**Table 1:** Demographic, diagnostic and clinical characteristics of adult patients participating in the study

|  |  |  |
| --- | --- | --- |
| **Parameter** | **Adult PCD patients (n=31)** | |
| ***Demographic information*** |  |  |
| Current Age\* | 33.6 (22.2-50.9) |  |
| Gender (female) | 18/31 | 58.1% |
| Age at Presentation\* | 23.79 (17.8-45.1) |  |
| Situs Abnormalities | 14/31 | 45.2% |
| ***Diagnostic characteristics*** |  |  |
| Nasal Nitric Oxide (nl/min)\* | 24.9 (13.0-48.0) |  |
| TEM result |  |  |
| Normal TEM | 8/31 | 25.8% |
| ODA+IDA | 9/31 | 29.0% |
| ODA only | 6/31 | 19.4% |
| CP/ IDA+MD | 8/31 | 25.8% |
| Other | 0/31 | 0% |
| HSVM result |  | |
| Normal HSVM | 0/31 | 0% |
| Immotile/Almost Immotile | 13/31 | 41.9% |
| Extremely stiff due to reduced ciliary bending | 8/31 | 25.8% |
| Stiff Beating pattern | 3/31 | 9.7% |
| Circular Pattern | 7/31 | 22.6% |
| Other | 0/31 | 0% |
| ***Clinical Characteristics*** |  |  |
| Chronic Rhinorrhoea | 31/31 | 100% |
| Chronic wet cough | 31/31 | 100% |
| History of NRDS | 14/31 | 45.2% |
| History of nasal polyps | 8/30 | 26.7% |
| History of Pneumonia | 12/29 | 41.4% |
| History of Heamoptysis | 3/31 | 9.7% |
| History of lung resection | 5/31 | 16.1% |

\* Median and Interquartile Range TEM: Transmission Electron Microscopy, HSVM: High Speed Video Microscopy, ODA+IDA: Combined Outer Dynein Arm defect and Inner Dynein Arm defect, ODA: Isolated Outer Dynein Arm defect, CP: Central Pair defect, IDA + MD: Inner Dynein Arm and Microtubular Disorganisation defect, NRDS: Neonatal Respiratory Distress Syndrome

**Table 2:** Internal consistency of QoL-PCD scales measured by Cronbach’s α and test–retest reliability measured by ICC.

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| **Scale** | **# Items** | **Scale Median**  **(IQR)** | **Cronbach’s α** | **ICC (95%)** |
| **Physical Functioning** | 5 | 80.0  (46.7-93.3) | 0.880 | 0.910  (0.803-0.959) |
| **Vitality** | 3 | 66.7  (44.4-77.8) | 0.463 | 0.817  (0.598-0.916) |
| **Emotional Functioning** | 5 | 86.7  (63.7-93.3) | 0.642 | 0.893  (0.766-0.951) |
| **Treatment Burden** | 4 | 66.7  (41.7-83.3) | 0.809 | 0.670  (0.167-0.869) |
| **Social Functioning** | 3 | 33.3  (0.0-66.7) | 0.681 | 0.768  (0.491-0.894) |
| **Role** | 4 | 66.7  (58.3-83.3) | 0.542 | 0.902  (0.785-0.955) |
| **Health Perspective** | 4 | 50.0  (41.7-66.7) | 0.639 | 0.670  (0.277-0.850) |
| **Upper Respiratory Symptoms** | 4 | 58.3  (41.7-83.3) | 0.687 | 0.853  (0.678-0.933) |
| **Lower Respiratory Symptoms** | 6 | 61.1  (44.4-72.2) | 0.784 | 0.895  (0.769-0.952) |
| **Hearing Symptoms** | 2 | 66.7  (58.3-100) | 0.661 | 0.838  (0.645-0.926) |

**Table 3:** Construct validity of QoL-PCD assessed through the association of specific QoL-PCD scale values with Gender, Age, FEV1 and FVC.

