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CASE REPORT

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Olfaction in primary ciliary dyskinesia

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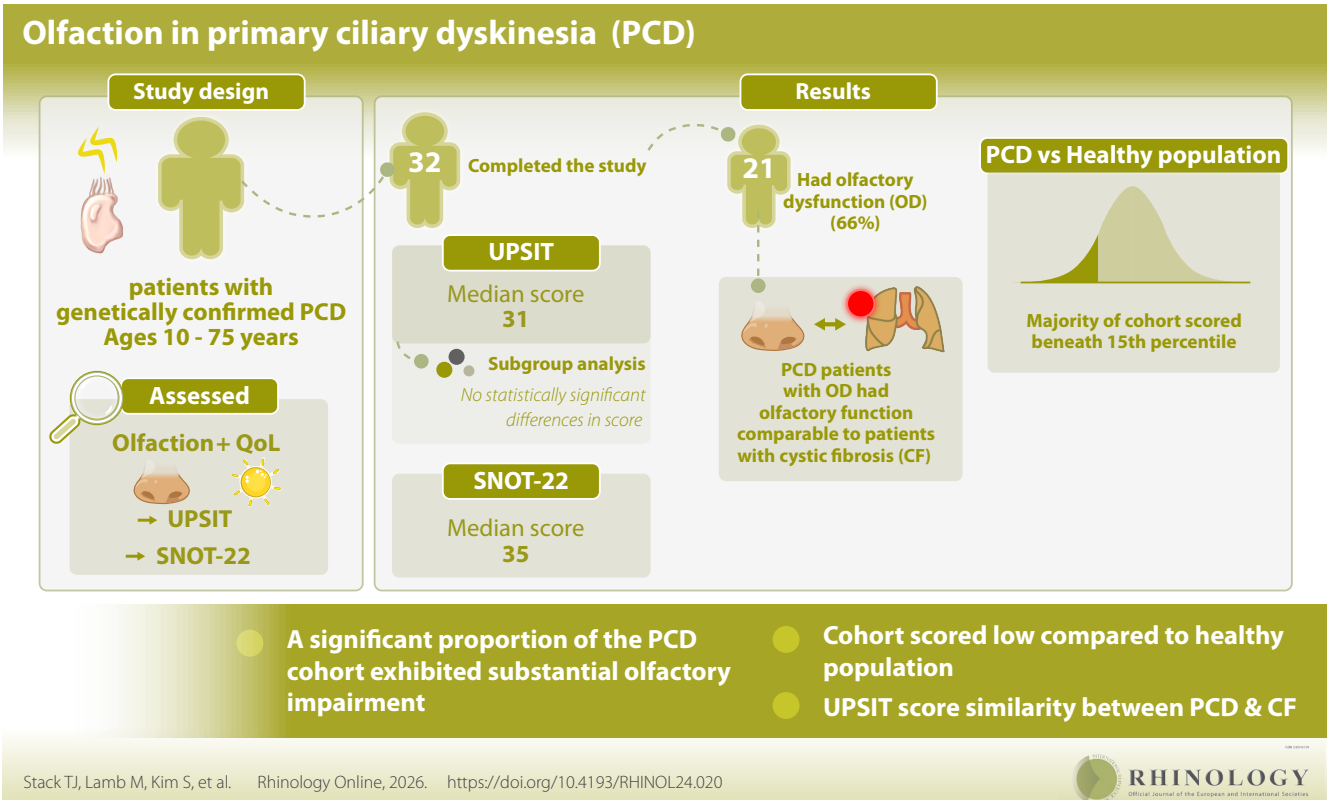

Abstract

Background: Primary ciliary dyskinesia (PCD) is an inherited ciliopathy characterized by bronchiectasis, organ laterality, and sinusitis. Chronic rhinosinusitis (CRS) is observed in most patients with PCD. In the general CRS population, olfactory dysfunction is a common complaint; however, there is little knowledge about olfaction dysfunction in PCD patients. This study explores the sinonasal impacts, specifically olfaction and sinonasal quality of life (QoL), in individuals with PCD.

Methodology: A cross-sectional study including patients with genetically confirmed PCD between ages 10 - 75 years. Olfaction and QoL were assessed by the University of Pennsylvania Smell Identification Test (UPSIT) and Sino-nasal Outcome Test (SNOT-22).

