THE UNIVERSITY OF IOWA | **STATE HYGIENIC LABORATORY**

NEWBORN SCREENING REPORT

319/384-5097 or 866/890-5965

Patient	Birth Date and Gender	Client Reference	Accession #
LAST42098567, FIRST42098567 198 33RD AVE SW ANYTOWN, IA 52999	2025-05-06 08:18 Female	42098567	78458
	Collected 2025-05-07 10:19	**Received** 2025-05-07 22:30	**Project**

Report To

ANYTOWN HOSPITAL
SEND-OUT LAB
1000 MEDICAL ST
ANYTOWN, IA 52999

Ordering Health Care Provider
WELBY, MARCUS

Primary Care Provider
WELBY, MARCUS

Sample Type
Blood spot specimen

Sample Note(s)

Screen	Birth Order	IA Barcode Number	Transfusion Interference	Weight at Collection	Guardian
Initial		IA7098422	No	2716 grams	MOMLAST42098567, MOMFIRST42098567

RESULTS OF ANALYSIS - FINAL REPORT

TEST	RESULT	ANALYSIS NOTE(S)
Congenital Adrenal Hyperplasia, Immunoassay		
Congenital adrenal hyperplasia interpretation	**Within Normal Limits**	
Congenital Hypothyroidism, Immunoassay		
Congenital hypothyroidism interpretation	**Within Normal Limits**	
Biotinidase Deficiency, Immunoassay		
Biotinidase deficiency interpretation	**Within Normal Limits**	
Galactosemia, Enzymatic Assay		
Galactosemia interpretation	**Within Normal Limits**	
Hemoglobinopathies, Various Methods		*1*
Hemoglobin disorders interpretation	**Within Normal Limits**	
Cystic Fibrosis, Various Methods		*2*
Cystic fibrosis interpretation	**Within Normal Limits**	
Expanded Screening Disorders, Tandem Mass Spectrometry		*3*
Fatty acid oxidation defects interpretation	**Within Normal Limits**	
Organic acidemias interpretation	**Within Normal Limits**	
Amino acidemias interpretation	**Within Normal Limits**	
Lysosomal Storage Disorders, Tandem Mass Spectrometry		*4*
Pompe disease interpretation	**Presumptive Positive - Program staff will contact with recommendations**	
Mucopolysaccharidosis Type 1 (MPS1) disease interpretation	**Presumptive Positive - See second tier test result**	
Mucopolysaccharidosis Type II (MPSII) disease interpretation	**Presumptive Positive - See second tier test result**	
Pompe Disease 2nd Tier NBS, Flow Injection Analysis-Tandem Mass Spectrometry		*5*
Pompe Disease 2nd Tier NBS interpretation	**Positive - see second tier results report attached to the sample in the OpenELIS Web Portal**	
Mucopolysaccharidosis (MPS) 2nd Tier NBS, LC-MS/MS		*6*
Mucopolysaccharidosis (MPS) 2nd Tier NBS interpretation	**Negative - see second tier results report attached to the sample in the OpenELIS Web Portal**	
X-linked Adrenoleukodystrophy, Tandem Mass Spectrometry		*7*
X-linked Adrenoleukodystrophy disease interpretation	**Within Normal Limits**	
Severe Combined Immunodeficiency, Real-Time PCR		*8*
Severe Combined Immunodeficiency (SCID) interpretation	**Within Normal Limits**	
Spinal Muscular Atrophy, Real-Time PCR		*9*
Spinal Muscular Atrophy (SMA) interpretation	**Within Normal Limits**	

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SAMPLE AND ANALYSIS NOTES

1. Core conditions screened: Sickle cell disease, Hemoglobin S/C disease, Hemoglobin S beta-thalassemia

2. Cystic Fibrosis Transmembrane Conductance Regulator (CFTR) gene variant testing was NOT performed on this sample because the Immunoreactive Trypsinogen (IRT) value was in the normal range.

3. Core conditions screened: Fatty acid oxidation defects: Carnitine uptake defect (Carnitine transport defect), Long-chain 3-hydroxy acyl-CoA dehydrogenase deficiency, Medium chain acyl-CoA dehydrogenase deficiency, Trifunctional protein deficiency, Very long-chain acyl-CoA dehydrogenase deficiency; Organic acidemias: Glutaric acidemia type I, 3-Hydroxy 3-methylglutaric aciduria, Isovaleric acidemia, 3-Methylcrotonyl-CoA carboxylase deficiency, Methylmalonic acidemia (methylmalonyl-CoA mutase, cobalamin disorders, vitamin B12 disorders), Beta-Ketothiolase deficiency, Propionic acidemia, Holocarboxylase synthetase deficiency; Amino acidemias: Argininosuccinic aciduria, Citrullinemia type 1, Homocystinuria, Maple Syrup Urine Disease, Classic Phenylketonuria, Tyrosinemia type I. The performance characteristics of this test were determined by the State Hygienic Laboratory. It has not been cleared or approved by the U.S. Food and Drug Administration.

4. Core conditions screened: Lysosomal Storage Disorders: Pompe disease, Mucopolysaccharidosis Type 1 (MPS1) disease, Mucopolysaccharidosis Type II (MPSII) disease. A Presumptive Positive interpretation for Pompe, MPS1, or MPSII will automatically reflex to second tier testing if a suitable sample is still available at SHL. A Borderline interpretation for Pompe will automatically reflex to second tier testing if a suitable sample is still available at SHL. If not, program staff will contact with recommendations for second tier testing. A second tier test interpretation will take precedence over the first tier screening interpretation. The performance characteristics of this test were determined by the State Hygienic Laboratory. It has not been cleared or approved by the U.S. Food and Drug Administration.

