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Health Status and Sociodemographic Characteristics of Adults Receiving a Cystic Fibrosis Diagnosis After Age 18 Years*

Eileen Widerman, PhD; Lois Millner, PhD; William Sexauer, MD, FCCP; and Stanley Fiel, MD, FCCP

Objectives: An increasing percentage of cystic fibrosis (CF) diagnoses are occurring in adulthood. The purpose of this study was to explore how age at diagnosis may be associated with selected disease and sociodemographic characteristics.

Design: The 1996 Cystic Fibrosis Foundation (CFF) Patient Registry data were analyzed to test for associations between age at diagnosis and selected variables. All cases involved individuals ≥ 18 years who were represented in the CFF Patient Registry for 1996. Patients were assigned to one of two groups: those diagnosed with CF after age 18 years (n = 786) and those diagnosed before 18 years (n = 6,641).

Results: In 1996, the incidence of late diagnosis was 7.8%, and the prevalence was 10.9%. The mean age of late diagnosis was 27 years. Respiratory symptoms most frequently led to late diagnosis. Patients receiving a late CF diagnosis were less likely to have alleles for Delta F508. There was no correlation between age at diagnosis and percent predicted FEV1, although patients in the late-diagnosis group were an average of 10 years older than those in the early-diagnosis group. Late diagnosis was associated with fewer complications, fewer hospitalizations, less oxygen use, fewer courses of home IV treatment, and less enzyme use. Women were most often diagnosed late. Men displayed more diversity in conditions leading to diagnosis. Psychosocially, those patients receiving late diagnoses were more likely to be college graduates, married, and employed full time. For those adults who died in 1996, there was a positive association between their age at diagnosis and age at death.

Conclusion: Those patients diagnosed with CF as adults differ, both medically and psychosocially, from those diagnosed at a younger age; these differences have implications for diagnosis, treatment, and education.

(CHEST 2000; 118:427–433)

Key words: cystic fibrosis; cystic fibrosis adult; cystic fibrosis diagnosis; cystic fibrosis late diagnosis

Abbreviations: CF = cystic fibrosis; CFF = Cystic Fibrosis Foundation; NCHS = National Center for Health Statistics

Cystic fibrosis (CF) is the most common life-threatening autosomal-recessive disorder in the white population. Although most cases are diagnosed in infancy or childhood, a small, and apparently increasing, percentage is diagnosed in adulthood.

Incidence and Prevalence of Late Diagnosis in CF

Some statistical models estimate the incidence of CF relative to live births among whites as 1:3,419, and among nonwhites as 1:12,163. The Cystic Fibrosis Foundation (CFF) maintains a registry of all patients seen at CFF-accredited care centers in the United States. In 1997, 4% of the 20,999 known patients had received their diagnoses as adults, and 918 new diagnoses of CF were reported, 65 of which (7%) were in men and women > 18 years of age. Among those patients aged ≥ 18 years in 1997, 11.5% had received their diagnoses as adults. Just 3 years earlier, new diagnoses in those patients > 18 years of age represented just 5.8% of the total of new diagnoses.

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**History and Background**

The first adult CF diagnosis was made in 1946. Since that time, a number of case studies of adults receiving a diagnosis of CF have appeared in the literature, with most published prior to the discovery of the CF gene. Their explanations for late diagnosis include the following: mild expression of the disease; unusual presentations; and the common perception of CF as a disease exclusive to childhood. Currently, missed diagnoses are more commonly attributed to the varied phenotypic expressions of hundreds of identified CF mutations.

**Relevant Literature**

Although CF medical caregivers anecdotally note that their patients who have received a late diagnosis have unique concerns, there have been few attempts to systematically describe this subpopulation of CF adults or to explore the issues that appear to be associated with late diagnosis. In a qualitative study of the phenomenon, Widerman found that her 36 subjects attributed their “missed” diagnoses to the following: physician error; mild or atypical symptoms; psychological factors (e.g., ignoring symptoms or fear); situational factors (e.g., living in a rural area or health maintenance organization restrictions); and/or moving or changing doctors frequently. Subjects described themselves as “different” from those patients receiving diagnoses in childhood and expressed a need for research-based information related to late diagnosis in CF. Widerman identified four “pathways to diagnosis,” which were determined according to the presence of symptoms and individuals’ consideration of the possibility of disease. These pathways were found to be associated with individuals’ postdiagnosis experiences and needs.