Results: Data included 32 participants who completed the study. The median UPSIT score was 31, (IQR = 6.5). The median SNOT-22 score was 35 (IQR = 25). Subgroup analysis revealed no statistically significant differences in UPSIT scores. The majority of the PCD cohort had olfactory dysfunction (66%, n = 21). PCD patients exhibited comparable olfactory function to those with cystic fibrosis. Compared to healthy population data, most of the cohort scored below the 15th percentile.

Conclusions: Previous studies suggest olfactory impairment secondary to PCD, however; objective data is limited on olfactory function in patients with PCD. Our data show a significant proportion of the PCD cohort exhibited substantial olfactory impairment. Additionally, the cohort scored relatively low compared to the healthy population. Our data suggest that UPSIT scores of the PCD cohort were similar to a cystic fibrosis cohort, suggesting that olfactory dysfunction is similar between these two mucociliary diseases.

Key words: sinusitis, olfaction disorders, nasal polyps

Introduction

Primary ciliary dyskinesia (PCD) is a genetic disorder characterized by bronchiectasis, organ laterality defects, and chronic rhinosinusitis (CRS) due to loss in mucociliary clearance [1]. CRS is nearly universal in PCD patients; however, little is known about olfaction in individuals with PCD. Given the negative impact of olfactory dysfunction (OD) on quality of life (QoL) [2], it is important to understand olfactory function in these individuals.

Although research is limited, studies suggest high rates of OD in patients with PCD-related CRS compared to CRS or healthy controls [3, 4]. This investigation aimed to characterize OD in PCD patients, quantifying the severity and impact on QoL compared to a healthy population. Secondarily, this data was compared with previously published cystic fibrosis (CF) olfaction data [5], as PCD and CF are genetic diseases that affect mucociliary clearance. By addressing this gap in knowledge, our findings will contribute to the comprehension of severity of OD in PCD.

Methods

Population and study design

This IRB-approved prospective cohort study included PCD patients aged 10 or older. Patients with neurodegenerative disease, head trauma, congenital conditions associated with anosmia, or COVID-19-related OD were excluded. Subjects completed the UPSIT and SNOT-22 questionnaires. Demographic, clinical information, and baseline sinus treatment regimens were abstracted. We also used prior data from a study focused on CF olfaction.

University of Pennsylvania Smell Identification Test

The primary endpoint of this study was the University of Pennsylvania Smell Identification Test (UPSIT) scores. The UPSIT is a 40-item "scratch and sniff" assessment. Scores range from 0-40, with lower scores indicating worse olfactory function [6].

Statistical analysis

Data normality was assessed using Shapiro-Wilk test, revealing a non-parametric distribution. Continuous variables were expressed as medians and interquartile range (IQR), while categorical variables were presented as absolute counts (n) and relative frequencies (%). UPSIT scores were categorized [6] and compared to baseline (treatment-naive) CF UPSIT scores, as reported in a previous study [5]. Data analysis was performed using RStudio (Version2023.06.0+421) (RStudio Inc., Boston, MA, USA).

Results

Of the 32 participants, 59% were female, 94% self-identified as white, 66% used a steroid nasal spray, and 44% had a history of sinus surgery. PCD demographic characteristics (ST1) were not statistically different than the CF group (ST2) [5]. The median UPSIT score was 31, (IQR = 6.5) and the median SNOT-22 score was 35 (IQR = 25) (Table 1). The majority of the PCD cohort had some degree of OD (66%, n=21), while 34% (n=11) exhibited normosmia (Table 2). On the SNOT-22, most respondents (59%) reported some degree of a 'problem' regarding decreased sense of taste/smell (mean=1.4, ST3). Of those who denied olfaction issues on the SNOT-22 (n=13), six had OD. Of those who reported a problem with smell (n=19), 15 had OD.

