



## Department of Health

ANDREW M. CUOMO  
Governor

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

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

December 7, 2017

Dear Health Care Provider,

***RE: Changes to the New York Newborn Screening Program's Cystic Fibrosis (CF) Screening Protocol***

Please be advised that for specimens received and tested as of December 1, 2017, the protocol for CF will change to reduce the number of CF referrals requiring follow-up testing. Please see changes to the screening algorithm below.

Previous Algorithm:

All specimens undergo an initial assessment for immunoreactive trypsinogen (IRT) level. Specimens with levels in the top 5% each day are screened by DNA analysis for a panel of 39 common CF variants. Infants identified with 1 or 2 mutations are referred for follow-up sweat testing. Infants with no mutations and an IRT in the top 0.1% [calculated from the previous 10 days IRT values] are also referred for follow-up sweat testing.

New Algorithm:

All specimens undergo an initial assessment for immunoreactive trypsinogen (IRT) level. Specimens with levels in the top 5% each day will be screened by DNA analysis for a panel of 39 common CF variants. Infants identified with 2 variants are referred for follow-up sweat testing. Specimens with only one variant or an IRT in the top 0.1% [calculated from the previous 10 days IRT values] will undergo third-tier screening by *CFTR* gene sequencing. Specimens identified with two CF variants will be referred for follow-up sweat testing. Specimens identified with one CF variant will be reported out as a likely carrier only, no further evaluation will be required. Genetic counseling is recommended for those identified as carriers. Specimens with elevated IRT and no mutations will be reported out as screen negative, no further evaluation will be required.

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