

CALIFORNIA DEPARTMENT OF PUBLIC HEALTH
NEWBORN SCREENING PROGRAM
850 MARINA BAY PARKWAY, ROOM F175
RICHMOND, CA 94804

T1 P1 121 (260) 051101238802



ALEX ESPINOSA, MD
2450 ASHBY AVE # 4NW
BERKELEY, CA 94705-2067

Newborn's Provider: ALEX ESPINOSA, MD

Testing Laboratory:
ALLIED LABORATORY

453 RAVENDALE DR, STE B, MOUNTAIN VIEW, CA 94043-5221



020212388021

If you have any questions regarding these screening outcomes, please contact the Newborn Screening Staff at Stanford University Medical Center at (650) 724-8120.

NEWBORN SCREENING RESULTS - INITIAL

BABY		
MARRUJO		
Gender:	Female	
NBS FORM #:	33240328	
Medical Record:	63262627	
BIRTH/COLLECTION INFORMATION		
	Date	Time
Baby's Birth:	07/27/20	2221
Specimen Collection:	07/29/20	0500
Age at Collection:	30 hour(s)	
Birth Weight:	2800 grams	
Ethnicity:		
Specimen Collection Site:	ALTA BATES SUMMIT MEDICAL CENTER	
Feeding Type:	Only Formula	
Newborn on TPN/Hypertal or Amino Acids: No		
MOTHER		
MARRUJO, JAZMYNE		
520 LOCUST ST		
SAN JOSE, CA 95110-2707		
Phone: (306) 809-0758		
Secondary Phone:		

SCREEN NEGATIVE RESULTS

Adrenoleukodystrophy (ALD)
Biotinidase Deficiency (BD)
Congenital Adrenal Hyperplasia (CAH)
Cystic Fibrosis (CF)
Galactosemia (GAL)
MS/MS Acylcarnitine Panel

MS/MS Amino Acid Panel (Including PKU)
Mucopolysaccharidosis I (MPS I)
Pompe Disease
Primary Congenital Hypothyroidism (PCH)
Severe Combined Immunodeficiency (SCID)
Spinal Muscular Atrophy (SMA)

HEMOGLOBIN PATTERN

FA

Usual hemoglobin pattern. These results assume no transfusion prior to testing and do not rule out the possibility of a thalassemia trait or rare hemoglobin variants.

INTERPRETATION

Marrujo, GIRL Jazmyne

MRN: 63262627 KAO, LILY CHING CHIUNG
7/27/2020 2 wks WUBESHET, MARAMAWIT
SEX: F NCU
MEDS, LABS CSN: 1045698110 7/27/2020

HAR: 304795632



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.

OFFICE USE ONLY:

