



National PCD Registry

Annual Report

Lay Summary

2024

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On behalf of the National PCD Centres

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Who took part?

This report is based on information from over 300 children and adults with PCD who were seen in specialist centres across the UK in 2024.

The table shows how many patients from each centre contributed data that year.

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

People who consented to participate in the PCD Registry UK study that year were included. The data was given by the team who looks after you and this information was then analysed to provide information we have put together in this report.

What do we know about the patients who took part?

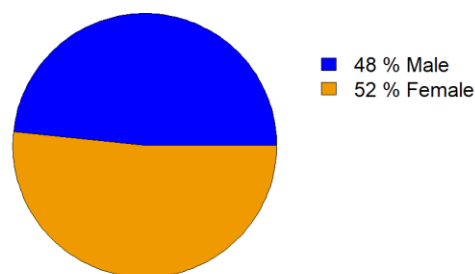
Gender

This pie chart shows the gender of patients who took part in the PCD Registry in 2024.

- Both men and women are represented
- PCD affects men and women equally

Figure 1:

This pie chart shows us that both men and women took part in the Registry. PCD affects both men and women equally.



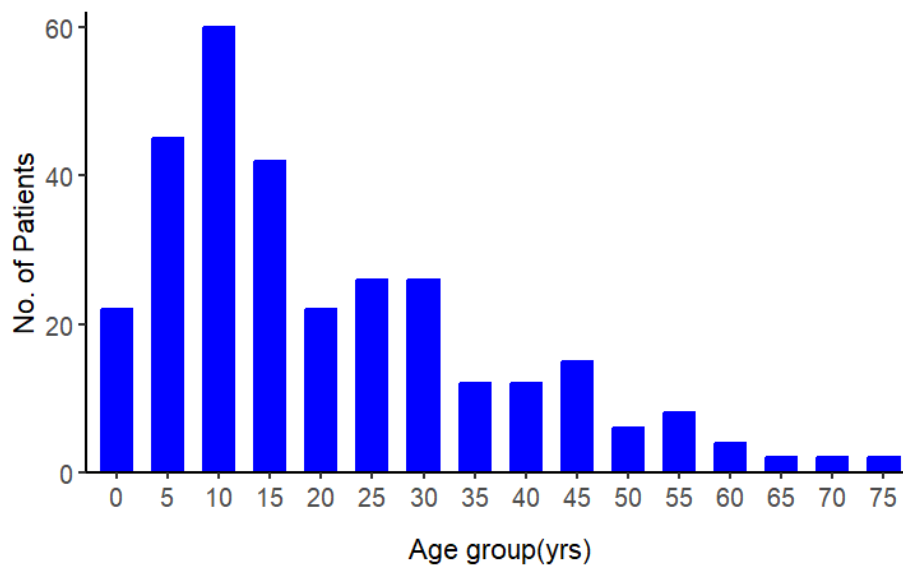
In short: The Registry included a balanced mix of male and female patients, reflecting how PCD occurs in both genders.

Age

This graph shows the ages of patients who took part in the Registry..

- Most participants are children aged 5–15 years
- PCD also affects adults, but the higher number of children likely reflects that specialist children's PCD centres contributed more data in 2024

Figure 2: This graph shows us the age of patients who took part.



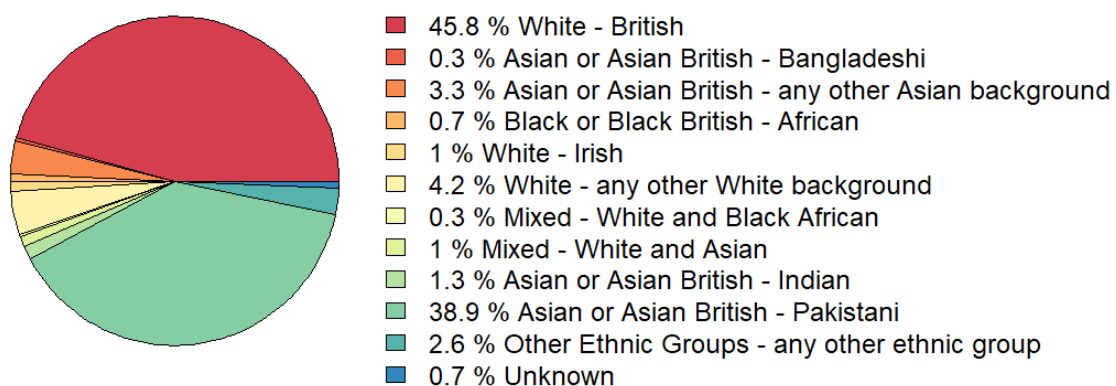
In short: The Registry included mostly children, though PCD can affect people of all ages.

Ethnicity

This pie chart shows the ethnic backgrounds of people with primary ciliary dyskinesia (PCD) in the registry.

- Just under half of the patients are White-British
- The rest come from a variety of other ethnic backgrounds

Figure 3: This pie chart shows the spread of ethnicity recorded for people with PCD. Just under half of the people included were of White-British origin and the remainder of other ethnic origin.



In short: PCD affects people from many different ethnic groups, with White-British being the largest single group in this dataset.

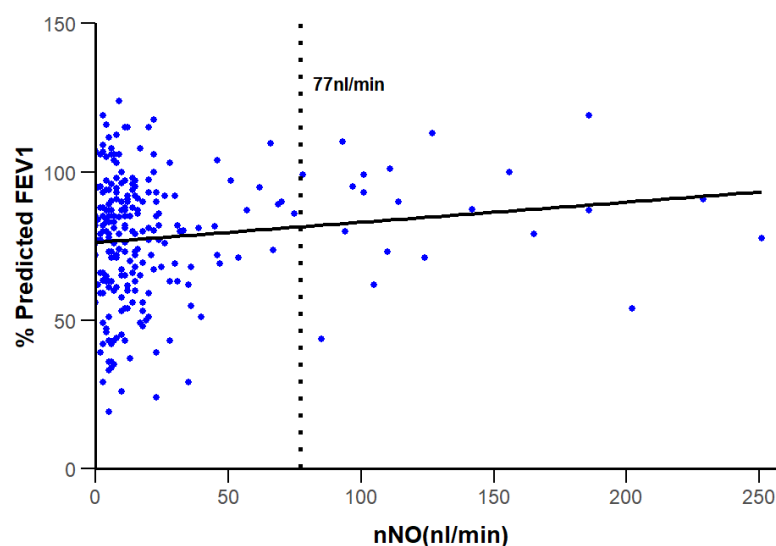
Tests used to diagnose PCD

Nasal nitric oxide (nNO)

This test measures nitric oxide (nNO) in the nose, which is usually checked when diagnosing PCD.

