Long-term Assessment of Quality of Life in Primary Ciliary Dysfunction
Time for New Tools?

To the Editor:

We deeply commend Quittner et al1 in a recent issue of CHEST (August 2014) on their effort in developing the first disease-specific patient-reported outcome (PRO) measure validating the Quality of Life Questionnaire-Bronchiectasis (QOL-B). This is an area that we actively pursued in primary ciliary dyskinesia (PCD), a genetic cause of chronic suppurative lung disease with great impact on health because of abnormal mucociliary clearance leading to recurrent airway infections and bronchiectasis.2-4 Sensitive measures for tracking PCD lung disease progression have serious limitations in clinical practice since changes in spirometry may not be apparent, and repeated high-resolution CT scans increase the risk of ionizing radiation exposure.5

In a prospective, 1-year study of 20 subjects with PCD (median age, 16.9 years; range, 12-33.4 years) we verified whether three of the most widespread PROs used for assessing quality of life (QoL) in respiratory disorders (St. George’s Respiratory Questionnaire [SGRQ], Leicester Cough Questionnaire [LCQ], and Medical Outcomes Study Short Form 36 [SF36]) correlated with spirometry or 6-min walk test (6MWT). Patients completed SGRQ, LCQ, and SF36 and performed spirometry and 6MWT at scheduled visits (T₀, T₁₂). Table 1 summarizes the main findings.

During the study period, three respiratory exacerbations5 (range, 0-7) that required systemic antibiotics occurred. Eight patients (40%) needed four or more antibiotic courses. At baseline, none of the PROs was related to age at diagnosis and age at symptoms onset.

FEV₁, FVC, FEV₁/FVC, and forced expiratory flow at 25% to 75% of FVC, as well as 6-min walk distance, were not significantly related to any of the QoL assessment tools at T₀ and T₁₂. Over the 12-month period, no significant changes were found in any of the QoL outcomes considered or in spirometry or 6MWT. Despite the small sample size, our results show that SGRQ, LCQ, and SF36 are unrelated to the commonly accepted disease outcomes in PCD. Indeed, we provide the first demonstration, to our knowledge, that these tools are also suboptimal to longitudinally track QoL in PCD.

No PCD-specific QoL questionnaire has ever been validated, and the sensitivity of currently used questionnaires in detecting long-term QoL changes has not been

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**TABLE 1** Patient-Reported Outcomes, 6-Min Walk Test, Spirometry, and Sputum Culture Results From Patients With PCD at the Study Time Points Over 1-Year Period

<table>
<thead>
<tr>
<th>Outcome</th>
<th>T₀</th>
<th>T₁₂</th>
<th>P Value *</th>
</tr>
</thead>
<tbody>
<tr>
<td>SGRQ total score</td>
<td>15.2 (4.2-65)</td>
<td>14.1 (2.3-50.8)</td>
<td>.7</td>
</tr>
<tr>
<td>LCQ total score</td>
<td>16.9 (6.8-20.9)</td>
<td>18.2 (10.8-21)</td>
<td>.2</td>
</tr>
<tr>
<td>SF36 PCS</td>
<td>50 (30.1-60)</td>
<td>51.4 (26.4-60)</td>
<td>.4</td>
</tr>
<tr>
<td>SF36 MCS</td>
<td>55.3 (22.1-62.2)</td>
<td>55.1 (38.4-64)</td>
<td>.6</td>
</tr>
<tr>
<td>6MWD, % predicted</td>
<td>77.5 (60-89)</td>
<td>72 (60-84)</td>
<td>.2</td>
</tr>
<tr>
<td>FEV₁, % predicted</td>
<td>86.5 (45-117)</td>
<td>87 (41-117)</td>
<td>.8</td>
</tr>
<tr>
<td>FVC, % predicted</td>
<td>93 (64-133)</td>
<td>98.5 (64-134)</td>
<td>.8</td>
</tr>
<tr>
<td>FEV₁/FVC, %</td>
<td>74.5 (52-85)</td>
<td>73.5 (51-99)</td>
<td>.9</td>
</tr>
<tr>
<td>FEF₂₅₋₇₅, % predicted</td>
<td>56 (16-90)</td>
<td>54 (13-120)</td>
<td>.8</td>
</tr>
<tr>
<td>Positive sputum cultures, %</td>
<td>70</td>
<td>30</td>
<td>.03</td>
</tr>
</tbody>
</table>

Data are presented as median and range values. 6MWD = 6-min walk distance; FEF₂₅₋₇₅ = forced expiratory flow at 25% to 75% of FVC; LCQ = Leicester Cough Questionnaire; MCS = mental component summary; PCD = primary ciliary dyskinesia; PCS = physical component summary; SF36 = Short Form-36; SGRQ = St. George’s Respiratory Questionnaire.

* Mann-Whitney U test or Fisher exact test.  
  † Including Haemophilus influenzae, Pseudomonas aeruginosa, Staphylococcus aureus, Streptococcus pneumoniae, and Aspergillus niger.
evaluated so far. Pending the validation of QOL-B for measuring symptoms, functioning, and QoL also in PCD, and for evaluating the efficacy of new therapies, caution in the use of tools that are not disease specific for PCD is mandatory. According to what was reported in patients with bronchiectasis, QOL-B might be considered as an efficacy end point also in PCD clinical trials. We believe the need for development of a PCD-specific instrument for longitudinal QoL assessment on larger study populations is urgent.

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References