Cystic Fibrosis Trust

UK Cystic Fibrosis Registry

Annual Data Report 2023 — Scotland

March 2025

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UK Cystic Fibrosis Registry
Annual Data Report 2023 — Scotland

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Acknowledgements

First and foremost, UK Cystic Fibrosis Registry team would like to thank people with cystic fibrosis and their families for their support, as well as anyone who has generously donated to Cystic Fibrosis Trust. We would also like to express our gratitude to the UK cystic fibrosis centres and clinics for their continued dedication to obtaining consent and submitting data to the Registry.

Contact information

For more information about this report, or the UK Cystic Fibrosis Registry, please contact us: **registry@cysticfibrosis.org.uk @CFTrust**

Cystic Fibrosis Trust

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Executive summary

Welcome to the 2023 UK CF Registry Annual Data Report for Scotland. I hope you find things that give cause for celebration and pause for thought on what might be done to make care better for people with CF moving forward.

There's a lot of data presented here – so some things that I've reflected on are summarised here:

The age distribution (Section 1.3) is shifting as we would expect – though within Scotland this is quite variable, particularly around adolescence with a larger proportion transitioning to adult care now than 10 years previously.

There has been an increase in the proportion of 10 to 14-year-olds in the overweight BMI category with nearly 1 in 4 above the 90th percentile for BMI. (Section 1.7)

For adults, the change observed has been even more stark over the past five years, with an increase in the proportion of people in the overweight and obese BMI categories. Going forward, proportionate and personalised advice on the role of integrated exercise and tailored dietary input for a generation predominantly exposed to modulator therapies, should gain greater clinical focus if people with cystic fibrosis are to lead healthy, longer lives.

Lung health, as measured by FEV_1, is making transformational leaps (Section 1.15). These are to be heralded. However, these figures include data for those who have no access to modulator therapies, for whom there is yet to be medications offering this same transformation. For those with access to modulator therapies, there needs to be a continual refresher that this transformation is reliant on, consistent adherence.

Frequent airway sampling continues to be a mainstay in paediatric care (94% had three samples or more) but has halved in adult care over the past five years (72% down to 35%). Drier airways (and less sputum for samples) are of course a good thing, but these data highlight the need to develop better methods of identifying and (re)defining chronic lower airway infection.

With a population thankfully living longer, we need Registry data to help inform and resolve any potential long-term impacts of early life exposures, such as high fat diets, gut inflammation, ototoxic medicines and burden of treatment on our community. Section 1.21 begins to build this picture – and clinical engagement with this data will be pivotal in identifying early signs of those longer-term impacts.

Mucoactive therapies do appear to be reducing in use, particularly for hypertonic saline in those over 16 years of age, where use has halved over the last five years. DNase use is however declining at a much slower pace as people evaluate their individual response to modulator therapies and the risk/benefit of stopping. Results from the Registry-based trial CFSTORM will help inform future care in this space.

CFTR modulator use is now standard of care for most people with CF (Section 1.35), however, there remains individuals with eligible genotypes who have no record of CFTR modulator use. Patients, parents and clinic staff will all wish to ensure that eligibility is reviewed in those not currently taking CFTR modulators, to ensure any prior assessment of eligibility remains correct.

Numbers are small, but the reduction in patients evaluated and receiving lung and liver transplantation over the past five years, paired with an increase in life expectancy, is really positive to see.

The proportion of people living with CF in Scotland with annual review data was 76% in 2023 - our learning is based on just over three-quarters of the population. Entering data into a data registry can be tough and time consuming, but the data summarised in this report will help clinicians improve care of their patients so that people with CF live better lives now and in the future. From all in the UK CF Registry team – thank you.

With very best wishes

Steve Cunningham
Professor of Paediatric Respiratory Medicine
University of Edinburgh

Introduction

This report is aimed at anyone who is interested in the health, care, and outcomes of people with cystic fibrosis (CF) in the UK. This includes people with CF, their families and clinical teams, healthcare managers, commissioners, and policy makers.

You can find a glossary of scientific and clinical terms on page 60.

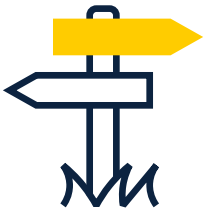
Cystic fibrosis

Cystic fibrosis is an inherited disease caused by a faulty version of a gene known as CFTR. The gene and the protein it makes help control the movement of salt and water in and out of cells. When the gene, and the protein it makes, is faulty, it can cause thicker mucus. One of the main areas affected is the lungs; over time this thick mucus blocks and damages airways, leading to infections and making it hard to breathe. People with CF may also develop other problems, such as liver disease or CF diabetes (CFD). Around 85% of people with CF also have difficulty digesting food.

UK Cystic Fibrosis Registry

The UK CF Registry has been sponsored and hosted by Cystic Fibrosis Trust since 2007. It is a database of consenting people with CF in the UK. The Registry collects demographic, treatment and health outcomes data. You can find a full list of the data items we collect at **cysticfibrosis.org.uk/registry**.

The purpose of the UK CF Registry is to improve the health of people with cystic fibrosis. This is done in a number of ways:



helping people with CF and their families understand CF, and make informed decisions

giving clinical teams the evidence they need to improve the quality of care

monitoring the safety and effectiveness of new treatments for cystic fibrosis

providing data for research to find out the best ways to treat cystic fibrosis

helping commissioners provide funding to NHS CF centres that is proportionate to their patients' disease severity

supporting clinical trials through feasibility studies and pragmatic data collection

Governance

The Registry Steering Committee (RSC) is responsible for making sure that the UK CF Registry is compliant with data protection legislation, and its Research Ethics Committee-approved Study Protocol. It also makes recommendations about the future development of the Registry.

The Registry Research Committee, which is a subcommittee of the RSC, assesses applications for data and guides the Registry research strategy.

Please see Appendix 1 of the UK Cystic Fibrosis Registry 2023 Annual Data Report.

Data are only recorded on the UK CF Registry if explicit written consent is given by the person with CF or, for a child, their parent or guardian.

When data are provided to third parties such as the NHS or university researchers, they are either anonymised (all identifiable data removed completely) or pseudonymised (all identifiable data replaced with a unique identification number). Pseudonymisation is used so that data can be traced back to what is in the 'live' database by the Registry team for the purposes of updating the data or answering queries. This means that the Registry data used for research, and the results cannot identify the people whose data are stored on the UK CF Registry.

If requests from pharmaceutical companies are granted, for research or submissions to regulators or the NHS, the data are analysed and aggregated by Registry statisticians and only summary data are provided.

Data collection

Data are entered onto the UK CF Registry by NHS employees at CF centres in the UK using a secure web portal.

Where can I find more information?

You can find out more about CF, and the UK CF Registry, at **cysticfibrosis.org.uk/registry**.

Section 1: Scotland-wide analysis

This section provides an overview of the cystic fibrosis (CF) population, health outcomes, and care in Scotland, with comparisons to the full CF population of the United Kingdom, including CF centres in England, Northern Ireland, Scotland, and Wales.

1.1 Summary of the UK Cystic Fibrosis Registry

	2023	
	UK	Scotland
CF patients registered[1]	11318	972
Excluding diagnoses that year	11146	956
CF patients with an annual review; n[2]	10344	723
Age in years; median[3]	22	22
All newly diagnosed patients (newborn screening and other)[4]	173	16
Number of patients born identified by newborn screening[4]	124	9
Age at diagnosis in months; median[3]	1.3	1.3
Adults aged 16 years and over; %[3]	63.7	459
Males; %[3]	53.3	53.9
Genotyped; %[3]	99.4	99.7
Total deaths reported during annual review year (%)[5]	49	6
Age at death in years; median (95% CI)[5]	46 (37, 55)	55*

Annual review: A Registry annual review form contains a combination of data relating to a person with CF's yearly annual review appointment at their CF centre, and their clinical care and health over the past 12 months.

Notes:

1 Number of patients diagnosed with CF, seen in the past two years, and alive at 1 January in the given year.

2 As patients newly diagnosed in a given year may not have their first annual review in the same year, the proportion with an annual review is calculated from the total registered excluding those diagnosed in the given year.

3 Calculated from patients with an annual review in the given year (see footnote 2 above).

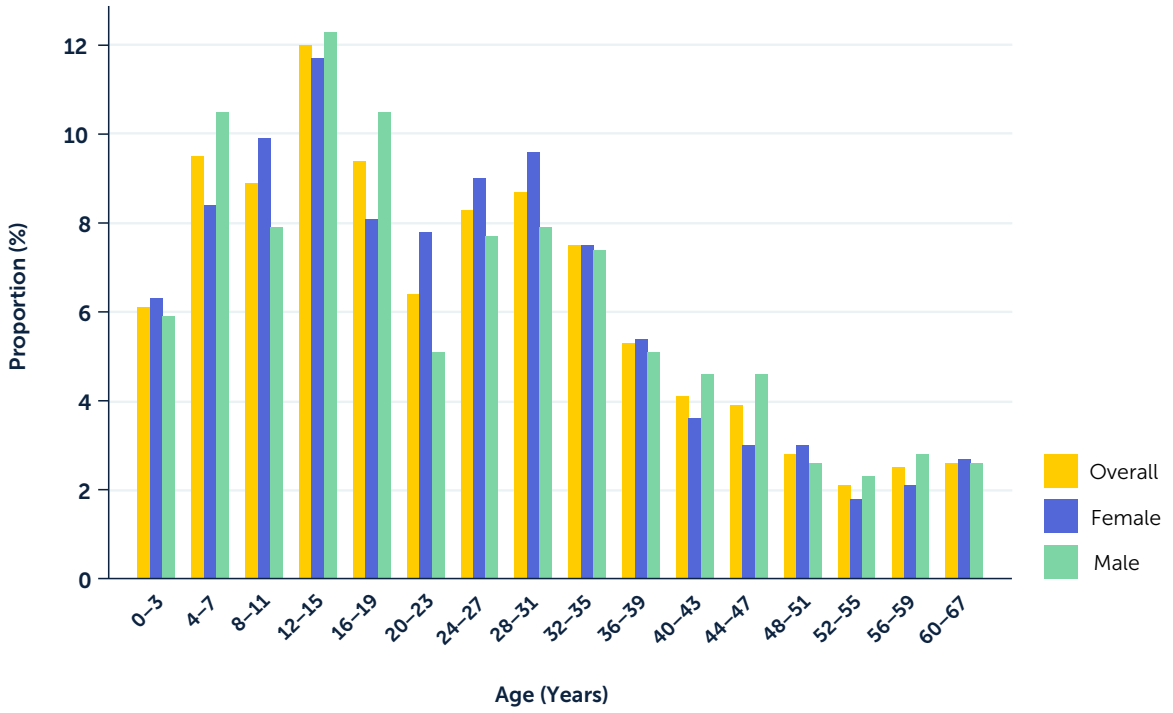
4 Calculated from all patients registered on the database.

5 Calculated from all registered patients who died in the given year.

* CI not provided due to small sample size.

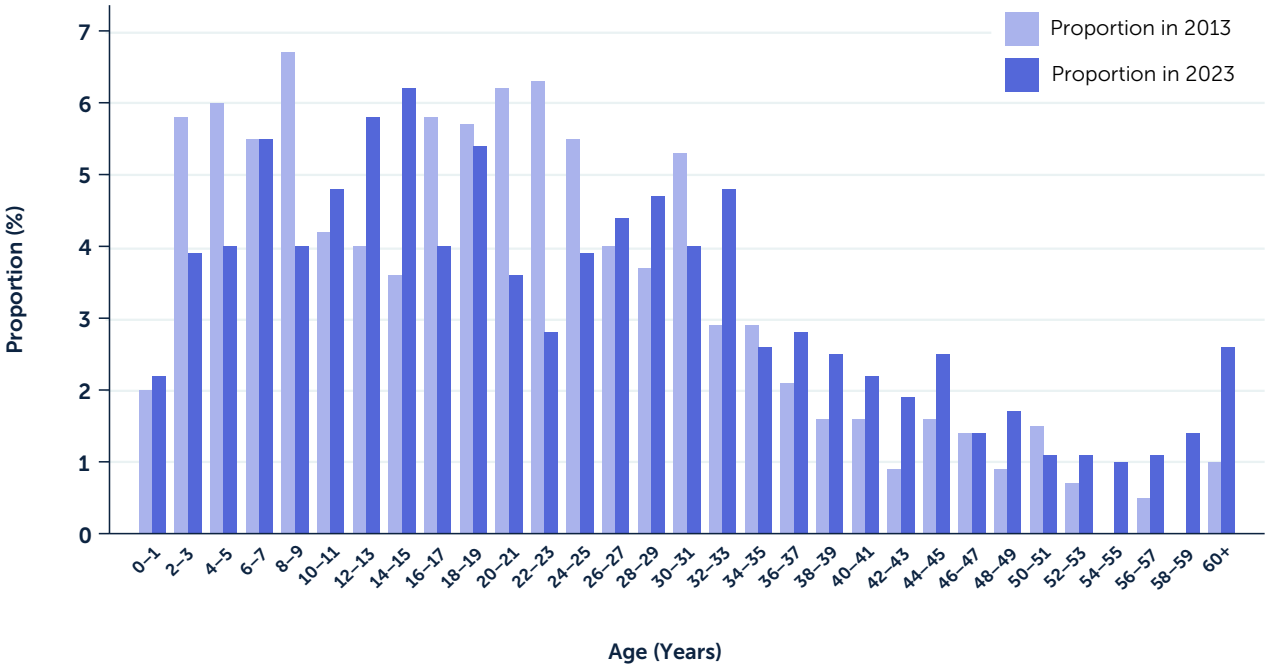
1.2 Age distribution by sex

The following chart shows the mix of ages by sex in the CF population in Scotland.



Age	All; n(%)	Females; n(%)	Males; n(%)
0-3	44 (6.1)	21 (6.3)	23 (5.9)
4-7	69 (9.5)	28 (8.4)	41 (10.5)
8-11	64 (8.9)	33 (9.9)	31 (7.9)
12-15	87 (12.0)	39 (11.7)	48 (12.3)
16-19	68 (9.4)	27 (8.1)	41 (10.5)
20-23	46 (6.4)	26 (7.8)	20 (5.1)
24-27	60 (8.3)	30 (9.0)	30 (7.7)
28-31	63 (8.7)	32 (9.6)	31 (7.9)
32-35	54 (7.5)	25 (7.5)	29 (7.4)
36-39	38 (5.3)	18 (5.4)	20 (5.1)
40-43	30 (4.1)	12 (3.6)	18 (4.6)
44-47	28 (3.9)	10 (3.0)	18 (4.6)
48-51	20 (2.8)	10 (3.0)	10 (2.6)
52-55	15 (2.1)	6 (1.8)	9 (2.3)
56-59	18 (2.5)	7 (2.1)	11 (2.8)
60-67	19 (2.6)	9 (2.7)	10 (2.6)
<16	264 (36.5)	121 (36.3)	143 (36.7)
≥16	459 (63.5)	212 (63.7)	247 (63.3)
<18	293 (40.5)	134 (40.2)	159 (40.8)
≥18	430 (59.5)	199 (59.8)	231 (59.2)
Overall	723	333	390

1.3 Age distribution of the UK CF population in 2013 vs 2023
N=723 in 2023, N=810 in 2013



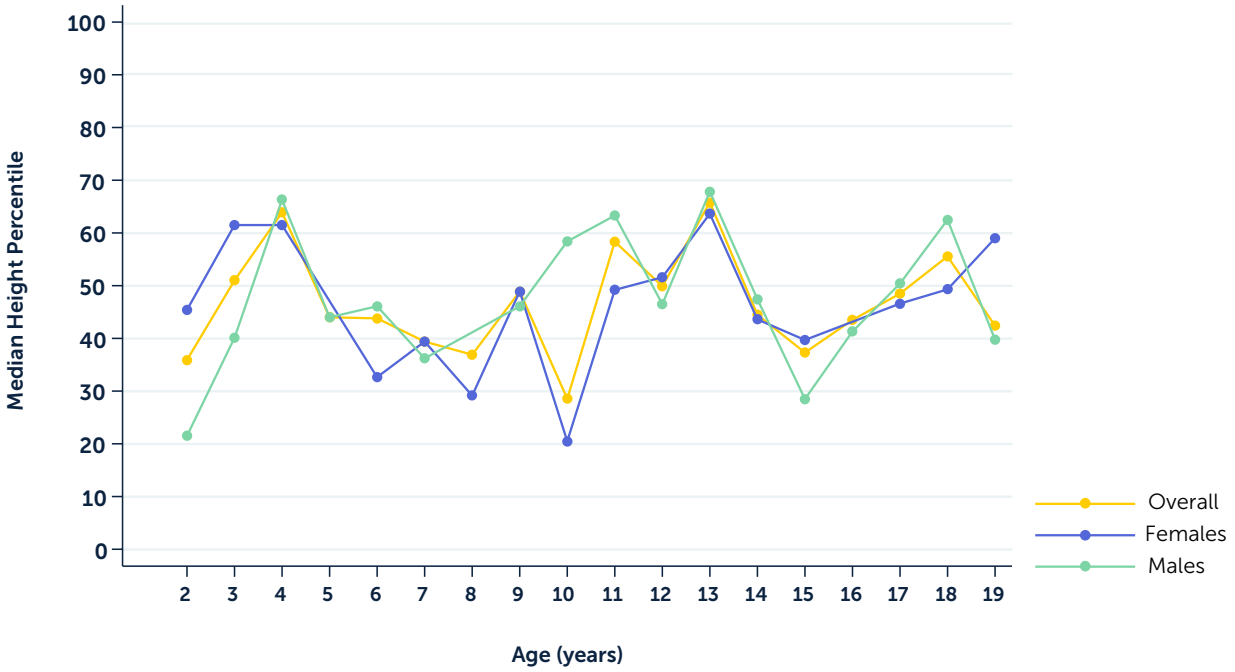
1.4 Ethnicity

Ethnicity n(%)	2013	2018	2023
Total	**810**	**819**	**723**
Total known[1]	**809**	**812**	**721**
White	788 (97.4)	796 (98.0)	697 (96.7)
Asian	14 (1.7)	9 (1.1)	18 (2.5)
Black	0 (0.0)	0 (0.0)	<5
Mixed	<5	<5	-*
Other	-*	-*	0 (0.0)

1 Proportions are calculated from total known ethnicities.
* Redacted to adhere to statistical disclosure guidelines.

1.5 Height percentiles of children and young people (<20 years)[1] N=332

The following chart and table show the height percentiles of people with CF, aged 19 and under, in relation to UK growth data for the general population. If a person with CF is on the 40th percentile, only 40% of people the same age are their height or shorter; 60% are taller.