Other researchers have studied those patients who have received a diagnosis of CF as adolescents and/or adults, both in the United States and in Europe. These studies, most of which are unpublished, have involved small samples but indicate, like the study of Widerman, that receiving a diagnosis of CF after childhood may result in a range of reactions and may necessitate significant changes in lifestyle.

**Purpose**

Although a late diagnosis of CF suggests milder disease and a longer than expected life span, there are no published data documenting differences, if any, in the health statuses and salient CF and demographic descriptors of those patients receiving diagnoses as adults.

The purpose of this study was to analyze the 1996 CFF Annual Registry data for the following reasons: (1) to determine how, if at all, men and women receiving a diagnosis of CF after age 18 differed from adults receiving a diagnosis before age 18 on selected CF-related and demographic characteristics that year; and (2) to explore the extent to which identified differences were associated with gender.

**Materials and Methods**

Approval was obtained from the CFF to analyze their 1996 Patient Registry data (released in Fall 1997), which were the most recent data available when the study was undertaken (n = 20,886). These data represent responses to two-page questionnaires completed and submitted annually by accredited CFF centers. Demographic, diagnostic, genetic, mortality, health status, and other related variables are assessed for each patient seen within the preceding 2 years.

The data were transferred to a mainframe computer for analysis using appropriate software (SAS; SAS Institute; Cary, NC), selecting as cases those involving patients aged ≥18 years (n = 7,427). These patients then were assigned to one of two groups: group ED (early diagnosis), comprising adults who had received diagnoses of CF at ≤17 years (n = 6,641); or group AD (adult diagnosis), comprising adults who received diagnoses of CF at ≥18 years (n = 786). These two groups then were compared. Variables created by the CFF were utilized where possible, and new variables were created as necessary to address the issues of interest in the study. $\chi^2$ analyses, t tests, and Pearson correlation were employed. In most cases, data were further analyzed by gender, in that gender has been shown to be associated with longevity in CF.

**Results**

**Incidence and Prevalence of Adult Diagnosis in 1996**

According to the 1996 CFF Registry summary data, there were 7,435 individuals with CF who were ≥18 years of age in the United States (data available for 7,427), representing 35.6% of the CFF Registry adult cases. Of those, 813 individuals (10.9%) had received diagnoses at ≥18 years of age (data available for 786). Among individuals receiving new diagnoses that year, 71 (7.8%) received diagnoses at age ≥18 years of age.

**Sociodemographic Description**

There were significant relationships between age at diagnosis and the following sociodemographic variables: current age, race, gender, educational attainment, marital status, and employment status. In 1996, patients in group AD were older (mean, 36.83; SD, 9.58) than those in group ED (mean, 26.34; SD, 7.01; p < 0.00001). Although the patients in group AD were predominantly white (97.51%), $\chi^2$...
testing revealed a significant relationship ($p = 0.011$) between race and age at diagnosis for Asians/Pacific Islanders (late diagnosis, 31.25%) compared to blacks (late diagnosis, 7.41%) and whites (late diagnosis, 10.66%).

Over one half of adults with CF in 1996 were men, yet women were more likely to receive a late diagnosis, as illustrated in Table 1.

A greater percentage of those patients in group AD than in group ED were college graduates (40.98% vs 28.41%, respectively; $p = 0.001$), were employed full time (48.46% vs 34.85%, respectively; $p = 0.001$), and were married (72.55% vs 34.54%, respectively; $p = 0.001$). These significant differences between groups ED and AD may reflect plans made without knowledge of CF, milder disease expression, or the average age difference of 10 years between groups.

In 1996, most adults with CF had medical insurance coverage (97.50%). Those in group ED were more likely to have state-based coverage than those in group AD (31.21% vs 24.55%, respectively; $p = 0.001$), which would be expected given the eligibility requirements of these programs. Among all adults with CF, 17.72% were members of a health maintenance organization.

**Diagnosis**

For all adults included in the 1996 CFF Registry, the mean age at diagnosis was 5.51 years. The mean age at diagnosis for group AD was 27.36 years for women and 27.20 for men (difference not significant). Again, $\chi^2$ analysis revealed that women were significantly more likely to be diagnosed late than men ($p = 0.001$).

There were significant associations between age at diagnosis and the condition suggesting the diagnosis, as illustrated in Table 2.

