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EDITORIAL

Adults with cystic fibrosis in Portugal: A first step towards improvement

The last two decades have seen improving survival in patients with cystic fibrosis (CF) with more patients living into adulthood.[1] The improvement in survival can be attributed to a number of factors including new medications, greater use of antibiotics, better nutrition, and the development of specialized centers where teams of caregivers provide expert evaluation and guidance. Yet despite such gains, our patients still die too young and their treatments come with considerable burden, so there remains great opportunity for further improvements. While we anticipate the development of newer therapeutic options, we can still strive to be even better with the tools we have now.

Quality improvement (QI) is a systematic and continuous process leading to measurable improvement in the health status of our patients. A key aspect of QI is measurement, meaning there must be access to data in order to understand the current state of affairs, to identify opportunities for improvement, and to assess the impact of the interventions. In this issue of the Portuguese Journal of Pulmonology, Silva and colleagues provide a glimpse into the current status of the adult cystic fibrosis population in Portugal.[2] The analysis describes the 89 adult patients who receive their care at three CF centers. The temptation is always to compare these observations to others, but it is imperative to avoid interpreting them in a manner in which we take credit for some and cast blame for others. Rather, the point is that it is merely a portrait of the current state upon which one can develop plans for improvement.

Comparison to other registries, such as the US CF Patient Registry,[1] is not always so easy since the latter reports data inclusive of all patients, not merely the adults. For example, in this report the median age of diagnosis is reported as 13 years in Portugal, whereas in the US the Registry reports it as 3.7 years. Presumably the median age of diagnosis in Portugal, were we to include all patients, is far younger, but it is notable that in Portugal there is a larger proportion (nearly 40%) of patient that are not diagnosed until adulthood. In the US, for example, <5% of patients are diagnosed in adulthood and generally because they have a less severe condition. A previous report have suggested that late diagnosis may define a population of long-term survivors,[3] but this cohort evaluated only those patients >40 years of age and with a median age of diagnosis of 48.5 years, so it is not the best comparison. Of note, the median age of diagnosis in the US is slowly creeping upward (3.1 years to 3.7 years in the last decade) but this is in an era of newborn screening where the diagnosis would be made in the first few months of life. This suggests that there is greater identification of older patients with milder manifestations; that is, that clinicians are now including CF in their differential diagnosis of patients with respiratory or other CF-related symptoms. Another potential explanation for the later diagnosis in Portugal is a larger percentage of milder CF mutations, demonstrated by the lower proportion of patients that are homozygous for F508del (nearly 50% in the US). However it is also possible that those patients with more severe mutations have a reduced survival and that this mutation analysis in the adults is not representative of the Portuguese pediatric CF population.

There are many more questions that could be explored in this population and establishing the database and making it transparent can now allow for generation of hypotheses leading to QI projects. A clear opportunity acknowledged by the authors is earlier diagnosis. Implementation of a newborn screening program would allow for earlier treatment interventions.[4] This is but one example, but that is how it starts. We applaud the authors and the Portuguese CF community for presenting their data and look forward to future revelations demonstrating clinical improvements.

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