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Introduction

Ear problems are common in primary ciliary dyskinesia (PCD), but little is known on their spectrum and severity. We aimed to characterise otologic disease in PCD patients using data from the ENT Prospective International Cohort of PCD patients (EPIC-PCD), a multicentre prospective observational clinical cohort.

Methods

We prospectively included patients with a routine ENT examination and a completed FOLLOW-PCD symptoms questionnaire at the same visit. We compared reported ear symptoms and otologic clinical findings between children and adults.

Results

We included 397 patients (211 males) with median age 15 years (range 0 – 73) from 12 centres from 10 countries (Table 1). Participants were diagnosed at a median age of 9 years (range 0-66). Adults reported more frequently ear pain and hearing problems, often daily, compared to children (Figure 1).

Eardrum examination (Figure 2) showed that abnormalities were not uncommon even though patients were examined usually outside of an acute infection. Otitis media with effusion (glue ear) was the most common finding seen in 96 (39%) children and 25 adults (18%), usually in both ears. Tympanic sclerosis was more commonly seen in adults compared to children (33 versus 10%). Tympanometry was performed in 216 patients and showed pathologic results (type B) in 49%. Audiometry was performed 273 patients and 44% showed hearing impairment in at least one ear, most commonly mild (Figure 3). Patient-reported did not correlate always with audiometry findings especially in cases of mild impairment. In addition, several patients reported frequent hearing problems even with a normal audiometry.

	Total N (%)	Children <18 y N (%)	Adults ≥18 y N (%)
N. of participants	397 (100)	256 (100)	140 (100)
Age (in years)	15.2 (0.2–72.4)	11.1 (0.12–17.9)	28.2 (18.0–72.4)
Male sex	211 (53)	140 (55)	71 (51)
Diagnosis			
Definite	251 (63)	154 (60)	97 (69)
Probable	125 (32)	88 (34)	37 (27)
Test results pending	21 (5)	15 (6)	6 (4)
Eardrum examination	384 (97)	248 (97)	136 (97)
Tympanometry	216 (54)	129 (50)	87 (62)
Audiometry	273 (69)	154 (60)	119 (85)

Table 1: General characteristics of participants in the EPIC-PCD study.

Definite diagnosis defined based on ERS guidelines

Conclusion

The EPIC-PCD is the largest prospective cohort assessing ENT features in PCD patients. We have found that ear problems do not disappear with age, but otologic disease progresses and acute problems such as otitis media are replaced with chronic issues such as tympanic sclerosis. Hearing impairment is common in all ages and patient perception often differs from audiometry results.

Our findings highlight the need of multidisciplinary care of PCD patients, including regular ENT follow-up and the importance of hearing assessment even when patients report no problems. Prospective standardised information on ear symptoms and examination can contribute to the better phenotypic characterisation of PCD.

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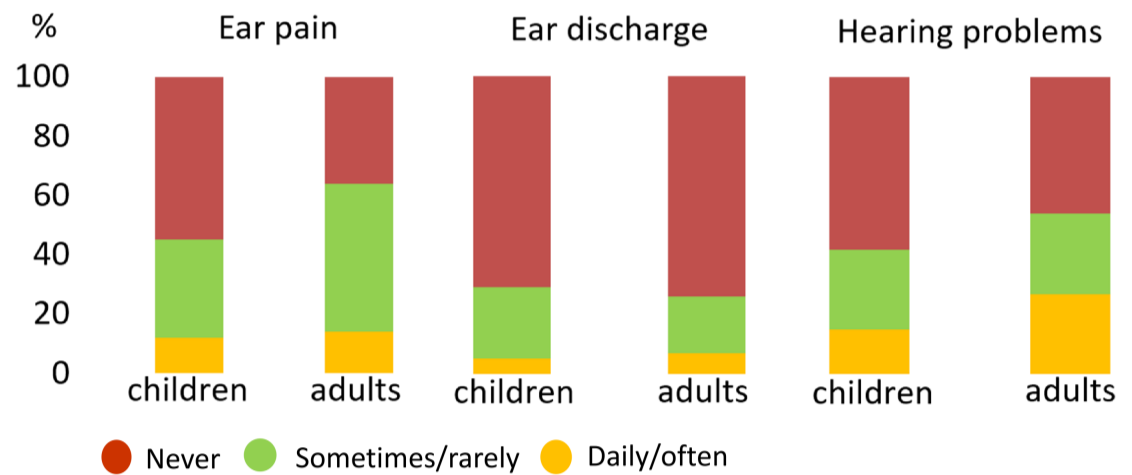


Figure 1: Patient-reported ear symptoms in PCD patients (N=397), overall and by age group.

