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

**NEWBORN SCREENING REPORT**

**605/773-3361**

| Patient | Birth Date and Gender | Client Reference | Accession # |
|---|---|---|---|
| LAST6274513, FIRST6274513<br><br>218 19TH AVE<br>ANYTOWN, SD 57999 | 2025-05-15 02:18<br>Male | 6274513 | 78468 |
| | **Collected**<br>2025-05-16 03:29 | **Received**<br>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 | Birth Order | IA Barcode Number | Transfusion Interference | Weight at Collection | Guardian |
|---|---|---|---|---|---|
| Initial | | IA9052753 | No | 2219 grams | MOMLAST6274513, MOMFIRST6274513 |

## 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** | **Borderline - See second tier test result** | |
| *Pompe Disease 2nd Tier NBS, Flow Injection Analysis-Tandem Mass Spectrometry* | | *5* |
| **Pompe Disease 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* | | *6* |
| **X-linked Adrenoleukodystrophy disease interpretation** | **Within Normal Limits** | |
| *Severe Combined Immunodeficiency, Real-Time PCR* | | *7* |
| **Severe Combined Immunodeficiency (SCID) interpretation** | **Within Normal Limits** | |
| *Spinal Muscular Atrophy, Real-Time PCR* | | *8* |
| **Spinal Muscular Atrophy (SMA) interpretation** | **Within Normal Limits** | |

## 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.

| Patient Name | Birth Date | Client Reference | Accession # |
|---|---|---|---|
| LAST6274513, FIRST6274513 | 2025-05-15 02:18 | 6274513 | 78468 |

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. 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.

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. 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.

7. 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. 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 15:25 MBH |
| 2. Congenital Hypothyroidism, Immunoassay | 2025-05-17 06:00 MBH | 10320 | 2025-05-21 15:25 MBH |
| 3. Biotinidase Deficiency, Immunoassay | 2025-05-17 06:00 MBH | 10320 | 2025-05-21 15:25 MBH |
| 4. Galactosemia, Enzymatic Assay | 2025-05-17 06:00 MBH | 10320 | 2025-05-21 15:25 MBH |
| 5. Hemoglobinopathies, Various Methods | 2025-05-17 06:00 MBH | 10320 | 2025-05-21 15:25 MBH |
| 6. Cystic Fibrosis, Various Methods | 2025-05-17 06:00 MBH | 10320 | 2025-05-21 15:25 MBH |
| 7. Expanded Screening Disorders, Tandem Mass Spectrometry | 2025-05-17 06:00 MBH | 10320 | 2025-05-21 15:25 MBH |
| 8. Lysosomal Storage Disorders, Tandem Mass Spectrometry | 2025-05-17 06:00 MBH | 10320 | 2025-05-21 15:25 MBH |
| 9. Pompe Disease 2nd Tier NBS, Flow Injection Analysis-Tandem Mass Spectrometry | 2025-05-18 10:30 MBH | 10415 | 2025-05-21 15:25 MBH |
| 10. X-linked Adrenoleukodystrophy, Tandem Mass Spectrometry | 2025-05-17 06:00 MBH | 10320 | 2025-05-21 15:25 MBH |
| 11. Severe Combined Immunodeficiency, Real-Time PCR | 2025-05-17 06:00 MBH | 10320 | 2025-05-21 15:25 MBH |
| 12. Spinal Muscular Atrophy, Real-Time PCR | 2025-05-17 06:00 MBH | 10320 | 2025-05-21 15:25 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, 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

| Patient Name | Birth Date | Client Reference | Accession # |
|---|---|---|---|
| LAST6274513, FIRST6274513 | 2025-05-15 02:18 | 6274513 | 78468 |

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. 