- In PCD, nasal nitric oxide is usually low, but about 7% of people in the Registry had a normal level
- The graph looks at whether nNO levels are linked to lung function
- No connection was found between nasal nitric oxide and lung function

Figure 4: nasal nitric oxide and lung function



In short: Most people with PCD have low nasal nitric oxide, but it doesn't seem to affect how well their lungs work.

Genetics and nasal nitric oxide

Primary ciliary dyskinesia (PCD) is an inherited condition that affects how the tiny hairs in your airways (cilia) work. It is caused by faulty genes passed down from your parents.

This report shows that most people with PCD have a confirmed genetic cause.

Some types of PCD are more likely to have normal nasal nitric oxide (nNO) levels, which is a test that can help identify PCD. The chart shows the number and percentage of patients with nNO levels above 77 nl/min, broken down by which gene they have.

In short: Different genetic types of PCD can affect nasal nitric oxide differently, and this chart shows how common normal nNO levels are for each gene.

Table 3: PCD is an inherited condition that affects the way your cilia work. PCD is passed down by inheriting a faulty gene. This report shows that most people have a confirmed genetic cause. There are certain types of PCD that are more likely to have a normal nasal nitric oxide. This table shows the number and percentage of patients with normal levels of nasal nitric oxide (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

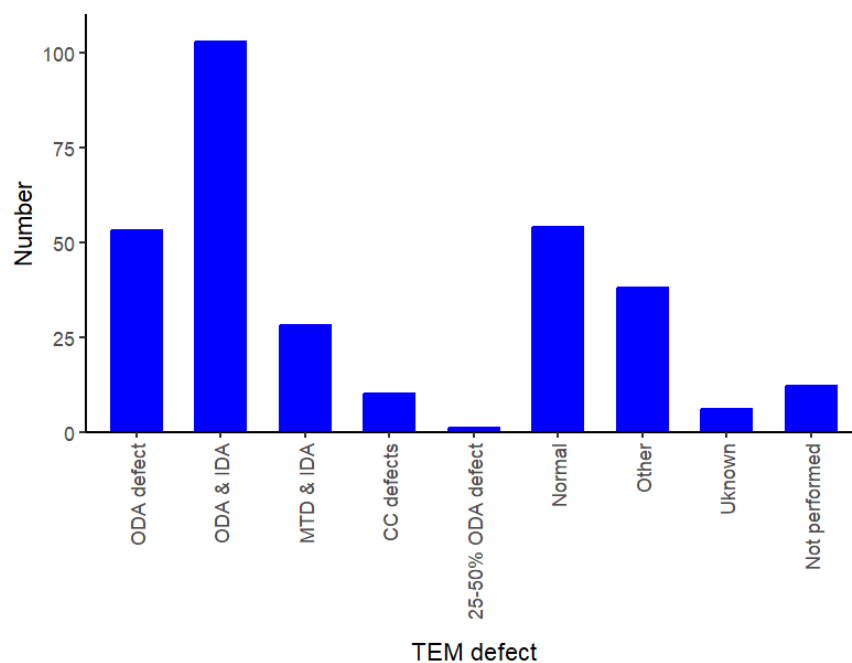
Transmission electron microscopy (TEM)

This shows the results of TEM (transmission electron microscopy) tests in people with primary ciliary dyskinesia (PCD). TEM is a special microscope that looks at the tiny structures inside the cilia (the tiny hairs in the airways).

The results show which parts of the cilia are affected:

- ODA (outer dynein arm) – one of the structures that helps cilia move
- IDA (inner dynein arm) – another structure that helps movement
- MTD (microtubular disarrangement) – the internal structure of the cilia is disorganized
- CC (central complex) – the central part of the cilia

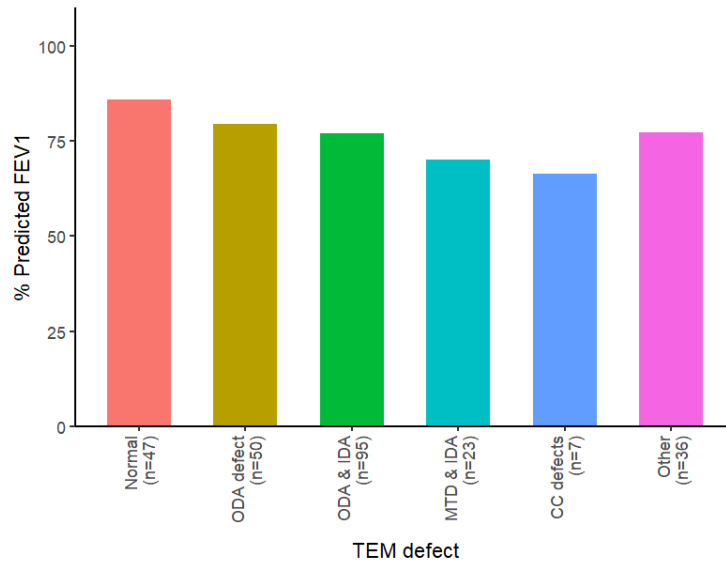
Figure 5: TEM findings in PCD patients enrolled in the National PCD Registry (ODA – outer dynein arm, IDA – inner dynein arm, MTD – microtubular disarrangement, CC - central complex)



In short: TEM helps doctors see which tiny parts of the cilia are abnormal in PCD, which can help confirm the diagnosis.

We also looked at how lung function compares with results of TEM.
The analysis showed **no significant differences** in lung function between the different TEM groups.

Figure 6: TEM findings and lung function in PCD



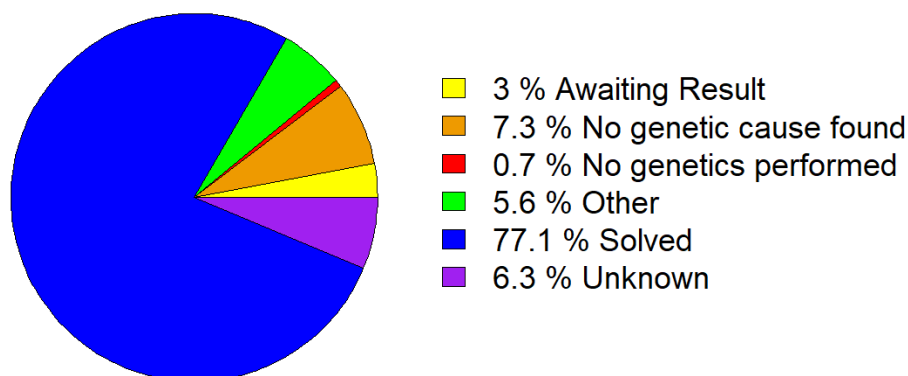
In short: The type of cilia defect seen under the microscope did not appear to affect lung function in this group of PCD patients.

PCD Genetics

This pie chart shows the genetic testing results for people with primary ciliary dyskinesia (PCD) in the Registry in 2024.

