New York Cystic Fibrosis Newborn Screening Consortium Quality Improvement: Focus on Parent and Pediatrician Education and Develop a Statewide Standard of Care for CF-Related Metabolic Syndrome infants.

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Abstract

Background: CFTR-Related Metabolic Syndrome/ Cystic Fibrosis Screen Positive, Inconclusive Diagnosis (CRMS/CFSPID) is the diagnosis in infants who have a positive Cystic Fibrosis (CF) newborn screen (NBS), 2 CF-causing mutations, and borderline or normal sweat test. NY State implemented a new CF NBS algorithm (IRT-DNA-SEQ) in December 2017 with significant improvement in positive predictive value. This algorithm also resulted in detection of more CRMS cases. For these infants repeat sweat testing is recommended at 6, 12, and 18 months to monitor for risk of rising sweat chloride over time and 6-48% of infants with CRMS develop clinical features suggestive of CF. Infants with CRMS and sweat test results in the normal range are often lost to follow and parents were unwilling to return for recommended repeat sweat testing during the statewide lockdown during the peak of COVID-19 pandemic. We recognized the practice gap exacerbated during the pandemic and underscore the importance of establishing a medical home in a CF Center for longitudinal care. **Methods:** Retrospective analysis of infants with CRMS from December 2017 to December 2020 were collected by 10 NY CF Centers and the NBS program with NYU as the data collection and statistical analysis site. Infants with CRMS without repeat sweat chloride testing at 6 months of age were considered lost to follow up, and their parents were contacted via mail or telephone. Families completed a questionnaire that was developed with the assistance of CF Voice to evaluate parental understanding of CRMS and the recommendation for repeat sweat chloride testing. Primary care providers (PCPs) caring for infants with CRMS were also contacted and provided educational materials about CRMS. A subcommittee of CF Center Directors met to develop a statewide approach for the management of infants with CRMS. **Results:** Of 350 infants diagnosed with CRMS, 179 (51.1%) infants were lost to follow up. As an outcome of this QI effort 31 (17.3%) were scheduled for repeat sweat tests and follow up at CF Centers. This QI effort explored the knowledge and practice gap among PCPs with limited understanding of the implications of a CRMS diagnosis. CF Center Directors subcommittee issued a consensus statement regarding evaluation and follow up for infants with CRMS in NY. **Conclusions:** This QI effort effectively recaptured infants with CRMS previously lost to follow up. Consensus recommendations for CRMS include annual visits until 2-6 years of age for repeat sweat testing and in adolescence to educate the patient about clinical and reproductive implications of CRMS.

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