



National PCD Registry

Annual Report

2024

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On behalf of the National PCD Centres

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Recruitment

306 PCD patients (adults and children) were enrolled in the National PCD registry and had 2024 data entered into it.

Table 1: numbers of PCD patients recruited to the national PCD registry in 2024 by PCD centre (UHS - University Hospital Southampton, RBH - Royal Brompton Hospital.)

PCD Centre	Number recruited
UHS, Adults	64
UHS, Paediatrics	43
Leeds, Paediatrics	78
Leeds, Adults	94
RBH, Paediatrics	14
RBH, Adult	4
Leicester, Paediatrics	9
Leicester, Adults	0
TOTAL	306

Data collection

Data was collected by the PCD centres at the time of their patients' routine clinical appointments, typically at their annual review, in accordance with the data coding manual for the PCD registry.

Data analysis

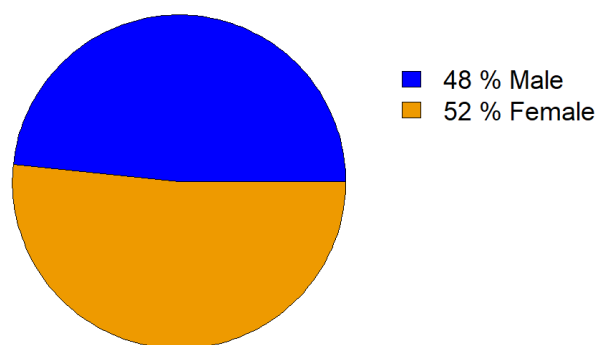
Similar to last is report, due to the relatively small number of patients included in this year's report, we have limited our analysis of the data to looking at some differences in FEV1, one of our important outcome measures, between some sub-groups. One of the most important factors effecting percent predicted FEV1 is the age of the patient, see figure 9. As such, in some of these analyses, the differences in FEV1 seen between groups may be confounded by any age differences between groups. As the available data was not powered for more complex regression analysis, to highlight this, the median age of the groups is shown to allow the reader to be considerate to this when view the data.

ANOVA analyses include a Bonferroni correction to account for the multiple fields analysed.

Demographics

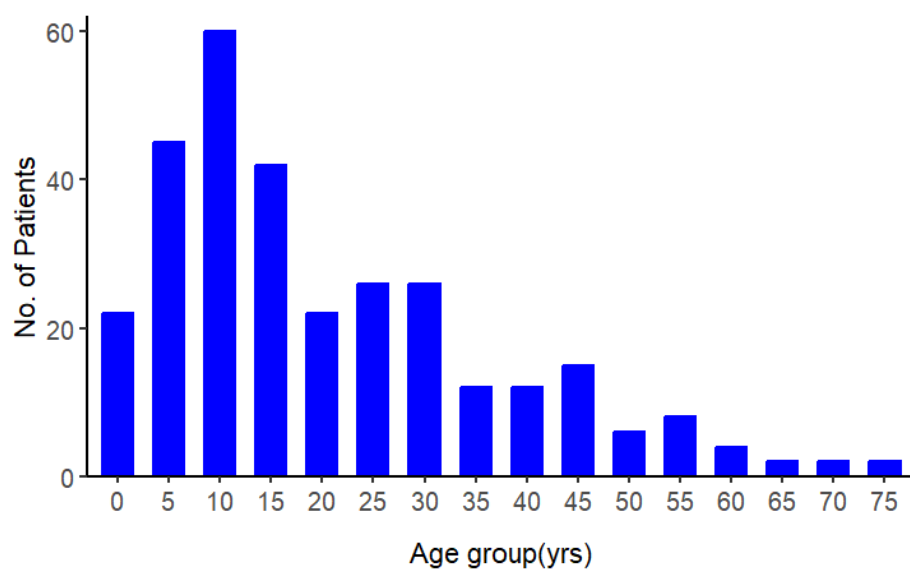
Gender

Figure 1: Pie chart of the gender of PCD patients enrolled in the National PCD Registry (n=306)



Age

Figure 2: Age distribution of PCD patients enrolled in the National PCD Registry (n=306)



Ethnicity

Figure 3: Pie chart of the ethnicity of PCD patients enrolled in the National PCD Registry

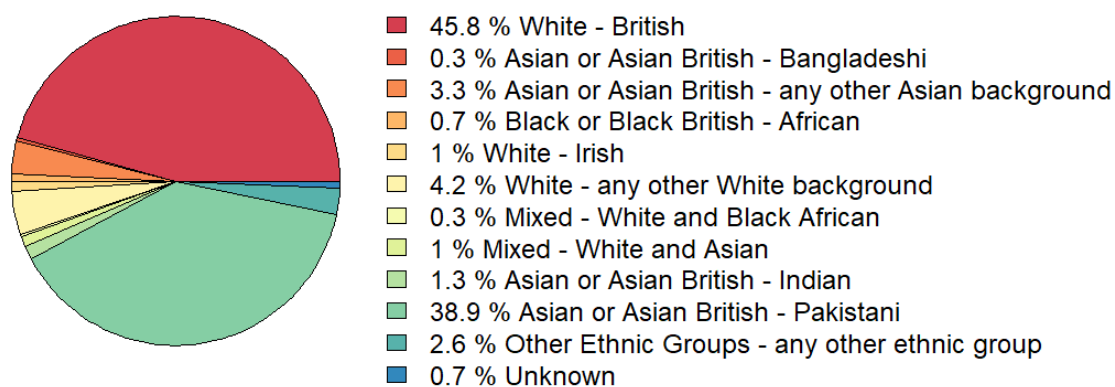


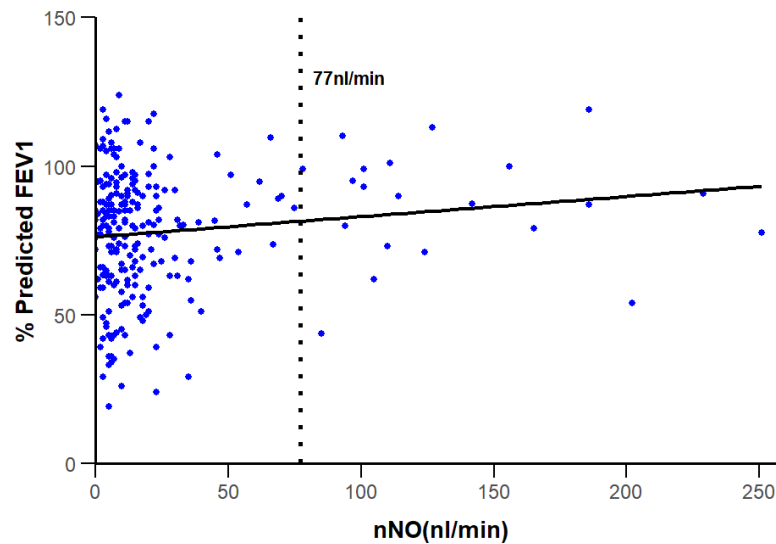
Table 2: Ethnicity of PCD patients recruited to National PCD Registry (n=306)