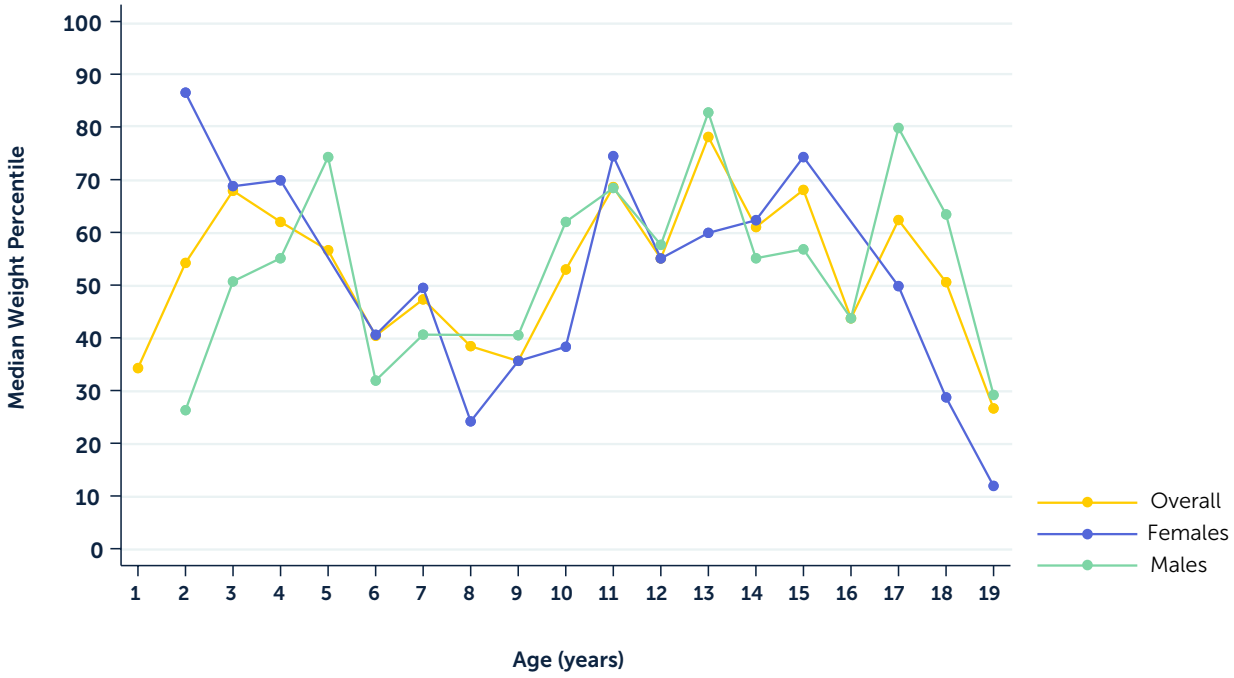


Age	Overall	Median	IQR	Female	Median	IQR	Male	Median	IQR
2	15	35.9	18.2-66.6	8	45.4	29.9-80.1	7	21.5	16.6-50.1
3	11	51.0	32.7-70.1	6	61.5	49.8-88.2	5	40.1	31.9-51.0
4	18	63.9	14.6-80.0	7	61.5	6.4-80.0	11	66.3	34.0-80.0
5	11	44.0	22.4-82.8	<5	-*	-*	7	44.0	22.4-82.8
6	23	43.8	15.0-76.5	10	32.7	19.2-53.0	13	46.1	15.0-76.5
7	17	39.4	23.4-72.5	7	39.4	29.9-72.5	10	36.2	20.7-80.3
8	13	36.9	18.8-46.4	10	29.2	18.8-46.4	<5	-*	-*
9	15	48.9	26.6-78.2	7	48.9	27.1-83.9	8	46.1	12.1-77.2
10	16	28.5	17.5-64.4	9	20.5	19.3-68.6	7	58.4	10.5-64.0
11	18	58.3	40.7-76.0	6	49.2	30.9-59.0	12	63.3	48.0-80.0
12	20	49.9	21.5-68.3	10	51.6	28.3-68.3	10	46.5	15.3-67.3
13	22	65.8	37.7-88.4	10	63.6	14.1-88.8	12	67.9	57.5-88.2
14	24	44.5	27.6-76.3	10	43.6	18.0-74.6	14	47.4	34.9-78.1
15	20	37.3	21.2-65.8	9	39.7	24.7-53.1	11	28.5	17.7-69.8
16	13	43.5	35.6-66.4	<5	-*	-*	9	41.3	35.6-61.4
17	16	48.5	28.1-64.6	9	46.6	29.0-59.7	7	50.4	27.2-65.2
18	22	55.5	18.4-67.7	9	49.3	13.7-65.3	13	62.5	39.9-67.7
19	17	42.4	12.3-59.0	5	59.0	2.1-59.0	12	39.8	13.6-59.4
Overall	**311****	**47.5**	**23.2-70.3**	**140**	**43.6**	**2.0-68.7**	**171**	**49.4**	**26.0-70.9**

1 Based on UK-WHO growth charts, 1990 (updated 1996).
* Redacted to adhere to statistical disclosure guidelines.
** Number with non-missing data and age 2 to 19 years.

1.6 Weight percentiles of children and young people (<20 years)[1] N=332

The following chart and table show the weight percentiles of people with CF, aged 19 and under, in relation to the UK growth data for the general population. If a person with CF is on the 40th percentile, only 40% of people the same age are their weight or lower; 60% weigh more.



	Overall			Female			Male		
Age	n	Median	IQR	n	Median	IQR	n	Median	IQR
1	12	34.2	9.3-59.7	<5	-*	-*	-*	.	13.9-59.7
2	17	54.2	26.4-86.5	9	86.5	54.2-92.8	8	26.2	11.4-62.8
3	11	67.9	39.8-80.7	6	68.8	64.5-80.7	5	50.7	36.2-68.6
4	18	62.0	43.6-81.4	7	69.9	27.3-84.7	11	55.1	43.6-81.4
5	11	56.6	21.8-80.1	<5	-*	-*	-*	74.3	21.8-85.5
6	23	40.4	10.4-62.2	10	40.6	31.6-56.9	13	31.9	5.6-62.2
7	17	47.3	32.6-82.1	7	49.5	33.4-96.9	10	40.6	28.7-82.1
8	13	38.4	20.6-71.2	-*	24.1	16.0-45.3	<5	-*	-*
9	15	35.6	17.5-69.3	7	35.6	29.4-69.3	8	40.5	16.5-63.7
10	16	53.0	25.1-62.5	9	38.3	20.7-57.7	7	62.0	45.8-65.8
11	19	68.6	25.3-92.9	6	74.4	43.9-79.7	13	68.4	24.4-92.9
12	20	55.0	15.4-86.4	10	55.0	15.4-86.9	10	57.7	32.4-76.0
13	21	78.1	54.3-93.7	9	59.9	24.2-90.0	12	82.8	64.3-94.2
14	24	61.0	53.3-77.9	10	62.3	60.0-77.5	14	55.1	27.0-90.6
15	20	68.1	33.8-79.2	9	74.3	53.0-79.4	11	56.8	23.6-79.0
16	13	43.7	39.1-71.8	<5	-*	-*	-*	43.7	42.5-82.7
17	16	62.3	43.6-85.1	9	49.8	41.0-70.4	7	79.8	50.3-88.7
18	22	50.6	27.3-79.8	9	28.7	8.9-63.9	13	63.4	28.2-88.7
19	17	26.6	11.9-41.8	5	11.9	3.0-34.0	12	29.2	16.4-46.7
Overall	**325****	**54.3**	**27.1-79.7**	**144****	**53.2**	**28.4-78.4**	**181****	**55.1**	**26.4-81.2**

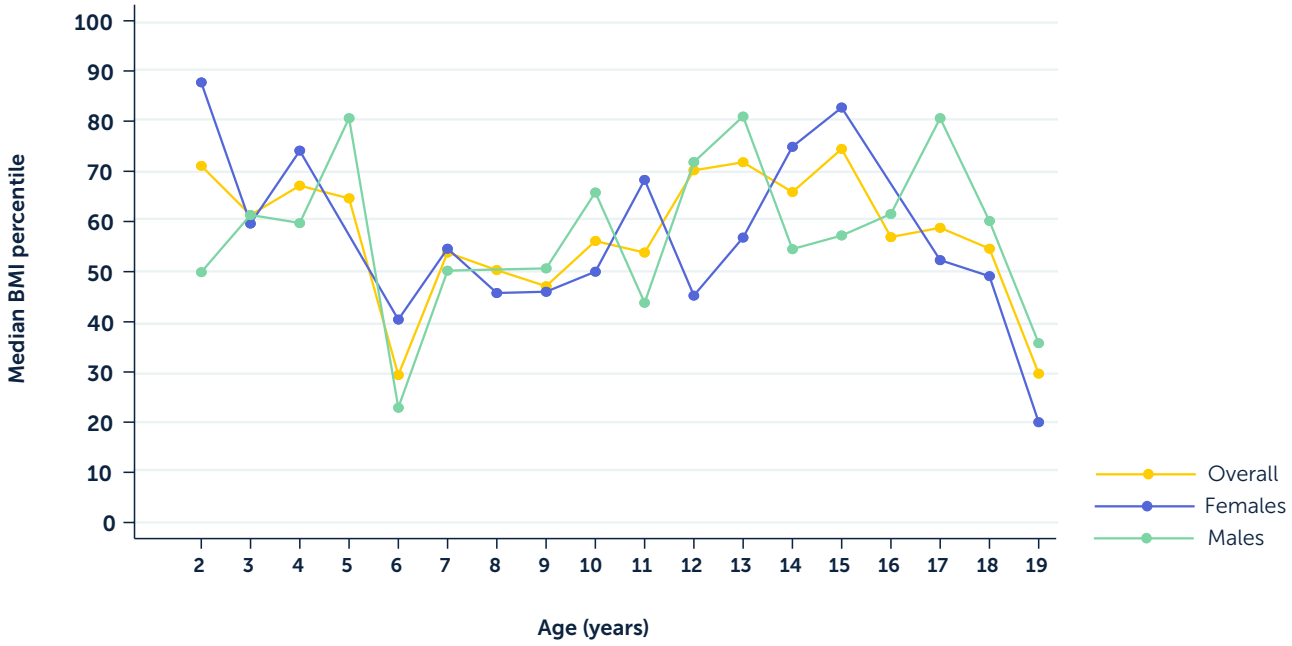
1 Based on UK-WHO growth charts, 1990 (updated 1996).

* Redacted to adhere to statistical disclosure guidelines.

** Number with non-missing data and age 2 to 19 years.

1.7a Body Mass Index (BMI) percentiles in children and young people (<20 years)[1]
N=332

The following chart and table show the BMI percentiles of people with CF, aged 19 and under, in relation to the UK growth data for the general population. If a person with CF is on the 40th percentile, it means that only 40% of the population at the same age are their BMI or lower; so 60% have a higher BMI.

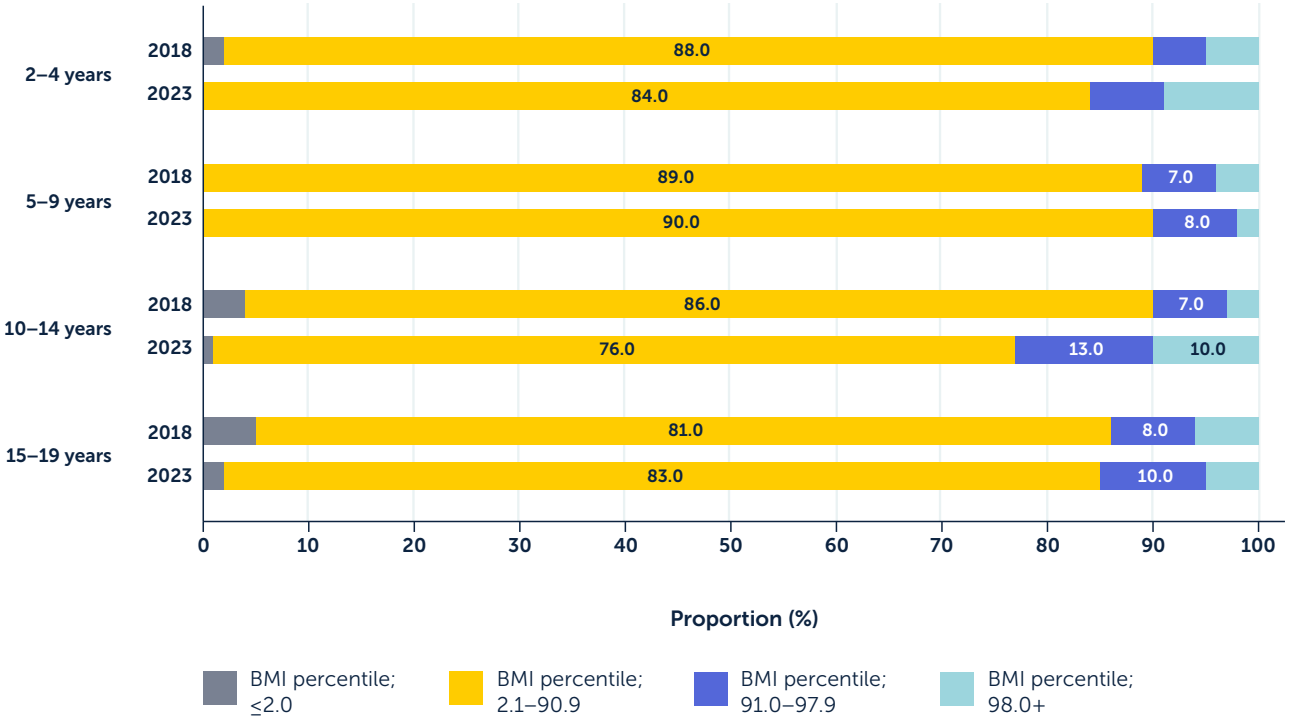


	Overall			Female			Male		
Age	n	Median	IQR	n	Median	IQR	n	Median	IQR
2	15	71.1	49.9-89.1	8	87.7	73.8-93.7	7	49.9	33.1-71.1
3	11	61.3	35.4-87.2	6	59.6	35.4-85.6	5	61.3	58.3-87.2
4	18	67.2	50.2-81.4	7	74.1	63.2-83.5	11	59.7	37.7-81.4
5	11	64.6	23.5-82.5	<5	-*	-*	-*	80.6	64.6-88.4
6	23	29.4	22.5-68.9	10	40.4	24.7-68.9	13	22.9	19.9-68.2
7	17	53.8	42.1-64.2	7	54.6	48.9-98.2	10	50.2	33.2-56.2
8	13	50.3	22.6-68.3	-*	45.8	15.1-60.4	<5	-*	-*
9	15	47.1	13.8-60.6	7	46.0	3.2-51.2	8	50.7	30.6-61.7
10	16	56.1	42.4-67.9	9	50.0	37.6-56.5	7	65.8	55.7-78.8
11	18	53.8	13.4-97.0	6	68.3	45.8-99.0	12	43.8	9.1-94.7
12	20	70.2	22.8-86.1	10	45.2	15.1-92.8	10	71.9	50.3-82.1
13	21	71.8	54.6-95.3	9	56.8	55.3-82.5	12	80.9	50.0-96.2
14	24	65.9	49.2-91.1	10	74.9	64.8-87.2	14	54.5	27.0-96.6
15	20	74.4	41.8-87.8	9	82.7	73.5-87.6	11	57.2	32.4-90.7
16	13	56.9	35.3-63.7	<5	-*	-*	-*	61.5	35.3-87.5
17	16	58.8	35.7-91.3	9	52.3	33.4-63.0	7	80.6	54.6-94.7
18	22	54.6	26.8-80.6	9	49.1	23.4-80.0	13	60.1	29.7-89.5
19	17	29.7	16.8-47.7	5	20.0	6.6-22.1	12	35.8	21.8-53.5
Overall	**310**	**56.8**	**30.5-82.9**	**139**	**56.3**	**29.8-82.7**	**171**	**58.0**	**30.5-83.4**

1 Based on UK-WHO growth charts, 1990 (updated 1996).
* Redacted to adhere to statistical disclosure guidelines.
** Number with non-missing data and age 2 to 19 years.

1.7b Body Mass Index (BMI) percentiles in children and young people (<20 years)[1] for 2018 and 2023

The following graph shows the change in BMI groups for children and young people with CF from 2018 to 2023.



			BMI category by age and year : n*(%)			
Age group	Year	Total number of people in each age group**	BMI percentile; ≤2.0	BMI percentile; 2.1-90.9	BMI percentile; 91.0-97.9	BMI percentile; 98.0+
2-4 years	2018	43	<5	38 (88.0)	<5	<5
	2023	44	0 (0.0)	37 (84.0)	<5	<5
5-9 years	2018	89	0 (0.0)	79 (89.0)	6 (7.0)	<5
	2023	79	0 (0.0)	71 (90.0)	6 (8.0)	<5
10-14 years	2018	101	<5	87 (86.0)	7 (7.0)	<5
	2023	99	<5	75 (76.0)	13 (13.0)	10 (10.0)
15-19 years	2018	63	<5	51 (81.0)	5 (8.0)	<5
	2023	88	<5	73 (83.0)	9 (10.0)	<5

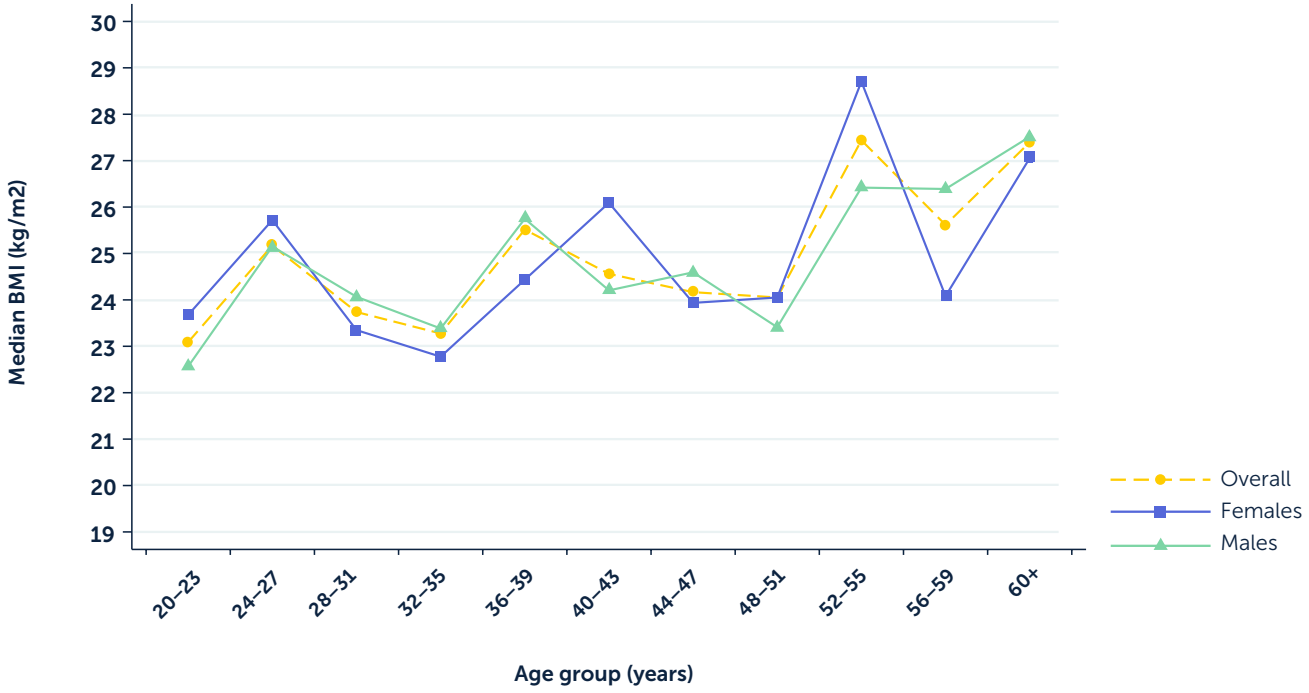
1 Based on UK-WHO growth charts, 1990 (updated 1996).

* Redacted to adhere to statistical disclosure guidelines.

** And with non-missing data.

1.8a Body Mass Index (BMI) in adults (≥20 years) N=391

The following chart and table show the BMI of people with CF aged 20 and over.

