Expanded Newborn Screening Programs

PerkinElmer Genetics’ state-of-the-art laboratory in Bridgeville, PA provides one of the most comprehensive newborn screening programs for clinically significant metabolic disorders in the world. The breadth of technology and services provided by our laboratory differentiates us from others performing similar testing. We are a leader in combining tandem mass spectrometry (MS/MS), molecular technologies and biochemical analysis with tightly integrated software to ensure timely and accurate results.

Primary Marker for Detection

Succinylacetone (SUAC) is the primary marker for Tyrosinemia Type 1 (Figure 1). Previously reported methods for SUAC detection require an additional extraction and derivatization process. Our work has resulted in the development of an MS/MS assay for the simultaneous extraction and measurement of amino acids, free carnitine, acylcarnitines, and succinylacetone from a single sample preparation. Including succinylacetone in the primary newborn screen along with amino acids and acylcarnitines improves sensitivity and specificity for Tyrosinemia Type-I.

Specimen Required

Dried blood spot with a minimum of 25 µL of blood on 903 filter paper.

Get the Benefits

• Fast turn-around time (24-48 hrs)
• Improved specificity for Tyrosinemia Type-I detection

For more information, please contact your PerkinElmer representative: 800-762-4000 or 203-925-4602.