



## Department of Health

ANDREW M. CUOMO  
Governor

HOWARD A. ZUCKER, M.D., J.D.  
Commissioner

SALLY DRESLIN, M.S., R.N.  
Executive Deputy Commissioner

October 18, 2016

Dear Health Care Provider,

**RE: 1. Changes to the New York Newborn Screening Program's reference range values for free carnitine (C0) and propionylcarnitine (C3) used for the evaluation of carnitine uptake disorder (CUD).  
2. Discontinuation of the use of methylmalonylcarnitine (C4DC) as a diagnostic marker for the evaluation of propionic acidemia and methylmalonic acidemia (PA/MMA).**

For specimens received on or after October 22, 2016, the level of propionylcarnitine (C3) will be used as a secondary marker for the evaluation of carnitine uptake disorder (CUD). C0 results greater than 10  $\mu\text{mole/L}$  will be considered screen negative. For C0 results between 8.00 and 10.00  $\mu\text{mole/L}$ , the C3 value will be used to further assess the result. If the C3 value is less than 1.00  $\mu\text{mole/L}$ , the result will be considered borderline and a repeat sample must be submitted. If the C3 result is greater than or equal to 1.00  $\mu\text{mole/L}$ , the sample will be considered screen negative for CUD and no further action will be necessary. Repeat specimens that show a second borderline result for C0 will result in referral to a Specialty Care Center. A review of confirmed cases of CUD from 2004 through 2015 showed that in all cases of confirmed CUD, the C3 value was less than 1.00  $\mu\text{mole/L}$ . This change should help eliminate approximately 25 to 30% of requests for repeat samples due to low carnitine.

For specimens received on or after October 22, 2016 we will no longer use methylmalonylcarnitine (C4DC) as a diagnostic marker for PA/MMA. A review of confirmed cases of PA/MMA did not show any cases of abnormal C4DC. A review of referrals based on an abnormal result for the C4DC marker, did not result in a diagnosis of PA or MMA. We will continue to report levels of C4DC when results for the primary marker (C3) and secondary marker (the ratio C3/C2) for PA/MMA are abnormal. This change will eliminate false positive results for PA/MMA due to an elevated result for C4DC.

As always, we will monitor test results over time and may adjust cut-offs based on long term studies. You will be informed if there are any additional changes.

Thank you for your cooperation in making New York State's Newborn Screening Program the best it can be. Questions can be directed to our main telephone number 518-473-7552.

Sincerely,

Michele Caggana, Sc.D., FACMG  
Director, Newborn Screening Program