



**Connecticut
Children's**
MEDICAL CENTER

Testimony of Craig Lapin, MD, Director, Central Connecticut Cystic Fibrosis Center and Associate Professor of Pediatrics, University of Connecticut Health Center to the Public Health Committee regarding Senate Bill 569, An Act Requiring New Infant Health Screening For Cystic Fibrosis

March 10, 2008

Senator Handley, Representative Sayers, Members of the Public Health Committee. Thank you for the opportunity to testify in support of Senate Bill 569, An Act Requiring New Infant Health Screening For Cystic Fibrosis. You will be hearing the personal impact that a delayed diagnosis has for families with cystic fibrosis. I would like to present the medical case to add cystic fibrosis to mandated newborn screening (NBS). CF is the most common lethally inherited disease in caucasians, although it also affects other ethnicities as well. It occurs 3 times more frequently than Phenylketonuria (PKU) and 50 times more than Maple Syrup Urine Disease (MSUD). It is more frequent than Sickle Cell Disease (SCD) that occurs in approximately 1 in 4000 newborns; CF occurs in 1 in 3000 newborns. PKU, MSUD, SCD are amongst the screened diseases here in Connecticut.

CF causes multiple problems but primary are respiratory and nutritional. There is extensive medical research that shows early diagnosis makes a significant difference in the health outcomes (and therefore lives) of patients with CF. In the short term – for infants diagnosed at less than 1 year of age because of CF symptoms (i.e. not by NBS) 26% were stunted (extremely short compared with normal population) versus 9% of those diagnosed by NBS, and 33% were grossly malnourished compared with 11% diagnosed by NBS. Significant CF infections were found twice as frequently (29% not NBS vs 15% NBS), and patients were hospitalized three times as much (64% not NBS vs 22% NBS). Of even greater concern, 5% of patients diagnosed with CF over a 3-year period had life-threatening malnutrition, compared with none by NBS.

In the long term, based on national CF registry data, as people with CF grow older, in every age group, those diagnosed by NBS are always at least half as likely to be malnourished compared with those diagnosed symptomatically. Those diagnosed by NBS are statistically less likely to be stunted, or to have the CF infections that have been shown to lead to more rapid pulmonary function decline and therefore decreased quality of life. By the second decade of life, people diagnosed by NBS are less likely to require hospitalization (thus decreasing cost of care).

Solid research, multiple studies, document that delayed diagnosis and malnutrition in this disease leads to failure to thrive, increased infections, a more rapid decline in lung

function, subsequent decreased quality of life, and decreased life span. In other words, people die earlier. Waiting until patients have symptoms of CF is associated with higher complications rates and morbidity compared to diagnosis by NBS. Data also suggests that patients diagnosed by NBS maintain better pulmonary functions, have less hospitalizations, and fewer CF-related infections. The Center for Disease Control (CDC) has determined that screening for CF is justified.

Our CF center has been part of the voluntary newborn screening program that has been extremely efficient and supportive of families for fifteen years, screening approximately 26,000 infants a year. Yale screens approximately 8,000 patients a year. We estimate there are approximately 10,000 Connecticut babies unscreened each year, or over 150,000 children over the 15-year period. Please, for the sakes of the infants, children, and adults with CF, mandate newborn screening for cystic fibrosis that extends the current program to all. Thank you.