

Patient LAST5230975, FIRST5230975			Birth Date and Gender 2025-05-15 06:34 Male		Client Reference 5230975		Accession # 78466	
219 17TH ST ANYTOWN, SD 57999			Collected 2025-05-16 07:15		Received 2025-05-17 02:00		Project	
Report To  ANYTOWN HOSPITAL SEND-OUT LAB 1000 MEDICAL ST ANYTOWN, SD 57999							Ordering Health Care Provider WELBY, MARCUS	
							Primary Care Provider WELBY, MARCUS	
							Sample Type Blood spot specimen	
							Sample Note(s)	
Screen Initial	Birth Order	IA Barcode Number IA8906235		Transfusion Interference No	Weight at Collection 3107 grams		Guardian MOMLAST5230975, MOMFIRST5230975	

## RESULTS OF ANALYSIS - FINAL REPORT

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

## SAMPLE AND ANALYSIS NOTES

- Core conditions screened: Sickie cell disease, Hemoglobin S/C disease, Hemoglobin S beta-thalassemia
- Cystic Fibrosis Transmembrane Conductance Regulator (CFTR) gene variant testing was NOT performed on this sample

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because the Immunoreactive Trypsinogen (IRT) value was in the normal range.

- 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.
- Core conditions screened: Lysosomal Storage Disorders: Pompe disease. A Presumptive Positive or Borderline interpretation 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.
- 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.
- 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.
- 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.
- 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	2025-05-17 06:00 MBH	10320	2025-05-21 13:50 MBH
2. Congenital Hypothyroidism, Immunoassay	2025-05-17 06:00 MBH	10320	2025-05-21 13:50 MBH
3. Biotinidase Deficiency, Immunoassay	2025-05-17 06:00 MBH	10320	2025-05-21 13:50 MBH
4. Galactosemia, Enzymatic Assay	2025-05-17 06:00 MBH	10320	2025-05-21 13:50 MBH
5. Hemoglobinopathies, Various Methods	2025-05-17 06:00 MBH	10320	2025-05-21 13:50 MBH
6. Cystic Fibrosis, Various Methods	2025-05-17 06:00 MBH	10320	2025-05-21 13:50 MBH
7. Expanded Screening Disorders, Tandem Mass Spectrometry	2025-05-17 06:00 MBH	10320	2025-05-21 13:50 MBH
8. Lysosomal Storage Disorders, Tandem Mass Spectrometry	2025-05-17 06:00 MBH	10320	2025-05-21 13:50 MBH
9. Pompe Disease 2nd Tier NBS, Flow Injection Analysis-Tandem Mass Spectrometry	2025-05-21 13:47 MBH	10415	2025-05-21 13:50 MBH
10. X-linked Adrenoleukodystrophy, Tandem Mass Spectrometry	2025-05-17 06:00 MBH	10320	2025-05-21 13:50 MBH
11. Severe Combined Immunodeficiency, Real-Time PCR	2025-05-17 06:00 MBH	10320	2025-05-21 13:50 MBH
12. Spinal Muscular Atrophy, Real-Time PCR	2025-05-17 06:00 MBH	10320	2025-05-21 13:50 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



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For questions about resubmission, results, referrals, and newborn screening procedures, contact the South Dakota Newborn Screening Program at 605/773-3361. 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. Disorder information is available at <https://doh.sd.gov/programs/newborn-screening/blood-spot/disorders/?pvs=21> . 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.