

Clinical Testing Newborn Screening

Tests performed in the Newborn Screening Section detect biochemical abnormalities associated with inherited metabolic disorders. These diseases are not the result of an outside causative agent or vector, but are, instead, the direct results of genetic or congenital deficiencies or variations. There is a charge for this service.

Specific Requirements: The specimens are dried blood spots (DBS) obtained from neonates who are at least 24 hours old. The blood must be collected directly onto the filter paper circles on the Illinois Department of Public Health Neonatal Screening Test Form, using heel pricks. Specimen collection instructions are located on the reverse side of the Illinois Department of Public Health's Neonatal Screening Test Form.

Information Required: Completed State of Illinois specified Neonatal Screening Test Form.

Criteria for Rejection: Specimens submitted on unapproved forms will be rejected.

See the Neonatal Screening Test Form for additional rejection criteria.

Link to Newborn Screening for Genetic/Metabolic Disorders at: <http://www.idph.state.il.us/HealthWellness/genetics.htm>

Table 13. Newborn Screening

Disease or Condition	Substance Assayed	Analytical Method
Biotinidase Deficiency	Biotinidase	Colorimetric Assay
Congenital Adrenal Hyperplasia	17- α -Hydroxyprogesterone	Fluoroimmunoassay
Galactosemia	Total Galactose and Galactose-1-Phosphate (GAL)	Continuous Flow Assay
	Galactose-1-Phosphate Uridyl Transferase (GALT)	
Congenital Hypothyroidism	Screen: Thyrotropin (TSH)	Fluoroimmunoassay
	2 nd Tier: Thyroxin (T4)	Fluoroimmunoassay
Amino Acid Disorders ¹	Amino Acids	Tandem Mass Spectrometry
Urea Cycle Disorders ¹	Amino Acids	Tandem Mass Spectrometry
Fatty Acid Oxidation Disorders ¹	Acylcarnitines	Tandem Mass Spectrometry
Organic Acid Disorders ¹	Acylcarnitines	Tandem Mass Spectrometry
Sickle Cell Disease and other Hemoglobinopathies	Abnormal Hemoglobins	High Performance Liquid Chromatography
Cystic Fibrosis	Screen: Immunoreactive Trypsin (IRT)	Fluoroimmunoassay
	2 nd Tier: DNA	Detection of Mutations in the CF Transmembrane Conductance Regulator Gene (CFTR) via Polymerase Chain Reaction (PCR) and Capillary Electrophoresis.
Abnormal findings are reported within one to four days, depending on condition. ¹ Details of Tandem Mass Spectrometry testing in Table 14.		

Performed at the Chicago Laboratory

LIMITATION OF TESTING: The purpose of the Newborn Screening Program in Illinois is to identify infants at risk for certain congenital conditions and in need of more definitive testing. Abnormal results always require medical evaluation. Results can be affected by: age at time of collection, feeding status, prematurity, low birth weight, transfusion, TPN, illness, medications, and collection and handling techniques. **As with any laboratory test false negative and false positive results are possible.** Newborn screening test results are insufficient information on which to base diagnosis or treatment.

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Table 14.IDPH – List of Disorders Detectable by Tandem Mass Spectrometry (MS/MS) ²

Type of Disorder	Name of Disorder	Abbreviation	Analyte (microMoles/L)	Comments
<i>Amino Acid</i>	Phenylketonuria	PKU	Phenylalanine	
	Maple syrup urine disease	MSUD	leucine/isoleucine and valine	
	Tyrosinemia type I and possibly type II	TYRO	tyrosine	Type I may not be detected in neonatal specimen.
	Homocystinuria	HCU	methionine	
	5-Oxoprolinuria	5OXP	5-oxoproline	
<i>Urea Cycle</i>	Citrullinemia	CIT	citrulline	CIT/ASA
	Argininosuccinic aciduria	ASA	citrulline	CIT/ASA
	Argininemia	ARG	arginine	ARG
<i>Organic Acid</i>	3-methylcrotonyl-CoA carboxylase deficiency	3MCC	C5-OH	3HMG/3MCC/3HGA/MCD
	3-hydroxy-3-methylglutaric-CoA lyase deficiency	3HMG	C5-OH	3HMG/3MCC/3 HGAJMCD
	3-methylglutaconic aciduria	3MGA	C5-OH	3HMG/3MCC/3HGAIMCD
	Multiple carboxylase deficiency	MCD	C5-OH	3FIMG/3MCC/3HGA/MCD
	2-methyl-butryryl-CoA dehydrogenase deficiency	2MBCD	C5	2MBCD/IVA
	Isovaleric acidemia	IVA	C5	2MBCD/IVA
	Methylmalonic acidemia	MMA	C3	PA/MMA
	Malonic aciduria	MA	C3-DC	
	Propionic acidemia	PA	C3	PA/MMA
	Beta-ketothiolase deficiency	BKT	C5:1, C5-OH	
Glutaric aciduria type I	GAI	C5-DC		
<i>Fatty Acid Oxidation</i>	Short chain acyl-CoA dehydrogenase	SCAD	C4	SCAD/IBCD
	Isobutyryl-CoA dehydrogenase deficiency	IBCD	C4	SCAD/IBCD
	Medium chain acyl-CoA dehydrogenase deficiency	MCAD	C6, C8, C10:1	
	Very long chain acyl-CoA dehydrogenase	VLCAD	C14:1, C14, C16	VLCAD
	Carnitine palmitoyl transferase deficiency type II	CPTII	C16, C18:1, C18	CACT/CPTII
	Trifunctional protein deficiency	TFPD	C16-OH, C18:1- OH	TFPD/LCHAD
	Long chain 3-hydroxy Acyl-CoA dehydrogenase	LCHAD	C16-OH, C18:1-OH	TFPD/LCHAD
	Glutaric Aciduria type II or multiple acyl-CoA dehydrogenase deficiency	GAI	C4, C5, C8:1, C8, C12, C14, C16, C5-DC	
	Carnitine uptake (transport) deficiency	CUD	C0 (low)	
	Carnitine/acylcarnitine translocase deficiency	CACT	C16, C18:1, C18	CACT/CPTII

² In some cases, additional metabolic disorders, not listed, may be detected