Respiratory conditions most frequently led to the CF diagnosis for all adults. However, $\chi^2$ testing showed that a significantly larger proportion of those patients in group AD received diagnoses based on respiratory symptoms ($p = 0.001$). Those patients in group ED demonstrated more diversity in the condition suggesting the diagnosis, most likely due to the number of conditions suggestive of CF that appear in infancy and childhood. Older patients more frequently received diagnoses due to the presence of nasal polyps and the results of genotyping. Although it seems probable that those patients with family histories of CF might receive diagnoses earlier, there was no difference between the groups for this variable. The finding that the category "other conditions" more often led to late diagnosis lends support to those who say that delayed confirmation of CF may be due to atypical presentations.

Interestingly, gender differences in conditions leading to diagnosis were found only among patients in group AD, as examined in Table 3. There were no gender differences related to conditions leading to diagnosis for group ED.

$\chi^2$ analysis indicated that men were significantly more likely to receive diagnoses as a consequence of genotyping ($p = 0.013$), perhaps suggested by infertility testing. The number of men diagnosed late as a result of infertility testing could not be determined from the CFF 1996 data set because this variable was not assessed at that time.

**CF Characteristics/Health Status**

**Genotype:** Over 60% (62.34%) of those patients in group AD had been genotyped, while 49.63% of those in group ED had been genotyped ($p = 0.001$). For those patients genotyped, Table 4 illustrates the significant association between age at diagnosis and the presence of the most common mutation associated with CF, Delta F508. Significantly higher percentages of patients in group AD than in group ED were either heterozygous for Delta F508 or did not carry the allele (85.31% vs 51.30%, respectively).

**Lung Function:** The percent predicted of FVC and FEV$_1$ scores was submitted to analysis to identify differences, if any, between groups AD and ED. Results appear in Table 5. There was a significant difference in FVC scores, with patients in group AD attaining slightly higher mean percent predicted values; FEV$_1$ scores did not appear to be significantly different. However, there was a 10.2-year mean age difference between the two diagnosis groups in 1996, and this difference had to be considered in evaluating these results. In other words, despite being considerably older, the AD group showed FEV$_1$ lung function test results similar to those of the ED group.

To further investigate the relationship between FEV$_1$ scores and age at diagnosis, additional $t$ tests were run. Specific age groupings within groups ED

<table>
<thead>
<tr>
<th>Table 1—Association Between Age at Diagnosis and Gender in Adults With CF*</th>
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</thead>
<tbody>
<tr>
<td>Age at Diagnosis, %</td>
</tr>
<tr>
<td>Gender</td>
</tr>
<tr>
<td></td>
</tr>
<tr>
<td></td>
</tr>
<tr>
<td>Male (n = 4,146)</td>
</tr>
<tr>
<td>Female (n = 3,281)</td>
</tr>
</tbody>
</table>

* $\chi^2 = 23.46; df = 1; p = 0.001$.  

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and AD were compared for the FEV$_1$ variable. Results showed that, for similar age groups, group AD consistently, and significantly, displayed higher FEV$_1$ scores. The ages investigated included ages 25 through 27 years ($p = 0.0007$), 30 through 32 years ($p = 0.0044$), 35 through 37 years ($p = 0.0130$), and ≥ 38 years ($p = 0.0334$).

For all adults with CF, there were significant differences in FEV$_1$ lung function by gender. Using the FEV$_1$ categories of the CFF for illness severity (normal, mild, moderate, and severe), $\chi^2$ analyses revealed that women were more highly represented than men in the moderate category (40.72% vs 33.80%, respectively) and the mild category (24.76% vs 15.33%, respectively). For both diagnosis groups, > 60% of those patients in the severe category were men. Similar findings related to gender were found using FVC categories of illness severity. Among those in group AD, more women appeared in the mild category ($p = 0.01$) and the normal category ($p = 0.02$). The opposite was found for those patients who received diagnoses at ≤ 18 years of age.

**Culture Results:** There were a number of significant associations between age at diagnosis and culture results, as illustrated in Table 6. Differences in exposure and treatment conditions related to age at diagnosis may account for these results. Those patients in group AD may have had fewer medical contacts, less exposure to others with CF, and fewer courses of antibiotics than those in group ED.

There were no significant associations between gender and culture results within group AD.