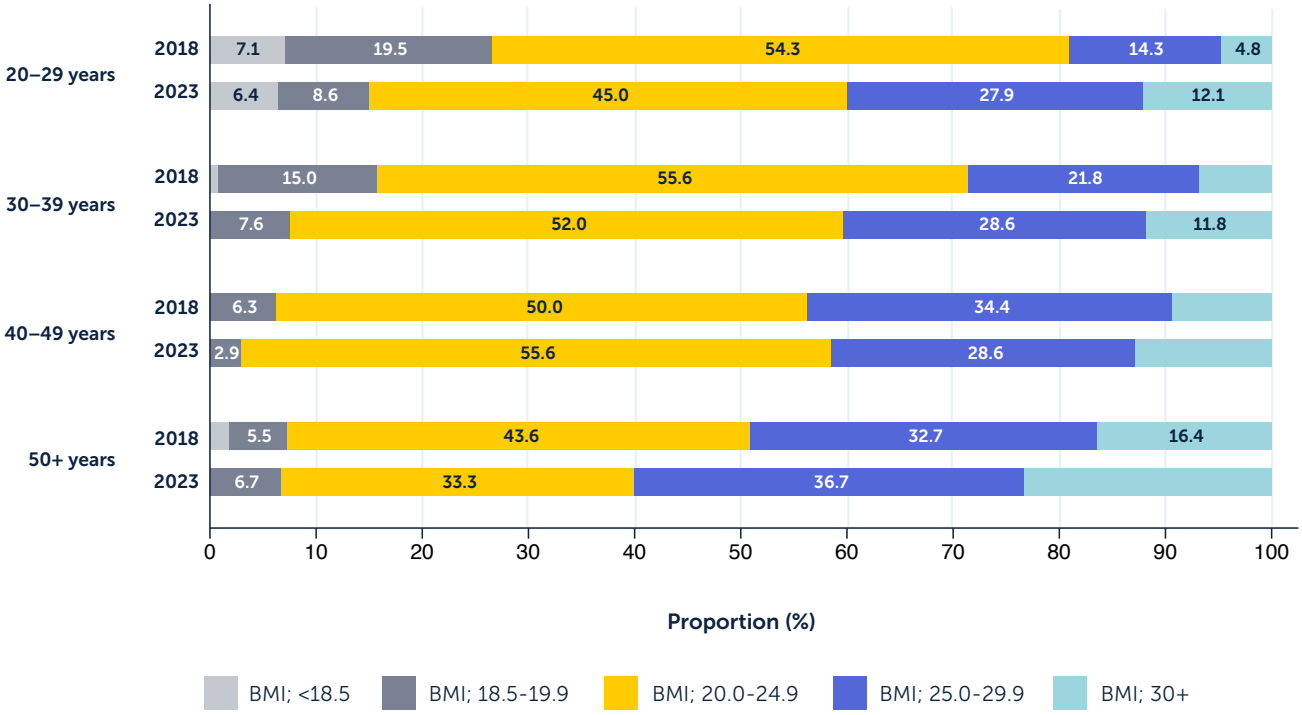


	Overall			Female			Male		
Age	n	Median	IQR	n	Median	IQR	n	Median	IQR
20-23	46	23.1	20.9-25.5	26	23.7	20.6-25.5	20	22.6	20.9-25.5
24-27	60	25.2	21.9-28.8	30	25.7	21.7-29.4	30	25.2	22.1-28.3
28-31	63	23.7	22.1-26.0	32	23.3	21.7-26.1	31	24.1	22.1-25.7
32-35	53	23.3	21.4-25.7	24	22.8	20.8-26.0	29	23.4	21.8-25.5
36-39	37	25.5	22.2-28.4	17	24.4	22.0-27.4	20	25.7	23.4-29.3
40-43	30	24.6	21.8-28.7	12	26.1	22.7-30.7	18	24.2	21.8-28.2
44-47	28	24.2	22.8-26.5	10	23.9	21.7-26.3	18	24.6	23.0-26.7
48-51	20	24.1	21.7-26.6	10	24.1	22.8-25.5	10	23.4	21.2-28.3
52-55	15	27.4	23.7-30.6	6	28.7	23.9-30.6	9	26.4	23.7-30.0
56-59	18	25.6	22.2-28.4	7	24.1	22.1-30.5	11	26.4	24.9-28.4
60+	19	27.4	24.4-32.2	9	27.1	24.4-34.2	10	27.5	25.2-29.5
Overall	**389****	**24.3**	**22.0-27.6**	**183****	**24.2**	**21.8-27.5**	**206****	**24.4**	**22.1-27.9**

** Number with non-missing data.

1.8b Body Mass Index (BMI) in adults for 2018 and 2023

The following graph shows the change in the proportion of people in each BMI group in 2018 and 2023.



			BMI category by age and year : n*(%)				
Age group	Year	Total number of people in each age group**	BMI; <18.5	BMI; 18.5-19.9	BMI; 20.0-24.9	BMI; 25.0-29.9	BMI; 30+
20-29 years	2018	210	15 (7.1)	41 (19.5)	114 (54.3)	30 (14.3)	10 (4.8)
	2023	140	9 (6.4)	12 (8.6)	63 (45.0)	39 (27.9)	17 (12.1)
30-39 years	2018	133	<5	20 (15.0)	74 (55.6)	29 (21.8)	-*
	2023	119	0 (0.0)	9 (7.6)	62 (52.1)	34 (28.6)	14 (11.8)
40-49 years	2018	64	0 (0.0)	<5	32 (50.0)	22 (34.4)	-*
	2023	70	0 (0.0)	<5	39 (55.7)	20 (28.6)	-*
50+ years	2018	55	<5	<5	24 (43.6)	18 (32.7)	9 (16.4)
	2023	60	0 (0.0)	<5	20 (33.3)	22 (36.7)	-*

** And with non-missing data.

1.9 Education and employment in adults (≥16 years) N=459

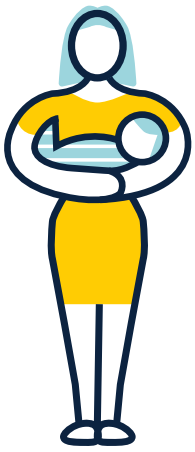
The following table shows how people with CF reported their education and employment status in 2023. Please note that the groups are not mutually exclusive; for example, someone may be a student as well as working part-time.

	Overall numbers of patients; n(%)	Male; n(%)	Female; n(%)
Numbers of patients; n	459	247	212
Number who completed questionnaire; n(%)	459 (100.0)	247 (100.0)	212 (100.0)
Full-time employment; n(%)	188 (41.0)	123 (49.8)	65 (30.7)
Part-time employment; n(%)	82 (17.9)	27 (10.9)	55 (25.9)
Student; n(%)	83 (18.1)	41 (16.6)	42 (19.8)
Homemaker; n(%)	11 (2.4)	<5	10 (4.7)
Unemployed; n(%)	67 (14.6)	38 (15.4)	29 (13.7)
Disabled; n(%)	<5	<5	<5
Retired; n(%)	17 (3.7)	13 (5.3)	<5
Volunteer; n(%)	<5	<5	<5
Unknown entered; n(%)	7 (1.5)	<5	<5
No. in work or study; n(%)	353 (76.9)	191 (77.3)	162 (76.4)

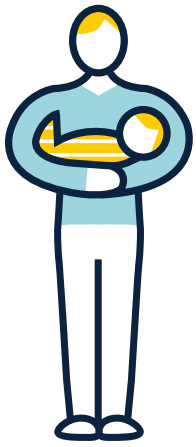
* Redacted to adhere to statistical disclosure guidelines

1.10 Parenthood

	2020	2021	2022	2023
Women with CF who had babies; n	<5	7	8	5
Men with CF who became fathers; n	0	<5	<5	<5



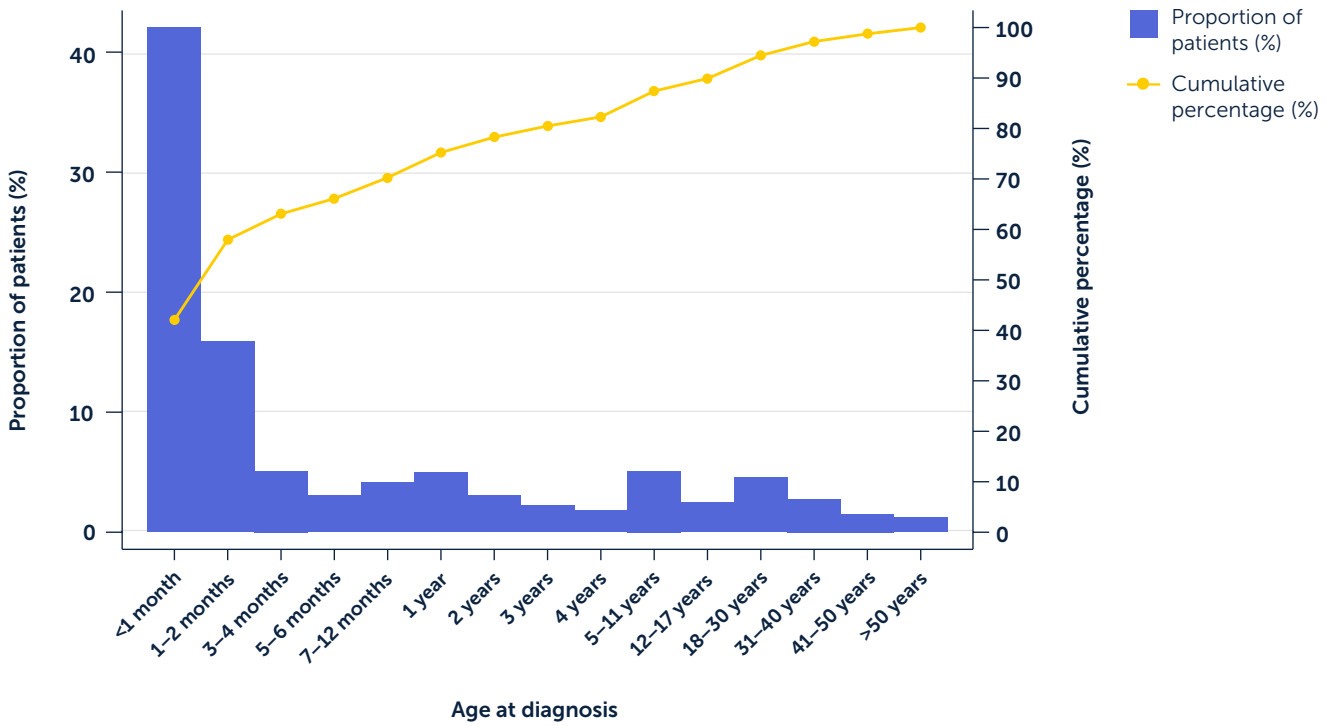
Five women with cystic fibrosis had babies in Scotland during 2023



Fewer than five men with cystic fibrosis became fathers in Scotland during 2023

Diagnosis of cystic fibrosis

1.11 Age at diagnosis
N=723



The median age at diagnosis for patients aged under 16 in 2023 is **21 days**.

Newborn screening for CF has been done routinely in the whole of the UK since mid-2007. It is part of the heel-prick blood-spot testing done at five to seven days of age. The blood sample is tested for a number of conditions, including cystic fibrosis. This means that more babies born after 2007 receive an early diagnosis than those born before.

A total of **nine patients** born in 2023 were identified by newborn screening (including those without complete data).

79 (10.9%) of Scottish CF patients were diagnosed at age 16 or over. Five new CF diagnoses were recorded in Scotland for people aged 16 or over during 2023.

1.12 Mode of presentation

The following tables show the top five most frequent modes of presentation for those diagnosed between 2013–2023 and those born between 2013–2023, excluding those recorded as being diagnosed through newborn screening (NBS) or genotype.

	All patients diagnosed 2013–2023	Age <16 at diagnosis	Age ≥16 at diagnosis
Total patients	**187**	**154**	**33**
Number diagnosed by newborn screening	134	134	0
Total non-NBS or Genotype	53	20	33
Presentation Type(for people not diagnosed by NBS or genotype)**	**Overall (n=53)**	**Age <16 at diagnosis (n=20)**	**Age ≥16 at diagnosis (n=33)**
Persistant or acute respiratory infection	14 (26.4)	<5	11 (33.3)
Meconium ileus	10 (18.9)	10 (50.0)	0 (0.0)
Bronchiectasis	8 (15.1)	0 (0.0)	8 (24.2)
Family history	5 (9.4)	<5	<5
Unknown	<5	0 (0.0)	<5

	All patients born 2013–2023
Total patients	**149**
Number diagnosed by newborn screening or genotype	131
Total non-NBS or Genotype	18
Presentation Type (for people not diagnosed by NBS or genotype)**	**Overall (n=18)**
Meconium ileus	10 (55.6)
Persistent or acute respiratory infection	<5
Failure to thrive/ malnutrition	<5
Family history	<5
Prenatal	<5

* Redacted to adhere to statistical disclosure guidelines
** percentages may not total to 100 because (a) only top five presentation types are reported and (b) multiple presentation types can be selected.

Lung health

For people with CF, thickened mucus in the lungs is linked to repeat or chronic infections. This can cause permanent damage, making it harder to breathe.

In CF, the condition of the lungs is often measured using FEV_1; the Forced Expiratory Volume of air in the first second of a forced exhaled breath. In this report, an FEV_1% predicted is based on the FEV_1 we would expect for a person without CF of the same age, gender, height, and ethnicity.

A person with CF who has FEV_1% predicted of 100% can breathe out the same amount of air in the first second of an exhaled breath as we would expect from a comparable person without cystic fibrosis. A person with FEV_1% predicted of 50% breathes out half the volume of air as a comparable person without cystic fibrosis.

For people with CF, an FEV_1% predicted of 85% or higher is the target, as this indicates normal or near-normal lung health. Each individual with CF will have their own FEV_1 target, based on their own lung function results and trends.

An aim of CF care is to prevent FEV_1% predicted from falling as much as possible, for as long as possible. This is often a team effort between people with CF, their family, and their medical team, which can include doctors, nurses, physiotherapists, dietitians, and psychologists.

The FEV_1% predicted values shown in this report are calculated using an equation called Global Lungs Initiative, or 'GLI'[1].

1 Quanjer PH et al. Eur respir J. 2012 40(6):1324-1343

1.13 FEV_1% predicted (GLI equations) at annual review in patients aged six years and older who have not had a lung transplant
N=628

People with CF who have had lung transplants are excluded, as their new 'non-CF' lungs may have lung health similar to a person without cystic fibrosis.

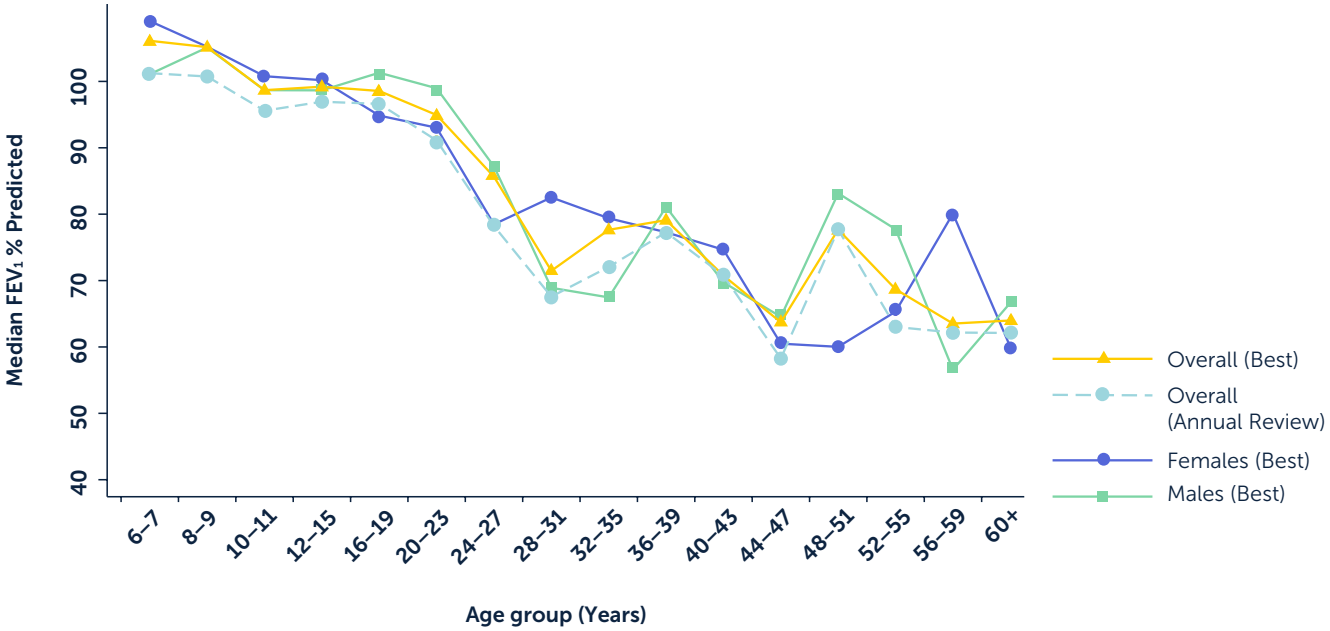
	Overall			Female			Male		
Age (yrs)	n	Median	IQR	n	Median	IQR	n	Median	IQR
6-7	37	101.2	91.2-114.6	15	103.9	94.9-116.8	22	97.3	89.9-111.4
8-9	28	100.7	95.9-109.4	17	99.9	96.3-106.8	11	102.8	93.6-113.5
10-11	32	95.6	84.8-101.2	14	94.7	91.0-100.8	18	96.7	83.8-101.6
12-15	83	96.9	88.1-106.3	38	97.5	89.3-109.7	45	95.7	88.1-104.2
16-19	64	96.6	86.6-104.3	24	90.6	84.6-98.9	40	99.0	89.3-106.5
20-23	46	91.1	63.6-103.5	26	89.6	63.6-103.6	20	92.8	58.7-101.7
24-27	58	78.2	61.5-96.0	29	74.0	56.8-95.9	29	80.9	70.8-96.0
28-31	62	67.6	43.3-88.3	32	67.1	42.1-89.6	30	67.8	48.7-81.9
32-35	50	72.1	56.1-85.6	23	75.3	65.3-83.7	27	65.6	46.0-91.9
36-39	35	77.3	48.1-89.6	15	72.1	48.1-85.9	20	79.1	52.4-91.5
40-43	24	70.8	55.0-83.5	10	73.6	54.8-80.3	14	67.3	55.2-87.1
44-47	26	58.3	51.3-84.2	9	57.8	51.3-93.7	17	58.3	52.7-81.9
48-51	17	77.7	41.3-87.0	8	57.5	39.2-80.1	9	83.2	71.3-94.2
52-55	15	63.0	57.8-91.0	6	61.8	57.8-91.0	9	68.7	61.4-83.4
56-59	14	62.2	41.2-80.1	6	77.9	63.5-80.1	8	53.8	38.1-71.4
60-63	11	55.6	45.6-67.8	-*	55.6	45.6-67.8	<5	-	-
64-67	186	97.6	89.4-107.0	85	98.8	90.8-109.7	101	96.6	86.9-106.3
68+	428	78.3	56.3-94.4	196	77.0	56.6-91.3	232	79.3	56.1-96.4
<16	206	97.6	89.4-107.0	94	97.8	90.0-107.9	112	97.5	89.3-107.0
≥16	396	77.6	55.1-93.0	185	76.0	55.6-91.4	211	78.3	53.1-95.3
Overall	**602****	**86.9**	**66.6-100.3**	**279**	**85.9**	**66.1-99.9**	**323**	**87.2**	**67.0-100.7**

* Redacted to adhere to statistical disclosure guidelines.

** Number with non-missing data.

1.14 Best FEV_1% predicted (GLI equations) in patients aged six years and older who have not had a lung transplant
N=628

For the best FEV_1 calculation, where best FEV_1% was missing or less than the FEV_1% at annual review, the annual review FEV_1% was used.

