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

NEWBORN SCREENING REPORT

605/773-3361

Patient	Birth Date and Gender	Client Reference	Accession #
LAST004, FIRST004 #34 1234 22ND ST ANYTOWN, SD 57999	2024-05-19 09:47 Female	1976056	32003
	Collected 2024-05-20 10:15	**Received** 2024-05-21 02:00	**Project**

Report To

ANYTOWN HOSPITAL
SEND-OUT LAB
1000 MEDICAL ST
ANYTOWN, SD 57999

Ordering Health Care Provider
DOE, JANE

Primary Care Provider
DOE, JOHN

Sample Type
Blood spot specimen

Sample Note(s)

Screen	Birth Order	IA Barcode Number	Transfusion Interference	Weight at Collection	Guardian
Initial		IA9064327	No	2263 grams	MOMLAST004, MOMFIRST004

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	**Within Normal Limits**	
X-linked Adrenoleukodystrophy, Tandem Mass Spectrometry		*5*
X-linked Adrenoleukodystrophy disease interpretation	**Within Normal Limits**	
Severe Combined Immunodeficiency, Real-Time PCR		*6*
SCID interpretation	**Within Normal Limits**	
Spinal Muscular Atrophy, Real-Time PCR		*7*
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.

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

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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. A Presumptive Positive or 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 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. 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.

6. 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. 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	2024-05-21 05:57 MBH	10320	2024-05-29 15:38 DU
2. Congenital Hypothyroidism, Immunoassay	2024-05-21 08:18 MBH	10320	2024-05-29 15:38 DU
3. Biotinidase Deficiency, Immunoassay	2024-05-21 05:14 MBH	10320	2024-05-29 15:38 DU
4. Galactosemia, Enzymatic Assay	2024-05-21 03:16 MBH	10320	2024-05-29 15:38 DU
5. Hemoglobinopathies, Various Methods	2024-05-21 11:43 MBH	10320	2024-05-29 15:38 DU
6. Cystic Fibrosis, Various Methods	2024-05-21 12:17 MBH	10320	2024-05-29 15:38 DU
7. Expanded Screening Disorders, Tandem Mass Spectrometry	2024-05-21 12:17 MBH	10320	2024-05-29 15:38 DU
8. Lysosomal Storage Disorders, Tandem Mass Spectrometry	2024-05-21 15:23 MBH	10320	2024-07-25 17:06 MBH
9. X-linked Adrenoleukodystrophy, Tandem Mass Spectrometry	2024-05-22 09:17 MBH	10320	2024-05-29 15:38 DU
10. Severe Combined Immunodeficiency, Real-Time PCR	2024-05-21 14:10 MBH	10320	2024-05-29 15:38 DU
11. Spinal Muscular Atrophy, Real-Time PCR	2024-05-21 14:25 MBH	10320	2024-05-29 15:38 DU

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 Certificate: 16D0709302

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.