**Complications:** $\chi^2$ testing established that a slightly higher, and significant, percentage of those patients in group AD than those in group ED experienced no complications during 1996 (75.83% vs 70.62%, respectively; $p = 0.002$). Complications that were significantly more prevalent in group ED included cirrhosis with portal hypertension ($p = 0.020$), diabetes ($p = 0.001$), and liver disease requiring consultation ($p = 0.020$). Pancreatitis was the only complication significantly more prevalent in group AD ($p = 0.001$). These findings may be related to the identified differences in genotype between the two groups. Research has shown that liver disease appears to be related to pancreatic insufficiency, and pancreatitis has been associated with mutations in the *CFTR* gene.$^{16,17}$ The actual num-

### Table 3—Gender Differences for Selected Conditions Leading to Late Diagnosis in Adults With CF (Group AD)

<table>
<thead>
<tr>
<th>Condition Suggesting Diagnosis*</th>
<th>Group ED &lt; 18 yr (n = 6,641)</th>
<th>Group AD &gt; 18 yr (n = 786)</th>
<th>p Value</th>
</tr>
</thead>
<tbody>
<tr>
<td>Acute or persistent respiratory symptoms (n = 4,327)</td>
<td>55.50</td>
<td>81.55</td>
<td>0.001</td>
</tr>
<tr>
<td>Failure to thrive malnutrition (n = 2,752)</td>
<td>40.05</td>
<td>11.70</td>
<td>0.001</td>
</tr>
<tr>
<td>Steatorrhea abnormal stools (n = 2,691)</td>
<td>38.34</td>
<td>18.45</td>
<td>0.001</td>
</tr>
<tr>
<td>Intestinal obstruction (n = 961)</td>
<td>13.64</td>
<td>7.00</td>
<td>0.001</td>
</tr>
<tr>
<td>Rectal prolapse (n = 246)</td>
<td>3.67</td>
<td>&lt; 0.01</td>
<td>0.001</td>
</tr>
<tr>
<td>Nasal polyps (n = 235)</td>
<td>2.23</td>
<td>11.07</td>
<td>0.001</td>
</tr>
<tr>
<td>Genotype† (n = 45)</td>
<td>&lt; 1</td>
<td>3.82</td>
<td>0.001</td>
</tr>
<tr>
<td>Liver problems (n = 51)</td>
<td>&lt; 1</td>
<td>1.65</td>
<td>0.001</td>
</tr>
<tr>
<td>Other§ (n = 121)</td>
<td>&lt; 1</td>
<td>8.02</td>
<td>0.001</td>
</tr>
</tbody>
</table>

*More than one condition may have been present.
†Reasons for genotype performance were not specified in CF Patient Registry.
§Conditions in this category were not specified.

### Table 4—The Association Between Delta F508 (Summary Data) and Age at Diagnosis for Adults With CF

<table>
<thead>
<tr>
<th>Summary Delta F508 Data</th>
<th>Group ED &lt; 18 yr (n = 3,296)</th>
<th>Group AD &gt; 18 yr (n = 490)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Homozygous (n = 1,677)</td>
<td>48.70</td>
<td>14.69</td>
</tr>
<tr>
<td>Heterozygous (n = 1,601)</td>
<td>30.50</td>
<td>61.02</td>
</tr>
<tr>
<td>Neither (n = 508)</td>
<td>11.80</td>
<td>24.29</td>
</tr>
</tbody>
</table>

*$\chi^2 = 207.59$; df = 2; $p = 0.001$. 
ber of those patients receiving diagnoses late who were treated for complications was quite low, with percentages ranging from 0.13% for gallbladder disease to 5.73% for diabetes.

National Center for Health Statistics Percentages for Height and Weight: \( \chi^2 \) tests revealed that, using National Center for Health Statistics (NCHS) standards, those patients in group AD were significantly taller (\( p = 0.001 \)) and heavier (\( p = 0.001 \)) than those in group ED. Within group AD, 51.59% of patients were at \( \leq 24\% \) of the NCHS standard weight (compared to 67.33% of those receiving a diagnosis in group ED); 28.23% were at \( \leq 24\% \) of the NCHS standard height (compared to 45.03% of those in group ED).

Enzyme Supplement Use: Table 7 shows the significant differences in reported use of enzymes for adults with CF by age at diagnosis. These data indicate that those patients in group ED were more likely to take pancreatic enzyme supplements. Because pancreatic sufficiency is thought to be related to genotype, the association between genotype and enzyme use was tested using \( \chi^2 \) analysis. There was, for both groups, a significant positive association between enzyme use and the presence of one or two alleles for Delta F508 (\( p = 0.001 \)). Of those patients in group AD not taking enzymes, only 1.32% were heterozygous for Delta F508.