Ethnicity	Number	% of whole
White - British	140	45.8
Asian or Asian British - Bangladeshi	1	0.3
Asian or Asian British - any other Asian background	10	3.3
Black or Black British - Caribbean	0	0
Black or Black British - African	2	0.7
Black or Black British - any other black background	0	0
Other Ethnic Groups - Chinese	0	0
Other Ethnic Groups - any other ethnic group	8	2.6
White - Irish	3	1.0
White - any other White background	13	4.2
Mixed - White and Black Caribbean	0	0.0
Mixed - White and Black African	1	0.3
Mixed - White and Asian	3	1.0
Mixed - any other mixed background	0	0.0
Asian or Asian British - Indian	4	1.3
Asian or Asian British - Pakistani	119	38.9
Unknown	2	0.7

Diagnostic investigations

Nasal nitric oxide (nNO)

Figure 4: Nasal NO (nl/min) vs mean % predicted FEV1 in PCD patients enrolled in the National PCD Registry



The correlation line was not found to be significant, $P=0.047$.

Of note 21 (6.9%) PCD patients within the registry had a nNO above 77nl/min, the commonly quoted cut off used to screen for the possibility of the diagnosis of PCD.

Nasal nitric oxide (nNO) by gene

Table 3: Number and percentage of patients with nasal nitric oxide levels over 77 nl/min by gene

Gene	No of patients	No of patients with nNO > 77nl/min	Percentage (%) of patients with nNO > 77nl/min
ARMC4	1	0	0
CCDC103	9	5	44.4
CCDC114	1	0	0
CCDC151	2	0	0
CCDC39	10	1	10
CCDC40	16	0	0
CCDC65 (aka DRC2)	3	0	0
CCNO	1	0	0
CFAP300 (aka C11orf70)	1	0	0
DNAAF1 (aka LRRC50)	2	0	0
DNAAF2 (aka KTU)	1	0	0
DNAAF3 (aka C19orf51)	3	0	0
DNAAF4 (aka DYX1C1)	11	1	9.1
DNAAF5 (aka HEATR2)	6	1	16.7
DNAAF6 (aka PIH1D3)	1	0	0
DNAH11	21	3	14.3
DNAH5	39	0	0
DNAH9	2	0	0
DNAI1	11	0	0
DNAI2	3	0	0
DRC1 (aka CCDC164)	3	1	33.3
Genetic testing not performed	1	0	0
Genetic testing undertaken but no causative gene found	20	4	20
HYDIN	12	1	8.3
LRRC56	1	1	100
LRRC6	12	1	8.3
MCIDAS	2	0	0
OFD1	2	1	50
Other	9	0	0
Await result	5	0	0
RPGR	1	0	0
RSPH1	4	2	50
RSPH4A	13	0	0
SPAG1	2	0	0
TTC12	1	0	0
Unknown	11	0	0
ZMYND10	1	0	0

Transmission electron microscopy (TEM)

Figure 5: TEM findings in PCD patients enrolled in the National PCD Registry (ODA – outer dynein arm, IDA – inner dynein arm, MTD – microtubular disarrangement, CC - central complex)

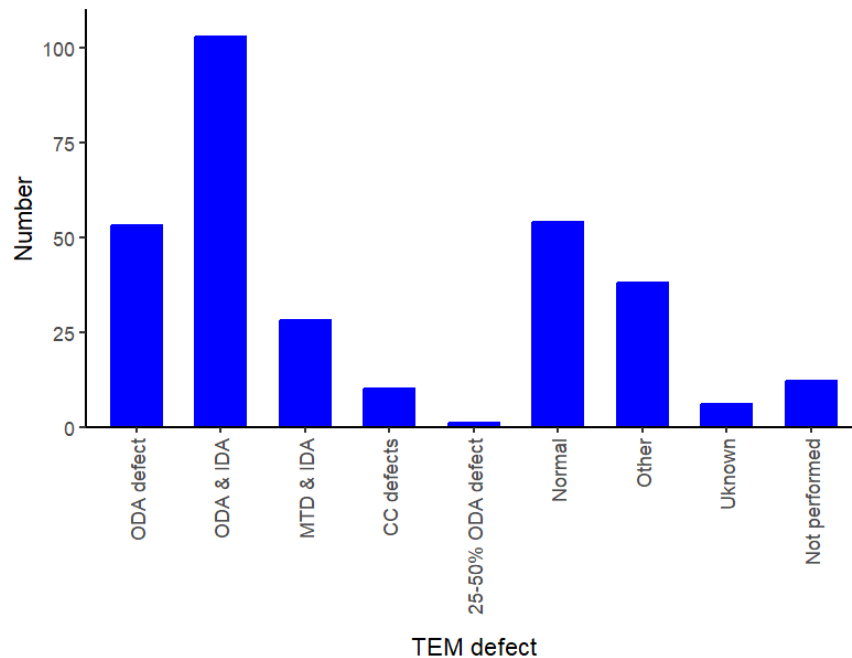
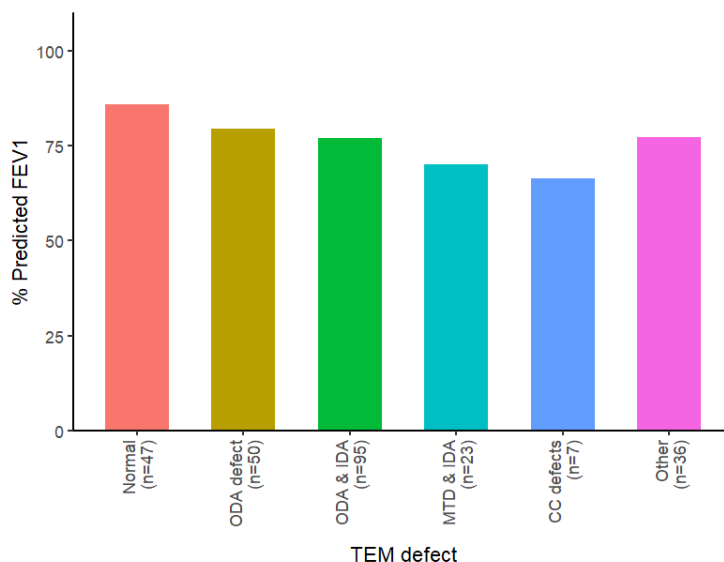


