Disorders Screened in New Jersey

In accordance with Chapter 24 of the Public Laws of 1988 (N.J.S.A. 26:2-110 and N.J.S.A. 26:2-111, New Jersey Administrative Code, Title 8, Department of Health, Chapter 18, Newborn Biochemical Screening Program), New Jersey has expanded its statewide system of newborn biochemical testing to include a total of 54 disorders, which, if not detected early, can cause severe health problems, interfere with mental development or even cause death.

New Jersey Newborn Biochemical Screening Program Disorders	
Disorder Name	**Abbreviation**
Fatty Acid Oxidation Disorders	
2,4 Dienoyl-CoA reductase deficiency	DE RED
Carnitine palmitoyltransferase I deficiency	CPT IA
Carnitine palmitoyltransferase II deficiency	CPT II
Carnitine acylcarnitine translocase deficiency	CACT
Carnitine uptake defect/Carnitine transport defect	CUD
Glutaric acidemia type II	GA II
Long-chain L-3 hydroxyacyl-CoA dehydrogenase deficiency	LCHAD
Long-chain acyl-CoA dehydrogenase deficiency	LCAD
Medium/Short-chain L-3 hydroxyacyl-CoA dehydrogenase deficiency	M/SCHAD
Medium-chain acyl-CoA dehydrogenase deficiency	MCAD
Medium chain ketoacyl-CoA thiolase deficiency	MCAT
Short-chain acyl-CoA dehydrogenase deficiency	SCAD
Trifunctional protein deficiency	TFP
Very long-chain acyl-CoA dehydrogenase deficiency	VLCAD
Organic acidemia Disorders	
2-Methyl-3-hydroxybutyric aciduria	2M3HBA
2-Methylbutyrylglycinuria	2MBG
3-Hydroxy-3-methylglutaric aciduia	HMG
3-Methylcrotonyl-CoA carboxylase deficiency	3MCC
3-Methylglutaconic aciduria	3MGA
beta-Ketothiolase deficiency	BKT
Glutaric acidemia type I	GA I
Isobutyrylglycinuria	IBG
Isovaleric acidemia	IVA
Malonic acidemia	MAL
Methylmalonic acidemia - Cobalamin A, B Disorders	CBL A,B

Methylmalonic academia with homocystinuria - Cobalamin C, D Disorders	CBL C,D
Methylmalonic acidemia – Methylmalonyl-CoA Mutase	MUT
Holocarboxylase synthase deficiency	MCD
Propionic acidemia	PROP
Amino Acid & Urea Cycle Disorders	
Argininemia	ARG
Argininosuccinic aciduria	ASA
Benign hyperphenylalaninemia	H-PHE
Biopterin defect of cofactor biosynthesis	BIOPT (BS)
Biopterin defect of cofactor regeneration	BIOPT (REG)
Citrullinemia, type I	CIT I
Citrullinemia, type II	CIT II
Homocystinuria	HCY
Hypermethioninemia	MET
Maple syrup urine disease	MSUD
Classic phenylketonuria	PKU
Tyrosinemia, type I	TYR I
Tyrosinemia, type II	TYR II
Tyrosinemia, type III	TYR III
Endocrine Disorders	
Congenital adrenal hyperplasia	CAH
Primary congenital hypothyroidism	CH
Metabolic Disorders	
Biotinidase deficiency	BIOT
Classic galactosemia	GALT
Galactoepimerase deficiency	GALE
Galactokinase deficiency	GALK
Other Disorders	
Cystic fibrosis	CF
Hemoglobin S/Beta-thalassemia	Hb S/β Thal
Hemoglobin S/C disease	Hb S/C
Other Hemoglobinopathies	Var Hb
Hemoglobin S/S Disease (Sickle cell anemia)	Hb S/S

Babies with abnormal screening results are aggressively followed by the Newborn Screening and Genetic Services Program in Special Child Health and Early Intervention Services to ensure that affected children and their families are linked with a primary care provider and the regional network of specialty care centers to receive timely and appropriate services.

Early identification of all disorders screened in the newborn period is only the first step in a successful newborn screening program. Additional resources and funding to ensure immediate access to confirmatory testing, follow-up, and assurance of appropriate and comprehensive treatment services are also required. Consultants have been identified to provide comprehensive services for the various disorders and state funding has been committed to provide a statewide safety net of pediatric metabolic treatment centers, cystic fibrosis care centers, sickle cell programs, endocrine specialists and regional biochemical genetics laboratories.

Additionally, affected children must be reported to the Special Child Health Services Registry which will further link the family to community-based, culturally competent, comprehensive case management and early intervention services.

If you have any questions or need additional information, please contact:

Lorraine Freed Garg, MD, MPH
E-mail: Lori.Garg@doh.state.nj.us
Medical Director
New Jersey Department of Health
Special Child Health and Early Intervention Services
Newborn Screening and Genetic Services Program
P.O. Box 364
Trenton, New Jersey 08625-0364
phone # (609) 984-0755

Scott M. Shone, Ph.D
E-mail: scott.shone@doh.state.nj.us
Research Scientist 1/Program Manager
New Jersey Department of Health
Newborn Screening Laboratory
Division of Public Health Infrastructure, Laboratories and Emergency Preparedness
P.O. Box 371
Trenton, New Jersey 08625-0371
phone # (609) 530-8361

More information for parents can be found on the following web sites:
www.babysfirsttest.org
www.newbornscreening.info/Parents/facts.html

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