

Patient LAST001, FIRST001 #34 1234 22ND ST ANYTOWN, AK 99999		Birth Date and Gender 2024-05-19 09:47 Female	Client Reference 56514678	Accession # 32000	
		Collected 2024-05-20 10:15	Received 2024-05-21 02:00	Project	
Report To	ANYTOWN HOSPITAL SEND-OUT LAB 1000 MEDICAL ST ANYTOWN, AK 99999			Ordering Health Care Provider DOE, JANE	
				Primary Care Provider DOE, JOHN	
				Sample Type Blood spot specimen	
				Sample Note(s)	
Screen Initial	Birth Order	IA Barcode Number IA5720981	Transfusion Interference No	Weight at Collection 2263 grams	Guardian MOMLAST001, MOMFIRST001

RESULTS OF ANALYSIS - FINAL REPORT

<u>TEST</u>	<u>RESULT</u>	<u>ANALYSIS NOTE(S)</u>
<i>Congenital Adrenal Hyperplasia, Immunoassay</i> Congenital adrenal hyperplasia interpretation	Within Normal Limits	
<i>Congenital Hypothyroidism, Immunoassay</i> Congenital hypothyroidism interpretation	Within Normal Limits	
<i>Biotinidase Deficiency, Immunoassay</i> Biotinidase deficiency interpretation	Within Normal Limits	
<i>Galactosemia, Enzymatic Assay</i> Galactosemia interpretation	Within Normal Limits	
<i>Hemoglobinopathies, Various Methods</i> Hemoglobin disorders interpretation	Within Normal Limits	1
<i>Cystic Fibrosis, Various Methods</i> Cystic fibrosis interpretation	Within Normal Limits	2
<i>Expanded Screening Disorders, Tandem Mass Spectrometry</i> Fatty acid oxidation defects interpretation	Within Normal Limits	3
	Organic acidemias interpretation	
	Amino acidemias interpretation	
<i>Severe Combined Immunodeficiency, Real-Time PCR</i> SCID interpretation	Within Normal Limits	4
<i>Spinal Muscular Atrophy, Real-Time PCR</i> SMA interpretation	Within Normal Limits	5
<i>CPT1A Arctic Variant (c.1436C>T), Real-Time PCR</i> CPT1A Arctic Variant (c.1436C>T) interpretation	Negative	4

SAMPLE AND ANALYSIS NOTES

- Core conditions screened: Sickle 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 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

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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.

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- 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

<u>TEST</u>	<u>ANALYZED</u>	<u>SITE</u>	<u>RELEASED</u>
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. Severe Combined Immunodeficiency, Real-Time PCR	2024-05-21 14:10 MBH	10320	2024-05-29 15:38 DU
9. Spinal Muscular Atrophy, Real-Time PCR	2024-05-21 14:25 MBH	10320	2024-05-29 15:38 DU
10. CPT1A Arctic Variant (c.1436C>T), Real-Time PCR	2024-05-23 09:17 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, procedures, and the CPT1A Arctic Variant, please contact the Alaska Newborn Screening Program at 907/334-2295 or email newborn.screening@alaska.gov.

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://www.shl.uiowa.edu/screening/newborn/disorderslist.xml> . 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.