Older patients offer insight into the future of cystic fibrosis (w/ Video)

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An emerging population of middle-aged cystic fibrosis patients contains significantly more females and includes a large proportion of patients who lived for decades without a diagnosis or specialized care, according to research published by researchers at National Jewish Health. The comprehensive analysis of this over-40 cohort, published online May 20 in the *American Journal of Respiratory and Critical Care*, may help guide future treatment of cystic fibrosis as survival past 40 becomes increasingly common.

"As both care and diagnosis have improved, cystic fibrosis has been transformed from a uniformly fatal childhood disease to a condition where survival to middle age and older is possible," said National Jewish Health pulmonologist and lead author Jerry Nick, MD. "We were surprised to discover that the majority of patients diagnosed as adults were females, representing a striking reversal of the "gender gap" in cystic fibrosis, and that they have delayed, but equally severe disease."

Cystic fibrosis (CF), an inherited disease of the lungs and digestive system, affects about 30,000 people in the United States and is the most common genetic disease among Caucasians. A single defective gene causes a missing or non-functional channel for chloride to travel into and out of cells. Lack of this channel disrupts the water balance in the lungs of CF patients, causing the development of thick, dehydrated mucus, which serves as a fertile environment for bacterial growth. Most patients die of respiratory failure brought on by repeated severe bacterial infections in the lungs.

In 1962 the median predicted survival for children with cystic fibrosis was 10 years. Today, it is 37, and children diagnosed today can expect to live into their 50s. Improved care is helping patients live longer. In addition, many more patients with non-traditional symptoms are being diagnosed for the first time as adults.

These two groups - those diagnosed as children and those diagnosed for the first time as adults - comprise two distinct populations of CF patients over 40. Those diagnosed as adults usually have a genetic mutation that produces a partially functional gene and a slightly different set of symptoms from those diagnosed as children. Most commonly, they have functional pancreases and fewer digestive problems.

Dr. Nick and his colleagues analyzed epidemiological and health data on 156 CF patients over 40 year of age who receive care at National Jewish Health, the largest adult cystic fibrosis clinic in the nation. In addition, data were analyzed on nearly 3,000 patients from around the nation who were included in the Cystic Fibrosis Foundation Patient Registry from 1992-2007.

The researchers found that the fate of females changes considerably in the older CF population. It has long been recognized that a "gender gap" is present in CF, favoring males. Historically, females have been diagnosed later, had a poorer prognosis, and survived fewer years than males.

Accordingly, Dr. Nick's analysis showed that fewer females diagnosed as children survived to age 40. However, among those diagnosed as adults, females represented a significant majority, accounting for 72 percent of patients in Colorado and 54 percent nationally. Among the adult diagnosed patients, females survived on average 9 to 14 years longer than males.

The complex factors that account for the differential fate of female CF patients is not understood, although Dr. Nick believes it could be a mixture of behavioral and biological factors.

Dr. Nick's findings also indicate that patients diagnosed as adults do not really have milder diseases -- as is commonly believed -- just a delayed onset of an equally severe form of the disease. Although patients diagnosed as adults live longer than those diagnosed as children, the adult-diagnosed patients lose lung function as rapidly those diagnosed in childhood, and approximately 85% die of respiratory failure or post-transplant complications.

Dr. Nick believes there is a significant number of adults whose CF remains undiagnosed. His analysis indicates that once those patients are accurately diagnosed, proper care can significantly improve their health. Patients diagnosed as adults and subsequently followed at a CF center reversed progressive lung function decline and improved their lung function for at least four years.

Older patients commonly do not get specialized CF care. It is generally recognized that the team approach to treatment provided by the 112 CF Foundation-accredited Care Centers results in better clinical outcomes. However, less than half of long-term CF survivors continued to be seen at CF Centers as they pass 40 years, with the fewest among the adult-diagnosed patients.

"In the coming years, more and more cystic fibrosis patients will be living into their 40s, 50s and beyond," said Dr. Nick. "Our findings concerning the role of gender, in survival, progression of disease, and type of care in current long-term survivors provides important insights that will help us prepare for better treatment of the steadily aging CF population."