Figure 6: TEM findings vs mean % predicted FEV₁ of PCD patients enrolled in the National PCD Registry (ODA – outer dynein arm, IDA – inner dynein arm, MTD – microtubular disarrangement, CC - central complex, *p value significant)



ANOVA analysis did not demonstrate any significant differences in mean predicted FEV₁ between the different TEM groups.

For reference the median age of the respective TEM defect groups are normal 16 years, ODA defect 19 years, ODA & IDA defect 16 years, MTD & IDA 16 years, CC defect 12 years.

PCD Genetics

Figure 7: Pie chart of the proportion of patients within the national PCD registry by whether they have a confirmed genetic diagnosis, are awaiting results of genetic testing, have had no genetic cause identified, have had no genetic tests performed or their status is unknown (n=301)

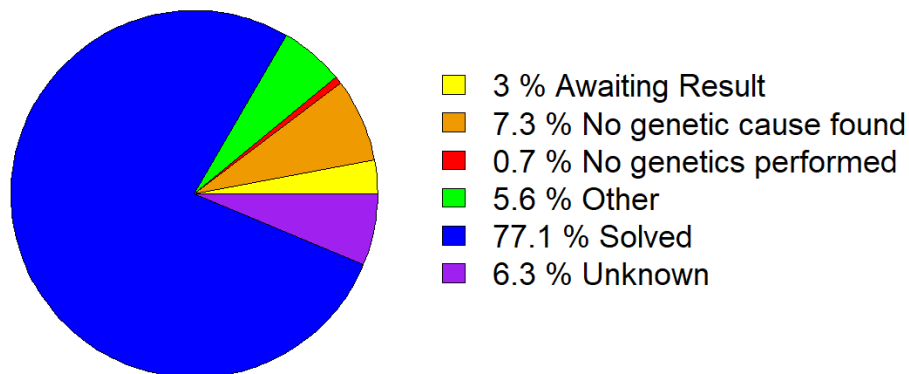
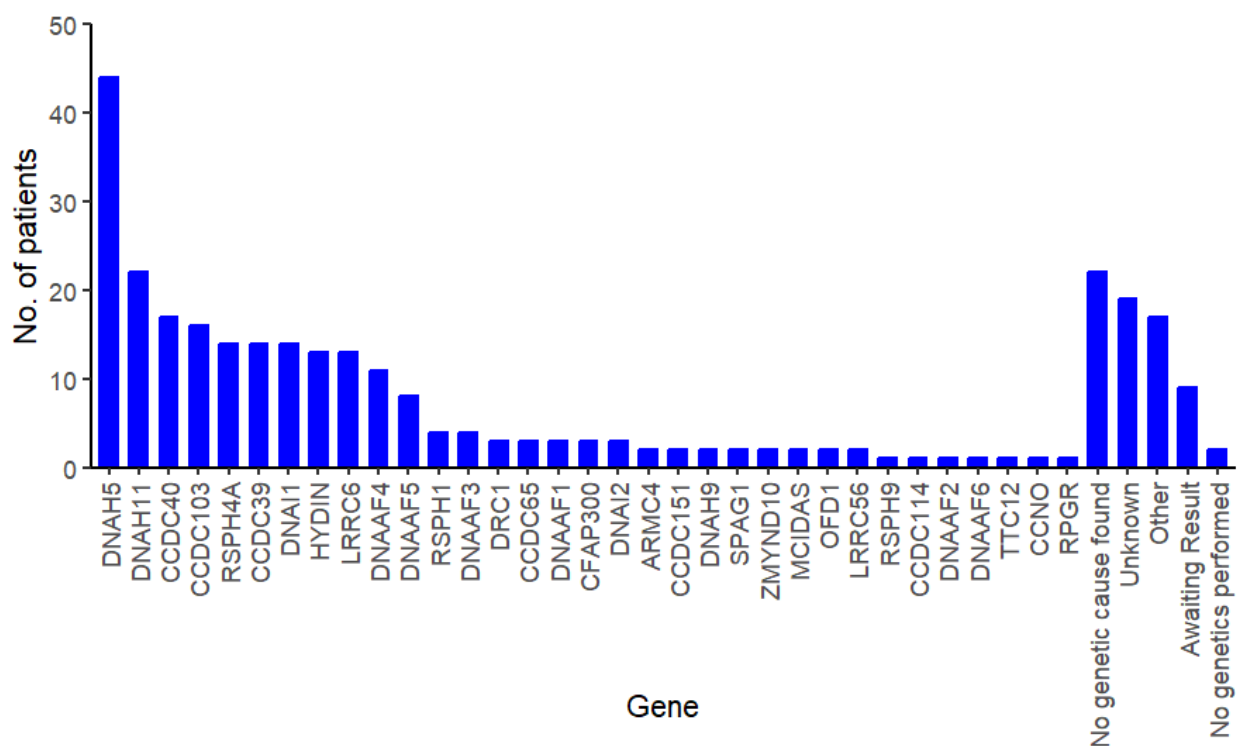


Figure 8: Genetics findings in PCD patients enrolled in the National PCD Registry (n=301)



Mean % predicted FEV1 values were compared for each of the genes using ANOVA analysis, which included a Bonferroni correction to allow for the multiple data fields analysed.

There were significant differences the mean % predicted FEV1 of patients with different PCD causing genes.

Table 4: Mean % predicted FEV1 for genes where significant differences were noted between groups.