- Most patients have a confirmed genetic diagnosis, meaning the gene causing PCD is known
- Some patients are still waiting for results
- A few patients have no gene identified yet

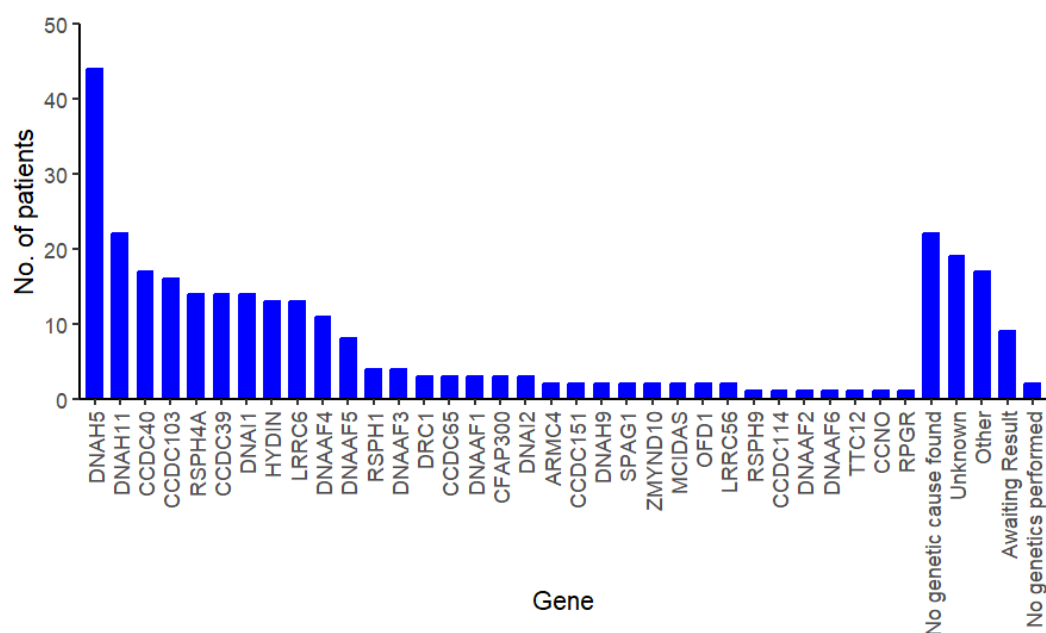
Figure 7: This pie chart shows that most people with PCD have a confirmed genetic diagnosis. Some patients are still waiting for results or no genes have been found yet.



In short: Most people with PCD in the registry know the genetic cause of their condition, but some are still waiting or have unknown results.

The next graph shows the different genetic causes of PCD found in patients who took part in the registry study in 2024.

Figure 8: This graph shows the genetic cause we have found in the patients who participated in the registry study in 2024.



In short: Mutations in some genes are more commonly seen in PCD than others. This graph gives an overview of which genes are responsible for PCD in this group of patients.

Lung function was compared for different genetic causes of PCD in the patients. This report shows that patients with certain genetic causes for PCD appear to have better lung function while other genes might be associated with lower lung function.

Table 4: Average lung function for certain genes

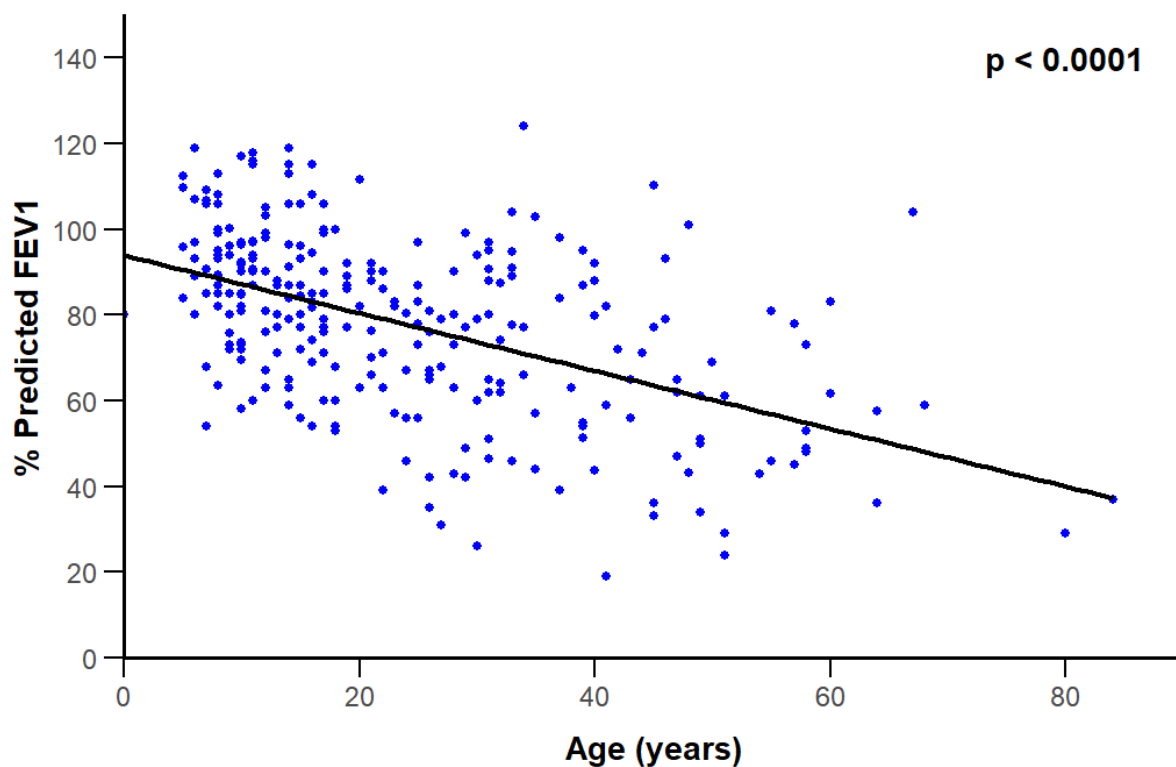
Gene	Mean % predicted FEV ₁
DNAH5	80.2
DNAH11	94.2
DNAAF4 (aka DYX1C1)	88.5
CCDC39	67.3
CCDC40	63.2
RSPH4A	57.9

Lung function

This graph shows how lung function changes over time in people with primary ciliary dyskinesia (PCD).

