INFORMATION**

| 3040-0010 | NeoBase Non-derivatized MSMS kit (outside the USA |
|-----------|---|
| 3040-001U | NeoBase Non-derivatized MSMS kit (in the USA)     |
| 3041-0020 | NeoBase Non-derivatized solutions                 |
| 3042-0020 | NeoBase Succinylacetone assay solution            |

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