Linear regression analyzed the correlation between the reported loss of smell/taste SNOT-22 question and UPSIT scores, showing that individuals who report no loss of smell or taste have a predicted UPSIT score of approximately 31.6 (coef = -1.98, R2=0.13, $p < 0.043$). Compared to previous data evaluating olfaction in a CF cohort (n = 34) [5], PCD patients' olfactory function was not significantly different (p=0.46) (Table 2). The median UPSIT score in the CF cohort was 29 (IQR=10), and median score for this PCD cohort was 31, (IQR=6.5). Percentiles, based on healthy population data, were determined for each individual UPSIT score (ST4). The median percentile for this PCD cohort was 6.5 (IQR=12), with the majority (23/32) scoring below 15th percentile.

Discussion

The primary objective of this study was to assess the olfactory function in individuals with PCD. Previous literature suggests OD rates of 52% [7] - 74% [3], consistent with our results. UPSIT scores did not differ significantly based on sex, ultrastructural features of cilia, or history of sinus surgery, suggesting that these factors may not be major determinants of OD in PCD patients.

By determining percentiles for each UPSIT score using healthy population data, the study provides a benchmark for evaluating

Table 1. PCD olfaction data. UPSIT scores were categorized into percentiles, which consider age and sex.

Characteristic	PCD Cohort, N = 32 [a]
UPSIT Score	31 (28, 34)
SNOT-22 Score	35 (22, 47)
SNOT-22 Item: Decreased sense of taste/smell	
No Problem (0)	13 (41%)
Very Mild Problem (1)	6 (19%)
Mild or slight Problem (2)	5 (16%)
Moderate Problem (3)	4 (13%)
Severe Problem (4)	4 (13%)
UPSIT Percentile	
< 5%	12 (38%)
5-15%	11 (34%)
15-25%	3 (9.4%)
25-35%	2 (6.3%)
Over 35%	4 (13%)

[a] Median (IQR); n (%). Percentile data is derived from healthy population data from the UPSIT manual nomogram [6]. Abbreviations: UPSIT, University of Pennsylvania Smell Identification Test; SNOT-22, Sino-Nasal Outcome Test-22; PCD, primary ciliary dyskinesia.

olfactory performance in PCD patients. The median percentile of 6.5, indicates that, on average, the PCD cohort scored low compared to the healthy population. A large cohort study (n=384) found that 24% of adults with PCD report subjective OD [8]; however, our findings suggest that a significant proportion of the PCD cohort exhibits substantial olfactory impairment, falling below the range of olfactory function observed in the healthy population. Furthermore, our data revealed that nearly half (6/13) of the PCD subjects who denied smell loss had hyposmia based on UPSIT scoring.

The PCD UPSIT scores showed similarities to a CF cohort with comparable distributions of sex and age. Our PCD-cohort median UPSIT score (31) is comparable to previously published CF-cohort UPSIT scores of 29 [5] and 30.6 [9], suggesting OD is similar between these two mucociliary diseases.

Despite the valuable insights gained from this study, the sample size was limited, potentially limiting the generalizability of the findings. Additionally, we used a published cohort of CF patients, and normative data used in the development of the UPSIT as a control group. Future studies with larger sample sizes and control groups could provide more comprehensive insights into OD in PCD.

Table 2. UPSIT score category comparisons between individuals with PCD and CF.

	CF, N = 34 [a]	PCD, N = 321	p-value [b]
UPSIT Category			0.5
Normosmia	9 (26%)	11 (34%)	
Mild Microsmia	6 (18%)	8 (25%)	
Moderate Microsmia	7 (21%)	6 (19%)	
Severe Microsmia	9 (26%)	3 (9.4%)	
Total Anosmia	3 (8.8%)	4 (13%)	

[a] n (%); [b] Fisher's exact test; The baseline CF scores were taken in individuals who had not received cystic fibrosis transmembrane conductance regulator (CFTR) modulator therapies. Existing guidelines categorize UPSIT scores into different olfactory diagnoses. For adults, the classifications include normosmia: UPSIT ≥34 (males) or ≥35 (females), mild microsmia: 30–33 (males) or 31–34 (females), moderate microsmia: 26–29 (males) or 26–30 (females), severe microsmia: 19–25, and total anosmia: ≤18). Abbreviations: UPSIT, University of Pennsylvania Smell Identification Test; CF, cystic fibrosis; PCD, primary ciliary dyskinesia.