5. The "Pompe Disease 2nd Tier NBS interpretation" will take precedence over the "Pompe disease interpretation". This test was developed and its performance characteristics determined by Mayo Clinic Laboratories, 200 First Street SW, Rochester, MN 55905 (CLIA Certificate: 24D0404292) in a manner consistent with CLIA requirements. This test has not been cleared or approved by the U.S. Food and Drug Administration.

6. The "Mucopolysaccharidosis (MPS) 2nd Tier NBS interpretation" will take precedence over the "Mucopolysaccharidosis Type 1 (MPS1) disease interpretation" or the "Mucopolysaccharidosis Type II (MPSII) disease interpretation". This test was developed and its performance characteristics determined by Mayo Clinic Laboratories, 200 First Street SW, Rochester, MN 55905 (CLIA Certificate: 24D0404292) in a manner consistent with CLIA requirements. This test has not been cleared or approved by the U.S. Food and Drug Administration.

7. Core condition screened: X-linked Adrenoleukodystrophy. The performance characteristics of this test were determined by the State Hygienic Laboratory. It has not been cleared or approved by the U.S. Food and Drug Administration.

8. The performance characteristics of this test were determined by the State Hygienic Laboratory. It has not been cleared or approved by the U.S. Food and Drug Administration.

9. Spinal Muscular Atrophy (SMA) screening uses a deletion in exon 7 of the survival motor neuron 1 (SMN1) gene to assess risk. 95% of SMA cases are caused by deletion of SMN1 exon 7. Thus, the screening assay may miss up to 5% of SMA cases that are not caused by SMN1 exon 7 deletion.

ANALYSIS INFORMATION

TEST	ANALYZED	SITE	RELEASED
1. Congenital Adrenal Hyperplasia, Immunoassay	2025-05-08 06:00 MBH	10320	2025-05-13 17:02 MBH
2. Congenital Hypothyroidism, Immunoassay	2025-05-08 06:00 MBH	10320	2025-05-13 17:02 MBH
3. Biotinidase Deficiency, Immunoassay	2025-05-08 06:00 MBH	10320	2025-05-13 17:02 MBH
4. Galactosemia, Enzymatic Assay	2025-05-08 06:00 MBH	10320	2025-05-13 17:02 MBH
5. Hemoglobinopathies, Various Methods	2025-05-08 06:00 MBH	10320	2025-05-13 17:02 MBH
6. Cystic Fibrosis, Various Methods	2025-05-08 06:00 MBH	10320	2025-05-13 17:02 MBH
7. Expanded Screening Disorders, Tandem Mass Spectrometry	2025-05-08 06:00 MBH	10320	2025-05-13 17:02 MBH
8. Lysosomal Storage Disorders, Tandem Mass Spectrometry	2025-05-08 06:00 MBH	10320	2025-05-13 17:02 MBH

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TEST	ANALYZED	SITE	RELEASED
9. Pompe Disease 2nd Tier NBS, Flow Injection Analysis-Tandem Mass Spectrometry	2025-05-09 10:30 MBH	10415	2025-05-13 17:02 MBH
10. Mucopolysaccharidosis (MPS) 2nd Tier NBS, LC-MS/MS	2025-05-09 10:30 MBH	10415	2025-05-13 17:02 MBH
11. X-linked Adrenoleukodystrophy, Tandem Mass Spectrometry	2025-05-08 06:00 MBH	10320	2025-05-13 17:02 MBH
12. Severe Combined Immunodeficiency, Real-Time PCR	2025-05-08 06:00 MBH	10320	2025-05-13 17:02 MBH
13. Spinal Muscular Atrophy, Real-Time PCR	2025-05-08 06:00 MBH	10320	2025-05-13 17:02 MBH

SITE(S) PERFORMING TESTING

10320 STATE HYGIENIC LABORATORY ANKENY, IOWA LABORATORIES COMPLEX, 2220 S ANKENY BLVD, ANKENY, IA 50023; Phone 515/725-1630; Fax 515/725-1650; Michael A. Pentella, Ph.D., D(ABMM), Director; CLIA ID Number 16D0709302

10415 MAYO CLINIC LABORATORIES, ROCHESTER MAIN CAMPUS, 200 FIRST STREET SW, ROCHESTER, MN 55905; CLIA Certificate: 24D0404292

For questions about resubmission and result interpretation please contact the medical consultants. During regular work hours (8:00 AM to 4:30 PM): 319/384-5097 or toll free in Iowa 866/890-5965. 24hr Geneticist on-call number: 319/356-1616. This is a screening test and not indicated for stand-alone purposes; results should be used in conjunction with other available laboratory and clinical information. A false negative or a false positive result must always be considered when screening; therefore, clinical findings and status should be considered whenever interpreting laboratory results. Newborn reference values may not be applicable to older infants, thus screening results should be interpreted with caution in such cases. Information on the conditions screened is available at https://hhs.iowa.gov/programs/programs-and-services/family-health/congenital-inherited-disorders/iowa-newborn-screening-program/iowa-newborn-screening-education/blood-spot-screening-information . The result(s) of this report relate only to the items analyzed. Where the laboratory has not been responsible for the sampling stage the results apply only to the sample as received. This report shall not be reproduced except in full without the written approval of the laboratory.