## NEONATAL SCREEN - Details (OLIVIA)

## **Component Results**

Component	Your Value	Standard Range	Flag
Reason For Lab Test	Your Value Initial screen		
Sample Condition	Your Value Acceptable		
Newborn Screening Report - Overall Interpretation	Your Value All screening is in range for the conditions tested		
Positive Markers	Your Value None		
Equivocal Markers	Your Value None		
Newborn Screening Short Narrative Summary	Your Value NBS Testing Lab - ALLIED LABORATORY ~453 RAVENDALE DR, STE B, MOUNTAIN VIEW, CA 94043-5221		
	Genetic Disease Laboratory - GDL 850 MARINA BAY PKWY, # G265, RICHMOND, CA 94804- 6403		
	Lab Director - Genetic Disease Laboratory, (510) 231-1790		
	If you have any questions regarding these screening outcomes, please contact the Newborn Screening Staff at Stanford University Medical		

Standard Range

Flag

Center at (650) 724-8120.

**Methods and Limitations:** 

Assays for BD, CAH, CF, GAL, MS/MS Acylcarnitine and Amino Acid Panels, PCH, hemoglobinopathies, and SCID were performed at the testing laboratory specified on the front of this report. Assays for ALD Tier-1, ALD Tier-2, CAH Tier-2, Pompe Disease Tier-1, MPS I Tier-1, and SMA were developed and/or optimized by the Genetic Disease Laboratory (GDL), and performed at GDL. Performance characteristics of these assays are determined by GDL. The SMA assay is designed to identify 95% of SMA patients who have homozygous deletions of the SMN1 gene on chromosome 5q. These assays have not been cleared or approved by the U.S. Food and Drug Administration (FDA). The FDA has determined that such clearance or approval is not necessary. The assays are used clinical purposes. They should not be regarded as investigational or for research. GDL is certified under the Clinical Laboratory **Improvement Amendments of** 1988 (CLIA-88) to perform high complexity genetic disease screening. **Attention Healthcare Provider:** 

Due to biological variability of newborns and differences in detection rates for the various disorders in the newborn period, the Newborn Screening Program will not identify all newborns with these conditions. While a positive screening result identifies newborns at an increased risk to justify a diagnostic work-up, a negative screening result does not rule out the possibility of a disorder. Health care providers should remain watchful for any sign or symptoms of these disorders in their patients. A newborn screening result should not be considered diagnostic, and cannot replace the individualized evaluation and diagnosis of an infant by a well-trained, knowledgeable health care provider.

Full NBS Summary For Display/Printing

**Your Value** 

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Component	Your Value	Standard Range	Flag
Conditions Tested For In NBS Study	Your Value SMA, X-ALD, GAA, MPS-I, 3- MCC, 3MGA, 5-OXO, ARG, CIT-I, CIT-II, CPT-Ia, CPT-II, GA-1, GA- 2, HHH, HMG, IVA, LCHAD, MAL, MCAD, MCD, MET, MSUD, NKHG, PKU, PRO I, TYR-1, TYR- II, VLCAD, BIO, CAH, CF, GALT, SCID SCAD or EMA or IBG or GA-2 (MADD), Hemoglobinopathies, Hypothyroidism, CUD, BKT, PA, MMA, 2M3HBA		
CA Accession Number	Your Value 282-66-024/21-2021-12		
State Printed On Filter Paper Card	Your Value CA		
Birth Weight	Your Value 3265 grams	Standard Range grams	
Birth Time	Your Value 0944		
Newborn Age In Hours	Your Value 25 hour(s)	Standard Range hour(s)	
Birth Plurality	Your Value Singleton		
Infant Factors That Affect Newborn Screening Interpretation	Your Value None		_ <del>_</del>
Feeding Types	Your Value Unknown		
CF NBS Comment	Your Value Negative		
Biotinidase NBS Comment	Your Value Negative		<u> </u>

Component	Your Value	Standard Range	Flag
Galactosemias NBS Comment	Your Value Negative		
Congenital Hypothyroidism NBS Comment	Your Value Negative		
Congenital Adrenal Hyperplasia NBS Comment	Your Value Negative		
Acylcarnitine NBS Comment	Your Value Negative		
Amino Acidemias NBS Comment	Your Value Negative		
SCID NBS Comment	Your Value Negative		
Adrenoleukodystrophy NBS Comment	Your Value Negative		
MPS I NBS Comment	Your Value Negative Interpretation Comments: The alpha-L-iduronidase (IDUA)enzyme activity is above the cut-off of the daily patient median - suggestive of screen negative for Mucopolysaccharidosis I (MPS I) disorder.	9	
Pompe NBS Comment	Your Value Negative Interpretation Comments: The acid alpha-glucosidase (GAA) enzyme activity is above the cut-off of the daily patient median - suggestive of screen negative for Pompe disease.		
Hemoglobin Pattern	Your Value FA		