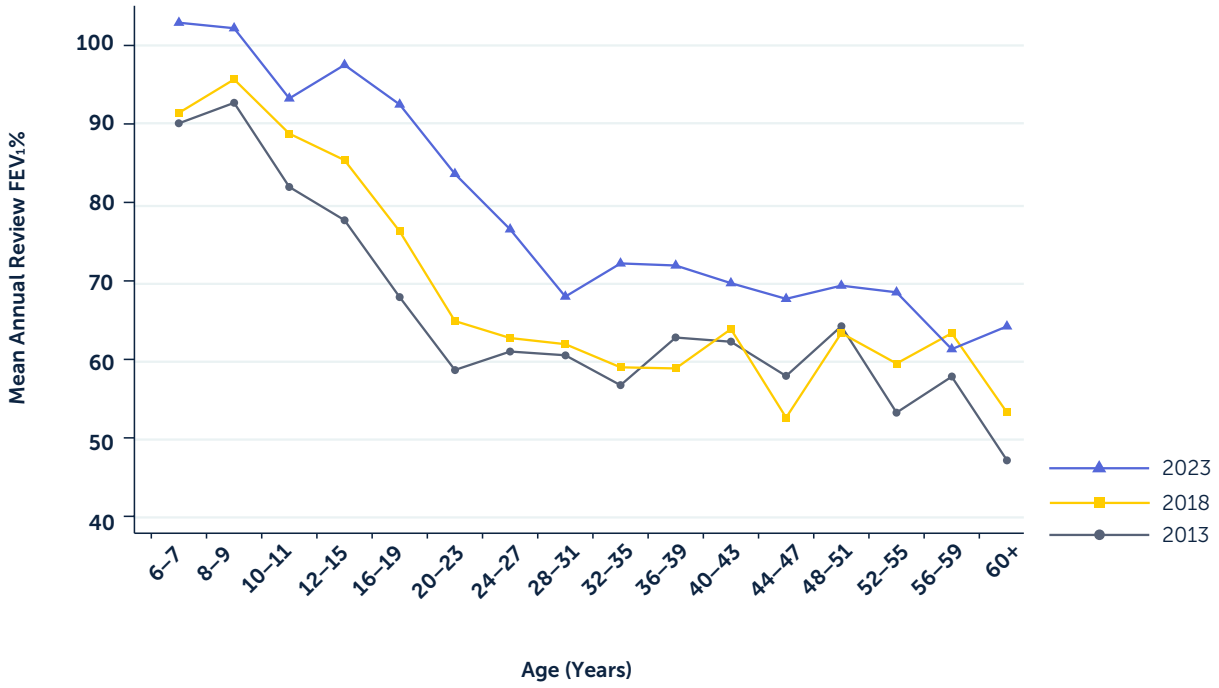


	Overall			Female			Male		
Age (yrs)	n	Median	IQR	n	Median	IQR	n	Median	IQR
6-7	40	106.1	96.0-114.4	17	109.1	104.2-114.6	23	101.1	93.8-114.3
8-9	28	105.2	98.6-112.1	17	105.2	98.7-109.5	11	105.1	96.6-113.5
10-11	33	98.7	93.0-104.6	14	100.8	91.0-105.1	19	98.7	93.5-103.6
12-15	86	99.2	90.4-108.8	39	100.2	91.0-110.6	47	98.7	89.4-107.2
16-19	68	98.6	89.3-106.9	27	94.8	87.1-102.5	41	101.3	93.0-110.3
20-23	46	94.9	75.2-104.7	26	93.0	84.9-103.6	20	99.0	67.8-107.2
24-27	59	85.6	68.9-96.4	29	78.5	61.2-95.9	30	87.2	77.6-96.4
28-31	62	71.5	52.3-94.0	32	82.5	52.7-98.2	30	68.9	52.3-86.0
32-35	51	77.7	56.1-94.8	24	79.4	68.6-87.3	27	67.5	46.6-95.6
36-39	35	79.1	59.9-94.7	15	77.3	59.9-88.3	20	81.1	56.7-97.4
40-43	24	70.8	55.0-88.4	10	74.6	54.8-82.7	14	69.7	55.2-89.3
44-47	26	63.7	52.7-91.8	9	60.5	55.9-93.7	17	64.5	52.7-86.9
48-51	17	77.7	46.8-87.6	8	60.0	46.8-81.8	9	83.2	77.7-94.2
52-55	15	68.7	62.3-91.0	6	65.3	62.3-91.0	9	77.7	63.0-89.7
56-59	15	63.5	43.3-83.9	7	80.1	63.5-83.9	8	56.6	39.2-73.9
60+	18	64.0	53.6-77.4	8	60.0	53.0-74.7	10	66.7	55.2-86.3
<16	187	101.1	93.0-110.6	87	104.1	93.5-111.2	100	100.4	92.6-109.9
≥16	436	82.9	61.5-98.2	201	82.7	63.6-95.9	235	83.1	59.2-99.5
<18	216	100.9	92.6-109.9	100	102.1	92.1-110.0	116	100.6	92.8-109.9
≥18	206	88.8	67.5-100.8	101	89.4	64.4-100.7	105	87.4	68.1-101.3
Overall	**623****	**91.0**	**70.4-103.1**	**288**	**90.8**	**71.2-103.4**	**335**	**92.0**	**68.7-102.8**

* Redacted to adhere to statistical disclosure guidelines.
** Number with non-missing data.

1.15 FEV_1% predicted (GLI equations) over time in patients aged six years and older who have not had a lung transplant N=628 in 2023, N=698 in 2018, N=604 in 2013**

The chart below shows how FEV_1% in 2023 compares to Registry data from 2013 and 2018. 2013 is shown as a comparator year.



Age (yrs)	n	2013 mean FEV_1%: Mean (SD)	n	2018 mean FEV_1%: Mean (SD)	n	2023 mean FEV_1%: Mean (SD)	p-values (t-test)***
6-7	45	90.1 (12.5)	45	91.4 (13.7)	37	102.9 (16.6)	<0.001
8-9	47	92.7 (10.6)	39	95.7 (15.9)	28	102.2 (10.0)	0.061
10-11	29	82.0 (12.8)	31	88.8 (15.8)	32	93.3 (12.4)	0.214
12-15	59	77.7 (21.5)	90	85.4 (16.1)	83	97.5 (13.9)	<0.001
16-19	97	67.9 (22.8)	51	76.3 (22.4)	64	92.5 (17.1)	<0.001
20-23	91	58.6 (23.7)	90	64.9 (26.4)	46	83.6 (25.3)	<0.001
24-27	71	61.0 (25.0)	77	62.7 (23.8)	58	76.6 (23.9)	0.001
28-31	59	60.5 (24.1)	62	61.9 (22.8)	62	68.0 (24.8)	0.160
32-35	36	56.7 (22.7)	58	59.0 (20.5)	50	72.2 (25.0)	0.003
36-39	23	62.8 (24.0)	34	58.8 (24.2)	35	72.0 (22.6)	0.023
40-43	15	62.2 (25.9)	27	63.9 (26.8)	24	69.7 (18.7)	0.375
44-47	19	57.9 (21.6)	14	52.6 (25.3)	26	67.7 (26.9)	0.091
48-51	13	64.2 (19.8)	25	63.4 (20.2)	17	69.4 (26.0)	0.402
52-55	6	53.2 (16.7)	20	59.5 (20.8)	15	68.5 (23.2)	0.232
56-59	6	57.8 (26.8)	7	63.3 (21.1)	14	61.3 (20.4)	****
60+	5	47.2 (23.9)	12	53.2 (26.0)	17	64.2 (20.4)	0.214
<16	180	85.4 (16.8)	205	89.2 (15.9)	180	98.6 (14.0)	N/A
≥16	441	61.4 (23.6)	477	63.2 (24.0)	428	75.1 (24.5)	N/A
<18	227	82.4 (19.1)	225	88.3 (16.5)	206	97.9 (14.3)	N/A
≥18	394	60.3 (23.6)	457	62.5 (23.9)	402	73.9 (24.5)	N/A

* Redacted to adhere to statistical disclosure guidelines.

** Due to missing data, means are calculated from a population of 608 in 2023, 682 in 2018 and 621 in 2013.

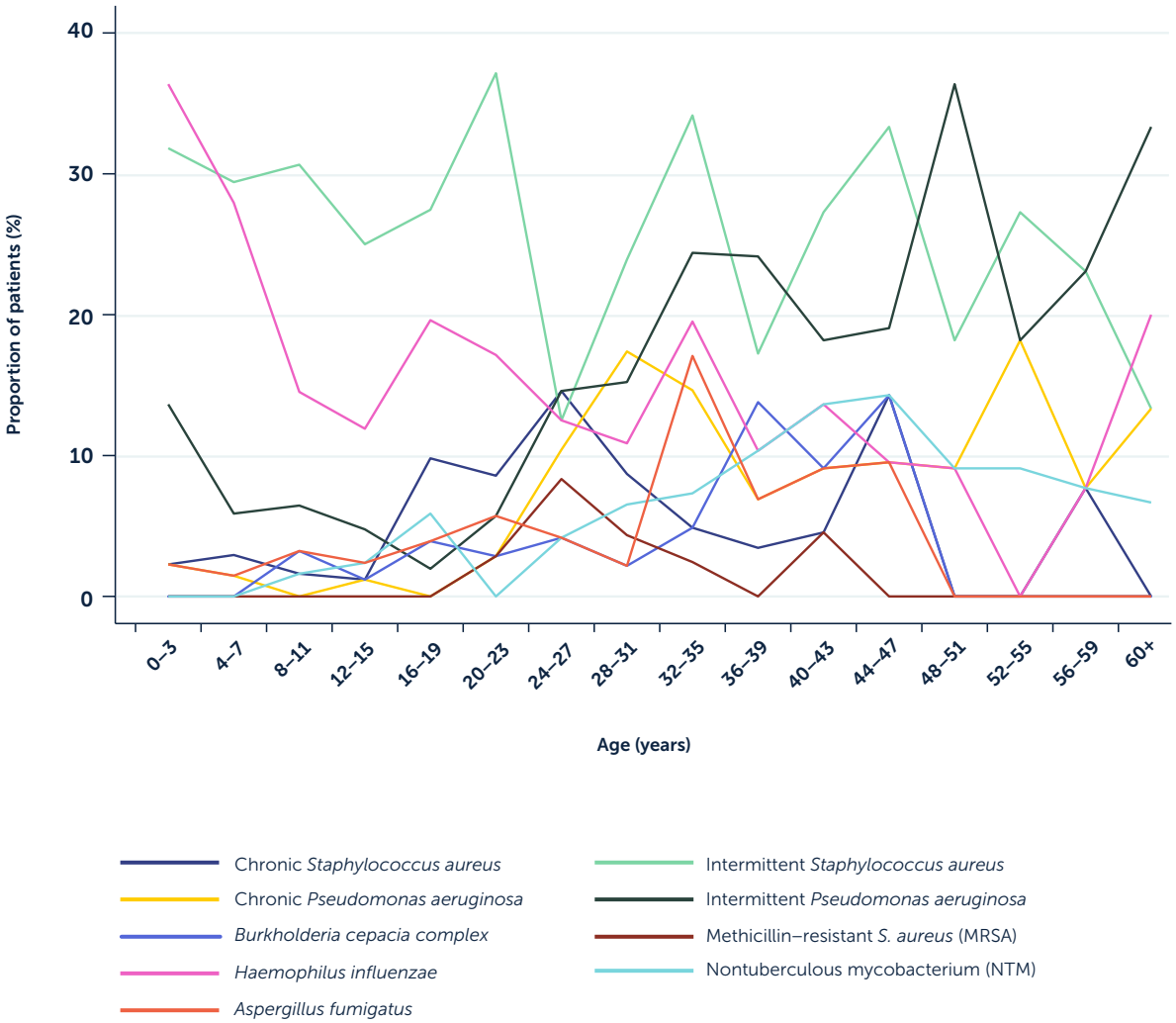
*** t-test comparing 2023 with 2018. If the p-value is less than 0.05 then the difference in the mean is statistically significant.

**** t-test not performed due to small numbers in these age groups.

Lung infections

Lung infections can permanently reduce lung function in people with cystic fibrosis. Some lung infections can become 'chronic', meaning that they can't ever be removed completely using medicines. All other infections are reported if they have occurred at least once as a positive growth in the 12 months prior to the patient's annual review data set.

1.16 Lung infections in 2023
N=601*



* Proportions are calculated from the number of patients with at least one sample taken in the relevant age group. Some proportions should be treated with caution due to the very small numbers of people with the infections in some age groups.

1.17 Lung infections in 2023
<16 years N=258; ≥16 years N=343

Infections in this table reflect bugs grown in the 12 months prior to the 2023 annual review. The UK CF Registry definition of 'chronic' is three or more isolates in the last 12 months.

	Paediatric Age Range (Years)				Overall
	0-3	4-7	8-11	12-15	Paediatric (<16 years)
Number in age range	44	69	64	87	264
Number who had culture taken*	44	68	62	84	258
Chronic *S. aureus* n(%)	<5	<5	<5	<5	5 (5.0)
Intermittent *S. aureus* n(%)	14 (14.0)	20 (20.0)	19 (19.0)	21 (21.0)	74 (74.0)
Chronic *P. aeruginosa* n(%)	<5	<5	0	<5	<5
Intermittent *P. aeruginosa* n(%)	6 (6.0)	<5	<5	<5	18 (18.0)
B. cepacia complex n(%)	0	0	<5	<5	<5
B. cenocepacia n(%)	0	0	0	0	0
B. multivorans n(%)	0	0	0	0	0
B. cepacia (other) n(%)	0	0	0	0	0
MRSA n(%)	0	0	0	0	0
H. influenza n(%)	16 (16.0)	19 (19.0)	9 (9.0)	10 (10.0)	54 (54.0)
NTM n(%)	0	0	<5	<5	<5
Aspergillus fumigatus n(%)	<5	<5	<5	<5	6 (6.0)

* Proportions are calculated from the number of people who were recorded as having at least one respiratory culture sample taken.

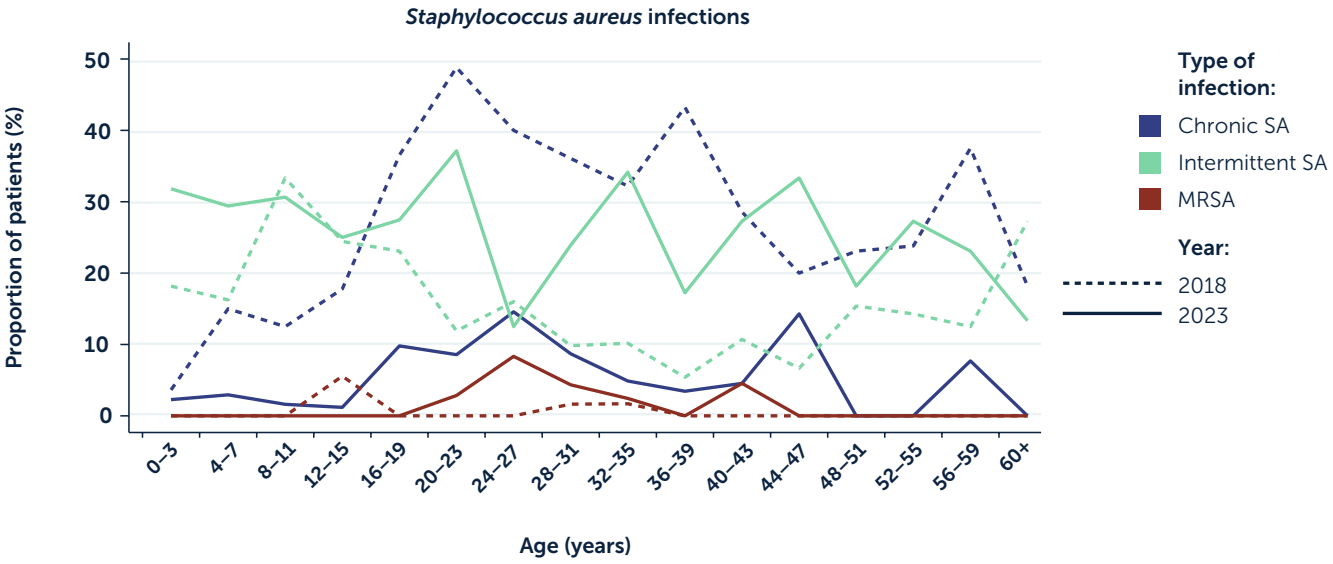
Lung infections in 2023 (contd.)
<16 years N=258; ≥16 years N=343

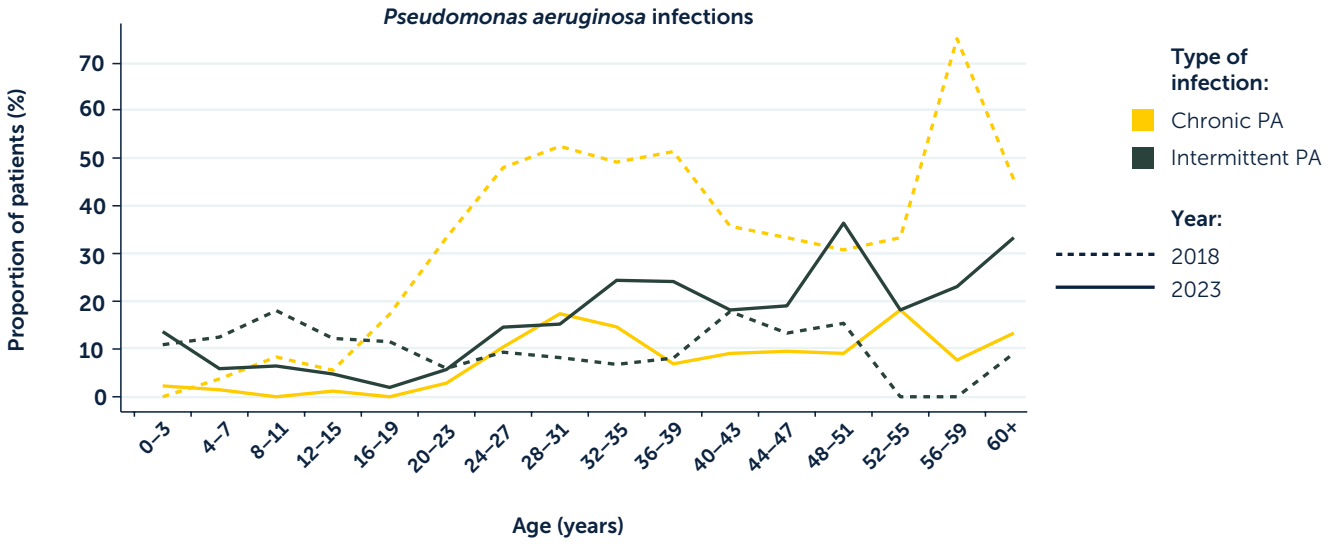
	Adult Age Range (Years)						Overall
	16-19	20-23	24-27	28-31	32-35	36-39	Adults (≥16 years)
Number in age range	68	46	60	63	54	38	459
Number who had culture taken*	51	35	48	46	41	29	343
Chronic *S. aureus* n(%)	5 (5.0)	<5	7 (7.0)	<5	<5	<5	27 (27.0)
Intermittent *S. aureus* n(%)	14 (14.0)	13 (13.0)	6 (6.0)	11 (11.0)	14 (14.0)	5 (5.0)	86 (86.0)
Chronic *P. aeruginosa* n(%)	0	<5	5 (5.0)	8 (8.0)	6 (6.0)	<5	32 (32.0)
Intermittent *P. aeruginosa* n(%)	<5	<5	7 (7.0)	7 (7.0)	10 (10.0)	7 (7.0)	56 (56.0)
B. cepacia complex n(%)	<5	<5	<5	<5	<5	<5	17 (17.0)
B. cenocepacia n(%)	0	0	0	0	<5	<5	5 (5.0)
B. multivorans n(%)	<5	<5	<5	0	<5	<5	10 (10.0)
B. cepacia (other) n(%)	0	0	0	<5	0	0	<5
MRSA n(%)	0	<5	<5	<5	<5	0	9 (9.0)
H. influenza n(%)	10 (10.0)	6 (6.0)	6 (6.0)	5 (5.0)	8 (8.0)	<5	48 (48.0)
NTM n(%)	<5	0	<5	<5	<5	<5	24 (24.0)
Aspergillus n(%)	<5	<5	<5	<5	7 (7.0)	<5	20 (20.0)

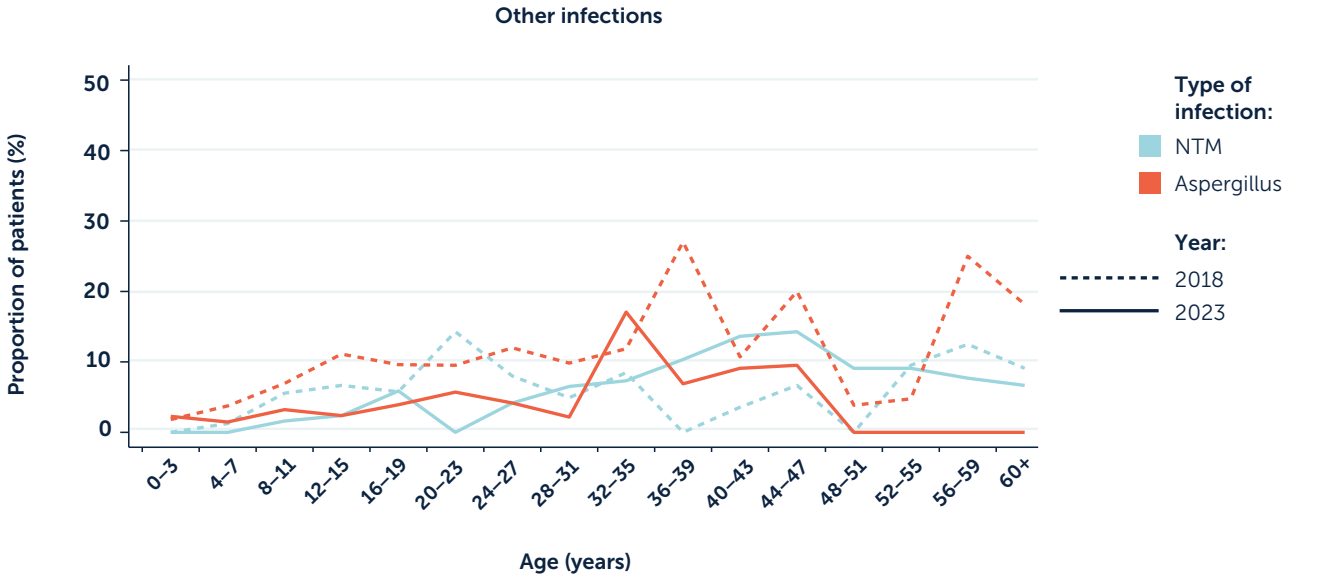
	Adult Age Range (Years)						Overall
	40-43	44-47	48-51	52-55	56-59	60+	Adults (≥16 years)
Number in age range	30	28	20	15	18	19	459
Number who had culture taken*	22	21	11	11	13	15	343
Chronic *S. aureus* n(%)	<5	<5	0	0	<5	0	27 (27.0)
Intermittent *S. aureus* n(%)	6 (6.0)	7 (7.0)	<5	<5	<5	<5	86 (86.0)
Chronic *P. aeruginosa* n(%)	<5	<5	<5	<5	<5	<5	32 (32.0)
Intermittent *P. aeruginosa* n(%)	<5	<5	<5	<5	<5	5 (5.0)	56 (56.0)
B. cepacia complex n(%)	<5	<5	0	0	0	0	17 (17.0)
B. cenocepacia n(%)	<5	0	0	0	0	0	5 (5.0)
B. multivorans n(%)	0	<5	0	0	0	0	10 (10.0)
B. cepacia (other) n(%)	0	0	0	0	0	0	<5
MRSA n(%)	<5	0	0	0	0	0	9 (9.0)
H. influenza n(%)	<5	<5	<5	0	<5	<5	48 (48.0)
NTM n(%)	<5	<5	<5	<5	<5	<5	24 (24.0)
Aspergillus n(%)	<5	<5	0	0	0	0	20 (20.0)

* Proportions are calculated from the number of people who were recorded as having at least one respiratory culture sample taken.