- Higher lung function is better, and scores over 85% are considered normal
- The data suggests that lung function tends to decrease as people get older
- This report mostly includes younger patients, so future reports with more adults will give a clearer picture

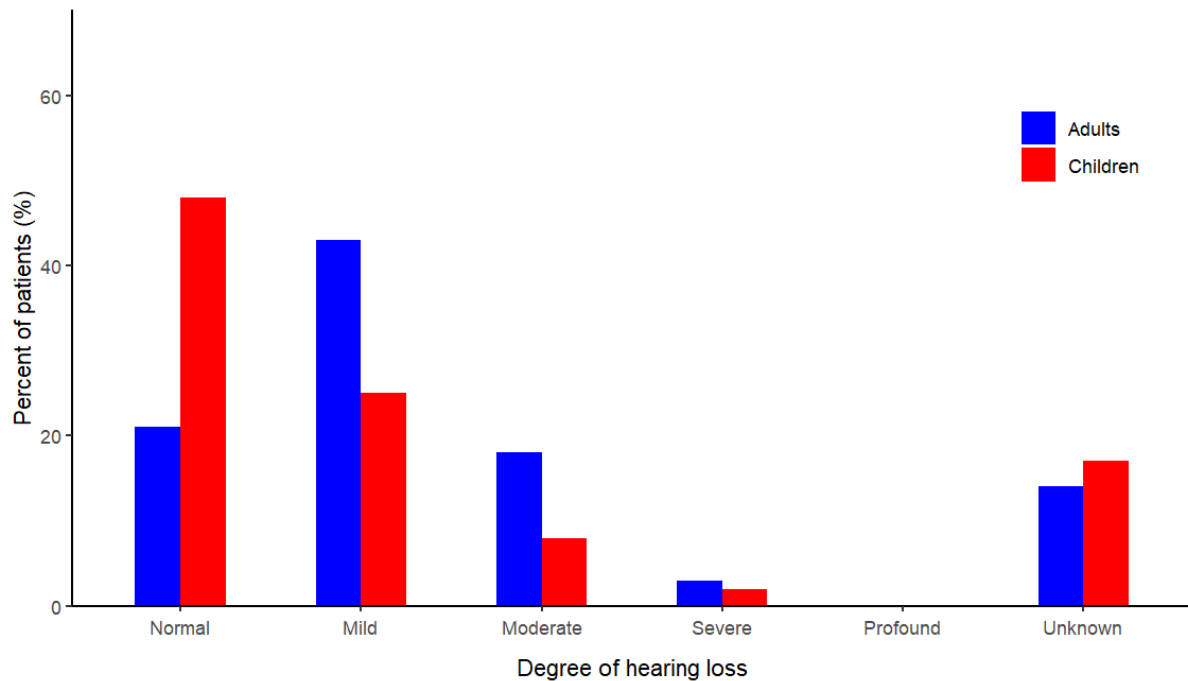
Figure 9: This graph looks at the average lung function over time.



In short: Lung function in PCD may decline with age, but more data from adults is needed to understand the full pattern.

Audiology

Figure 10: This shows results of hearing tests done for children and adults with PCD. Most people had normal or mild hearing loss. Hearing problems are common in PCD and the extent of hearing loss is different between individuals.



Hearing aids: Approximately 1 in 5 people with PCD in the Registry in 2024 needed hearing aids

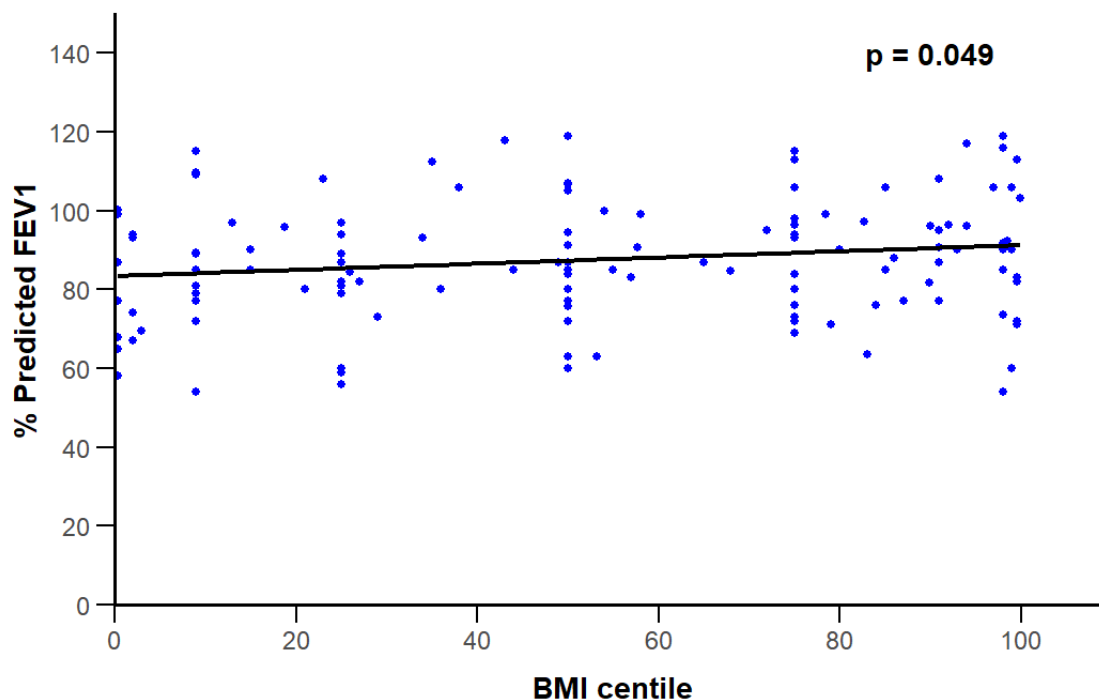
In short: While many PCD patients hear normally or only slightly less well, some may have more significant hearing issues.

Nutrition

This graph shows how weight (nutrition) is related to lung function in people with PCD.

- The trend line shows that people with better nutrition tend to have better lung function
- This highlights the importance of eating well to help keep lungs healthy

Figure 11: This graph looks at the relationship between weight and lung function. The trend line found that people with better nutrition had better lung function. This shows the importance of eating well to keep your lungs healthier.



In short: Good nutrition is linked to better lung function in people with PCD.

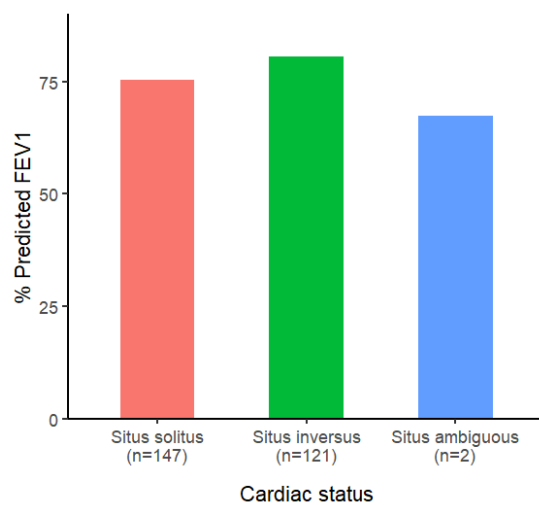
Cardiology

Cardiac situs

We looked at lung function (measured by a test called FEV1) in people with PCD, comparing groups based on how their internal organs are arranged (called cardiac situs). The three groups were:

- Situs solitus (organs in the usual position)
- Situs inversus (organs are reversed, like a mirror image)
- Situs ambiguous (organs are arranged differently or irregularly)

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



In short: We found that lung function was similar in people with situs solitus and those with situs inversus, with no meaningful difference between the two groups.

