



## ATTACHMENT A - Delaware

### Disorders Included in the StepOne® Newborn Screening Panel

#### Disorders Detected by Other Technologies

Biotinidase Deficiency	Galactosemia**
Complete Deficiency	Galactokinase Deficiency
Partial Deficiency	Galactose-1-Phosphate Uridyltransferase Deficiency
Congenital Adrenal Hyperplasia**	Galactose-4-Epimerase Deficiency
Salt Wasting 21-Hydroxylase Deficiency	Severe Combined Immunodeficiency (SCID)
Simple Verilizing 21-Hydroxylase Deficiency	Sickle Cell & other Hemoglobinopathies
Congenital Hypothyroidism**	Hemoglobin S, S/C, S/Beta-Thalassemia, C, & E Diseases
Cystic Fibrosis (not valid after 90 days of age)*	Spinal Muscular Atrophy (SMA)

The analyses conducted by PerkinElmer Genetics produce results that can be used by qualified physicians in the diagnosis of disorders described herein. Evidence of these conditions will be detected in the vast majority of affected individuals; however, due to genetic variability, age of the patient at time of specimen collection, quality of the specimen, health status of the patient, and other variables, such conditions may not be detected in all affected patients.

**ATTACHMENT B**  
**PerkinElmer Genetics Second Tier Reflex Testing Menu**

PerkinElmer Genetics uses combinations of assays in a multi-tier approach that optimizes detection of abnormal results. Positive DNA identification for many disorders further speeds definitive diagnosis and implementation of critical therapies.

**Biochemical Second Tier Testing**

**Disorder**

**Testing Approach**

Congenital Adrenal Hyperplasia

First Tier; 17-OH P Second Tier; Extracted 17-OH P on all elevated.

Congenital Hypothyroidism

First Tier; either T4 or TSH. Second Tier TSH with a primary T4.