1.18 Lung infections in 2018 and 2023







1.19 Respiratory culture sample type

Overall	2018	2023
Number of people with an annual review (n)	819	723
Number of people with at least 3 samples of any type taken n(%)*	648 (79.1)	411 (56.8)
Number of people with at least 1 sample of any type taken n(%)*	774 (94.5)	604 (83.5)
Sample type**		
Sputum; n(%)	571 (73.8)	347 (57.5)
Cough; n(%)	332 (42.9)	379 (62.7)
Bronchoalveolar lavage; n(%)	17 (2.2)	19 (3.1)
Age <16 years	**2018**	**2023**
Number of people with an annual review (n)	304	264
Number of people with at least 3 samples of any type taken n(%)*	280 (92.1)	249 (94.3)
Number of people with at least 1 sample of any type taken n(%)*	297 (97.7)	261 (98.9)
Sample type**		
Sputum; n(%)	105 (35.4)	57 (21.8)
Cough; n(%)	284 (95.6)	259 (99.2)
Bronchoalveolar lavage; n(%)	16 (5.4)	14 (5.4)
Age>=16 years	**2018**	**2023**
Number of people with an annual review (n)	515	459
Number of people with at least 3 samples of any type taken n(%)*	368 (71.5)	162 (35.3)
Number of people with at least 1 sample of any type taken n(%)*	477 (92.6)	343 (74.7)
Sample type**		
Sputum; n(%)	466 (97.7)	290 (84.5)
Cough; n(%)	48 (10.1)	120 (35.0)
Bronchoalveolar lavage; n(%)	<5	<5

1.20 Non-tuberculous mycobacteria (NTM) or atypical mycobacteria

NTM is slow to grow and takes time to treat. It may be present for several years before eradication, or may never be cleared. In the table below, 'prevalence' represents all people reported in that year as having a positive culture. 'Incidence' represents all positive cultures in individuals that have not reported having any in the previous two years of data.

	2021 (n=777)	2022 (n=727)	2023 (n=723)
NTM prevalence; n(%)	28 (3.6)	24 (3.3)	27 (3.7)
On NTM treatment in the given year; n (% of NTM prevalence in given year)	11 (39.3)	5 (20.8)	10 (37.0)
NTM incidence	13 (1.8)	17 (2.5)	18 (2.6)
M. abscessus prevalence	14 (1.8)	<5	5 (0.7)
M. abscessus incidence	<5	<5	<5

* % is of those people with an annual review.

** Patients can have more than one sample taken so the % total may not add up to 100%.

Complications

1.21 Complications in 2023

The number shown is for a complication that has been present in the preceding 12 months.

Complications	Overall	<16 years	≥16 years
Respiratory related			
Nasal polyps requiring surgery	15 (1.9)	<5	-*
Sinus disease	82 (10.2)	0	82 (17.9)
Asthma	45 (5.6)	<5	-*
ABPA	18 (2.2)	<5	-*
Haemoptysis (massive, severe and/or moderate)	8 (1.0)	0	8 (1.7)
Massive haemoptysis	<5	0	<5
Pneumothorax requiring chest tube	0	0	0
Cardiac complications			
Tachyarrhythmia	0	0	0
Bradycardia	0	0	0
Cardiac arrest	<5	0	<5
Cardiomyopathy	<5	0	<5
Congenital heart disease	0	0	0
Heart failure	0	0	0
Ischaemic heart disease	0	0	0
Valvular disease	0	0	0
Other	<5	0	<5
Pancreas and hepatobiliary disease			
Raised liver enzymes	44 (5.5)	6 (2.3)	38 (8.3)
Liver disease	144 (17.9)	25 (9.5)	119 (25.9)
Cirrhosis with no portal hypertension	8 (1.0)	<5	-*
Cirrhosis with portal hypertension	20 (2.5)	<5	-*
Gall bladder disease requiring surgery	11 (1.4)	<5	-*
Pancreatitis	10 (1.2)	0	10 (2.2)
Upper gastrointestinal (GI)			
Gastro-oesphageal reflux disease (GORD)	207 (25.8)	7 (2.7)	200 (43.6)
Peptic ulcer	0	0	0
GI bleed (varices as source)	<5	0	<5
GI bleed (non varices as source)	<5	0	<5
Lower gastrointestinal			
Intestinal obstruction	<5	<5	0
DIOS	53 (6.6)	<5	-*
Fibrosing colonopathy / colonic stricture	0	0	0
Rectal prolapse	0	0	0
Renal			
Kidney stones	<5	<5	<5
Renal failure	6 (0.7)	0	6 (1.3)
Musculoskeletal			
Arthritis	8 (1.0)	<5	-*
Arthropathy	24 (3.0)	<5	-*
Bone fracture	<5	0	<5
Osteopenia	92 (11.5)	<5	-*
Osteoporosis	41 (5.1)	0	41 (8.9)
Other			
Cancer confirmed by histology	<5	0	<5
Port inserted or replaced	9 (1.1)	<5	-*
Depression	9 (1.1)	0	9 (2.0)
Hearing loss	9 (1.1)	<5	-*
Hypertension	17 (2.1)	0	17 (3.7)
Urinary incontinence	15 (1.9)	<5	-*
Faecal incontinence	6 (0.7)	<5	-*
Postural anomaly	13 (1.6)	<5	-*

* Redacted to adhere to statistical disclosure guidelines.

1.22 Incidence of complications

The table below describes new cases of a complication that have not been reported for an individual in at least the previous two years.

	2023		
	Overall (n=723)	<16 years (n=264)	≥16 years (n=459)
ABPA; n (%)	5 (0.7)	0	5 (1.1)
Cirrhosis - no portal hypertension; n (%)	<5	0	<5
Cirrhosis - with portal hypertension; n (%)	6 (0.8)	<5	<5
Cancer confirmed by histology; n (%)	<5	0	<5

1.23 CF diabetes**
N=581

Cystic fibrosis diabetes (CFD) is common in adults and adolescents with cystic fibrosis. This is because, for many people with CF, the pancreas does not work properly. This can mean that not enough insulin is produced, or it may not work properly, causing CFD. CFD is different from type 1 and type 2 diabetes, but has features of both.

	All ≥10 years (n=581)	10-15 years (n=122)	≥16 years (n=459)
On CFD treatment; n(%)			
Of those on treatment	95 (16.4)	12 (9.8)	83 (18.1)
Insulin[1]; n(%)	90 (94.7)	11 (91.7)	79 (95.2)
CFD Screening; n(%)			
Yes	283 (48.7)	75 (61.5)	208 (45.3)
Screening type; n(%)			
Continous glucose monitoring[2]; n(%)	81 (28.6)	22 (29.3)	59 (28.4)
Oral glucose tolerance test[2]; n(%)	238 (84.1)	51 (68.0)	187 (89.9)
Not screened (other)	125 (21.5)	<5	-*
Not screened (known CFRD)	165 (28.4)	39 (32.0)	126 (27.5)
Unknown	8 (1.4)	-*	<5

1 Proportion of patients on treatment.
2 Proportion of patients screened.
* Redacted to adhere to statistical disclosure guidelines.
** Alternatively known as CF related diabetes.

Antibiotics

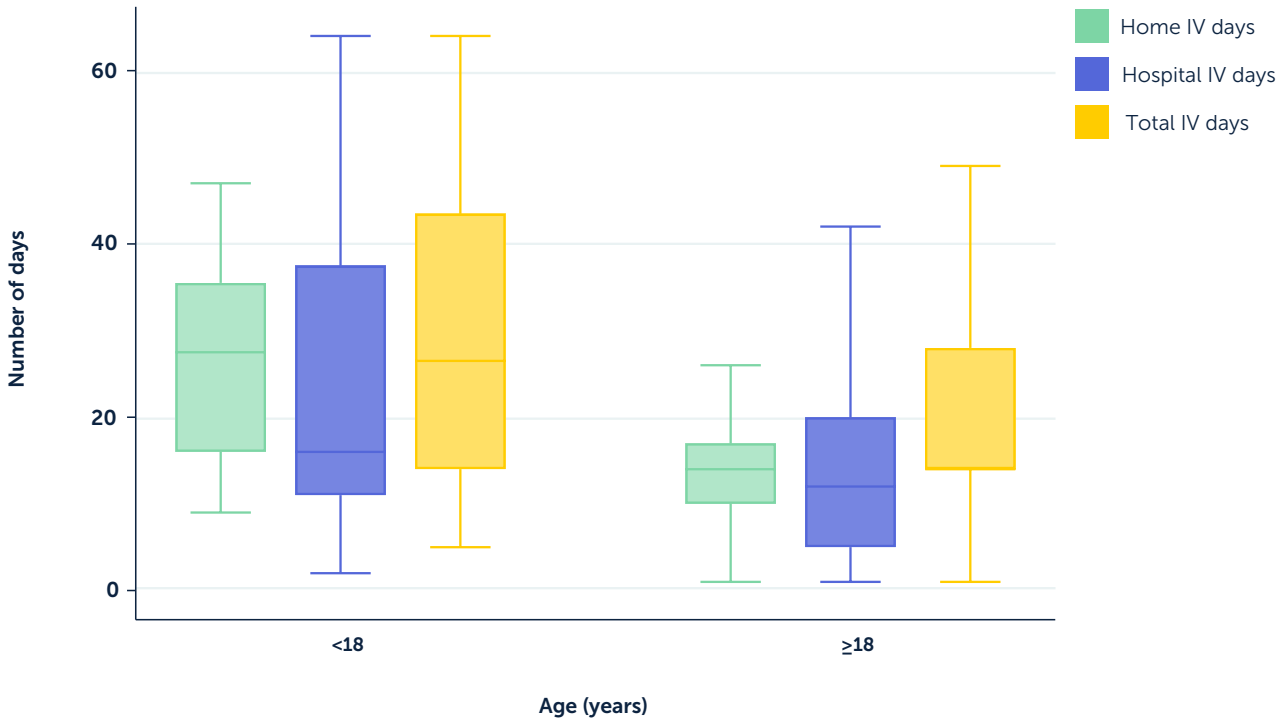
1.24 Intravenous (IV) antibiotics
N=723

When someone with CF becomes unwell with an infection, they might be prescribed IV antibiotics. IV antibiotics are given to the patient through their veins. This treatment can take a number of days and might take place as a hospital inpatient, or at home.

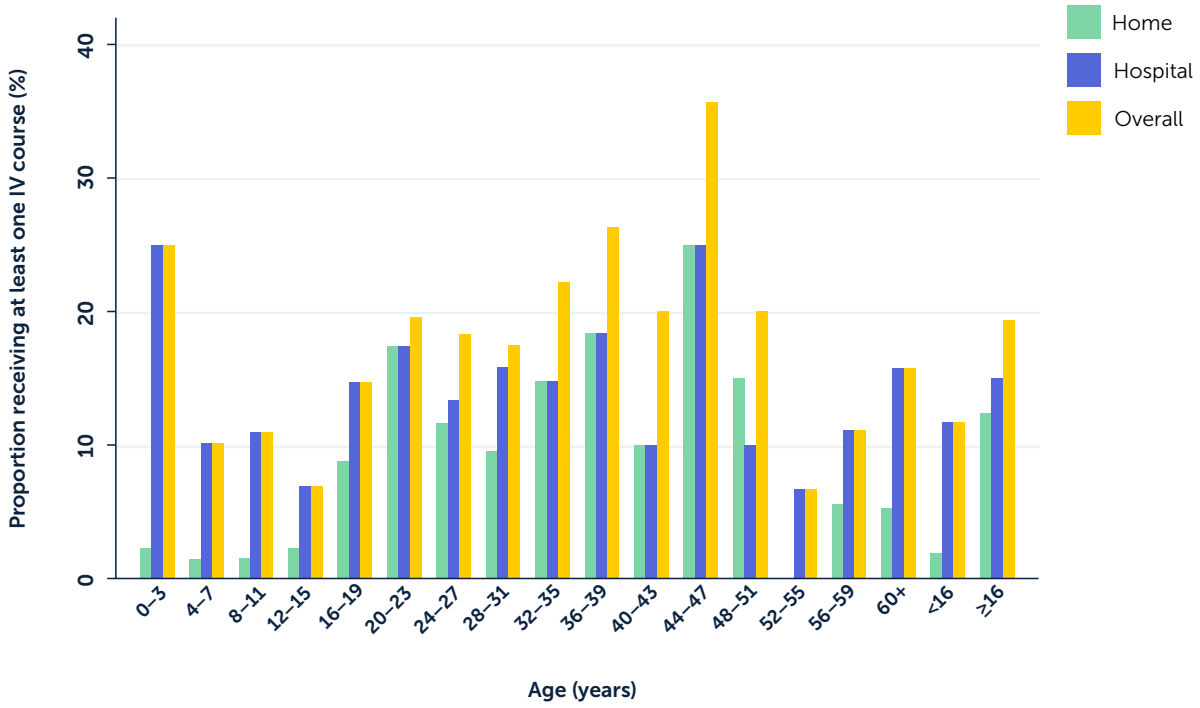
		Home		Hospital		Total	
Age	n	Patients n(%)	Median days (IQR)*	Patients n(%)	Median days (IQR)*	Patients n (%)	Median days (IQR)*
0-3	44	<5	*	11 (25.0)	15 (14-38)	11 (25.0)	15 (14-39)
4-7	69	<5	*	7 (10.1)	17 (7-64)	7 (10.1)	26 (14-64)
8-11	64	<5	*	7 (10.9)	16 (13-37)	7 (10.9)	21 (13-56)
12-15	87	<5	*	6 (6.9)	20 (9-32)	6 (6.9)	30 (27-41)
16-19	68	6 (8.8)	12 (10-36)	10 (14.7)	10 (3-21)	10 (14.7)	18 (14-52)
20-23	46	8 (17.4)	11 (6-19)	8 (17.4)	10 (7-16)	9 (19.6)	21 (14-28)
24-27	60	7 (11.7)	14 (7-19)	8 (13.3)	11 (6-24)	11 (18.3)	14 (14-41)
28-31	63	6 (9.5)	18 (13-23)	10 (15.9)	6 (4-35)	11 (17.5)	28 (14-40)
32-35	54	8 (14.8)	14 (12-14)	8 (14.8)	14 (8-15)	12 (22.2)	14 (14-22)
36-39	38	7 (18.4)	13 (10-14)	7 (18.4)	6 (1-28)	10 (26.3)	14 (14-19)
40-43	30	<5	*	<5	*	6 (20.0)	14 (14-14)
44-47	28	7 (25.0)	14 (4-42)	7 (25.0)	13 (3-42)	10 (35.7)	14 (13-56)
48-51	20	<5	*	<5	*	<5	*
52-55	15	0 (0.0)	n/a	<5	*	<5	*
56-59	18	<5	*	<5	*	<5	*
60+	19	<5	*	<5	*	<5	*
<16	264	5 (1.9)	29 (26-35)	31 (11.7)	16 (12-37)	31 (11.7)	26 (14-42)
≥16	459	57 (12.4)	14 (10-17)	69 (15.0)	12 (5-21)	89 (19.4)	14 (14-28)
<18	293	8 (2.7)	28 (16-36)	36 (12.3)	16 (11-38)	36 (12.3)	26 (14-44)
≥18	430	54 (12.6)	14 (10-17)	64 (14.9)	12 (5-20)	84 (19.5)	14 (14-28)
Overall	**723**	**62 (8.6)**	**14 (10-22)**	**100 (13.8)**	**14 (6-28)**	**120 (16.6)**	**14 (14-38)**

* Summary statistics not provided for very small samples.

This box plot graph illustrates the spread of the number of days on IV antibiotics in the Scottish CF population, stratified by age. A guide on how to correctly interpret this box plot graph can be found on page 51.



The bar graph below summarises the proportion of people receiving at least one course of IV antibiotics across different age groups within the Scottish CF population. Overall, the proportion of patients receiving at least one IV course at home was 8.6% and in hospital was 13.8%. The proportion receiving any IVs was 16.6%.



1.25 Inhaled antibiotic use
N=723

	2023		
	Overall	<16 years	≥16 years
Number of patients	723	264	459
Tobramycin solution; n(%)	35 (4.8)	13 (4.9)	22 (4.8)
Other aminoglycoside; n(%)	<5	0	<5
Colistin; n(%)	47 (6.5)	21 (8.0)	26 (5.7)
Promixin; n(%)	25 (3.5)	5 (1.9)	20 (4.4)
Aztreonam; n(%)	12 (1.7)	0	12 (2.6)
Colistimethate (DPI); n(%)	30 (4.1)	<5	-*
Tobramycin Inhalation Powder; n(%)	16 (2.2)	<5	-*
Levofloxacin; n(%)	<5	0	<5
At least one of the above; n(%)	**144 (19.9)**	**34 (12.9)**	**110 (24.0)**

1.26 Inhaled antibiotic use among people with chronic *Pseudomonas aeruginosa*

The consensus view in the UK is that 90% of people chronically infected with *P. aeruginosa* should be prescribed at least one of the above inhaled antibiotics.

	2023		
	Overall	<16 years	≥16 years
Patients with chronic P. aeruginosa	**35**	**3**	**32**
Tobramycin solution; n(%)	<5	<5	<5
Other aminoglycoside; n(%)	0	0	0
Colistin; n(%)	10 (28.6)	<5	-*
Promixin; n(%)	<5	0	<5
Aztreonam; n(%)	<5	0	<5
Colistimethate (DPI); n(%)	<5	0	<5
Tobramycin Inhalation Powder; n(%)	<5	0	<5
Levofloxacin; n(%)	<5	0	<5
At least one of the above; n(%)	**23 (65.7)**	**<5**	**20 (62.5)**

* Redacted to adhere to statistical disclosure guidelines.

1.27 Long-term azithromycin use

Azithromycin is an antibiotic with some anti-inflammatory properties. It is recommended for long-term use as a prophylactic antibiotic in people with chronic *Pseudomonas aeruginosa* infection.

		Number of patients on azithromycin; n	Patients with chronic *P. aeruginosa*; n(%)	Patients without chronic *P. aeruginosa*; n(%)
2013	**0-3 years**	<5	<5	<5
	4-15 years	59	8 (13.6)	51 (86.4)
	≥ 16 years	326	176 (54.0)	150 (46.0)
2018	**0-3 years**	<5	0	<5
	4-15 years	69	10 (14.5)	59 (85.5)
	≥ 16 years	343	161 (46.9)	182 (53.1)
2023	**0-3 years**	-*	<5	<5
	4-15 years	-*	<5	55 (98.2)
	≥ 16 years	224	29 (12.9)	195 (87.1)

1.28 Flucloxacillin use

Flucloxacillin is an antibiotic that is used prophylactically to prevent infection with bacteria.