Other CF Care Variables: Patients in group AD had fewer hospitalizations (\( p = 0.0001 \)) in 1996 than those in group ED. Those patients in group AD who were hospitalized had a mean duration hospital stay that was a week less than those in group ED (\( p = 0.0004 \)). Patients in group AD also had fewer office visits (\( p = 0.0025 \)) and fewer mean days of IV treatment (\( p = 0.037 \)) than did those in group ED during 1996. There were no significant differences between groups ED and AD for oxygen use, supplemental feeding, or transplantation.

Longevity

Among the 391 reported deaths across all age groups in 1996, there was a significant positive correlation between age at diagnosis and age at death (\( r = 0.57; p = 0.0001 \)). Twenty-four of the 283 adults who died in 1996 were members of group AD, and there was also a positive, significant relationship for this group between age at diagnosis and age at death (\( r = 0.60; p = 0.0001 \)). The adult death rate was 3.81% that year. For patients in group AD, the death rate was 3.05%, and for those in group ED it was 3.90%.

For all those patients with CF, the mean age at death for 1996 was 28.74. The mean age at death for those within group ED was 27.53 years (SD, 7.48 years); the mean age for those within group AD was 41.88 years (SD, 9.76 years) (\( p = 0.0001 \)). There was no significant gender difference in mean age at death for those patients in group AD (men, 41.85 years; women, 41.90 years).

Cause of Death: Regardless of age at diagnosis, respiratory conditions were the primary cause of death in adulthood for approximately 75% of those

<table>
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<tr>
<th>Table 5—The Relationship between Age at Diagnosis and Lung Function in Adults With CF*</th>
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<tbody>
<tr>
<td>Lung Function Indicator</td>
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<tr>
<td>-------------------------</td>
</tr>
<tr>
<td>FEV1, % predicted</td>
</tr>
<tr>
<td>FVC, % predicted</td>
</tr>
<tr>
<td>*Values given as mean (SD), unless otherwise indicated.</td>
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<tr>
<th>Table 6—Significance Differences in Culture Results Related to Age at Diagnosis in Adults With CF</th>
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<tbody>
<tr>
<td>Culture Result</td>
</tr>
<tr>
<td>----------------</td>
</tr>
<tr>
<td>Pseudomonas aeruginosa (n = 4,737)</td>
</tr>
<tr>
<td>Burkholderia cepacia (n = 391)</td>
</tr>
<tr>
<td>Normal flora (n = 236)</td>
</tr>
<tr>
<td>Nontuberculous mycobacterium (n = 85)</td>
</tr>
<tr>
<td>Klebsiella (n = 40)</td>
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<tr>
<th>Table 7—Use of Enzyme Supplements by Age at Diagnosis for Adults With CF*</th>
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<tr>
<td>Age at Diagnosis, %</td>
</tr>
<tr>
<td>-------------------</td>
</tr>
<tr>
<td>Use of Enzyme Supplements</td>
</tr>
<tr>
<td>Use pancreatic enzymes</td>
</tr>
<tr>
<td>Do not use pancreatic enzymes</td>
</tr>
<tr>
<td>*( \chi^2 = 376.66; \text{df} = 1; p = 0.001 ).</td>
</tr>
</tbody>
</table>
who died in 1996, followed by deaths related to transplantation (approximately 15%). There were no significant differences between groups ED and AD related to cause of death, and there were no significant gender differences within the groups.

**Discussion**

The results of this study suggest that those patients who received diagnoses of CF as adults are a unique subgroup of adults with CF. Adults who received diagnoses late, as represented in the 1996 CFF Registry, were more likely to be college graduates, employed full time, and married. They experience fewer complications, culture higher rates of normal flora, and live longer than do those who receive diagnoses under age 18 years. The average age at diagnosis of group AD in this study (27 years) is one explanation for these differences. Life plans and decisions were made without awareness (or confirmation) of having a chronic, progressive disease. Another explanation might be that the milder CF expression found among those patients in group AD allowed them to be more active.

Similar to the findings of Lester et al., there was a strong association between genotype and age-at-diagnosis in this study. It is certainly arguable that the less common mutations more often carried by those patients receiving diagnoses late could be responsible for the greater variance in conditions leading to diagnosis and the overall better indexes of health (i.e., hospitalizations, office visits, complications experiences, lung function, etc) found among those patients in group AD in this study. Research is suggesting genetic links for nontuberculous mycobacteria and pancreatitis. However, as researchers caution, relationships between the CFTR genotype and clinical manifestations of CF are multiple and complex.

