Genetic and Reproductive Knowledge Among Adolescents and Adults With Cystic Fibrosis

To the Editor:

Studies have shown that cystic fibrosis (CF) patients have limited knowledge of the genetics of CF. Previously, there was limited need to communicate this information: few CF patients lived to adulthood, and most who did could not reproduce. Genetic counseling focused on the patient’s parents, who were counseled about the recurrence risk at the time of the diagnosis. Today, CF is a disease of adulthood. In 2002, > 40% of CF patients in the United States were > 18 years old; by 2010, it will be > 50%. Together with advances in assisted reproductive technology, reproduction and recurrence risk are now important issues for adolescent and young adult CF patients.

A 19-item questionnaire was developed from the results of prior semistructured interviews with 18 CF patients aged 16 to 25 years. Knowledge-based questions (medical issues, inheritance, and reproductive options/risks) as well as communication patterns (preferred resources for learning about CF and preferred people with whom to talk about reproductive issues) were addressed. Recruited from the University of Alabama CF clinic population, 51 patients aged 15 to 29 years (mean, 21 years), 24 male (47%) and 27 female (53%), completed the questionnaire. The study was approved by the University of Alabama Institutional Review Board.

Regarding autosomal recessive inheritance, only 33% knew that two carriers have a 25% chance of having a child with CF, and 25% knew that two carriers have a 50% chance of having a child who is a carrier. However, 82% knew that two carriers could have a child who did not have CF, and 52% knew that two carriers could have a child who did not carry CF.

On their own reproductive risks, 59% knew that a CF patient had a 0% chance of having a child with CF if their partner was not a carrier, but only 26% knew that all their children would be carriers even if their partner was not a carrier. In the scenario of a CF patient with a CF carrier partner, 44% knew that a child had a 50% chance of having CF, and 24% knew that a child had a 50% chance of being a CF carrier.

Most patients knew about their reproductive potential, as 96% responded that CF patients are able to have children. However, when asked about whether the chance for having children was different for male and female patients with CF, 65% answered that it was more difficult for men, 8% that it was more difficult for women, and 27% answered “not sure.” While 62% reported that they knew that there were options for male CF patients who wanted to have children, only 26% knew of assisted reproductive technology.

Despite widespread availability, the lack of knowledge of adolescents and young adults with CF about the genetics of their disease continues. Furthermore, these patients are unaware of both modern technologies that could enable them to have biological children and the risk of those children having CF. This study illustrates the changing needs of patient education as medical knowledge progresses. CF patients would benefit from further genetic knowledge and counseling to enable them to make informed decisions about reproduction as they mature into adulthood.

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