	2018		2023	
Age	**Total patients**	**Patients on prophylactic flucloxacillin; n(%)**	**Total patients**	**Patients on prophylactic flucloxacillin; n(%)**
0-3	55	35 (63.6)	44	23 (52.3)
4-7	82	27 (32.9)	69	21 (30.4)
8-11	75	21 (28.0)	64	18 (28.1)
12-15	92	31 (33.7)	87	12 (13.8)
16-19	53	11 (20.8)	68	<5
20-23	91	22 (24.2)	46	<5
24-27	82	16 (19.5)	60	11 (18.3)
28-31	65	8 (12.3)	63	6 (9.5)
32-35	64	5 (7.8)	54	<5
36-39	41	<5	38	<5
40-43	31	0	30	0
44-47	18	0	28	0
48-51	26	<5	20	0
52-55	23	<5	15	<5
56-59	8	0	18	<5
60+	13	0	19	0
<16 years	304	114 (37.5)	264	74 (28.0)
≥16 years	515	69 (13.4)	459	31 (6.8)
<18 years	325	116 (35.7)	293	76 (25.9)
≥18 years	494	67 (13.6)	430	29 (6.7)
Overall	**819**	**183 (22.3)**	723	105 (14.5)

* Redacted to adhere to statistical disclosure guidelines.

1.29 Inhaled bronchodilators and corticosteroids

Age	Total patients	Patients on inhaled bronchodilators; n(%)	Patients on inhaled corticosteroids; n(%)	Patients on inhaled combination corticosteroids/ bronchodilators; n(%)
<6 years	73	6 (8.2)	<5	<5
6 - ≤16 years	204	42 (20.6)	23 (11.3)	11 (5.4)
6 - ≤18 years	242	58 (24.0)	29 (12.0)	13 (5.4)
<16 years	264	42 (15.9)	27 (10.2)	11 (4.2)
≥16 years	459	290 (63.2)	98 (21.4)	85 (18.5)
<18 years	293	54 (18.4)	29 (9.9)	14 (4.8)
≥18 years	430	278 (64.7)	96 (22.3)	82 (19.1)
Overall	**723**	**332 (45.9)**	**125 (17.3)**	**96 (13.3)**

Mucoactive therapies

1.30 Mannitol

	2018		2023	
Age	Total patients	Patients on Mannitol; n(%)	Total patients	Patients on Mannitol; n(%)
0-3	55	0	44	0
4-7	82	0	69	0
8-11	75	0	64	0
12-15	92	0	87	0
16-19	53	0	68	0
20-23	91	0	46	0
24-27	82	0	60	0
28-31	65	<5	63	0
32-35	64	<5	54	0
36-39	41	<5	38	<5
40-43	31	<5	30	0
44-47	18	0	28	0
48-51	26	<5	20	<5
52-55	23	0	15	0
56-59	8	0	18	0
60+	13	0	19	0
<16 years	304	0	264	0
≥16 years	515	9 (1.7)	459	<5
<18 years	325	0	293	0
≥18 years	494	9 (1.8)	430	<5
Overall	**819**	**9 (1.1)**	**723**	**<5**

1.31 DNase

	2013		2018		2023	
Age	Total patients	Patients on DNase; n(%)	Total patients	Patients on DNase; n(%)	Total patients	Patients on DNase; n(%)
0-3	70	<5	55	<5	44	6 (13.6)
4-7	101	11 (10.9)	82	22 (26.8)	69	22 (31.9)
8-11	82	22 (26.8)	75	32 (42.7)	64	29 (45.3)
12-15	61	27 (44.3)	92	51 (55.4)	87	40 (46.0)
16-19	99	41 (41.4)	53	24 (45.3)	68	39 (57.4)
20-23	97	42 (43.3)	91	57 (62.6)	46	18 (39.1)
24-27	75	29 (38.7)	82	39 (47.6)	60	28 (46.7)
28-31	67	25 (37.3)	65	42 (64.6)	63	31 (49.2)
32-35	49	12 (24.5)	64	26 (40.6)	54	27 (50.0)
36-39	26	7 (26.9)	41	13 (31.7)	38	14 (36.8)
40-43	19	<5	31	14 (45.2)	30	10 (33.3)
44-47	22	7 (31.8)	18	7 (38.9)	28	11 (39.3)
48-51	20	6 (30.0)	26	12 (46.2)	20	7 (35.0)
52-55	7	<5	23	8 (34.8)	15	<5
56-59	7	0	8	<5	18	8 (44.4)
60+	7	<5	13	5 (38.5)	19	<5
<16 years	314	63 (20.1)	304	106 (34.9)	264	97 (36.7)
≥16 years	496	179 (36.1)	515	249 (48.3)	459	199 (43.4)
<18 years	363	86 (23.7)	325	115 (35.4)	293	118 (40.3)
≥18 years	447	156 (34.9)	494	240 (48.6)	430	178 (41.4)
Overall	**810**	**242 (29.9)**	**819**	**355 (43.3)**	**723**	**296 (40.9)**

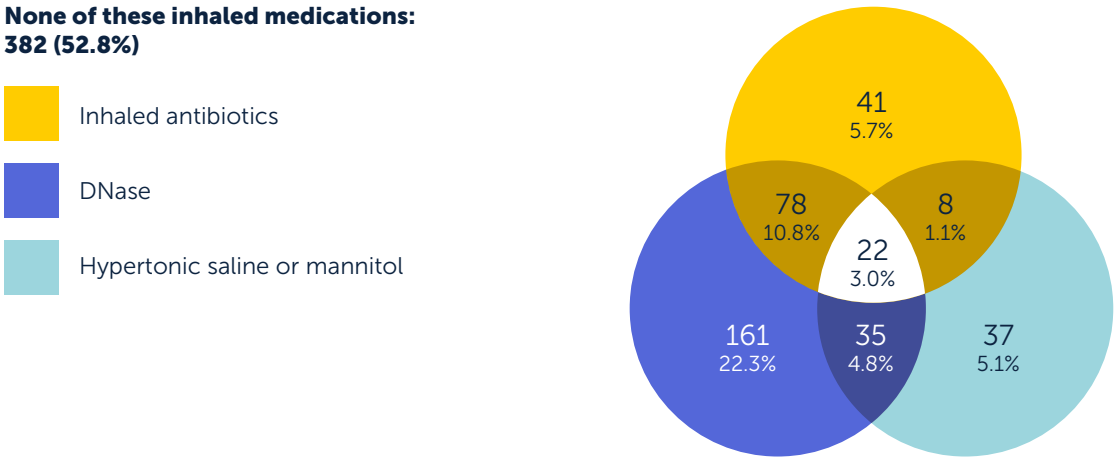
1.32 Hypertonic saline

This treatment helps to thin mucus so that it is easier to cough out of the body.

	2013		2018		2023	
Age	Total patients	Patients on hypertonic saline; n(%)	Total patients	Patients on hypertonic saline; n(%)	Total patients	Patients on hypertonic saline; n(%)
0-3	70	<5	55	7 (12.7)	44	<5
4-7	101	6 (5.9)	82	8 (9.8)	69	18 (26.1)
8-11	82	10 (12.2)	75	17 (22.7)	64	17 (26.6)
12-15	61	12 (19.7)	92	32 (34.8)	87	20 (23.0)
16-19	99	26 (26.3)	53	16 (30.2)	68	10 (14.7)
20-23	97	25 (25.8)	91	26 (28.6)	46	5 (10.9)
24-27	75	9 (12.0)	82	16 (19.5)	60	7 (11.7)
28-31	67	7 (10.4)	65	10 (15.4)	63	6 (9.5)
32-35	49	5 (10.2)	64	13 (20.3)	54	6 (11.1)
36-39	26	0	41	5 (12.2)	38	<5
40-43	19	<5	31	<5	30	<5
44-47	22	<5	18	<5	28	0
48-51	20	<5	26	<5	20	<5
52-55	7	<5	23	<5	15	0
56-59	7	0	8	<5	18	0
60+	7	<5	13	0	19	<5
<16 years	314	29 (9.2)	304	64 (21.1)	264	58 (22.0)
≥16 years	496	81 (16.3)	515	96 (18.6)	459	42 (9.2)
<18 years	363	46 (12.7)	325	72 (22.2)	293	66 (22.5)
≥18 years	447	64 (14.3)	494	88 (17.8)	430	34 (7.9)
Overall	**810**	**110 (13.6)**	**819**	**160 (19.5)**	**723**	**100 (13.8)**

1.33 Burden of treatment

The Venn diagram shows how many people with CF are on one or more inhaled therapies and the combinations they take. A total of 382 (52.8%) people in Scotland are on no inhaled therapies.



CFTR modulators

During 2023 the CFTR modulators (CFTRm) were made available to people with cystic fibrosis under a managed access agreement.

Ivacaftor

In 2023 ivacaftor had approval for use in people aged 4 months and older with at least one copy of a CFTR "gating" variant and for people with the R117H variant.

Lumacaftor/ivacaftor

Lumacaftor / ivacaftor is licensed for use in the UK for people with CF aged one and over with two copies of the F508del variant.

Tezacaftor/ivacaftor

Tezacaftor / ivacaftor is licensed for use in people with CF aged six years and over who have two copies of the F508del variant, or a single copy of F508del and one of 14 "residual" function variants.

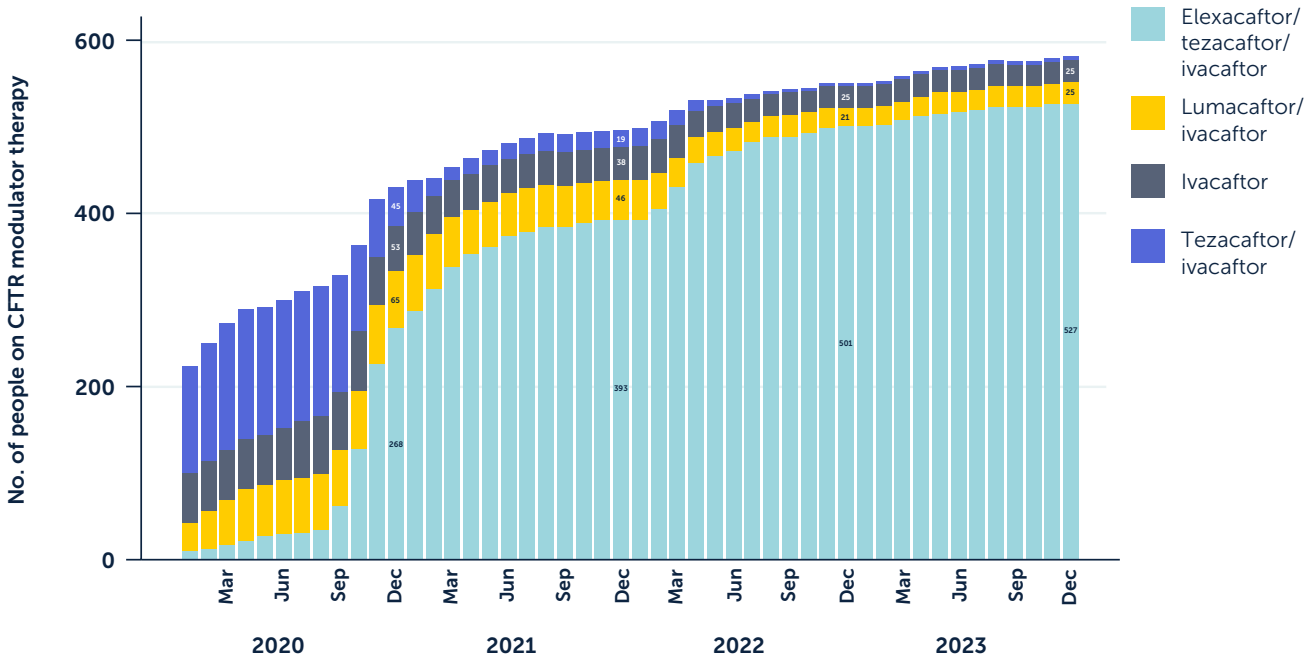
Elexacaftor/tezacaftor/ivacaftor

Elexacaftor/ tezacaftor/ ivacaftor was available for people with CF aged six years and over who have two copies of the F508del variant, or a single copy of F508del and one minimal function variant. In November 2023, this was extended to include children aged two and over. NHS commissioning statements adopted across the UK support the prescribing of CFTR modulators "off label"; the arrangement varies slightly across devolved nations but covers the 177 CFTR variants on an approved "FDA list", and in some devolved nations, responsive genotypes identified through the "French Compassionate Use Programme".

Access arrangements for the CFTR modulators prior to 2023 can be found in previous annual reports and on our website here: **cysticfibrosis.org.uk/treatmentsandmedication**.

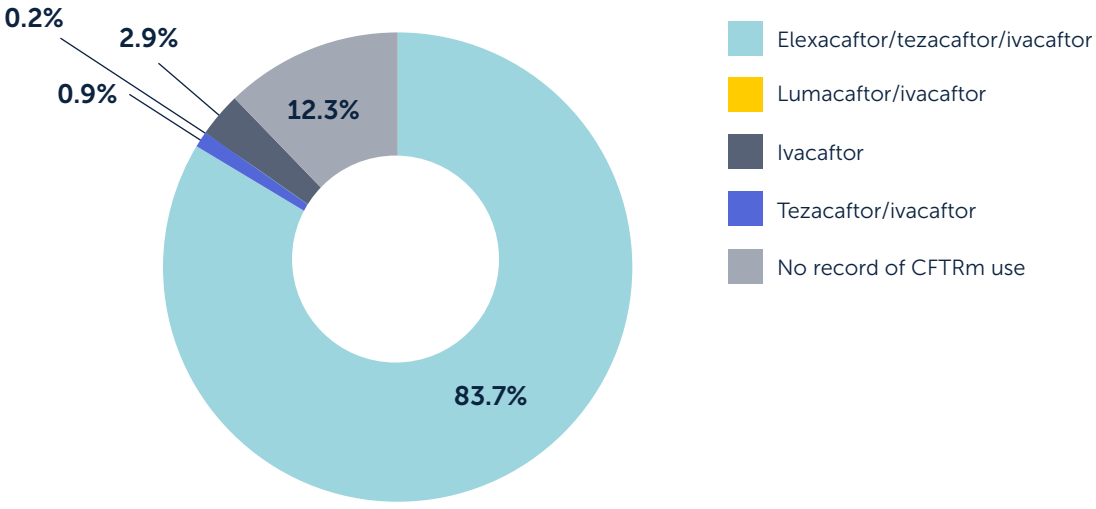
1.34 CFTR modulator use in 2023

The graph below shows the number of people taking each drug by month. Where people switched modulators, the most recent prescription is counted. Only patients who had an annual review are counted. By December, 581 people were taking a CFTR modulator in Scotland.



1.35a CFTR modulator use in all people aged six years and older[1] N=657

The chart shows the distribution of CFTR modulators taken in those aged 6 and over as of 31/12/2023. The last CFTR modulator as of 31/12/2023 is shown in the chart. 15.6% of people aged 6 and over had no record of any CFTRm use on the Registry.

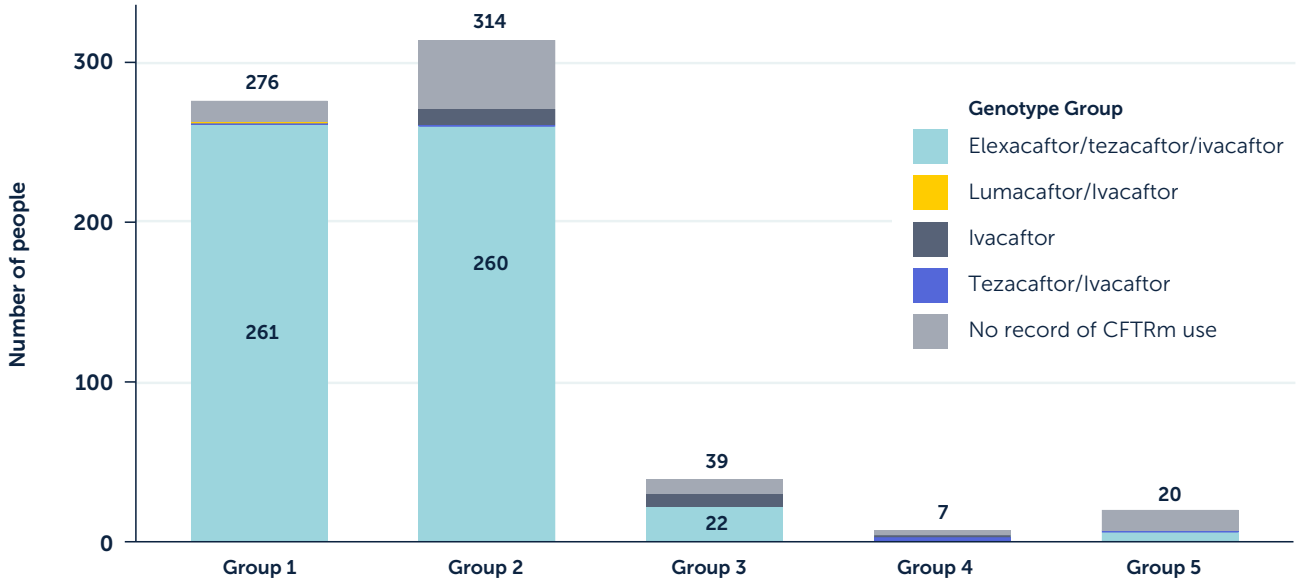


[1] Fewer than 5 people excluded because their last recorded CFTRm treatment was a part of a drug trial where the specific drug was unknown.

1.35b CFTR modulator use in all people aged six years and older by genotype group[1,2]
N=657

The chart below shows the most recent CFTR modulator recorded for people aged six and older and according to their genotype group as defined in the table below. The full list of CFTR variants included within these groups can be found in appendix 3.

Some people with a potentially responsive variant (Groups 1 to 4) have no recorded use of any CFTR modulator on the Registry. There could be several reasons for this: for example, a person may have had a transplant or there could be a data entry error. There are a small number of people[3] thought to have non-responsive genotypes (group 5) who are currently taking or have previously taken a modulator. It is not possible to tell from the Registry record why they received a CFTR modulator.



Genotype Group Definitions	
Group 1	F508del Homozygous
Group 2	F508del Heterozygous
Group 3	no F508del, but ETI* responsive variant as defined by FDA list (3a**) or French Compassionate Use Programme list (3b**)
Group 4	no F508del, no ETI* responsive variant as defined by FDA list (3a**) or French Compassionate Use Programme list (3b**), but at least 1 variant from lists 3c** or 3d**
Group 5	no F508del, no ETI* responsive variant as defined by FDA list (3a**) or French Compassionate Use Programme list (3b**), no variant from lists 3c** or 3d**

[1] Fewer than five patients were excluded because their last recorded CFTRm treatment was as part of a drug trial and the specific drug was unknown

[2] Age as of 31/12/2023

[3] For number and % details see Appendix 3 Table 3e

* ETI is Elexacaftor/tezacaftor/ivacaftor

** see Appendix 3

1.36 Demographic charateristics for people aged six years and older, by genotype group and CFTR modulator use[1,2]

	All potential responders*		Likely non-responders**[4]
	CFTRm use recorded[3]	No record of CFTRm use[3]	No record of CFTRm use[3]
Number of individuals (n)	568	68	13
Male n (%)	311 (54.8)	31 (45.6)	7 (53.9)
Ethnicity			
White n (%)	559 (98.4)	63 (92.7)	7 (53.9)
Asian n (%)	-***	<5	-***
Black n (%)	0	0	<5
Mixed n (%)	<5	<5	0
Other n (%)	0	0	0
Age (years)			
Mean (sd)	27 (15)	33 (17)	23 (11)
Median (IQR)	25 (15, 36)	30 (16, 47)	20 (16, 31)

[1] Fewer than five patients were excluded because their last recorded CFTRm treatment was as part of a drug trial and the specific drug was unknown

[2] Age as of 31/12/2023

[3] "CFTRm record/no record of CFTRm use" defined as if a patient had any CFTRm record as of 31/12/2023.

[4] Details for seven people considered non-responders who had a record of CFTRm use are not included

* Defined at least one F508del or ETI responsive variant as defined by FDA list (3a) or French Compassionate Use list (3b) or variant from lists 3c or 3d.

** Defined as no F508del, no ETI responsive variant as defined by FDA list (3a) or French Compassionate Use list (3b), and no variant from lists 3c or 3d.

*** Redacted to adhere to statistical disclosure guidelines.

Other therapies

1.37 Physiotherapy

Physiotherapy helps people with CF clear sticky mucus from their lungs.