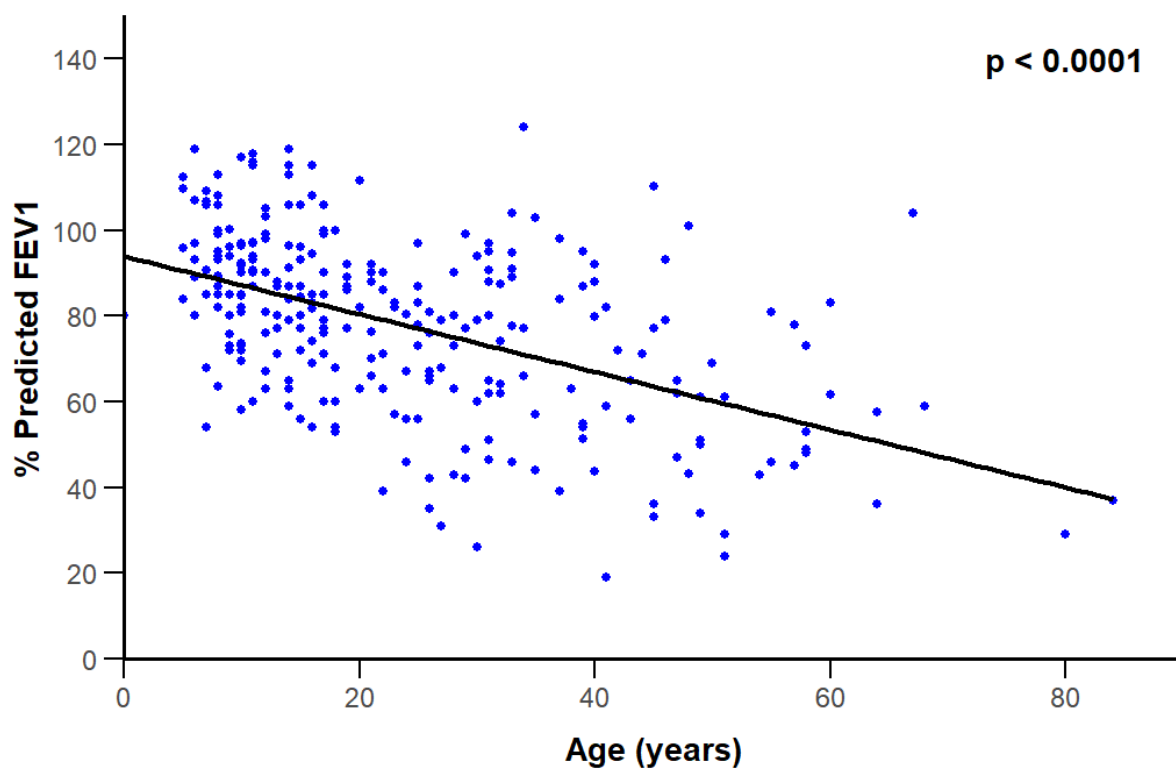
Gene	Mean % predicted FEV ₁
DNAH5	80.2
DNAH11	94.2
DNAAF4 (aka DYX1C1)	88.5
CCDC39	67.3
CCDC40	63.2
RSPH4A	57.9

Table 5: P values of differences in mean % predicted between FEV1 when comparing the gene in the row and the column.

Gene	RSPH4A	CCDC39	CCDC40
DNAH5	0.034		
DNAH11	<0.001	0.038	<0.001
DNAAF4 (aka DYX1C1)	0.017		

Lung function

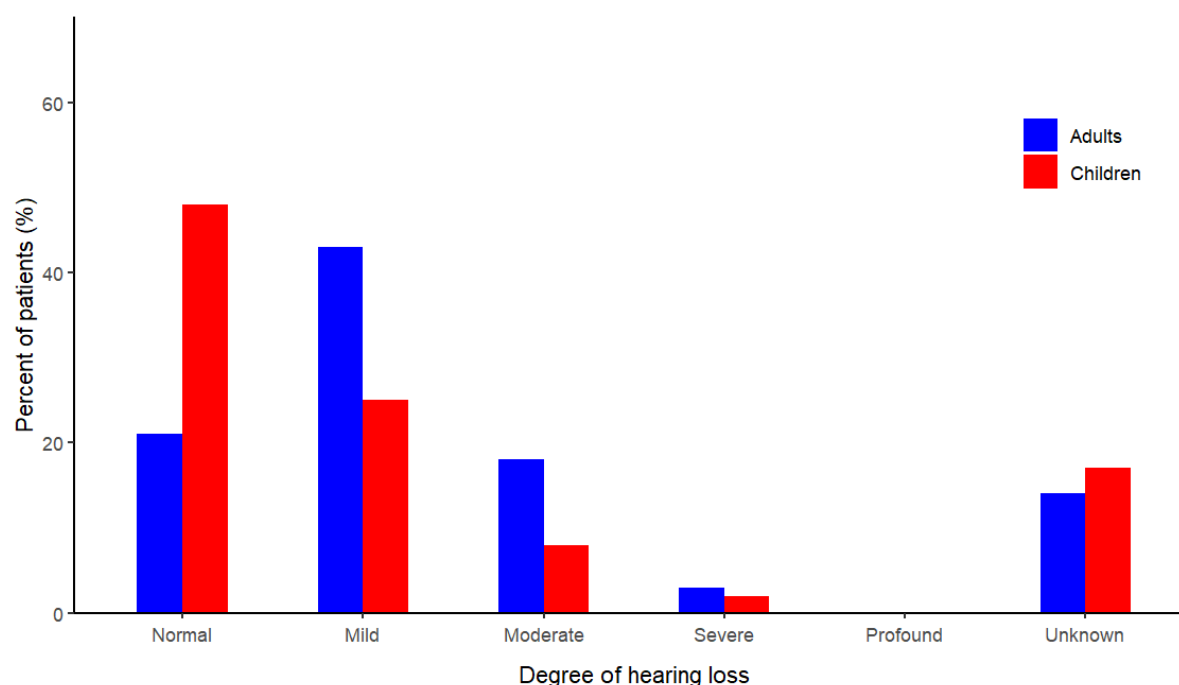
Figure 9: Mean % predicted FEV1 vs age of PCD patients enrolled in the National PCD Registry



Whilst this was cross sectional data, we note that there was a significantly lower % predicted FEV1 seen in those with a higher age, $p < 0.0001$. The gradient of the correlation line suggested that for every year of age gained there was a 0.7 decrease in % predicted FEV1.