Gender differences, both within the subpopulation of those receiving diagnoses as adults as well as between those receiving diagnoses who were > 18 years of age and < 18 years of age are also worthy of note. Men with CF have long been recognized as having milder disease expression and longer life expectancy. This study found that this association did not hold for those receiving diagnoses late. Women were more likely to receive diagnoses late and to receive them due to respiratory conditions. Women were also more likely to evidence milder disease as categorized by the CFF using FVC and FEV₁ scores. Although the CFF Registry did not assess congenital bilateral absence of the vas deferens as a reason for diagnosis, it would nevertheless be expected that a number of men in group AD would have received diagnoses via this route. Since men in whom congenital bilateral absence of the vas deferens was diagnosed present with mild, or no, lung disease, their presence in sufficient numbers should have skewed the gender comparisons of health indicators to favor men. Interestingly, this did not occur.

Since there is no evidence of gender differences in genotype, identified differences may be due to sociocultural factors. As children, boys with symptoms would be taken by parents for care; in adulthood, men are characterized as being more hesitant to seek medical care, perhaps explaining why women would have milder disease on diagnosis. It has been argued that physicians attribute less importance to women’s complaints than to those of men, possibly accounting for why it takes longer for CF to be diagnosed in women, although the milder disease found among women who received late diagnoses may also be a factor.

The study’s findings provide empirical support for what persons who are diagnosed late and their physicians have long hypothesized: those who receive diagnoses late are “different” from those who receive diagnoses in preadulthood.

**Limitations of the Study**

Although the CFF Patient Registry data yielded a comprehensive and systematic description of adults who received diagnoses of CF after age 18, there are limitations to this study. The database did not include individuals who received medical care outside the CFF network of specialty centers. In 1997, Widerman found that 13.8% of the 36 men and women she interviewed did not receive care from a CFF center and that an additional number only attended a CFF center yearly for assessment. Those not included in the CFF database may differ in important sociodemographic and/or CF characteristics from those who were. Second, the cross-sectional study design permitted associations only. Cause-and-effect relationships could not be established, and the results apply solely to the 1996 CFF Patient Registry participants as a group. Third, because the data are submitted by CF centers and are taken from medical records (which enhances validity for some variables), their accuracy and completeness are not confirmed by patients. Finally, the study was limited to the variables assessed by the CFF Registry instrument.

**Conclusions and Recommendations**

**Implications of Study**

CF caregivers need to be aware that adults who receive late diagnoses of CF have unique character-
istics that suggest unique needs in terms of education, treatment, and support. Psychosocially, those who receive diagnoses as adults must come to terms with having a life-threatening disease and must reconcile CF treatment and care with the demands of established lifestyles. Educationally, they are likely to require assistance applying CF information grounded in the conditions of those receiving diagnoses in childhood or infancy to adult medical situations. Medically, they need treatment that acknowledges their gender-related and other differences in CF expression and progression. The fact that a significant number of patients who are diagnosed late are married suggests that treating these patients should involve both recognizing how families respond to the diagnosis and recommending CF testing and/or screening for relatives.

**Recommendations**

1. Non-CF physicians should be reminded of the following: that a diagnosis of CF is possible in an adult; that patients who are part of racial and ethnic minorities can have CF; and that adults with CF can present with a range of symptoms, conditions, and/or complications, some of which are gender-related.

2. This study explored late diagnosis in adulthood, which was arbitrarily defined as age ≥18 years. Yet, between 1993 and 1996, the rate of CF diagnosis in adolescents (aged 12 through 17 years) increased from 8.8 to 13.9% as compared to 6.0 to 7.4% for those ≥18 years (S. Fitzsimmons, PhD; personal communication; March 24, 1998). This phenomenon is worthy of study, particularly to identify the differences, if any, among CF and other characteristics of adolescents and adults who received late diagnoses and patients <12 years of age who received diagnoses of CF.

**ACKNOWLEDGMENTS:** The authors acknowledge Barbara Palys, Chairperson, International Association of CF Adults, for her assistance in editing and guiding the study from the perspective of those receiving late diagnoses of CF. Roslyn Goren, MA, Manager, Statistical Applications, Temple University, provided invaluable statistical consultation and guidance. The support of the CFF for generously sharing the Patient Registry Data is also greatly appreciated.

**REFERENCES**