	Overall (n=723)	<16 years (n=264)	≥16 years (n=459)	<18 years (n=293)	≥18 years (n=430)
Active Cycle of Breathing Techniques	53 (7.3)	8 (3.0)	45 (9.8)	8 (2.7)	45 (10.5)
Assisted autogenic drainage	80 (11.1)	5 (1.9)	75 (16.3)	9 (3.1)	71 (16.5)
Autogenic drainage	99 (13.7)	<5	98 (21.4)	<5	98 (22.8)
Exercise; of which:	72 (10.0)	7 (2.7)	65 (14.2)	10 (3.4)	62 (14.4)
Exercise listed as only airway clearance technique	48 (6.6)	5 (1.9)	43 (9.4)	7 (2.4)	41 (9.5)
Forced expiration	12 (1.7)	0 (0.0)	12 (2.6)	0 (0.0)	12 (2.8)
High Pressure PEP	14 (1.9)	12 (4.5)	<5	12 (4.1)	<5
Manual techniques (percussion over pressures vibrations)	<5	<5	<5	<5	<5
Oscillating PEP	54 (7.5)	37 (14.0)	17 (3.7)	43 (14.7)	11 (2.6)
PEP	269 (37.2)	182 (68.9)	87 (19.0)	195 (66.6)	74 (17.2)
Postural drainage	<5	0 (0.0)	<5	0 (0.0)	<5
VEST	<5	0 (0.0)	<5	0 (0.0)	<5
Other	12 (1.7)	<5	9 (2.0)	<5	8 (1.9)
None	53 (7.3)	8 (3.0)	45 (9.8)	10 (3.4)	43 (10.0)

Note that these techniques are not mutually exclusive and represent primary and secondary forms of physiotherapy.

1.38 Primary or secondary airway clearance technique

	Overall (n=723)	<16 years (n=264)	≥16 years (n=459)	<18 years (n=293)	≥18 years (n=430)
Active cycle of breathing techniques	79 (10.9)	12 (4.5)	67 (14.6)	13 (4.4)	66 (15.3)
Assisted autogenic drainage	108 (14.9)	21 (8.0)	87 (19.0)	25 (8.5)	83 (19.3)
Autogenic drainage	171 (23.7)	23 (8.7)	148 (32.2)	26 (8.9)	145 (33.7)
Exercise	428 (59.2)	122 (46.2)	306 (66.7)	146 (49.8)	282 (65.6)
Forced expiration	42 (5.8)	7 (2.7)	35 (7.6)	9 (3.1)	33 (7.7)
High pressure PEP	14 (1.9)	12 (4.5)	<5	12 (4.1)	<5
Manual techniques (percussion over pressures vibrations)	6 (0.8)	<5	<5	<5	<5
Oscillating PEP	77 (10.7)	43 (16.3)	34 (7.4)	52 (17.7)	25 (5.8)
PEP	309 (42.7)	196 (74.2)	113 (24.6)	210 (71.7)	99 (23.0)
Postural drainage	<5	<5	<5	<5	<5
VEST	<5	0 (0.0)	<5	0 (0.0)	<5
Other	110 (15.2)	87 (33.0)	23 (5.0)	89 (30.4)	21 (4.9)

1.39 Exercise testing

Exercise testing provides valuable information on an individual's physical abilities which gives insights into prognosis or oxygen requirements. Physiotherapists and exercise specialists use test results to individualise exercise programmes and target specific needs. Results of exercise tests can also be motivating to the individual and can be used to set future exercise goals.

Exercise Test	Overall (n=723)	<16 years (n=264)	≥16 years (n=459)	<18 years (n=293)	≥18 years (n=430)
Yes*	153 (17.3)	25 (8.5)	128 (21.6)	32 (9.7)	121 (21.8)
No	426 (48.1)	155 (52.7)	271 (45.8)	169 (51.2)	257 (46.2)
Not known or missing	144 (16.3)	84 (28.6)	60 (10.1)	92 (27.9)	52 (9.4)
Type of exercise test[1,2]					
CPET	17 (11.1)	17 (68.0)	0 (0.0)	17 (53.1)	0 (0.0)
Shuttle test	10 (6.5)	5 (20.0)	5 (3.9)	5 (15.6)	5 (4.1)
Step test	38 (24.8)	0 (0.0)	38 (29.7)	<5	37 (30.6)
6 minute walk test	8 (5.2)	<5	<5	<5	<5
Other	12 (7.8)	0 (0.0)	12 (9.4)	<5	-*
Missing	78 (51.0)	<5	74 (57.8)	-*	69 (57.0)

* Exercise test represents all types of testing listed including Cardiopulmonary Exercise Test (CPET), shuttle test, 6 minute walk test, step test and other test.

[1] Proportion of patients who answered Yes above.

[2] More than one type of test can be recorded so % total may not sum to 100%.

1.40 Oxygen and non-invasive ventilation

	Overall (n=723)	<16 years (n=264)	≥16 years (n=459)	<18 years (n=293)	≥18 years (n=430)
Non invasive ventilation (NIV); n (%)	<5	0 (0.0)	<5	0 (0.0)	<5
Any oxygen use; n (%)	23 (3.2)	<5	-*	<5	-*
Among those who had oxygen use:					
Continuously	<5	0 (0.0)	<5	0 (0.0)	<5
Nocturnal or with exertion	9 (39.1)	0 (0.0)	9 (47.4)	0 (0.0)	9 (47.4)
As required (PRN)	<5	0 (0.0)	<5	0 (0.0)	<5
With exacerbation	6 (26.1)	<5	<5	<5	<5

1.41 Transplants

Lung transplantation has been available to people with CF for almost 30 years. Today, the most common operation carried out is a double-lung transplant, or 'bilateral sequential lung transplant'. The following table gives information about transplant activity over time.

	2018	2019	2020	2021	2022	2023
Patients evaluated; n	19	21	11	5	10	3
Patients accepted; n	7	10	2	3	4	3
Patients receiving transplants; n	<5	5	0	1	1	1
Bilateral lung	<5	<5	0	1	0	0
Liver	<5	<5	0	0	0	0
Other	<5	0	0	0	1	1

1.42 Feeding

Supplementary feeding, often using a nasogastric (via the nose) or gastrostomy (via the abdomen) tube directly to the stomach, is considered when a person with CF has poor weight gain, or progressive weight loss, despite efforts to increase oral intake.

Year		Overall	<16 years	≥16 years	<18 years	≥18 years
2013	**Total; n**	**810**	**314**	**496**	**363**	**447**
2013	Any supplemental feeding; n(%)	199 (24.6)	64 (20.4)	135 (27.2)	78 (21.5)	121 (27.1)
2013	Oral; n(%)	155 (19.1)	50 (15.9)	105 (21.2)	59 (16.3)	96 (21.5)
2013	Nasogastric tube; n(%)	9 (1.1)	0 (0.0)	9 (1.8)	<5	7 (1.6)
2013	Gastrostomy tube/Button; n(%)	46 (5.7)	15 (4.8)	31 (6.2)	20 (5.5)	26 (5.8)
2013	Jejunal; n(%)	0 (0.0)	0 (0.0)	0 (0.0)	0 (0.0)	0 (0.0)
2013	Total Parenteral Nutrition (TPN); n(%)	0 (0.0)	0 (0.0)	0 (0.0)	0 (0.0)	0 (0.0)
2018	**Total; n**	**819**	**304**	**515**	**325**	**494**
2018	Any supplemental feeding; n(%)	191 (23.3)	62 (20.4)	129 (25.0)	65 (20.0)	126 (25.5)
2018	Oral; n(%)	140 (17.1)	39 (12.8)	101 (19.6)	42 (12.9)	98 (19.8)
2018	Nasogastric tube; n(%)	11 (1.3)	<5	-*	<5	-*
2018	Gastrostomy tube/Button; n(%)	33 (4.0)	15 (4.9)	18 (3.5)	15 (4.6)	18 (3.6)
2018	Jejunal; n(%)	0 (0.0)	0 (0.0)	0 (0.0)	0 (0.0)	0 (0.0)
2018	Total Parenteral Nutrition (TPN); n(%)	0 (0.0)	0 (0.0)	0 (0.0)	0 (0.0)	0 (0.0)
2023	**Total; n**	**723**	**264**	**459**	**293**	**430**
2023	Any supplemental feeding; n(%)	123 (17.0)	39 (14.8)	84 (18.3)	45 (15.4)	78 (18.1)
2023	Oral; n(%)	72 (10.0)	21 (8.0)	51 (11.1)	23 (7.8)	49 (11.4)
2023	Nasogastric tube; n(%)	<5	<5	<5	<5	0 (0.0)
2023	Gastrostomy tube/Button; n(%)	14 (1.9)	7 (2.7)	7 (1.5)	9 (3.1)	5 (1.2)
2023	Jejunal; n(%)	0 (0.0)	0 (0.0)	0 (0.0)	0 (0.0)	0 (0.0)
2023	Total Parenteral Nutrition (TPN); n(%)	0 (0.0)	0 (0.0)	0 (0.0)	0 (0.0)	0 (0.0)

1.43 Pancreatic enzyme supplementation

Year		Overall	<16 years	≥16 years	<18 years	≥18 years
2013	**Total; n**	**810**	**314**	**496**	**363**	**447**
2013	Pancreatic enzyme supplements; n(%)	676 (83.5)	267 (85.0)	409 (82.5)	311 (85.7)	365 (81.7)
2018	**Total; n**	**819**	**304**	**515**	**325**	**494**
2018	Pancreatic enzyme supplements; n(%)	650 (79.4)	233 (76.6)	417 (81.0)	250 (76.9)	400 (81.0)
2023	**Total; n**	**723**	**264**	**459**	**293**	**430**
2023	Pancreatic enzyme supplements; n(%)	555 (76.8)	198 (75.0)	357 (77.8)	222 (75.8)	333 (77.4)

* Redacted to adhere to statistical disclosure guidelines

Genotypes*

Genotypes are part of the genetic makeup of an individual that usually control a particular characteristic, known as a phenotype. For people with CF, their genotype reveals which variants of the CF gene causes their cystic fibrosis. Everyone living with CF has two variants of the gene for CFTR; one on each allele. One is inherited from their mother, and one from their father. If both variants (or genotypes) are the same, the person is said to be homozygous. Someone who has two different variants is heterozygous.

Data completeness	n(%)
Patients genotyped with at least one variant recorded	968 (99.6)
Patients genotyped with both variants recorded	960 (98.8)
F508del mutations	
Homozygous F508del	406 (41.8)
Heterozygous F508del	470 (48.4)

1.44 Variant combinations in Scotland

This table shows the proportion (%) of patients with the most common mutation combinations. For example, 7.5% of the Scottish population have one copy of F508del and one copy of G551D in their genotype.

	Mutation 1							
Mutation 2	F508del	R117H	G551D	G542X	621+1G->T	Other	Unknown	Total
	(%)							
F508del	41.8							41.8
R117H	6.7	0.1						6.8
G551D	7.5	0.1	0.2					7.8
G542X	5.0	0.2	0.1	0.1				5.5
621+1G->T	0.6	0.0	0.1	0.0	0.0			0.7
Other	27.9	0.9	1.6	0.9	0.3	4.5		36.2
Unknown	0.6	0.1	0.1	0.0	0.0	0.0	0.4	1.2
Total	**90.1**	**1.4**	**2.2**	**1.0**	**0.3**	**4.5**	**0.4**	**100**

* In this section, we include everyone who is registered (see table 1.1) and where mutations are available.

1.45 CFTR variants in the Scottish population

The table below shows the number of people with CF who carry at least one of each mutation. The groups are not mutually exclusive, as people with heterozygous mutations appear twice in the table.

These are the 20 most common mutations in the Scottish population. The full list of recorded mutations can be found in Appendix 2.

.

Nucleotide	Protein	Legacy name	n	%
c.1521_1523delCTT	p.Phe508del	F508del	876	90.1
c.1652G->A	p.Gly551Asp	G551D	95	9.8
c.350G->A	p.Arg117His	R117H	79	8.1
c.1624G->T	p.Gly542X	G542X	62	6.4
c.200C->T	p.Pro67Leu	P67L	54	5.6
c.3454G->C	p.Asp1152His	D1152H	23	2.4
c.1679G->C	p.Arg560Thr	R560T	18	1.9
c.1585-1G->A		1717-1G->A	18	1.9
c.1477C->T	p.Gln493X	Q493X	15	1.5
c.2657+5G->A		2789+5G->A	14	1.4
c.3909C->G	p.Asn1303Lys	N1303K	12	1.2
c.1135G->T	p.Glu379X	E379X	12	1.2
c.489+1G->T		621+1G->T	10	1.0
c.3717+12191C->T		3849+10kbC->T	10	1.0
c.1364C->A	p.Ala455Glu	A455E	9	0.9
c.3528delC	p.Lys1177SerfsX15	3659delC	9	0.9
c.1558G->T	p.Val520Phe	V520F	8	0.8
c.178G->T	p.Glu60X	E60X	8	0.8
c.3140-26A->G		3272-26A->G	7	0.7
c.2657+2_2657+3insA		2789+2insA	7	0.7

Section 2: Centre-level analysis

Cystic fibrosis care in Scotland is led by eight regional centres, two stand-alone clinics and three networked clinics. The breakdown of centres and clinics delivering paediatric and adult care is shown below:

	Paediatric	Adult	Total
Centres	5	3	8
Stand-alone clinics	1	0	1

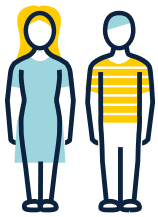
Section 2 shows analysis of data for individual CF centres. This allows people with CF, their families, and healthcare providers to review a centre's use of some medications and outcome data alongside national averages. This transparency is intended to help improve standards of care overall.

Lots of different factors can affect the outcomes of people with CF in centres, not all of which are within a centre's control. This might include the economic profile of the area, the age at which the person with CF was diagnosed and referred to the centre, and certain patient characteristics such as their gender, as well as facilities, care pathways, and the medical team providing care.

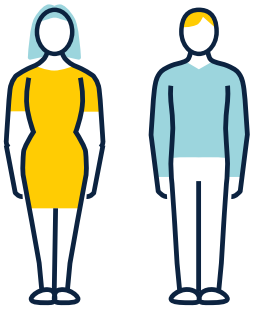
If a person with CF or a member of their family has questions about the results for their CF centre or clinic, they should discuss this with their CF team.

Full tables of the data are shown in Appendix 1.

Key



Paediatric centre

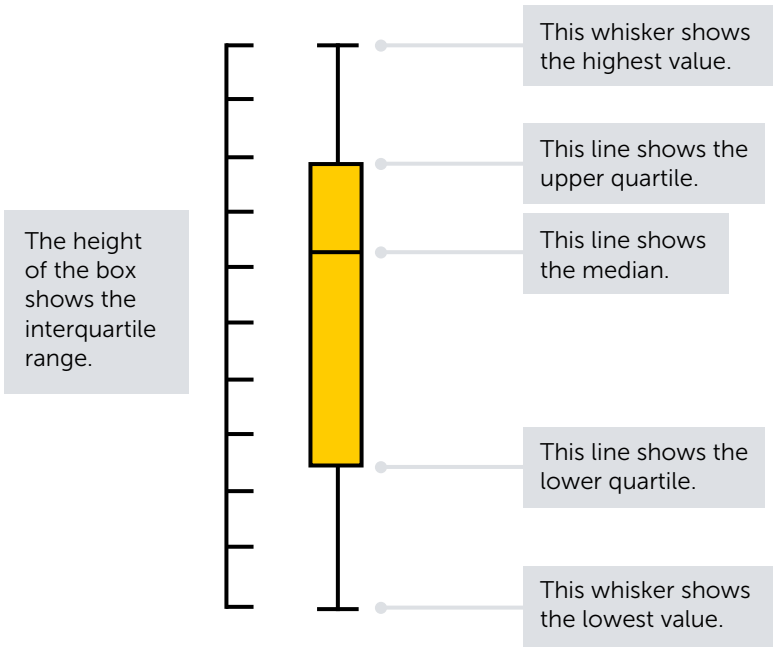


Adult centre

A guide to the charts

Some of the data in this section are shown as 'box plots'.

Box plots



- The 'box' shows the middle half of the data for that centre, going from the first quartile to the third quartile. The longer the box, the more varied the data for that centre.
- The horizontal line within the box shows the median result for that centre.
- The 'whiskers' above and below the box show the highest and lowest values for that centre, excluding any outliers.
- The position of the box between the whiskers shows any skew in the data. If a box is towards the top of the whisker, more of the people for this centre were recorded at the high end of the scale.

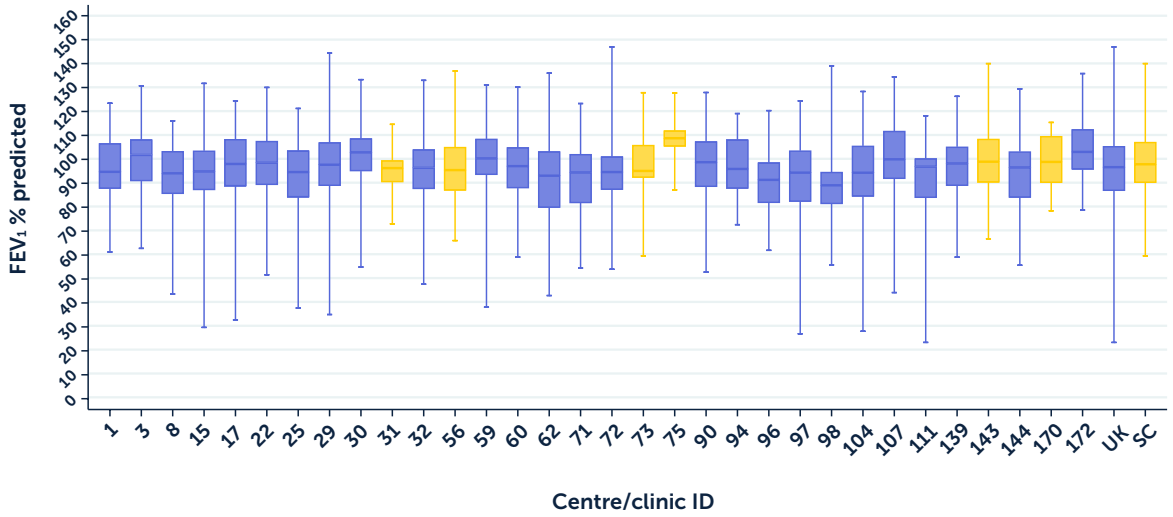
Section 2a: Paediatric centre analysis

This section shows results for the five paediatric centres with their network clinics, and two stand-alone clinics.