Component	Your Value	Standard Range	Flag
Hemoglobin Disorders NBS Comment	Your Value		
	<b>Usual hemoglobin</b>		
	pattern.}These results assume		
	no transfusion prior to testing		
	and do not rule out the		
	possibility of a thalassemia		
	trait or rare hemoglobin		
	variants.		
Cystic Fibrosis NBS Interpretation	Your Value		
	In range		
Biotinidase Deficiency	Your Value		
	In range		
Galactosemias	Your Value		
	In range		
Congenital Hypothyroidism	Your Value		
	In range		
Congenital Adrenal Hyperplasia	Your Value		
	In range		
Acylcarnitine	Your Value		
	In range		
SCID NBS Interpretation	Your Value		
	In range		
Adrenoleukodystrophy NBS Interpretation	Your Value		
	In range		
MPS I NBS Interpretation	Your Value		
	In range		
Pompe NBS Interpretation	Your Value		
	In range		
MPS I Alpha-L-iduronidase	Your Value	Standard	
	<b>6.78</b> umol/L/h	Range	
		>=1.1916	
		umol/L/h	

Component	Your Value	Standard Flag Range
Pompe Acid alpha glucosidase	<b>Your Value</b> <b>12.07</b> umol/L/h	Standard Range >=2.2428 umol/L/h
IRT Trypsinogen Immunoreactive	<b>Your Value</b> <b>44.78</b> ng/mL	Standard Range <69 ng/mL
TSH - Thyrotropin	<b>Your Value</b> <b>2.73</b> mIU/L	Standard Range <29 mIU/L
Galactose 1 Uridyl Transferase	Your Value 274.75 enzyme units	Standard Range >50 enzyme units
Biotinidase	<b>Your Value</b> <b>59.82</b> ERU	Standard Range >10 ERU
17-OH PROGESTERONE	<b>Your Value</b> <b>4.09</b> nmol/L	Standard Range <85 nmol/L
GLYCINE	<b>Your Value</b> <b>509</b> umol/L	Standard Range umol/L
Alanine+Beta Alanine+Sarcosine	<b>Your Value</b> <b>315</b> umol/L	Standard Range <1000 umol/L
Valine	<b>Your Value</b> <b>86.3</b> umol/L	Standard Range umol/L

Component	Your Value	Standard Flag Range
Valine/Phenylalanine Ratio	Your Value 1.87202 {Ratio}	Standard Range <3.5
		{Ratio}
Alloisoleucine+Isoleucine+Leucine+Hydroxyproline	Your Value 93.9 umol/L	Standard Range <250 umol/L
Alloisoleucine+Isoleucine+Leucine+Hydroxyproline/Alanine	Your Value 0.30 {Ratio}	Standard Range <1.1 {Ratio}
Phenylalanine	<b>Your Value</b> <b>46.1</b> umol/L	Standard Range <165
		umol/L
Phenylalanine/Tyrosine Ratio	Your Value 0.51 {Ratio}	Standard Range <2.4
		{Ratio}
Tyrosine	Your Value 90.8 umol/L	Standard Range <850 umol/L
Succinylacetone	Your Value 0.23 umol/L	Standard Range <4.5
		umol/L
Methionine	<b>Your Value</b> <b>16</b> umol/L	Standard Range 8 - 100 umol/L
Citrulline	Your Value 20 umol/L	Standard Range 5 - 60 umol/L

Component	Your Value	Standard Flag Range
Citrulline/Arginine Ratio	Your Value 3.27 {Ratio}	Standard Range <6 {Ratio}
Asparagine+Ornithine	<b>Your Value</b> <b>93</b> umol/L	Standard Range <800 umol/L
Ornithine/Citrulline Ratio	<b>Your Value 4.59</b> {Ratio}	Standard Range {Ratio}
Arginine	<b>Your Value</b> 6 umol/L	Standard Range <50 umol/L
Arginine/Ornithine Ratio	Your Value 0.07 {Ratio}	Standard Range <1.4 {Ratio}
Proline	<b>Your Value</b> 137 umol/L	Standard Range <1500 umol/L
5-Oxoproline+Pipecolate	<b>Your Value 26</b> umol/L	Standard Range umol/L
C-0 (CARNITINE, FREE)	<b>Your Value</b> <b>22.8</b> umol/L	Standard Range 6 - 125 umol/L
C-0/C16+C18 (Carnitine, Free/C16+C18 Ratio)	Your Value 9.80 {Ratio}	Standard Range <75 {Ratio}
C2 (Acetylcarnitine)	<b>Your Value</b> <b>19.4</b> umol/L	Standard Range 11 - 80 umol/L

Component	Your Value	Standard Flag Range
C3 (Propionylcarnitine)	Your Value 2.9 umol/L	Standard Range <6.3
		umol/L
C3/C2 (Propionylcarnitine/Acetylcarnitine)	Your Value 0.15 {Ratio}	Standard Range <0.3 {Ratio}
C3-DC+C4-OH (Malonylcarnitine+3-OH Butyrylcarnitine)	Your Value 0.070 umol/L	Standard Range <0.4 umol/L
C3-DC/C10 (Malonylcarnitine/Decanoylcarnitine)	Your Value 1.40000 {Ratio}	Standard Range <5.2 {Ratio}
C4 (Butyrylcarnitine+Isobutyrylcarnitine)	Your Value 0.340 umol/L	Standard Range <1.7 umol/L
C5 (Isovalerylcarnitine+Methylbutyrylcarnitine)	Your Value 0.070 umol/L	Standard Range <1 umol/L
Isovaleryl+Methyl Butyrylcarnitine/C3	Your Value 0.02414 {Ratio}	Standard Range <0.45 {Ratio}
C5:1 (Tiglylcarnitine)	Your Value 0.010 umol/L	Standard Range <0.5 umol/L
C5-OH (3-OH Isovalerylcarnitine)	Your Value 0.100 umol/L	Standard Range <0.85 umol/L