|  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- |
| **Parameter** | **Physical Functioning** | | | **Social Functioning** | | **Lower Respiratory Symptoms** | |
| **Value** | **Sig.** | **Value** | | **Sig.** | **Value** | **Sig.** |
| **Male** | 86.7  (60.0-93.3) | 0.060 | 44.4  (11.1-66.7) | | 0.395 | 66.7  (52.8-75.0) | 0.258 |
| **Female** | 60.00  (38.3-88.3) | 27.8  (0.00-55.6) | | 61.1  (37.5-68.1) |
| **<33.6 years\*** | 80.00  (54.9-93.3) | 0.280 | 50.00  (33.3-66.7) | | 0.022 | 63.9  (45.8-76.4) | 0.232 |
| **>33.6 years** | 60.0  (40.0-93.3) | 22.2  (0.00-44.4) | | 61.10  (33.3-66.7) |
| **<-2 FEV1\*** **z-score** | 56.7  (34.9-80.0) | 0.023 | 27.8  (0.00-52.80) | | 0.316 | 50.00  (38.9-65.3) | 0.016 |
| **≥-2 FEV1** **z-score** | 93.3  (66.7-93.3) | 44.4  (22.2-66.7) | | 66.7  (61.1-77.8) |
| **<-1.47 FVC\*** **z-score** | 60.00  (33.3-80.0) | 0.110 | 31.3  (0.0-55.6) | | 0.367 | 55.6  (38.9-66.7) | 0.112 |
| **≥- 1.47 FVC** **z-score** | 86.7  (48.3-93.3) | 38.8  (13.9-66.7) | | 66.70  (51.4-77.8) |
| \*Median Age, Median FEV1 Z-Score, Median FVC Z-score | | | | | | | |

**Table 4:** Correlation coefficients between scales from QoL-PCD and the scales of the generic SF-36 HRQoL questionnaire

|  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- |
|  | **SF36**  **Physical**  **Functioning** | **SF36**  **Role**  **Physical** | **SF36**  **Bodily**  **Pain** | **SF36**  **General**  **Health** | **SF36**  **Vitality** | **SF36**  **Social**  **Functioning** | **SF36**  **Role**  **Emotional** | **SF36**  **Mental**  **Health** |
| **Physical Functioning** | 0.776\* | 0.645\* | 0.351 | 0.407\* | 0.665\* | 0.650 | 0.605\* | 0.648\* |
| **Vitality** | 0.804\* | 0.696\* | 0.541\* | 0.298 | 0.703\* | 0.710 | 0.733\* | 0.588\* |
| **Emotional Functioning** | 0.675\* | 0.475\* | 0.406\* | 0.333 | 0.711\* | 0.621 | 0.530\* | 0.706\* |
| **Treatment Burden** | 0.113 | -0.077 | 0.013 | -0.061 | -0.015 | -0.050 | 0.066 | 0.033 |
| **Role** | 0.509\* | 0.469\* | 0.495\* | 0.288 | 0.460\* | 0.497\* | 0.403 | 0.403 |
| **Social Functioning** | 0.325 | 0.240 | 0.251 | 0.132 | 0.175 | 0.238 | 0.157 | 0.224 |
| **Health Perspective** | 0.307 | 0.209 | 0.268 | 0.461\* | 0.244 | 0.190 | 0.084 | 0.212 |
| **Upper Respiratory** | 0.534\* | 0.577\* | 0.352 | 0.478\* | 0.521\* | 0.489\* | 0.401 | 0.485\* |
| **Lower Respiratory** | 0.619\* | 0.644\* | 0.649\* | 0.635\* | 0.682\* | 0.571\* | 0.519 | 0.706\* |
| **Hearing Symptoms** | 0.226 | 0.295 | 0.207 | 0.291 | 0.168 | 0.180 | 0.241 | 0.034 |
| \*Significant at the 0.05 confidence level, Highlighted cells correspond to a-priori assumed positive relationships | | | | | | | | | |