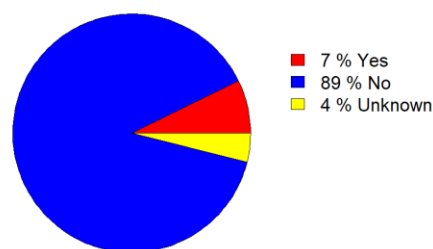
Congenital heart disease (CHD)

This chart shows how many people with PCD have a heart condition they were born with (called congenital heart disease, or CHD), compared to those who do not.

It divides patients into two groups:

- Those with CHD (a heart problem present from birth)
- Those without CHD

Figure 13: Pie chart of the presence of CHD or not in PCD patients enrolled in the PCD Registry in 2024

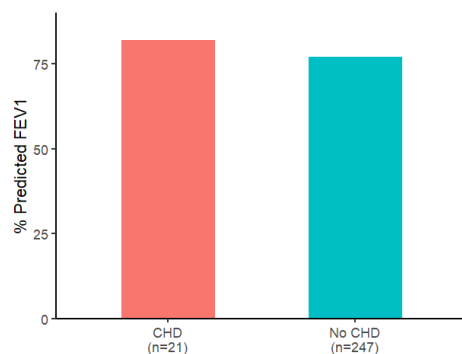


In short: The chart gives an overview of how common congenital heart disease is among people in the PCD Registry. Most people with PCD did not have CHD.

We also compared lung function (measured by FEV1) in people with PCD who have a congenital heart condition (CHD) and those who do not.

The results show the average lung function in each group, helping us see whether having a heart condition is linked to differences in breathing ability.

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



In short: There was no meaningful difference in lung function between those with an underlying heart condition to those who do not.

Microbiology

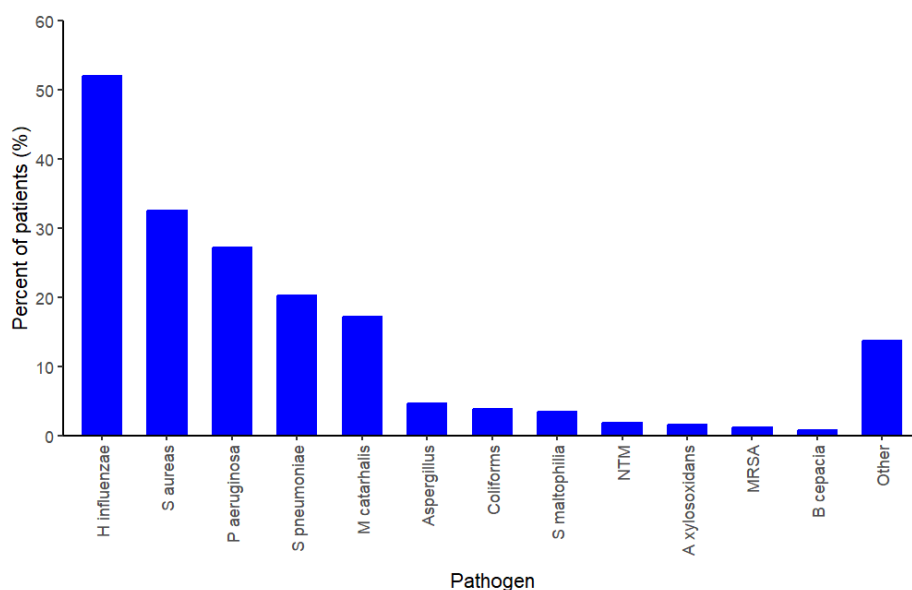
This section shows the percentage of people in the National PCD Registry in 2024 who had different types of bacteria ('germ' or respiratory pathogens) found in their airways over the course of a year.

It's important to note that some patients had more than one type of germ detected during the year, so they may be counted in more than one group.

The chart includes common bacteria, as well as an "Other" group. This "Other" category includes less common bacteria such as:

- Enterobacter
- Other types of Pseudomonas
- Other types of Streptococcus
- Candida (a type of yeast)
- Serratia

Figure 15: Graph showing types of bacteria found



In short: The chart shows how common different airway bacteria are found in people with PCD, and that some people had multiple infections in the same year.

We also looked at *Pseudomonas aeruginosa* in more detail. We compared lung function (measured by FEV1) in people with PCD in 2024, looking at those who had a long-term presence of this bacteria (were 'colonised' with it) and those who did not.

"Colonised" means the bacteria is **regularly found in the airways**, even if it isn't always causing symptoms.

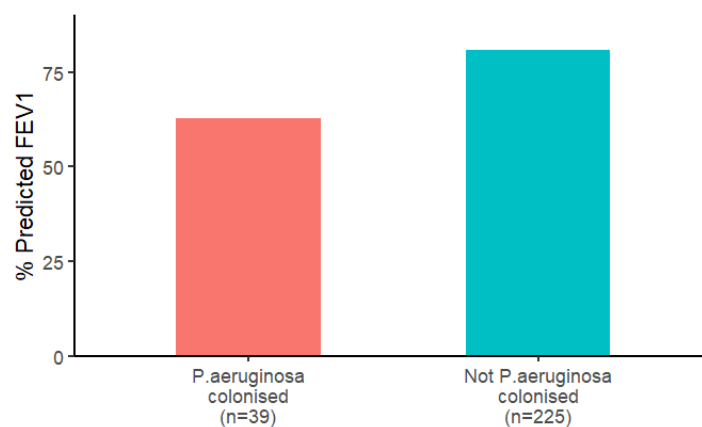
We found that in 2024, 42 people in the National PCD Registry were colonised with *Pseudomonas aeruginosa* in their lungs (. Of these, 39 had lung function results available.

We found that people who were colonised with this bacteria had lower lung function on average compared to those who were not:

- Lung function (FEV1) was 62.8% in the colonised group

Lung function (FEV1) was 80.8% in the non-colonised group. However, importantly, the people in the colonised group were also significantly older than those in the non-colonised group. We know that lung function is lower as people with PCD get older which might explain why the lower lung function seen in the colonised group was lower. For reference, the median age in the colonised group was 38.5 years and was 15 years in the non-colonised group.