Audiology

Figure 10: Audiology results in PCD adults and children enrolled in the National PCD Registry. Bar chart shows the proportion of patient's (divided into adults and children) with the degree of hearing loss including those with unknown results.

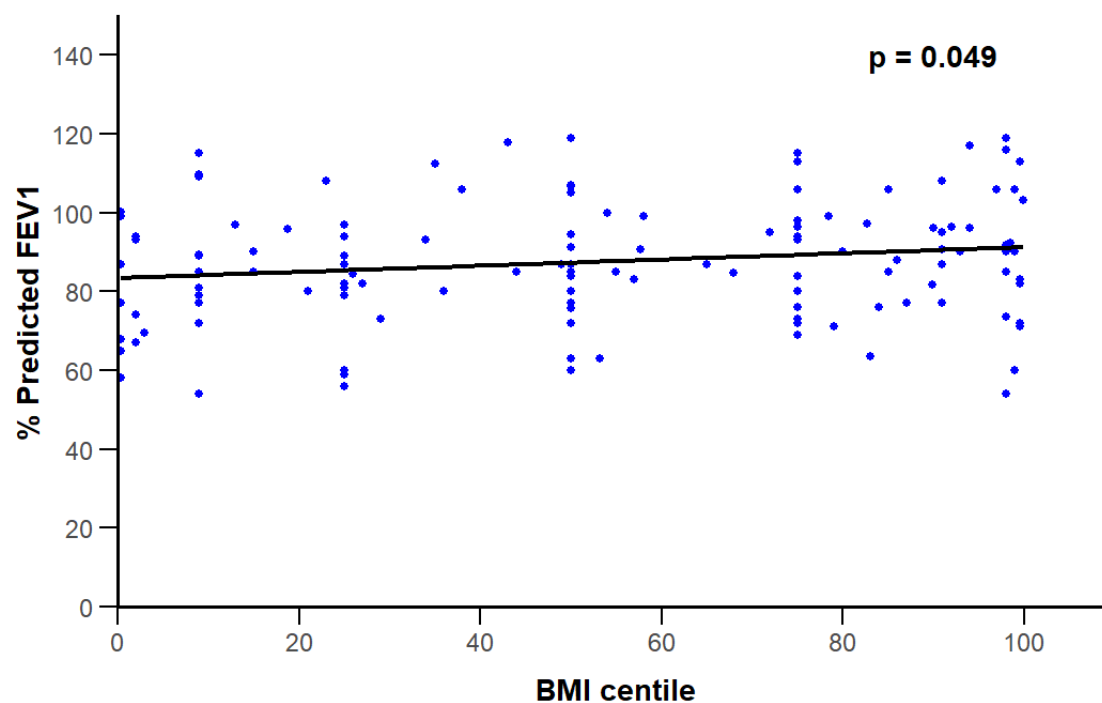


Hearing aids:

There were 69 (22.5%) patients prescribed hearing aids at the time of annual review.

Nutrition

Figure 11: Mean % predicted FEV1 vs BMI centile of children (<18 years) enrolled in the National PCD Registry

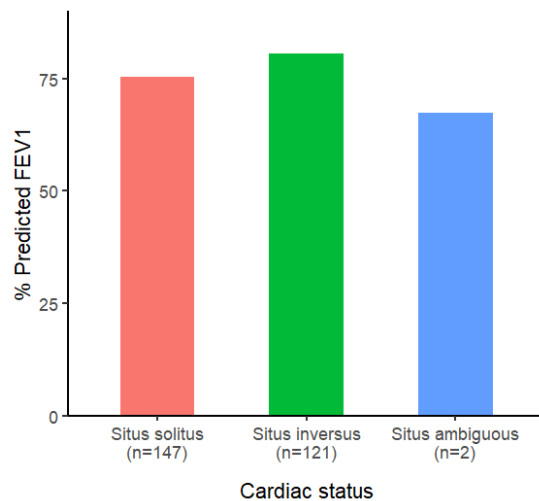


The correlation line was found to be significant, $P=0.049$

Cardiology

Cardiac situs

Figure 12: Mean % predicted FEV1 by cardiac situs (solitus, inversus & ambiguous) in PCD patients enrolled in the National PCD Registry



No significant difference was seen in the mean % predicted FEV1 of those with situs solitus and those with situs inversus. For reference the median age of the respective cardiac situs groups are situs solitus 20.5 years, situs inversus 16 years, situs ambiguous 16 years.

Congenital heart disease (CHD)

Figure 13: Pie chart of the presence of CHD or not in PCD patients enrolled in the PCD Registry

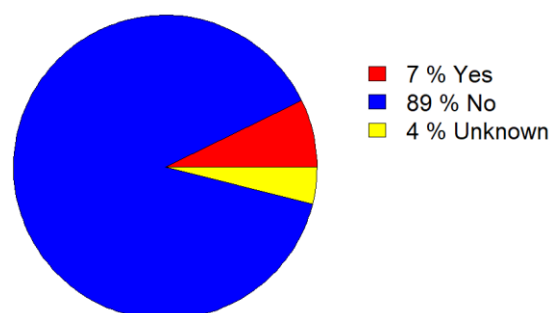
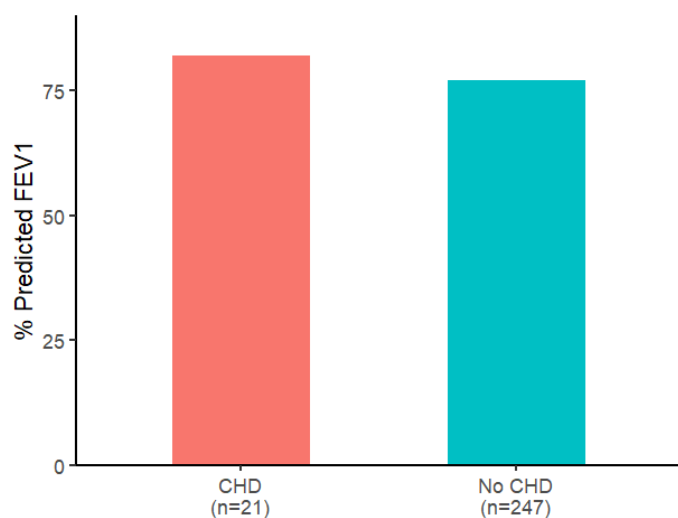


Figure 14: Mean % predicted FEV1 in those with and without congenital heart disease in PCD patients enrolled in the National PCD Registry



No significant difference was seen in the mean % predicted FEV1 of those with and without congenital heart disease. For reference, the median age of the CHD group was 15 years, and the no CHD group was 18 years.

