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Using Serum Alpha-Fetoprotein as a Diagnostic Marker in Screening Neonates for Cystic Fibrosis

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Background and Public Health Significance: Cystic fibrosis (CF) is a lethal autosomal recessive disorder. CF occurs with an incidence of 1 in 3,300 in the US. Approximately 1 in 29 Caucasians carry a CF mutation. The $\Delta F508$ mutation is present on at least one allele in 70% of all patients. It is thought that early diagnosis of CF by newborn screening in New York can help improve the quality of life for those affected by permitting early treatment and genetic counseling for the family.

Research Objectives: Currently CF screening, based on detection of elevated levels of immunoreactive trypsinogen (IRT) in infants' blood samples, has a 95% false positive rate. The objective of this study is to determine if the specificity and sensitivity of the test can be improved by analyzing serum alpha-fetoprotein (AFP) concentrations in neonates with CF.

Experimental Methods: Serum AFP concentrations were measured in 2,144 dried blood spots of 1-7 day-old infants collected from the NYS Newborn Screening Program using the Beckman Access Immunoassay System. Blood spots were selected based on IRT levels. Other demographic data was collected for further analysis.

Results: Initial data analysis showed no correlation between IRT and AFP serum concentrations ($R^2=0.0011$). No significant differences were observed in AFP concentration when the data were stratified by IRT level. AFP concentrations differed significantly when zero versus 1 or 2 mutation carriers were compared ($p=0.0087$). When stratified by mutation status, there was only a significant difference in AFP concentration between individuals having no mutations and those heterozygous positive for CF ($p=0.0143$).

Discussion: AFP may predict CFTR mutation status, however the pool of confirmed CF patients in this sample is low. To address this, approximately 200 blood specimens from CF patients collected since 1982 from the California Newborn Screening Program will be analyzed to increase statistical power and further investigate the relationship between AFP and IRT.