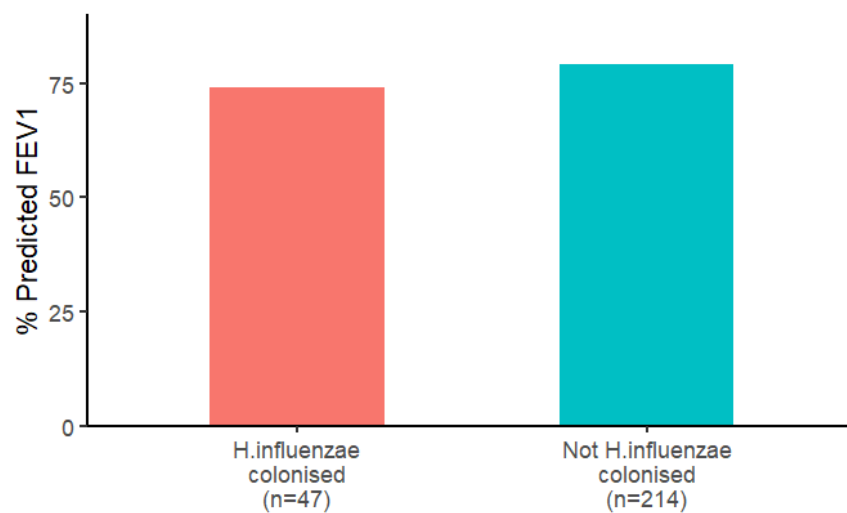
Figure 16: Graph comparing lung function between *Pseudomonas aeruginosa* colonised and non-colonised groups



In short: PCD patients with long-term *Pseudomonas aeruginosa* in their lungs had lower lung function, but they were also older, which may be an important factor to consider.

Another bacteria we looked into was *Haemophilus influenzae*. We compared lung function (measured by FEV1) in people with PCD in 2024, looking at those who had a long-term presence of a bacteria called *Haemophilus influenzae* and those who did not.

Figure 17: Graph comparing lung function between *Haemophilus influenzae* colonised and non-colonised groups



In short: We found no meaningful difference in lung function between the colonised and non-colonised groups for *Haemophilus influenzae*.

Radiology

We compared lung function (measured by FEV1) in people with PCD who had normal/clear chest X-rays and those with chest X-rays that had changes present (abnormal).

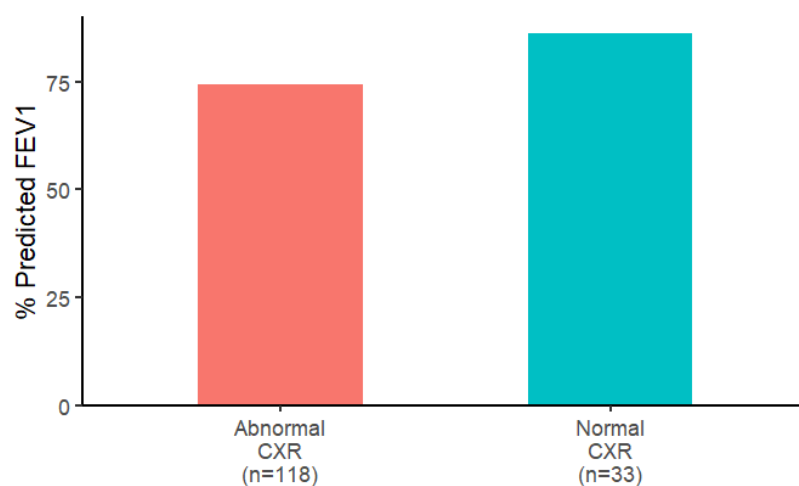
We found that people with abnormal chest X-rays had a lower lung function on average:

- **74.3%** in the abnormal group
- **86.1%** in the normal group

For context:

- The median age of the abnormal X-ray group was 16 years
- The median age of the normal X-ray group was 10 years

Figure 18: Lung function values based on if chest x-ray was normal or not



In short: PCD patients with abnormal chest X-rays tended to have lower lung function, although they were also slightly older on average.

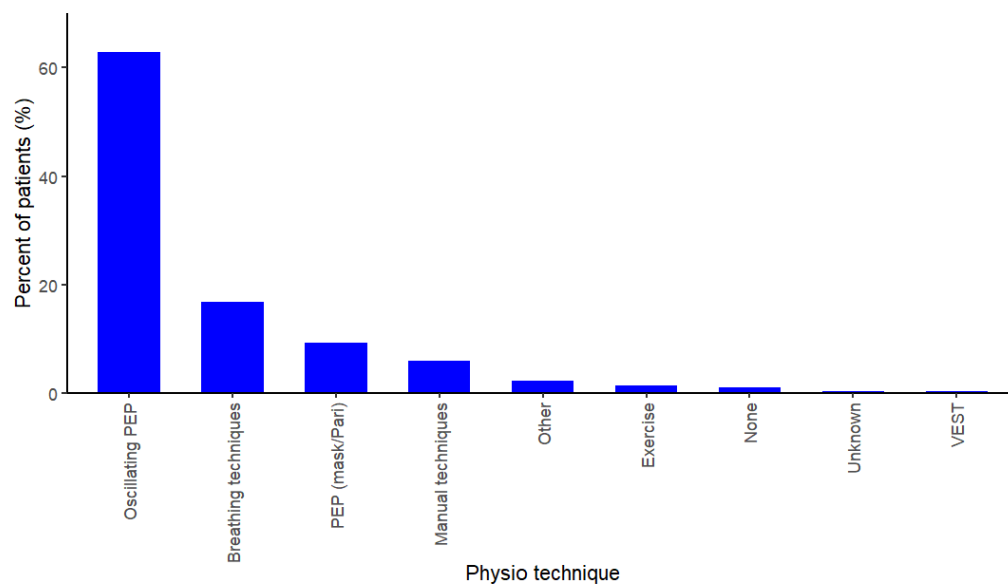
Treatments

Physiotherapy

This shows the main type of airway clearance physiotherapy that is recommended for people with PCD in the National PCD Registry in 2024.

Airway clearance physiotherapy helps clear mucus from the lungs to reduce infections and improve breathing and is the most important regular treatment that people with PCD should undertake.

