Newborn Screening Program Office: ..... (404) 657-4143

Georgia Public Health Laboratory: ..... (404) 327-7950

Voice Response Registration: ..... (404) 327-6800

Screening cards/envelopes: ..... (404) 327-7920/7921

Management and Treatment Resources

Emory University Division of Medical Genetics Newborn Screening Follow-up Program: ..... (404) 727-0486/0488

Metabolic Nutritionist: ..... (404) 727-2866

Geneticist-on-Call: ..... (404) 701-0532

Medical College of Georgia, Pediatric Sickle Cell Clinic

NBS Program Coordinator: ..... (706) 721-6251

On-call MD: ..... (706) 721-3893

GA Comprehensive Sickle Cell Center/Grady Health System

NBS Program Coordinator: ..... (404) 616-5990

On-Call MD: ..... (404) 616-3572

Sickle Cell Foundation of Georgia. ..... (404) 755-1641

or (800) 326-5287 (tollfree)

Gerogia Department of Human Resources
Division of Public Health
Two Peachtree Street, NW, 11th Floor
Atlanta, Georgia 30303-3142

http://health.state.ga.us/programs/nsmscd/

www.marchofdimesga.com/

Georgia
Newborn Screening
Program

The Newborn Screening Program is a legislatively mandated program. Its mission is to provide early detection of inherited genetic disorders for the prevention of morbidity and mortality attributed to these disorders.

Newborn Screening is an essential, preventive public health function to identify at risk infants in the first few days of life so that early intervention can be implemented to prevent severe mental retardation, chronic disability or death.

The cost of these disorders when left untreated is enormous, both in human suffering and in economic terms. Georgia law (O.C.G.A. 31-12-6 and O.C.G.A. 31-12-7) directs that a statewide network for genetic services be developed as a cooperative effort between public health, appropriate medical centers and private practitioners.

Physician Pocket Reference

March of Dimes

DHR

DISORDER/INCIDENCE	DEFECT	INDICATOR	SYMPTOMS	TREATMENT
Biotinidase Deficiency 1:60,000 (general population)	Enzyme deficiency, unable to recycle and produce free biotin	Absent or decreased biotinidase activity	Seizures, hypotonia, apnea, skin rash/infection, developmental delay, alopecia, deafness, blindness, metabolic acidosis, coma, death	Pharmacological doses of oral biotin (5-20 mg/day).
Congenital Adrenal Hyperplasia (CAH) 1:19,000 (GA)	Deficiency of 21-hydroxylase, unable to make cortisol and aldosterone. Many variant forms exist.	Elevated 17-hydroxyproges-terone	Abnormal electrolytes: low sodium, elevated potassium, hypoglycemia, dehydration, cardiac arrhythmia, death, ambiguous genitalia in females, progressive virilization in both sexes	Replace cortisol and aldosterone, salt supplementation in some, surgical correction for females
Galactosemia 1:40,000 (GA)	Deficiency of Galactose-1-Phosphate Uridyl Trasnferase (GALT), unable to convert galactose to glucose. Many variant forms exist.	Absent or reduced GALT enzyme activity. Elevated total galactose metabolites	Neonatal jaundice, vomiting, lethargy, diarrhea, liver damage, death from E.Coli sepsis, cataracts development delay, hepatomegaly, Fanconi's syndrome, growth failure	Eliminate galactose and lactose from the diet. Soy formulas in infancy. Lactose and galactose free solid foods and medications.
Hemoglobinopathies 1:1,300 (GA)	Abnormal hemoglobin production	Abnormal hemoglobin fractions	Hemolytic anemia, strokes, splenic sequestration, recurrent pain episodes, acute chest syndrome, life-threatening infections, hand-foot syndrome, tissue infarction/organ damage and failure	Prophylactic penicillin, immunizations, transfusions
Homocystinuria 1:350,000 (GA)	Deficiency of cystathionine synthetase, unable to metabolize methionine and homocystine	Elevated methionine	Mental retardation, seizures, behavior disorder, thromboses, dislocated lenses, tall/thin stature	Life long low methionine diet with cystine supplementation. Pyridoxine (Vitamin B6) supplementation also if responsive. Special formulas and "medical" foods.
Hypothyroidism 1:4,000 (GA)	Absent, hypoplastic, or dysfunctional thyroid gland. About 20% are genetic in origin	Low or normal thyroxine (T4) with elevated thyroid stimulating hormone (TSH)	Prolonged neonatal jaundice, poor muscle tone, constipation, lethargy, feeding problems, large tongue, dry and mottled skin, distended abdomen, umbilical hernia	Thyroid hormone replacement (L-Thyroxine)
Maple Syrup Urine Disease (MSUD) 1:130,000 (GA)	Deficiency of the BCKD enzyme complex, unable to metabolize the branched chain amino acids (leucine, isoleucine, and valine)	Elevated branched chain amino acids	Acidosis, hypertonia, seizures, vomiting, apnea, coma, severe mental retardation, neurological impairment, death. Ear wax/urine smells like maple syrup	Life long diet low in leucine, isoleucine, and valine. Thiamine (Vitamin B1) supplementation if responsive. Special formulas and "medical" foods
Medium Chain Acyl-CoA Dehydrogenase (MCAD) Deficiency 1:15,000 (US)	Enzyme deficiency, unable to metabolize fat for energy in the absence of glucose	Abnormal medium chain acylcarnitines	Hypoglycemia, hyperammonemia, vomiting, lethargy, coma, apnea, cardiac arrest, sudden unexplained death	Regular feedings to avoid fasting, low fat diet, oral L-carnitine supplementation
Phenylketonuira (PKU) 1:17,000 (GA)	Deficiency of phenylalanine hydroxylase, unable to convert phenylalanine to tyrosine	Elevated phenylalanine	Severe mental retardation, eczema, seizures, decreased pigmentation, behavior disorder, "mousey" odor	Life long low phenylalanine diet, tyrosine supplementation. Special formulas and "medical" foods.
Tyrosinemia 1:400,000 (GA)	Deficiency of Fumarylacetoacetate hydrolase (FAH), unable to metabolize tyrosine. Other variant forms exist.	Elevated Tyrosine	Hepatic damage, liver cirrhosis and failure, Fanconi syndrome, growth failure, hepatomegaly, hepatic carcinoma, thrombocytopenia.	Life long low phenylalanine and tyrosine diet. Special formulas and "medical" foods.