Component	Your Value	Standard Flag Range
Glutamine+3-OH Hexanoylcarnitine	<b>Your Value</b> <b>0.110</b> umol/L	Standard Range <0.5 umol/L
C5-DC/C3-DC (Glutarylcarnitine/Malonylcarnitine Ratio)	Your Value 1.57143 {Ratio}	Standard Range >0.5 {Ratio}
C6 (Hexanolycarnitine)	Your Value 0.030 umol/L	Standard Range <0.95 umol/L
C8 (Octanoylcarnitine)	Your Value 0.050 umol/L	Standard Range <0.6 umol/L
C8/C10 (Octanoylcarnitine/Decanoylcarnitine Ratio)	Your Value 1.00 {Ratio}	Standard Range {Ratio}
C8:1 (Octenoylcarnitine)	<b>Your Value</b> <b>0.120</b> umol/L	Standard Range <0.65 umol/L
C10 (Decanoylcarnitine)	Your Value 0.050 umol/L	Standard Range <0.65 umol/L
C10:1 (Decenoylcarnitine)	Your Value 0.050 umol/L	Standard Range <0.45 umol/L
C12 (Dodecanoylcarnitine)	Your Value 0.050 umol/L	Standard Range <2 umol/L

Component	Your Value	Standard Flag Range
C12:1 (Dodecenoylcarnitine)	Your Value 0.04 umol/L	Standard Range umol/L
C14 (Tetradecanoylcarnitine)	Your Value 0.120 umol/L	Standard Range <1.2 umol/L
C14:1 (Tetradecenoylcarnitine)	Your Value 0.050 umol/L	Standard Range <0.8 umol/L
C14:1/C12:1 (Tetradecenoylcarnitine/Dodecenoylcarnitine)	Your Value 1.25 {Ratio}	Standard Range {Ratio}
C14:2 (Tetradecadienoylcarnitine)	Your Value 0.02 umol/L	Standard Range umol/L
C14-OH (3-OH Tetradecanoylcarnitine)	Your Value 0.010 umol/L	Standard Range <0.2 umol/L
C16 (Palmitoylcarnitine)	Your Value 1.600 umol/L	Standard Range <10 umol/L
C16:1 (Palmitoleylcarnitine)	<b>Your Value</b> <b>0.150</b> umol/L	Standard Range <1.4 umol/L
C16-OH (3-OH Palmitoylcarnitine)	Your Value 0.010 umol/L	Standard Range <0.1 umol/L
C16-OH/C16 (3OH-Palmitoylcarnitine/Palmitoylcarnitine)	Your Value 0.00625 {Ratio}	Standard Range <0.07 {Ratio}

Component	Your Value	Standard Range	Flag
C18 (Stearoylcarnitine)	<b>Your Value</b> <b>0.420</b> umol/L	Standard Range <3.5 umol/L	
C18:1 (Oleoylcarnitine)	Your Value 0.730 umol/L	Standard Range <7 umol/L	
C18:2 (Linoleoylcarnitine)	<b>Your Value</b> <b>0.24</b> umol/L	Standard Range umol/L	
C18-OH (3-OH Stearoylcarnitine)	Your Value 0.010 umol/L	Standard Range <0.1 umol/L	
C18:1-OH (3-OH Oleoylcarnitine)	<b>Your Value</b> <b>0.010</b> umol/L	Standard Range <0.1 umol/L	
(TREC) T-CELL RECEPTOR EXCISION CIRCLE, PCR	Your Value 401 copies/uL	Standard Range >18 copies/uL	
Lysophosphatidylcholine ALD-1	<b>Your Value</b> <b>0.340</b> umol/L	Standard Range <0.42 umol/L	
Discharge Provider Name	Your Value SUDHARANI NARASIMHAN		
Discharge Provider ID	Your Value 1366512592		
Discharge Provider Address	Your Value 751 S BASCOM AVE SAN JOSE CA 95128-2604 USA		

Component	Your Value	Standard Flag Range
SMN1 Homozygous Deletion Analysis	Your Value Exon 7 Present	Standard Range
		Exon 7 Present
SMA NBS Interpretation	Your Value In range	
SMA NBS comment	Your Value SMA Negative,	

## **General Information**

Ordered by Susan Abraham, MD

Collected on 10/07/2021 11:05 AM from Foot, Left (Blood)

Resulted on 10/20/2021 5:00 AM

Result Status: Final result

This test result has been released by an automatic process.

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