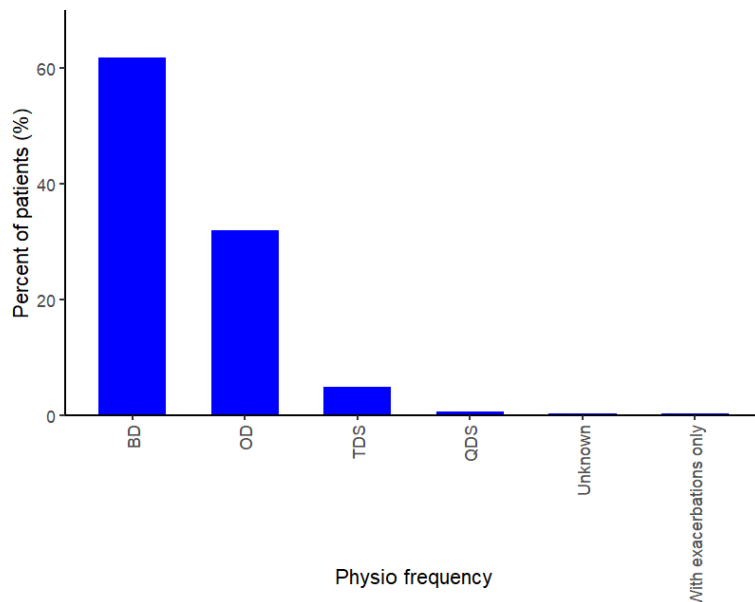
Figure 19: Type of airway clearance physiotherapy recommended to PCD patients



In short: The graph below shows that oscillating PEP, breathing techniques and PEP were the most common types of airway clearance recommended by professionals.

The next graph shows how often airway clearance physiotherapy is recommended.

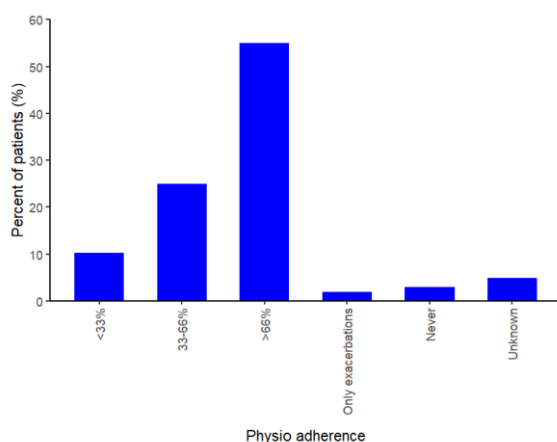
Figure 20: Frequency of airway clearance physiotherapy recommended to PCD patients enrolled in the National PCD Registry



In short: Most people with PCD were recommended to complete physiotherapy twice daily.

The next graph looks at how well people with PCD followed the recommended schedule for airway clearance physiotherapy.

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



In short: The majority of people with PCD report completing their physiotherapy on a daily basis.

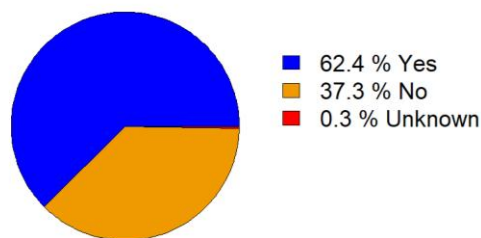
Mucolytics

Hypertonic saline nebulisation

This shows the percentage of people with PCD in the National PCD Registry who were prescribed nebulised hypertonic saline.

Hypertonic saline is a saltwater solution breathed in through a nebuliser that helps loosen mucus in the lungs, making it easier to clear.

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

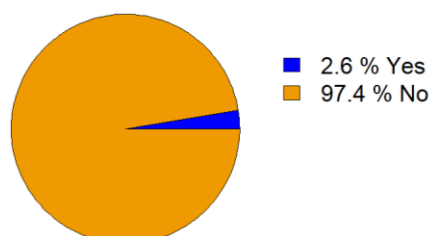


In short: Most people with PCD were prescribed nebulised hypertonic saline in 2024.

Dornase alfa nebulisation

This looks at the percentage of people with PCD who were prescribed dornase alfa to inhale through a nebuliser. Dornase alfa is a medication that thins mucus in the lungs, making it easier to cough up and clear.

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



In short: Most people with PCD were not prescribed dornase alfa in 2024.

Other treatments

Prophylactic antibiotics

This looks at how many people with PCD were prescribed antibiotics to stop them having frequent infections and which antibiotics were commonly used for this.

One hundred thirty-one patients were prescribed oral prophylactic antibiotics and 31 were prescribed nebulised prophylactic antibiotics. Nebulised antibiotics are commonly used for people who grow *Pseudomonas aeruginosa*.

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

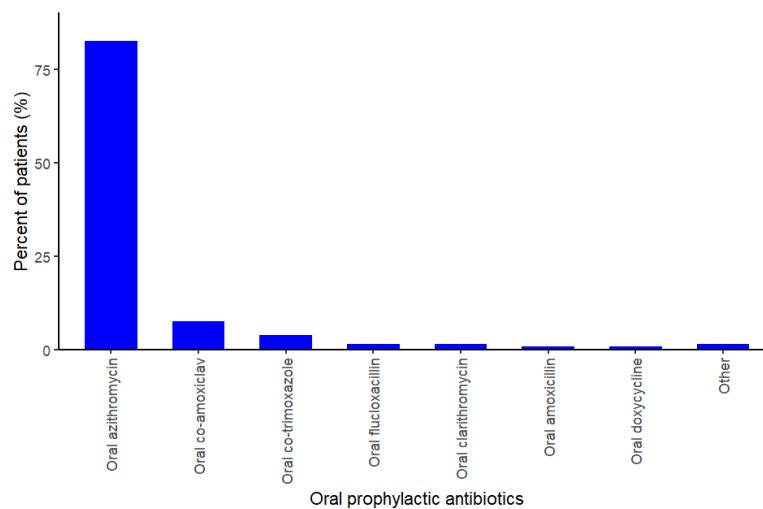
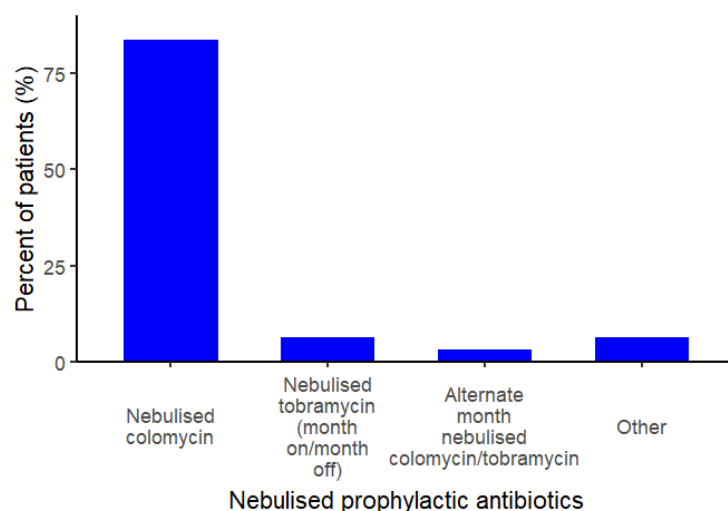


Figure 25: Percentage prescribed which nebulised prophylactic antibiotics in PCD patients enrolled in the National PCD Registry



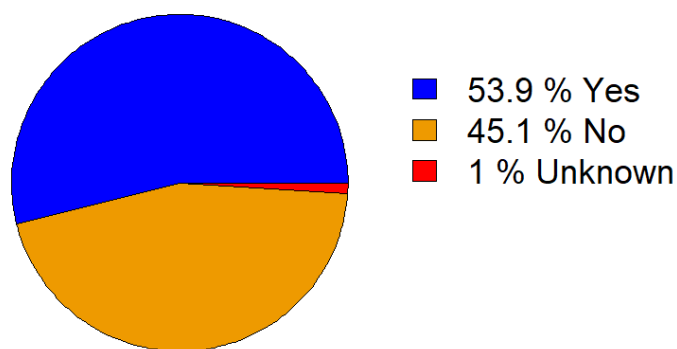
In short: Azithromycin was the commonest oral antibiotic used. For those with pseudomonas, the commonest nebulised antibiotic was colomycin.

