NEWBORN SCREENING PANEL

CLINICIANS TECHNICAL GUIDE

AGCT GENOMICS newborn screening panel (NBS) screens out following mentioned disorders based on latest technology in newborns.

Fatty Acid Oxidation disorders

Carnitine/Acylcarnitine Translocase Deficiency (Translocase)
Carnitine/Acylcarnitine Transferase Deficiency Type I1
3-Hydroxy Long Chain Acyl-CoA Dehydrogenase Deficiency (LCHAD)
2,4-Dienoyl-CoA Reductase Deficiency1
Medium Chain Acyl-CoA Dehydrogenase Deficiency (MCAD)
Multiple Acyl-CoA Dehydrogenase Deficiency (MADD or Glutaric Acidemia-Type II)
Neonatal Carnitine Palmitoyl Transferase Deficiency Type II (CPT-II)
Short Chain Acyl-CoA Dehydrogenase Deficiency (SCAD)
Short Chain Hydroxy Acyl-CoA Dehydrogenase Deficiency (SCHAD)
Trifunctional Protein Deficiency (TFP Deficiency)
Very Long Chain Acyl-CoA Dehydrogenase Deficiency (VLCAD)

Organic Acid Disorders

3-Hydroxy-3-Methylglutaryl-CoA Lyase Deficiency (HMG)
Glutaric Acidemia Type I (GA 1)
Isobutyryl-CoA Dehydrogenase Deficiency
Isovaleric Acidemia (IVA)
Acute onset
Chronic
2-Methylbutyryl-CoA Dehydrogenase Deficiency
3-Methylcrotonyl-CoA Carboxylase Deficiency (3MCC Def.)
3-Methylglutaconyl-CoA Hydratase Deficiency
Mitochondrial Acetoacetyl-CoA Thiolase Deficiency (3-Ketothiolase Def.)
Propionic Acidemia (PA)
Acute onset
Late onset
Malonic Aciduria
Methylmalonic Acidemias
Methylmalonyl-CoA Mutase Deficiency 0
Methylmalonyl-CoA Mutase Deficiency +
Some Adenosylcobalamin Synthesis Defects
Maternal Vitamin B12 Deficiency
Multiple CoA Carboxylase Deficiency

Amino Acid Disorders

Argininemia
Argininosuccinic Aciduria (ASA Lyase Deficiency)
Acute onset
Late onset
Carbamoylphosphate Synthetase Deficiency1
Citrullinemia (ASA Synthetase Deficiency)
Acute onset
Late onset
Homocystinuria
Hypermethioninemia
Hyperammonemia, Hyperornithinemia, Homocitrullinuria Syndrome (HHH) 1
Hyperornithinemia with Gyral Atrophy1
Maple Syrup Urine Disease (MSUD)
Classical MSUD
Intermediate MSUD
5-Oxoprolinuria (Pyroglutamic Aciduria)1
Phenylketonuria (PKU)
Classical PKU
Hyperphenylalaninemia
Biopterin Cofactor Deficiencies
Tyrosinemia
Transient Neonatal Tyrosinemia
Tyrosinemia Type I (Tyr I)2
Tyrosinemia Type II (Tyr II)
Tyrosinemia Type III (Tyr III)

OTHER OBSERVATIONS

Hyperalimentation
Liver Disease
Medium Chain Triglyceride Oil Administration
Presence of EDTA Anticoagulants in blood specimen
Treatment with Benzoate, Pyvalic Acid, or Valproic Acid
Carnitine Uptake Deficiency

Biotinidase Deficiency

Complete Deficiency
Partial Deficiency

Congenital Adrenal Hyperplasia

Salt Wasting 21-Hydroxylase Deficiency
Simple Verilizing 21-Hydroxylase Deficiency

Congenital Hypothyroidism

Cystic Fibrosis (not valid after 90 days of age)*

Galactosemia

Galactokinase Deficiency
Galactose-1-Phosphate Uridyltransferase Deficiency
Galactose-4-Epimerase Deficiency

Glucose-6 Phosphate Dehydrogenase Deficiency

Sickle Cell & other Hemoglobinopathies

Hemoglobin S, S/C, S/Beta-Thalassemia, C, & E Diseases

The analyses conducted by AGCT GENOMICS 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

***For information on DNA Carrier Testing for children over 90 days of age, please call 92-313-1513151**