

MEMORANDUM

DATE: April 10, 2024

TO: South Carolina Newborn Screening Community

- FROM: Jenny Meredith, Ph.D., HCLD(ABB) SC DHEC Public Health Laboratory (PHL) Director
- **SUBJECT:** South Carolina (SC) Newborn Screening (NBS) Program's New Disorders, Analytes/Analyte ratios and updated Expected Range

Effective April 22, 2024, the SC Newborn Screening Panel is expanding to include the following:

- Two new disorders: X-Linked Adrenoleukodystrophy (X-ALD) and Argininemia;
 - X-ALD will display in an additional row on the lab report
 - o Argininemia will be within the Amino Acid Panel on the lab report
- Additional analytes and analyte ratios to aid in the screening of existing NBS disorders (see Table 1).
- The expected range for the C0 analyte will be updated (see Table 2); C0 is used to screen for Carnitine Uptake Deficiency (CUD).

These changes are part of the SC NBS Program's commitment to continuous improvement. The SC NBS Program monitors test results and determines if adjustments are needed based on population studies.

Please direct any questions to Beth Bair, MS, Chemistry Division Director at bairea@dhec.sc.gov; 803-896-0991 or Tanya Spells, MS, MT(ASCP), NBS Follow up Program Director at spellsty@dhec.sc.gov; 803-898-0619.

Thank you to all our providers and collaborators for your attention to this important update.

Table 1. New analytes and analyte ratios

Analyte	Target Disorder	Expected Range (µmol/L)	
C26:0-LPC (C26:0 lysophosphatidylcholine)	X-ALD	1 st tier < 0.40	2 nd tier < 0.20
Arginine (ARG)	Argininemia	< 80.00	
Argininosuccinc Acid (ASA)	Argininosuccinic Aciduria	< 1.00	
C14OH (3-Hydroxy- tetradecanoyl carnitine)	Long Chain L-3 Hydroxyacyl-CoA dehydrogenase deficiency (LCHAD); Trifunctional protein deficiency (TFP)	< 0	.10
C16:1 (Hexa-decenoyl carnitine)	LCHAD, TFP, Very long-chain acyl-CoA dehydrogenase deficiency (VLCAD)	< 1.00	
C18OH (3-Hydroxy- octadecanoyl carnitine)	LCHAD, TFP	< 0	.10
C18:2-OH (3-Hydroxy- octadecadienoyl carnitine)	LCHAD, TFP	< 0	.04
Analyte ratios	Target Disorder	Expecte	d Range
C18OH/C18	LCHAD, TFP	< 0.08	
Citrulline/Arginine	Citrullinemia Types I and II	< 15.00	
ASA/Arginine	ASA	< 0.30	
Leucine/Alanine	Maple Syrup Urine Disease (MSUD)	< 1.00	
Arginine/Ornithine	Argininemia	< 1.00	
Ornithine/Citrulline	Citrullinemia Types I and II	> 1.00	

Table 2. Updated analyte Expected Range

	Current	Updated Expected
Analyte	Expected Range	Range (µmol/L)
	(µmol/L)	
C0 (Free Carnitine)	> 11.40	> 9.00