Nasal douching

This section shows the percentage of people with PCD in the National PCD Registry who were prescribed nasal douching.

Nasal douching involves rinsing the nose with a saltwater solution to help clear mucus, reduce infections, and improve breathing.

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



In short: The chart shows that just over half of people with PCD were prescribed nasal douching.

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.

Glossary of terms

Diagnostic test:

- **Dynein arms:** Microscopic (tiny) structures within cilia that enable them to beat. If they are absent or not constructed properly the cilia will usually be static (not moving) or twitching.
- **Microscopic:** Anything that is so tiny that it is invisible to the naked eye and can only be seen with special microscopes.
- **Nasal nitric oxide (nNO) test:** A simple breathing test to measure the level of nitric oxide in the nose and sinuses. In PCD, nNO tends to be lower than in the general population, but not always.
- **ppb:** Parts per billion, a unit used to measure nNO levels. This is sometimes converted to nanolitres per min (nL/min).
- **Transmission electron microscopy (TEM):** A two-dimensional (2D) examination of cells at high magnification to examine the internal structure and identify defects within the cilia's ultrastructure which is carried out routinely for all patients who have PCD testing. Around 15% of people with PCD have normal TEM results.
- **Ultrastructure:** The fine structure within the cilia, made up of microtubules, dynein arms and other microscopic structures.
- **DNA:** The genetic code that determines all the characteristics of a living thing. It is like an 'instruction book' to determine what we look like and how we function. 'Spelling mistakes' (known as 'mutations', see below) within the code can cause PCD.
- **Gene:** A unit of DNA that controls the development of one or more characteristics such as eye/ hair colour or how our cilia work.
- **Genetics:** The study of genes and how they are inherited. In relation to PCD, genetics is usually describing the test or result which finds the gene responsible for causing PCD. We only know about approximately two thirds of PCD causing genes which means a negative result does not mean you don't have PCD, although it does make this less likely.
- **Mutation:** If we think of our DNA as an instruction book, a mutation within a gene is like a 'spelling mistake'. Some of these mutations cause no issues whilst others can cause genetic conditions such as PCD. You normally need to inherit a mutation from both parents to have PCD.

PCD Management terms

Cardiac

- **Cardiac:** Term referring to the heart.
- **Congenital:** Present from birth
- **Dextrocardia:** Heart situated on the right side of the chest
- **Heterotaxy:** Birth defect that involves heart and other organs. 'Hetero' means 'different' and 'taxy' means 'arrangement'. Heterotaxy therefore means 'different or unusual placement' (of the organs). There are different forms of heterotaxy which can involve the formation and placement of the heart, stomach, intestines, liver, spleen and lungs.
- **Situs inversus (totalis):** Complete mirror image placement of the heart and other organs.
- **Situs solitus:** Normal organs arrangement.

Testing/Investigations

- **Audiology:** Hearing test. There are different ways to assess hearing depending on age. Testing is painless. The frequency of your hearing tests will be different depending on age and previous issues.
- **Culture:** Grow or multiply. Used in PCD diagnostics regarding the growing of new cilia to analyse – ALI culture. Also in relation to sputum samples/ microbiology where the laboratory examine bugs grown from cough swab or sputum.
- **Microbiology:** The study of small organisms (bugs) such as bacteria, viruses, fungi etc. Sometimes used in the clinic to refer to the results of cough swab/ sputum culture or the laboratory who run the tests.
- **Oral flora:** Sometimes called normal flora. This refers to the normal bugs we grow in our mouths that don't cause us a problem.
- **Pathogenic:** Causing or capable of causing disease. Respiratory pathogens refer to bugs that grow within the lungs or nose/sinuses, that might cause people to be unwell. You may also see this term in the genetic diagnosis of PCD, referring to mutations/spelling mistakes within the gene that might cause PCD.
- **Sputum sample:** This is a sample of mucus coughed up from the chest that can be sent to the laboratory to test for bugs that might grow there. Sputum, mucus and phlegm are often used to mean the same thing.
- **X-ray:** Images/pictures of the inside of the body.
- **Lung Function Test:** These are a range of tests that look at how well your lungs are working to help you breathe.
 - **FEV1/ Forced expiratory volume in 1 second:** This is the amount of air that you can blow out in the first second during your spirometry test.
 - **FVC/ forced vital capacity:** This is the total amount of air that you can breathe out in one blow during your spirometry test.

Medications

- **Antibiotics:** A drug used to treat infections caused by bacteria rather than viruses.
- **DNase (Dornase Alfa):** A type of enzyme that breaks down the DNA and therefore reduces the thickness of the mucus making it easier to clear. This is delivered as a nebuliser.
- **Hypertonic saline:** Special salty water nebuliser prescribed before or during physiotherapy
- **Nasal rinse/douche:** Washing salty water solution into the nose and sinuses to clear them of mucus, sometimes referred to as sinus rinse/douche.
- **Nebuliser/nebs:** A fine mist of medication that is inhaled into the lungs.
- **Prophylactic:** Medication taken to help prevent illness.
- **Medication abbreviations:**
 - OD: Once a day
 - BD: twice a day
 - TDS: three times a day
 - QDS: four times a day
 - PRN: as needed
 - Nocte: at night

Physiotherapy

- **Acapella:** Airway clearance device used for physiotherapy (oscillating PEP)
- **Aerobika:** Airway clearance device used for physiotherapy (oscillating PEP)
- **Autogenic drainage:** Breathing technique for airway clearance physiotherapy.
- **Bubble Pep:** Airway clearance device used for physiotherapy (blowing bubbles in a bottle of water)
- **Nebuliser/Compressor:** The box that powers a nebuliser.
- **Expectorate:** To cough out sputum / mucus.
- **Forced expiratory technique (FET):** Short sharp or long deep “huff” or breath out like you are trying to steam up a mirror.
- **Oscillating PEP:** A PEP device which also vibrates.
- **PEP/PEP device:** Positive expiratory pressure. Something making it harder to breath out. A device which provides resistance as you breath out through it.