Microbiology

Figure 15: Percentage of patients enrolled in the National PCD Registry. culturing each respiratory pathogens during the year (Other includes *Enterobacter* species, other *Pseudomonas* species, other *streptococcus* species, *candida* & *Serratia* species.) Of note, some patients cultured more than 1 pathogen during that year.

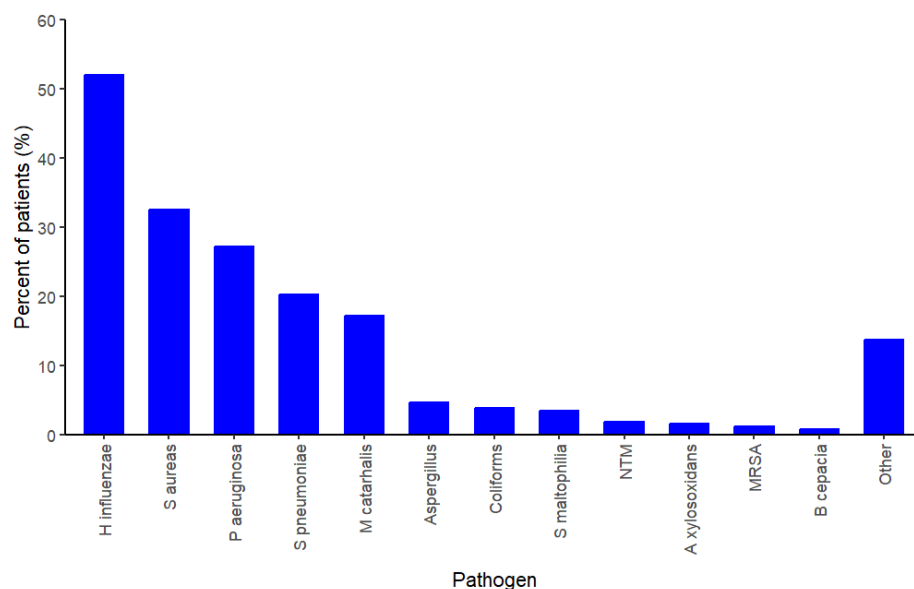
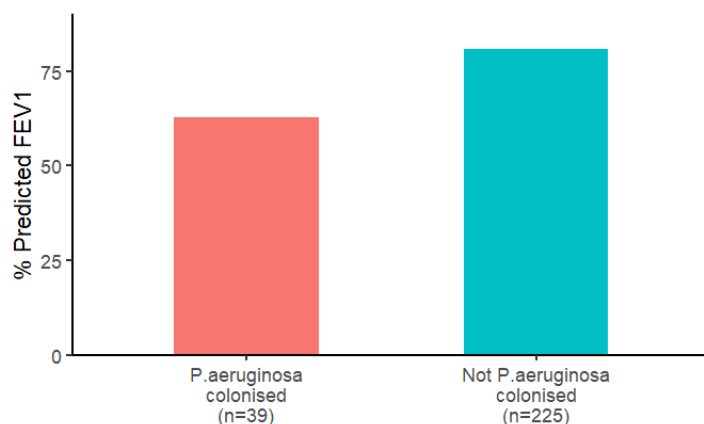


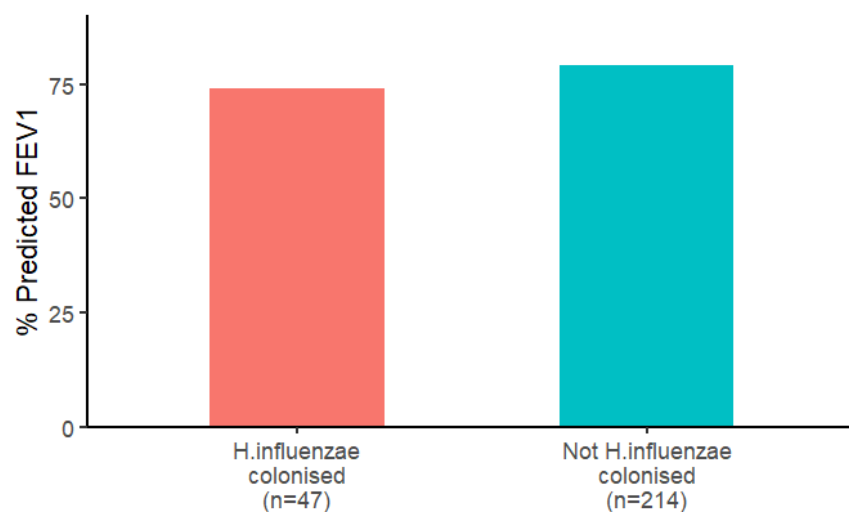
Figure 16: Mean % predicted FEV1 in those PCD patients enrolled in the National PCD Registry in 2024 who were colonised* with *Pseudomonas aeruginosa* compared to those who were not.



42 patients recruited to the national PCD registry in 2024 were colonised* with *P. aeruginosa*, 39 of whom had lung function data available. Those who were colonised with *P. aeruginosa* had a significantly lower mean % predicted FEV1 than those who were not (62.8% vs 80.8%, p value <0.01).

Importantly, for reference, the median age of the Pa colonised group was 38.5 years and the not Pa colonised group was 15 years.

Figure 17: Mean % predicted FEV1 in those PCD patients enrolled in the National PCD Registry in 2024 who were colonised* with *Haemophilus influenzae* compared to those who were not.