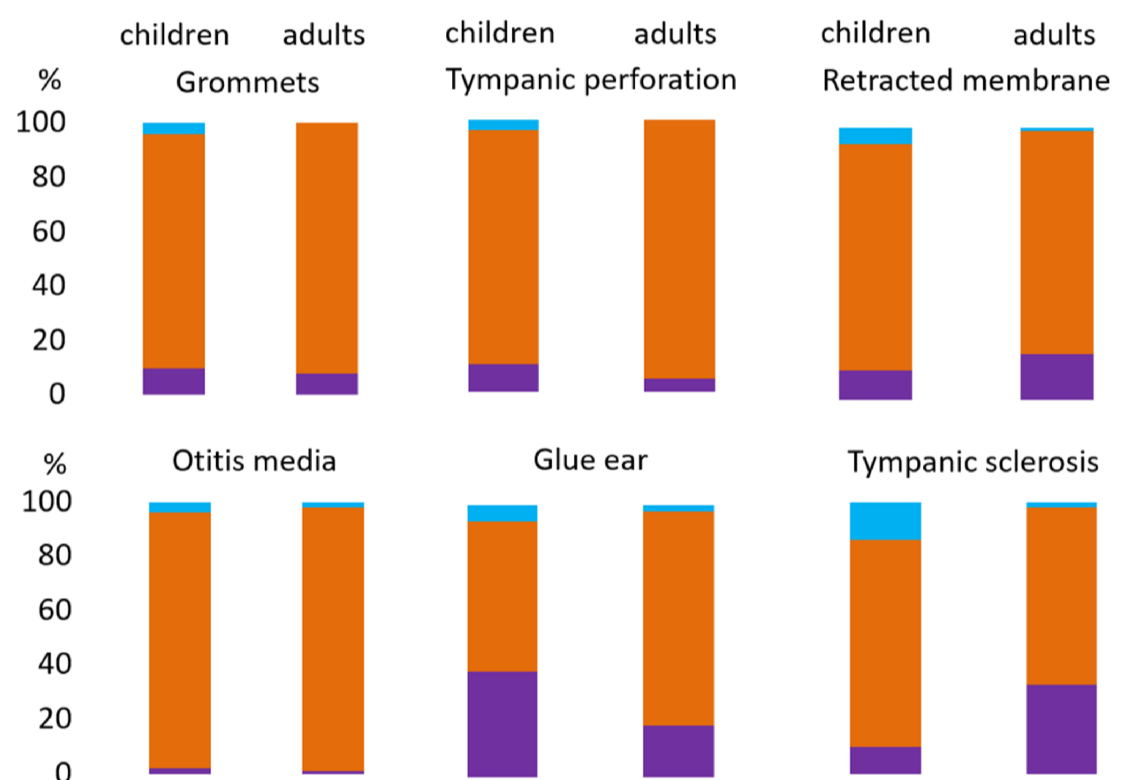


Figure 2: Eardrum examination findings in PCD patients (N=384), overall and by age group. We excluded 13 visits where eardrums were not assessed.

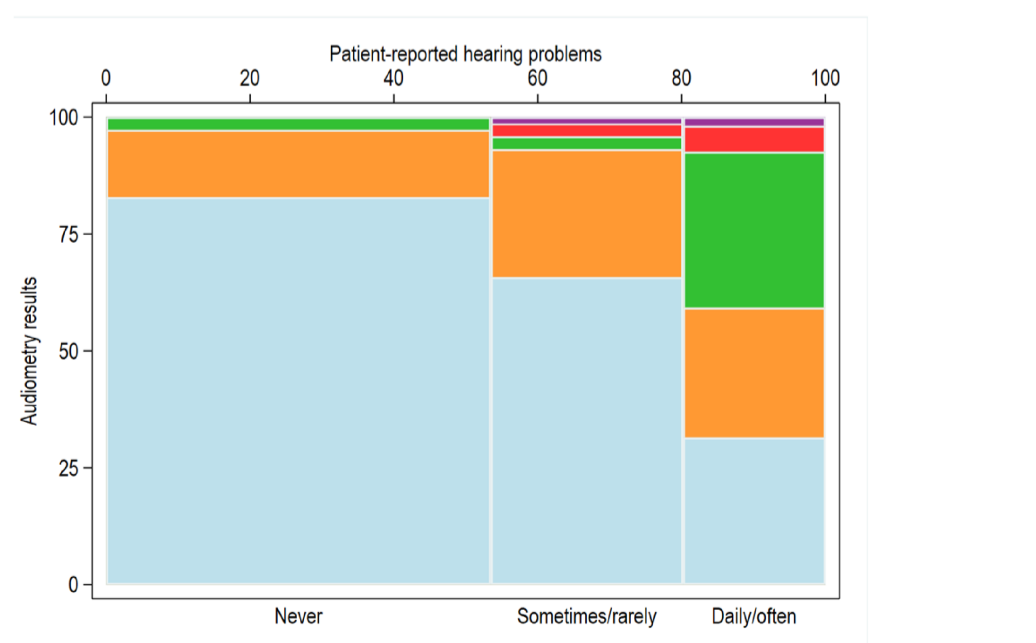


Figure 3: Correlation between patient-reported hearing problems and audiometry results in PCD patients (N=273)

■ Profound hearing loss (>81 dB)
■ Severe hearing loss (61-80 dB)
■ Moderate hearing loss (41-59 dB)
■ Mild hearing loss (26-40 dB)
■ Normal hearing