Determination of Cystic Fibrosis Frequency Among Newborns Diagnosed with Meconium Ileus and other Bowel Obstruction

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Background: The purpose of this study is to determine the incidence of Cystic Fibrosis (CF) among newborns with meconium ileus (a form of neonatal bowel obstruction). It has long been believed that upwards of 80% of newborns with meconium ileus have CF; however, recently 65% of newborns who have been reported to the New England Newborn Screening Program (NENSP) as having meconium ileus have screened negative for CF by the state newborn screen.

Objectives: This project is aimed at determining the explanation for this apparent discrepancy, and at confirmation that CF newborn screen negative infants with bowel obstruction have had further testing to confirm or deny a CF diagnosis. Upon its completion we hope that this project will further aid in our understanding of CF, its clinical manifestations and the effectiveness of the newborn screening program. In addition, it will educate both the subspecialist and primary care physicians regarding follow-up of neonates with diagnoses of bowel obstruction and/or CF.

Methods: This study will involve reviewing the charts of Massachusetts newborns born between 02/1/1999 and 01/25/2005 who were reported to the NENSP as having meconium ileus or other bowel obstruction (n ~150). The chart of each subject will be reviewed for clinical, laboratory, pathologic, and radiologic evidence that supports or refutes the diagnosis of both meconium ileus and CF.

Results: Research completed thus far has identified these babies by review of the newborn screening database. The charts of babies born at UMMHC (n 22) were reviewed and indicate that only rarely is the diagnosis of meconium ileus made without proper clinical, radiologic and/or pathologic data. However, a trend noticed in the UMMHC babies was that a general diagnosis of bowel obstruction and/or meconium plugs (n 15) was more common, and only two babies were officially given the diagnosis of meconium ileus. Mutational analysis and/or sweat testing reveied three babies with the diagnosis of CF. Of the two babies diagnosed with MI, one was later diagnosed with CF. Review of UMMHC charts also showed a lack of appropriate out-patient follow-up; three babies underwent a sweat test to confirm or deny the diagnosis of CF.

Preliminary Conclusion: Review of newborns at UMMHC indicate that the diagnosis of MI was only made when sufficient and appropriate evidence was present; the majority of babies were
diagnosed with alternative forms of bowel obstruction, most notably meconium plugs. Of the babies diagnosed with MI only one of the two were subsequently diagnosed with CF, however the insufficient number of cases makes statistical review impossible at this time. The most notable conclusion that has been revieled by babies born at UMMHC was that only three babies received appropriate follow-up. Any newborn diagnosed with bowel obstruction should undergo a sweat test to confirm or deny the diagnosis of CF; this is particularly important given that the genetic testing only uncovers approximately 90% of cases. A number of studies have shown the importance of early diagnosis of CF, particularly in regards to early childhood growth, thus appropriate follow-up testing is critical for each of these babies.