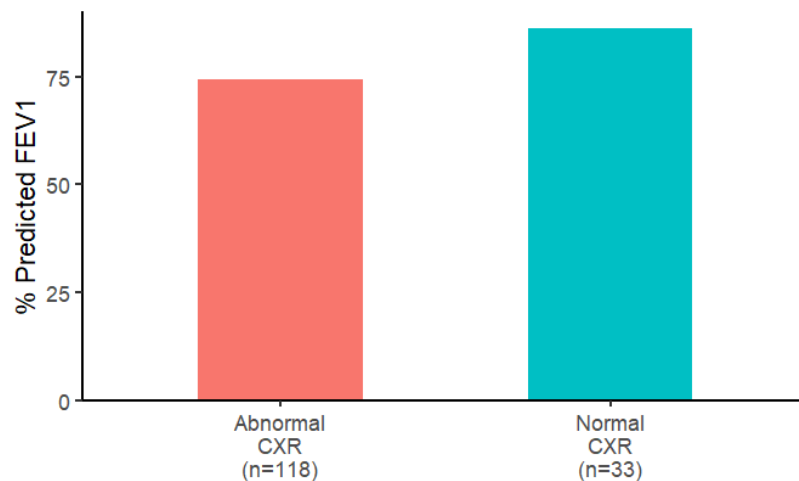


No significant difference in mean % predicted FEV1 was noted between these 2 groups.

(*For reference colonisation was defined by the standard Leeds Criteria, positive microbiology on 50% or more of the samples collected in the previous 12 months.)

Radiology

Figure 18: Mean % predicted FEV1 in those PCD patients enrolled in the National PCD Registry with normal and abnormal chest x-rays.



Those with abnormal chest x-rays had significantly lower mean % predicted FEV1 than those with normal ones (mean 74.3% vs 86.1%, *P=0.02). For reference, the median age of the abnormal CXR group was 16 years and the normal CXR group was 10 years.

Treatments

Physiotherapy

Figure 19: Primary type of airway clearance physiotherapy recommended to PCD patients enrolled in the National PCD Registry

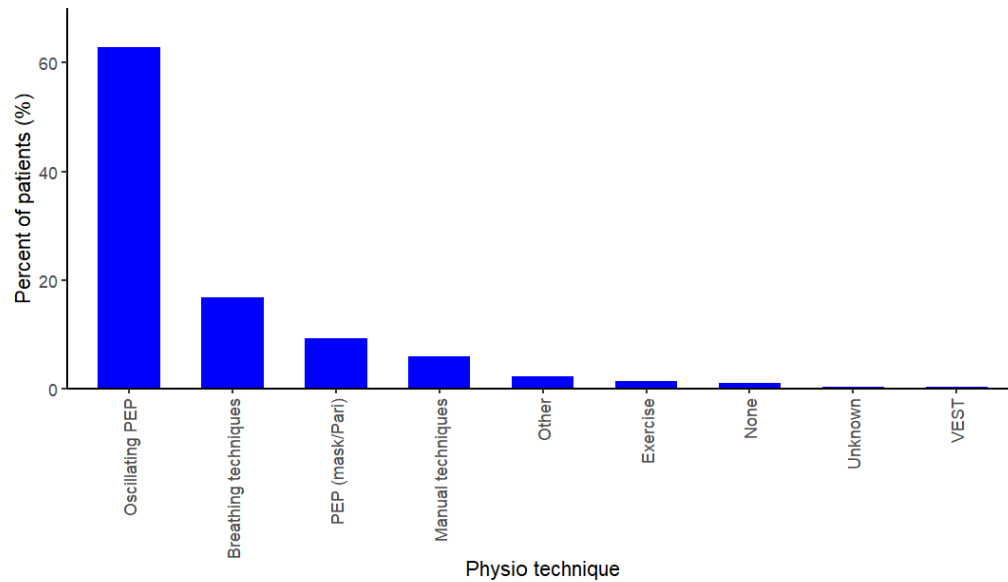


Figure 20: Frequency of airway clearance physiotherapy recommended to PCD patients enrolled in the National PCD Registry

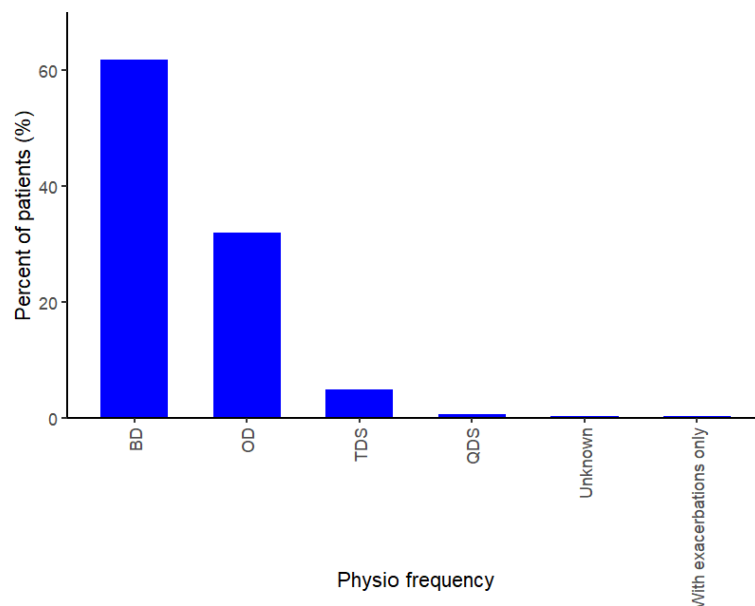
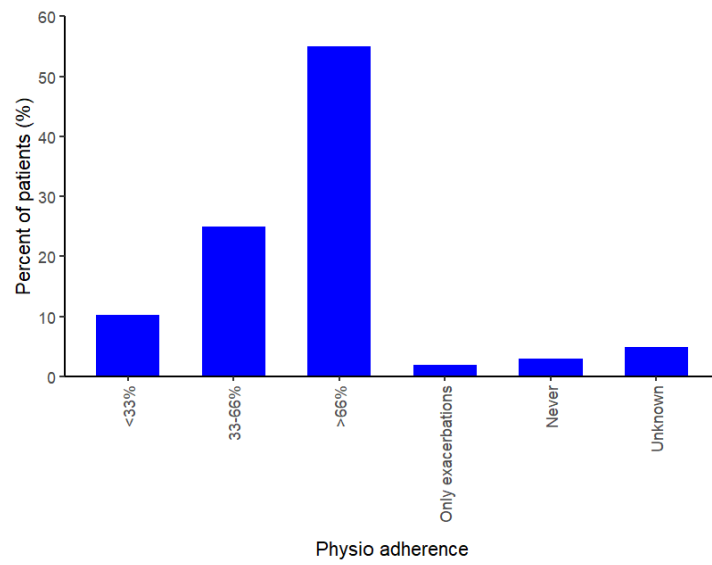