212-57-030/21-2020-12 08/03/20 R003 238050328

Acylcarnitine	Expected Range	Value	Flag	Amino Acid	Expected Range	Value	Flag
FC	> 6 to < 125 µmol/L	27.3 µmol/L		Glycine		481 µmol/L	
FC / (C16 + C18:1) Ratio	< 75	6.80		Alanine	< 1000 µmol/L	373 µmol/L	
C-2	> 11 to < 80 µmol/L	32.1 µmol/L		Valine		109.5 µmol/L	
C-3	< 6.3 µmol/L	2.7 µmol/L		Valine / Phenylalanine Ratio	< 3.5	1.70827	
C03 / C02 Ratio	< 0.3	0.08		Leucine/Isoleucine	< 250 µmol/L	117.6 µmol/L	
C-3DC	< 0.38 µmol/L	0.150 µmol/L		Leucine/Alanine Ratio	< 1.1	0.32	
C03DC / C10 Ratio	< 5.2	3.00000		Phenylalanine	< 165 µmol/L	64.1 µmol/L	
C05DC / C03DC Ratio	> 0.6	0.86667		Phenylalanine/Tyrosine Ratio	< 2.4	0.61	
C-4	< 1.7 µmol/L	0.220 µmol/L		Tyrosine	< 850 µmol/L	104.7 µmol/L	
C-5	< 1 µmol/L	0.150 µmol/L		Succinylacetone	< 4.5 µmol/L	0.23 µmol/L	
C05 / C03 Ratio	< 0.45	0.05556		Methionine	> 8 to < 100 µmol/L	19 µmol/L	
C-5:1	< 0.5 µmol/L	0.010 µmol/L		Citrulline	> 5 to < 60 µmol/L	13 µmol/L	
C-5OH	< 0.85 µmol/L	0.140 µmol/L		Citrulline/Arginine Ratio	< 6	2.18	
C-5DC	< 0.5 µmol/L	0.130 µmol/L		Ornithine	< 800 µmol/L	89 µmol/L	
C-6	< 0.95 µmol/L	0.050 µmol/L		Ornithine/Citrulline Ratio		6.72	
C-8	< 0.6 µmol/L	0.070 µmol/L		Arginine	< 50 µmol/L	6 µmol/L	
C08 / C10 Ratio		1.40		Arginine/Ornithine Ratio	< 1.4	0.07	
C-8:1	< 0.65 µmol/L	0.140 µmol/L		Proline	< 1500 µmol/L	180 µmol/L	
C-10	< 0.65 µmol/L	0.050 µmol/L		5-Oxoproline		18 µmol/L	
C-10:1	< 0.45 µmol/L	0.070 µmol/L					
C-12	< 2 µmol/L	0.090 µmol/L					
C-12:1		0.06 µmol/L					
C-14	< 1.2 µmol/L	0.280 µmol/L					
C14:1	< 0.8 µmol/L	0.110 µmol/L					
C14:1 / C12:1 Ratio		1.83					
C14:2		0.03 µmol/L					
C-14OH	< 0.2 µmol/L	0.020 µmol/L					
C-16	< 10 µmol/L	2.490 µmol/L					
C-16:1	< 1.4 µmol/L	0.270 µmol/L					
C-16OH	< 0.1 µmol/L	0.030 µmol/L					
C16OH / C16 Ratio	< 0.07	0.01205					
C-18	< 3.5 µmol/L	0.780 µmol/L					
C-18:1	< 7 µmol/L	1.530 µmol/L					
C-18:2		0.33 µmol/L					
C-18OH	< 0.1 µmol/L	0.010 µmol/L					
C-18:1OH	< 0.1 µmol/L	0.030 µmol/L					

Very Long Chain Fatty Acid Tier 1 (ALD)*

Disorder: Analyte	Expected Range	Value	Flag
ALD: C26	<0.42 µmol/L	0.270 µmol/L	

Lysosomal Storage Diseases*

Disorder: Analyte	Expected Range	Value	Flag	Interpretation Comments
Pompe: Acid alpha-glucosidase (GAA)	>=2.2968 µmol/L/h	9.98 µmol/L/h		The acid alpha-glucosidase (GAA) enzyme activity is above the cut-off of the daily patient median - suggestive of screen negative for Pompe disease.
MPS I: Alpha-L-iduronidase (IDUA)	>=1.0728 µmol/L/h	6.38 µmol/L/h		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.

Neuromuscular Disorders*

Disorder: Analyte	Expected Range	Value	Flag	Interpretation Comments
SMA: SMN1	Exon 7 Present	Exon 7 Present		

Marrujo, GIRL Jazmyne

MRN: 63262627

7/27/2020 2 wks

SEX: F

MEDS. LABS

KAO, LILY CHING CHIUNG

WUBESHET, MARAMAWIT

NICU

CSN: 1045699110 7/27/2020

HAR: 304795832



*Genetic Disease Laboratory: GDL 850 MARINA BAY PKWY, # G265, RICHMOND, CA 94804-6403

Director: Rasoul Alikhani Koupaie, PhD., Genetic Disease Laboratory, (510) 231-1790

Methods and Limitations:

Assays for ALD Tier-1, ALD Tier-2, CAH Tier-2, Pompe Tier-1, MPS I Tier-1 and SMA were developed and/or optimized by the California Department of Public Health Genetic Disease Laboratory Branch (GDLB). Performance characteristics of these assays are determined by GDLB. The SMA assay is designed to identify 95% of SMA patients who have homozygous deletion/mutation of SMN1 gene in 5q chromosome. 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 for clinical purposes. They should not be regarded as investigational or for research. GDLB is certified under the Clinical Laboratory Improvement Amendments of 1988 (CLIA-88) to perform high complexity genetic disease screening.