Conclusion

Our study sheds light on the OD experienced by individuals with PCD. The majority of PCD patients exhibited some degree of OD in PCD-related CRS appears comparable to CF-related CRS. More research is needed to understand the impact of OD on PCD patients and to develop interventions that cater to their unique needs for an improved quality of life.

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Ethics approval and consent to participate

Study approval statement: This study protocol was reviewed and approved by the University of North Carolina at Chapel Hill Office of Human Research Ethics approval number #22-2104. *Consent to participate statement*: The ethics approval board determined that phone consent was appropriate for this remote study. Verbal, informed consent was obtained from participants (or their parent/legal guardian/next of kin) to participate in the study. Written consent was not obtained for participation in this study, and this was approved by UNC-Chapel Hills IRB.

Consent for publication

Not applicable.

Authorship contribution

TJS: initial manuscript draft, data analysis, data collection; ML: manuscript revision, data collection; SK: manuscript revision, data analysis; IM, BDT, CKC, CSE, KS, NC, SD, BAS: manuscript revision; AJK: project conceptualization, initial manuscript draft, manuscript revision.

Availability of data and materials

The datasets used and/or analysed during the current study are available from the corresponding author upon reasonable request.

Conflict of interest

The authors declare that they have no competing interests.

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SUPPLEMENTARY MATERIAL

Table S1. Baseline characteristics for the PCD cohort.

Characteristic	N = 32 [a]
Sex	
Female	19 (59%)
Male	13 (41%)
Age	23 (19, 34)
Race	
Multiple Races	1 (3.1%)
Black or African American	1 (3.1%)
White	30 (94%)
Ethnicity	
Hispanic, Latino, or Spanish Origin	2 (6.3%)
Non-Hispanic, Latino or Spanish Origin	27 (84%)
Unknown	3 (9.4%)
Ultrastructural Feature of Cilia by Electron Microscopy	
IDA/MTD	10 (31%)
Normal	1 (3.1%)
ODA	15 (47%)
ODA mutation with normal ultrastructure	2 (6.3%)
ODA/IDA	4 (13%)
Rinses	
Budesonide	2 (6.3%)
Certirizine	2 (6.3%)
Saline	13 (41%)
Tobramycin	1 (3.1%)
Spray	
Steroid	21 (66%)
History of Sinus Surgery	14 (44%)

[a] n (%); Median (IQR); Abbreviations: PCD, primary ciliary dyskinesia; ODA, outer dynein arm; IDA, inner dynein arms; MTD, microtubular disorganization.

Table S2. The PCD and CF cohorts were comparable in age and sex, however the CF cohort had a higher percentage of those with sinus surgery ($p = 0.045$).

Characteristic, n (%)	PCD	CF	p-value*
Age (>21)	17 (60%)	24 (71%)	0.43
Sex, female	19 (59%)	13 (38%)	0.21
History of sinus surgery	14 (44%)	24 (71%)	0.045

*Fisher's Exact Test. Abbreviations: CF, cystic fibrosis; PCD, primary ciliary dyskinesia.

Table S3. PCD SNOT-22 scores by domain.

Symptom	Mean Score
Need to blow nose	2.8
Nasal Blockage	2.8
Sneezing	1.3
Runny nose	2.6
Cough	3.1
Post-nasal discharge	2.1
Thick nasal discharge	2.3
Ear fullness	2
Dizziness	0.8
Ear pain	1.1
Facial pain/pressure	1.5
Decreased sense of smell/taste	1.4
Difficulty falling asleep	1.4
Wake up at night	1.4
Lack of a good night's sleep	1.6
Wake up tired	2.1
Fatigue	1.9
Reduced productivity	1.3
Reduced concentration	1.3
Frustrated/restless/irritable	0.9
Sad	1
Embarrassed	1.1

Table S4. Raw UPSIT scores for each PCD patient and the respective percentiles based on the UPSIT manual normogram.

UPSIT	Percentile (%)
33	17
28	5
34	17
11	5
11	5
29	5
37	50
35	41
37	54
9	5
31	10
31	6
34	6
34	11
27	5
35	31
31	6
28	14
24	5
19	5
35	14
32	6
31	7
36	31
35	23
37	50
29	5
28	5
25	5
13	5
34	9
32	10

Abbreviations: PCD, primary ciliary dyskinesia.