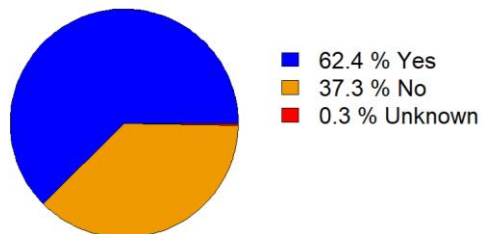
Figure 21: Patient reported adherence to recommended frequency of airway clearance physiotherapy in PCD patients enrolled in the National PCD Registry



Mucolytics

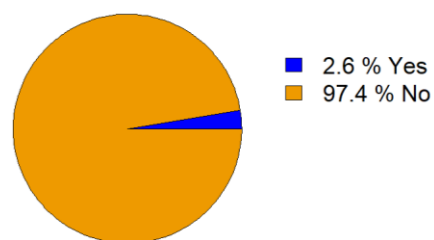
Hypertonic saline nebulisation

Figure 22: Pie chart of percentage prescribed nebulised hypertonic saline in PCD patients enrolled in the National PCD Registry



Dornase alpha nebulisation

Figure 23: Pie chart of percentage prescribed nebulised Dornase alpha nebulisation in PCD patients enrolled in the National PCD Registry



Other treatments

Prophylactic antibiotics

One hundred thirty-one patients were prescribed oral prophylactic antibiotics and 31 were prescribed nebulised prophylactic antibiotics.

Figure 24: Percentage prescribed which oral prophylactic antibiotics in PCD patients enrolled in the National PCD Registry

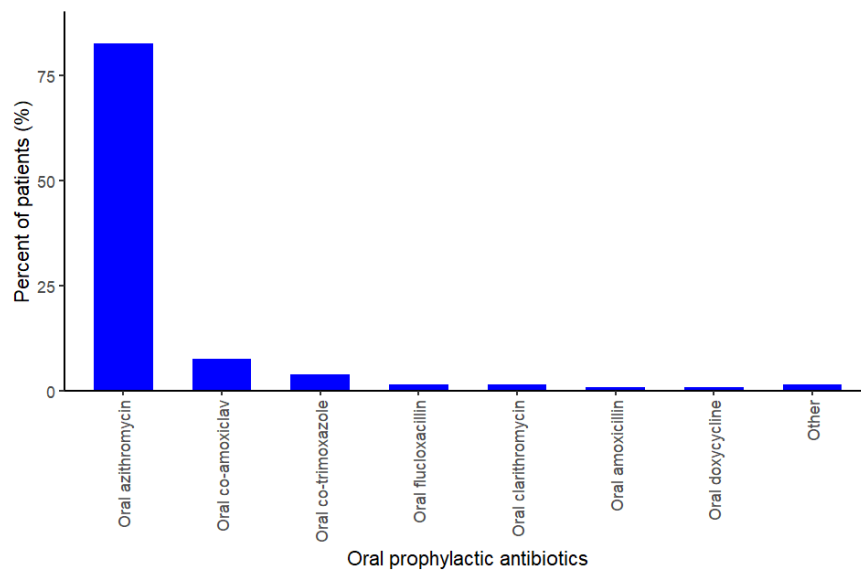
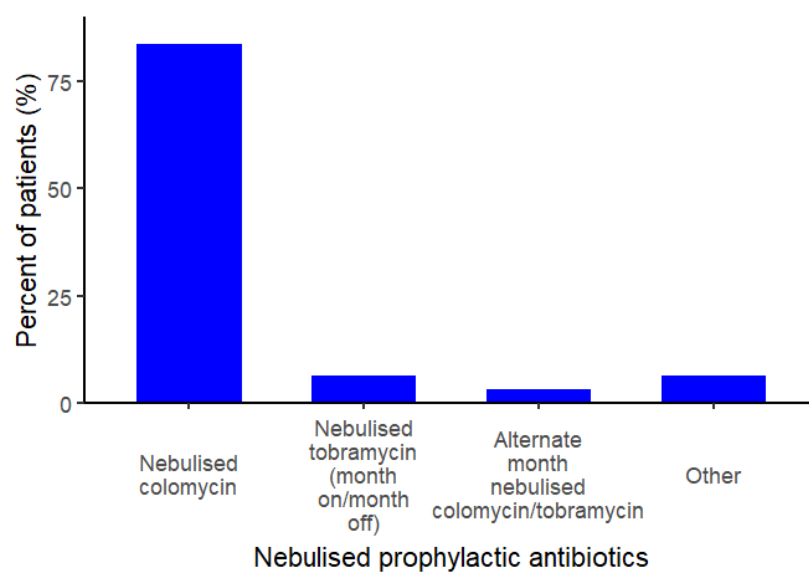
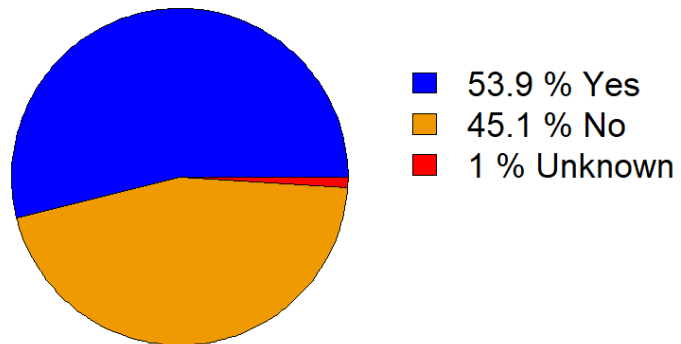


Figure 25: Percentage prescribed which nebulised prophylactic antibiotics in PCD patients enrolled in the National PCD Registry



Nasal douching

Figure 26: Pie chart of percentage prescribed nasal douching in PCD patients enrolled in the National PCD Registry



Acknowledgements

Our thanks for the support and guidance of the members of the PCD Registry research and steering committee.

Our thanks also to all the members of the PCD centre teams that have uploaded these data to the registry and also to the patient's for agreeing to take part in the national PCD registry.