Wisconsin State Laboratory of Hygiene
UNIVERSITY OF WISCONSIN–MADISON

Newborn Screening Laboratory
465 Henry Mall
Madison, WI 53706-1578
Ph: 608-262-6547 Fax: 608-262-5494

Laboratory Report

James J. Schauer, Ph.D., PE, MBA, Director
Errin C. Rider, Ph.D., D(ABMM),M(ASCP)CM,
Director of Clinical Laboratory Services

Submitted By: REPORT SUBMITTER NBS [2130]
Ordered By: TEST, DOCTOR

Report Date:
6/7/2022

Newborn Screening Report Submitter
123 Street Address
City WI 53706

ATTENTION:
Recommendation may be included

SUBMITTER PROVIDED INFORMATION

Name	Sex	Birth	Gest. Age (w)	Birthwt. (g)
NBS, TEST BABY 1234567890	Female	5/2/2022 0600	40	3000

Birth Facility	Mother or Guardian	Primary Care Provider
NOT PROVIDED	NBS,TEST MOM	TEST, DOCTOR

Specimen Details

	Type	Collected	Received
22NB000255	Dried Blood Spot	5/4/2022 0700	5/9/2022 0929
	Repeat Specimen	**Age at Collection**	**NBS Card No.**
		49 hrs	U123456

NEWBORN SCREENING SUMMARY (Final result)

	Result	Reference Value(s)
Aminoacidopathies	Screen negative	
Fatty Acid Oxidation (FAO) Disorders	Screen negative	
Organic Acidemias (OA)	Screen negative	
Biotinidase Deficiency	Screen negative	
Congenital Hypothyroidism	Screen negative	
Hemoglobinopathies	Screen negative	
Galactosemia	**SCREEN POSITIVE**	
Congenital Adrenal Hyperplasia (CAH)	Screen negative	
Cystic Fibrosis (CF)	Screen negative	
Severe Combined Immune Deficiency (SCID)	Screen negative	
Spinal Muscular Atrophy (SMA)	Screen negative	
Pompe Disease	Screen negative	

Comments:
A screen negative result indicates a low risk for the associated screened condition, but the risk is not zero. A screening test result should not be used for diagnosis. When clinical symptoms are indicated, appropriate diagnostic testing should be arranged regardless of NBS test results.

The screening tests performed on this specimen were intended for newborns. Interpretations are based on birthweight, gestational age, and age at collection. Interferences such as transfusion, parenteral nutrition/supplementation, prenatal steroid exposure, and antibiotic therapies may affect the screening test results.

The screening tests for aminoacidopathies, fatty acid oxidation disorders, organic acidemias, biotinidase deficiency, severe combined immune deficiency, spinal muscular atrophy, and the analysis of some included CFTR gene variants were developed and performance characteristics determined by WSLH. These tests have not been cleared or approved

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	Result	Reference Value(s)

by the FDA. The laboratory is regulated under CLIA as qualified to perform high-complexity testing. These tests are used for clinical purposes and should not be regarded as investigational or for research.

For details on the conditions screened, biomarkers, test methods, and reference values, refer to the WSLH website at www.slh.wisc.edu/newborn.

Galactosemia Screen (Final result)

	Result	Reference Value(s)
Galactose-1-Phosphate Uridyl Transferase (GALT)	**2.0**	>3.0 U/g Hb
Galactosemia	**SCREEN POSITIVE**	

Comments:
INTERPRETATION: The absence of galactose-1-phosphate uridyl transferase (GALT) enzyme activity indicates this child is at high risk for Galactosemia.

RECOMMENDATION: PERFORM CONFIRMATORY TESTING.
A medical geneticist from one of the following facilities will contact you to discuss patient care and appropriate confirmatory testing.

Children's Hospital of Wisconsin, Milwaukee; 414-266-2471 (Physician Referral Services)
University of Wisconsin American Family Children's Hospital, Madison; 608-263-3260 (Physician Access Center)

Condition(s) Screened	Method(s)	Biomarker(s)	Reference Value(s)
Aminoacidopathies	1st Tier: MS/MS 2nd Tier: LC-MS/MS	Amino acids	See: www.slh.wisc.edu/newborn
Fatty Acid Oxidation (FAO) Disorders	1st Tier: MS/MS 2nd Tier: LC-MS/MS	Acylcarnitine profile	See: www.slh.wisc.edu/newborn
Organic Acidemias (OA)	1st Tier: MS/MS 2nd Tier: LC-MS/MS	1st Tier: Acylcarnitine profile 2nd Tier: Organic acids	See: www.slh.wisc.edu/newborn
Galactosemia	1st Tier: Enzyme assay 2nd Tier: Enzyme assay	1st Tier: Galactose-1-phosphate uridyl-1-transferase (GALT) activity 2nd Tier: Total galactose	1st Tier: >3.0 U/g Hb 2nd Tier: <6.0 mg/dL
Biotindase Deficiency	Enzyme assay	Biotinidase activity	Present
Congenital Hypothyroidism (CH)	Immunoassay	Thyroid Stimulating Hormone (TSH)	0-96h: <50 uIU/mL 97-312h: <17 uIU/mL ≥313h: <15 uIU/mL
Congenital Adrenal Hyperplasia (CAH)	1st Tier: Immunoassay 2nd Tier: LC-MS/MS	1st Tier: 17-Hydroxyprogesterone (17-OHP) 2nd Tier: steroid profile	Age- and birthweight-dependent (See: www.slh.wisc.edu/newborn)
Hemoglobinopathies	1st Tier: IEF 2nd Tier: HPLC	Hemoglobin (Hgb) fractions	Presence of fetal and adult Hgb
Cystic Fibrosis (CF)	1st Tier. Immunoassay 2nd Tier. Next Generation Sequencing	1st Tier: Immunoreactive Trypsinogen (IRT) 2nd Tier: *CFTR* gene variants	1st Tier: <170 ng/mL 2nd Tier: None detected
Severe Combined Immune Deficiency (SCID)	Real-Time PCR	T-cell Receptor Excision Circles (TREC)	≤1.079 MoM
Spinal Muscular Atrophy (SMA)	Real-Time PCR	Functional *SMN1* exon 7	Present
Pompe Disease	Enzyme assay and MS/MS	Acid alpha glucosidase (GAA) activity	>15% of the daily median

END OF REPORT