SCREENING FOR SEVERE COMBINED IMMUNODEFICIENCY (SCID)

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 the laboratory differentiates PerkinElmer Genetics from others performing similar testing. They are currently regarded as a pioneer in combining tandem mass spectrometry (MS/MS), molecular technologies and biochemical analysis with tightly integrated software to ensure timely and accurate results.

Characteristics of SCID

Severe Combined Immunodeficiency (SCID) is a group of disorders characterized by the absence of both humoral and cellular immunity. The defining characteristic for SCID is always a severe defect in T-cell production and function, with defects in B-lymphocytes as a primary or secondary problem and, in some genetic types, in natural killer (NK) cell production as well.

Molecular Technologies

PerkinElmer Genetics' molecular testing lab is equipped with automated liquid handlers, which are able to do DNA extraction and set up polymerase chain reaction (PCR) reactions with a large number of samples. It also has a number of thermal cyclers for both conventional and real-time quantitative PCR reactions. The laboratory is currently using genotyping assays for primary screening of glucose-6-phosphate dehydrogenase (G6PD) deficiency and for secondary confirmation of some metabolic disorders detected by biochemical or tandem mass spectrometry assays.

Real-Time Quantitative Assay

T-cell Receptor Excision Circles (TRECs) are circular DNA fragments generated during T-cell receptor rearrangement. In healthy neonates, TRECs are made in large numbers, while in infants with SCID they are barely detectable. At PerkinElmer Genetics, real-time quantitative PCR assay is used to determine the TREC copy number in blood, which can be used to distinguish T-cell lymphopenic SCID infants from healthy babies.

Specimen Required

Dried blood spot with a minimum of 25 µL of blood on 903, 226 or other FDA cleared filter paper.

Get the Benefits

- HRSA recommended for the Primary Panel
- Fast turn-around time (72 hrs)
- Early detection
- One of the only labs where SCID screening is available

For more information, please contact your PerkinElmer representative: 866-463-6436 or 412-220-2300.

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