Key

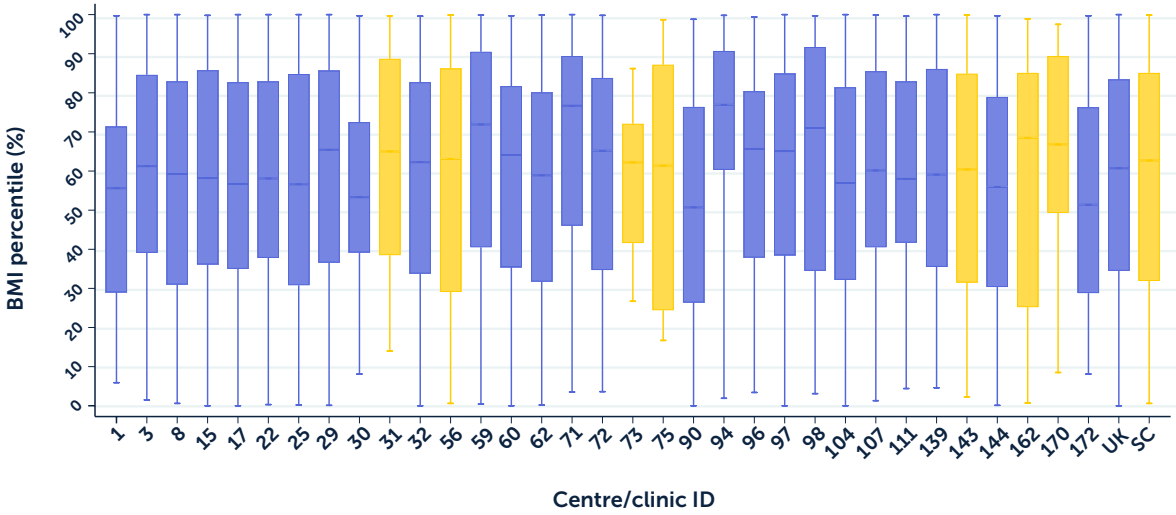
● Services in the UK ● Services in Scotland

2.1 FEV_1% predicted (GLI equations) among patients aged six and older by paediatric centre/clinic (without a history of lung transplant)



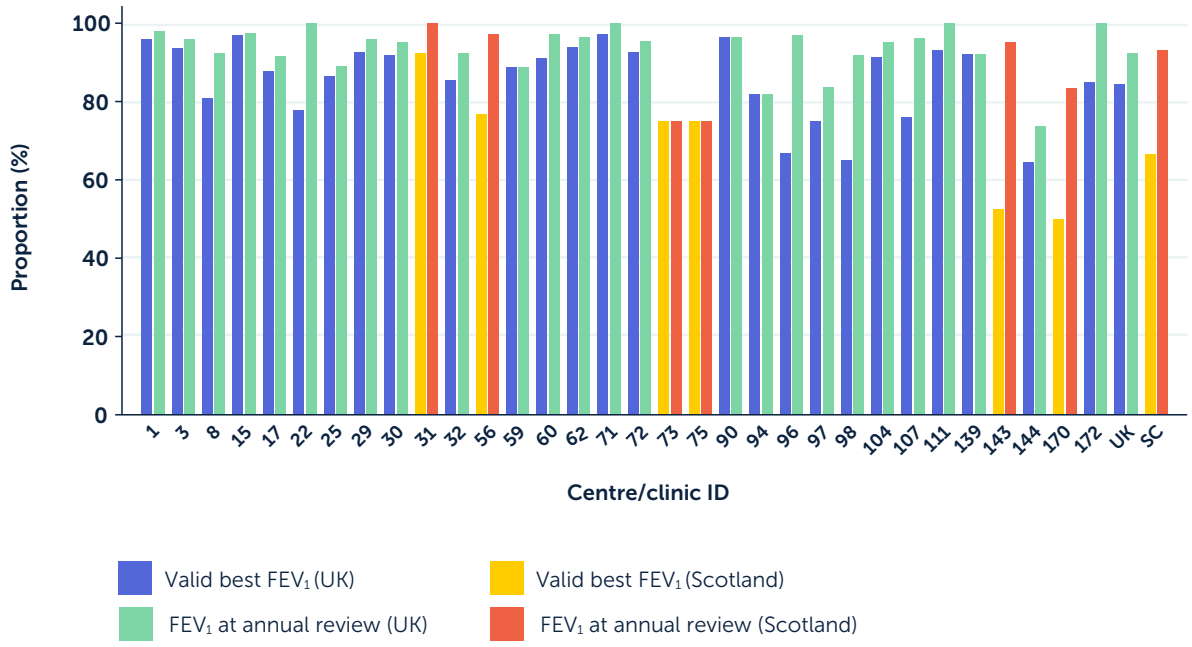
The mean FEV_1% predicted of patients attending paediatric centres/clinics in Scotland is 98.3% predicted (IQR: 90.3-107.2).

2.2 Body Mass Index (BMI) percentile among patients aged two to 15 years by paediatric centre/clinic



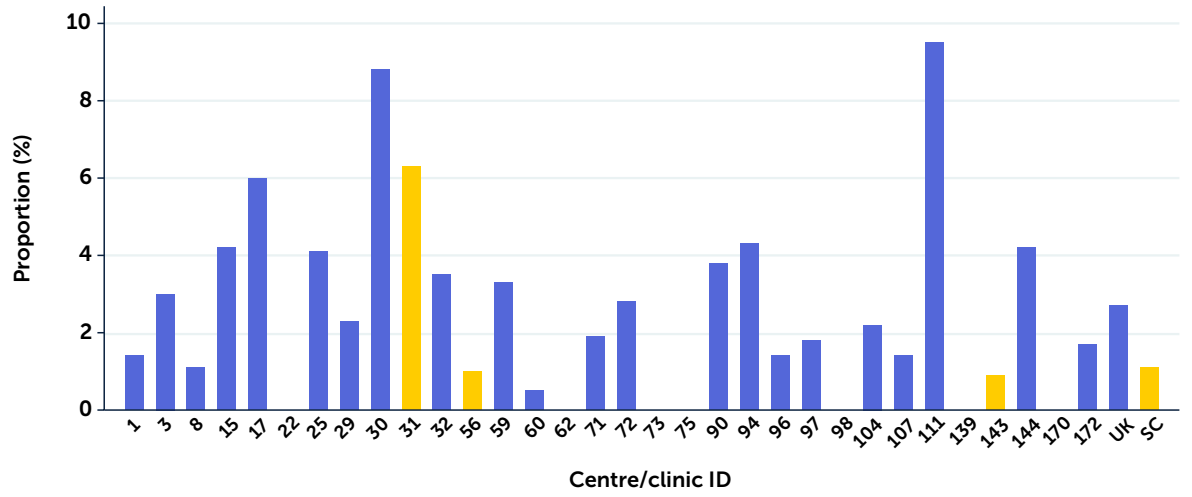
The median BMI percentile of patients attending paediatric centres/clinics in Scotland is 58.3 (IQR: 32.4-83.4).

2.3 Data completeness by paediatric centre/clinic*

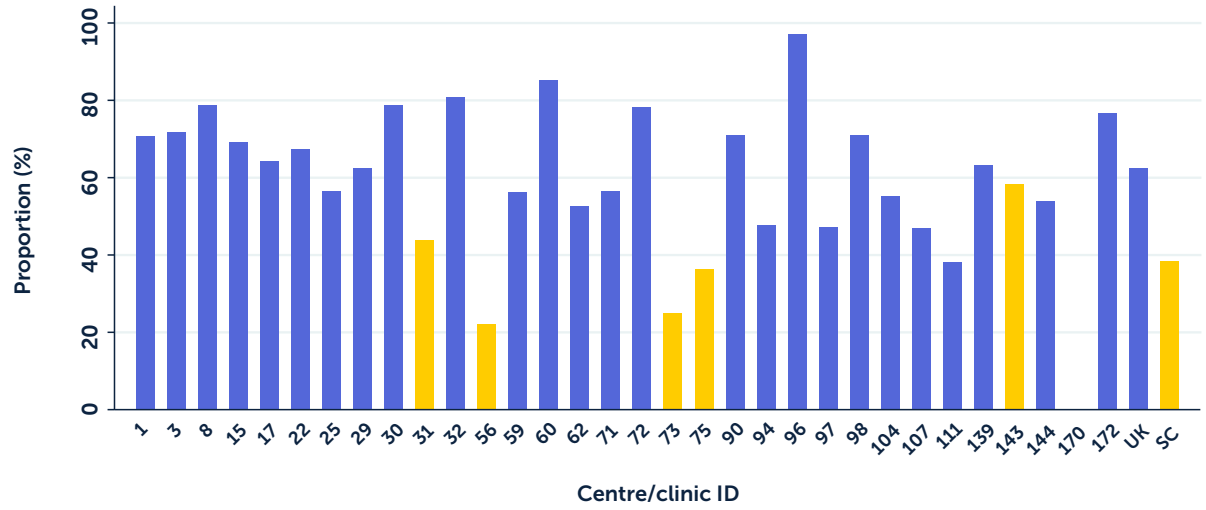


* The chart above shows the proportion of patients who had a valid best FEV_1% and an FEV_1% at annual review, excluding patients under six years of age. Best FEV_1% was considered valid if it was not missing, and the percent predicted was not more than 0.5% lower than the annual review value. For some patients there may be medical reasons why FEV_1 could not be taken, so centres may not be able to get 100% completeness.

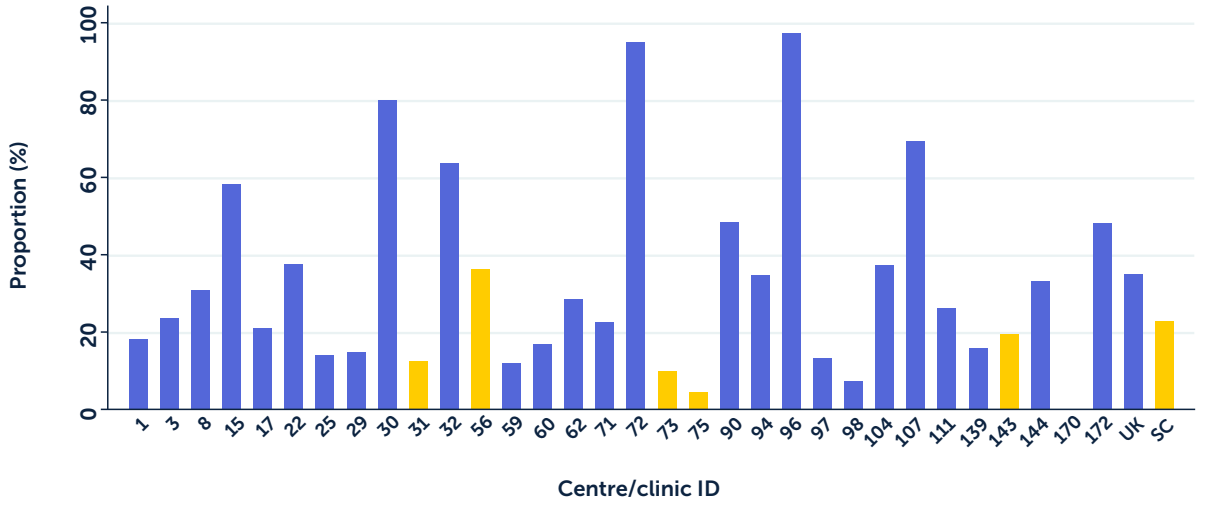
2.4 Proportion of patients with chronic *Pseudomonas aeruginosa* by paediatric centre/clinic



2.5 Proportion of patients receiving DNase treatment by paediatric centre/clinic



2.6 Proportion of patients receiving hypertonic saline treatment by paediatric centre/clinic



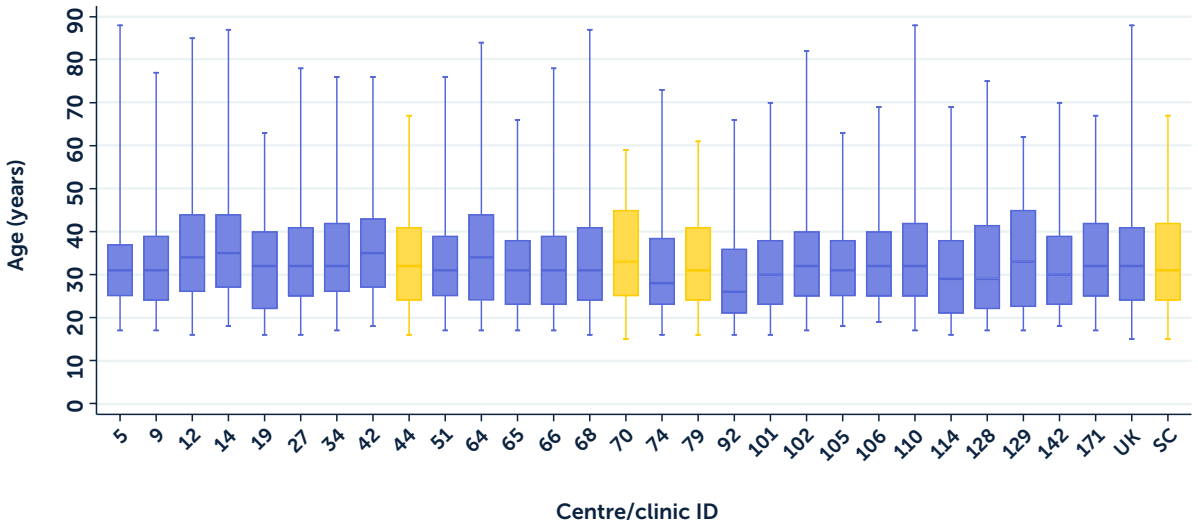
Section 2b: Adult centre analysis

This section shows results for the three adult centres with their network clinics.

Key

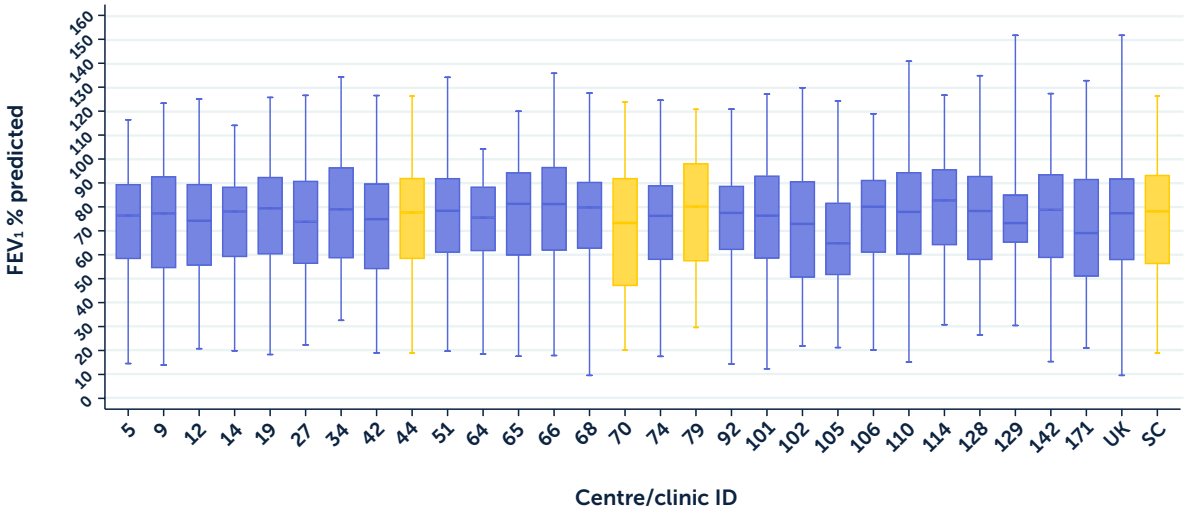
● Services in the UK ● Services in Scotland

2.7 Age distribution by adult centre/clinic



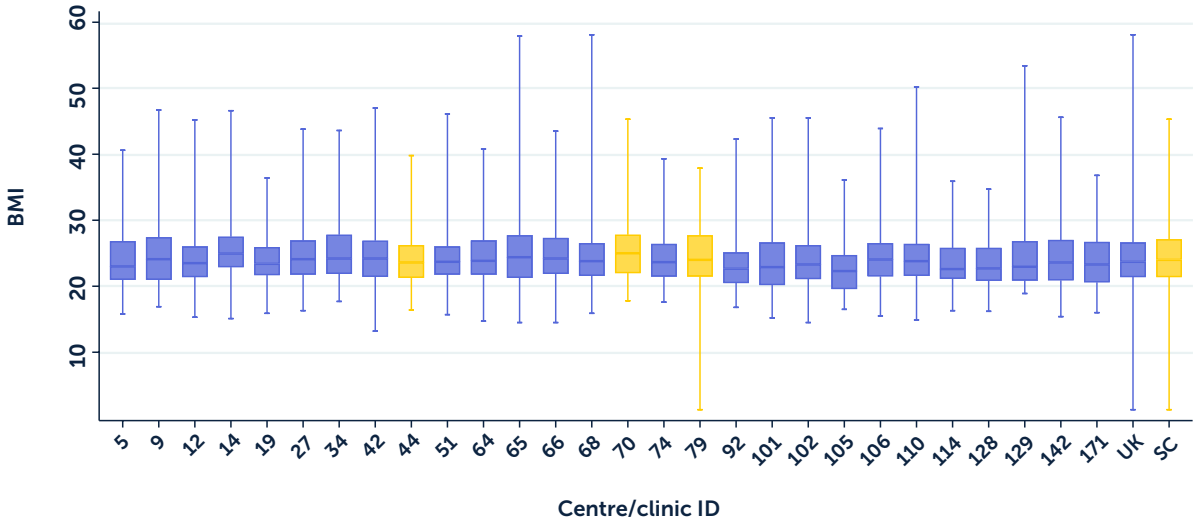
The median age of patients attending adult services in Scotland is 31 years (IQR: 24-42).

2.8 FEV_1% predicted (GLI equations) by adult centre/clinic (without a history of lung transplant)



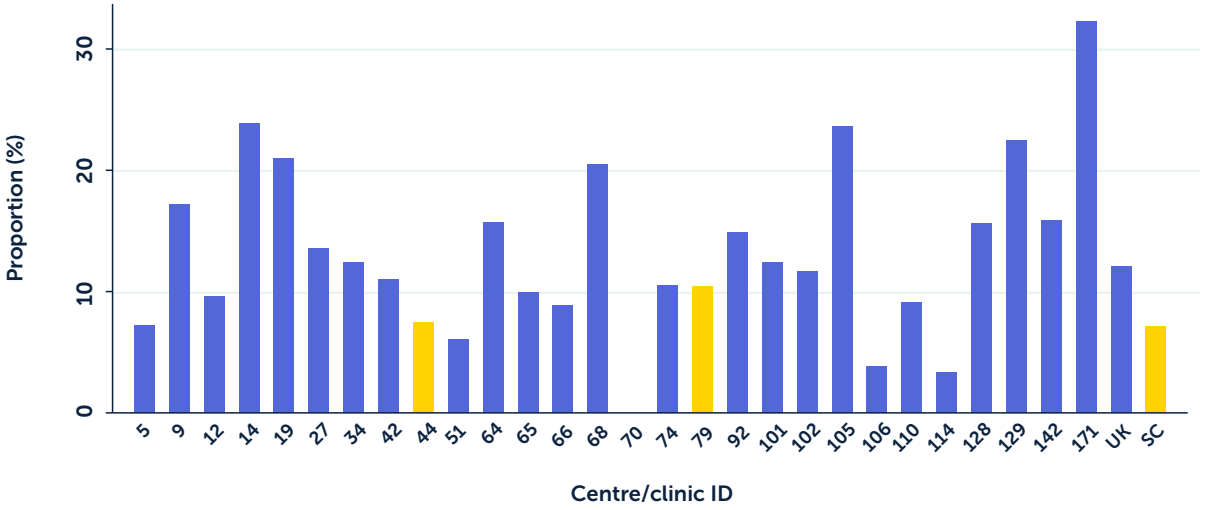
The median FEV_1 % predicted of patients attending adult services in Scotland is 78.1% (IQR 56.1-93.3).

2.9 Body Mass Index (BMI) distribution among patients aged 16 years and older by adult centre/clinic



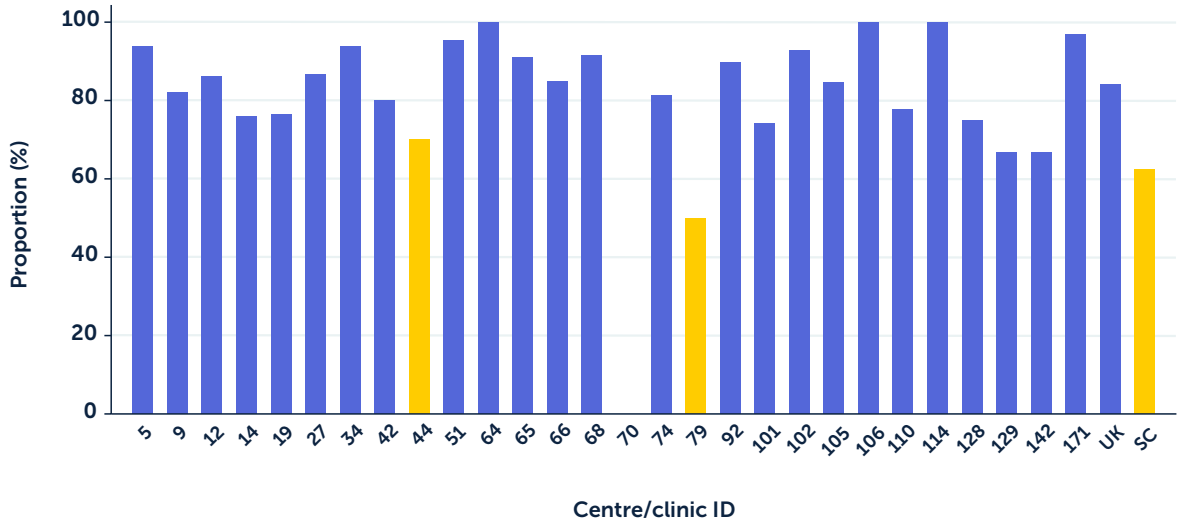
The median BMI of patients attending adult services in Scotland is 24.0 (IQR: 21.4-27.1).

2.10 Proportion of patients with chronic *Pseudomonas aeruginosa* by adult centre/clinic



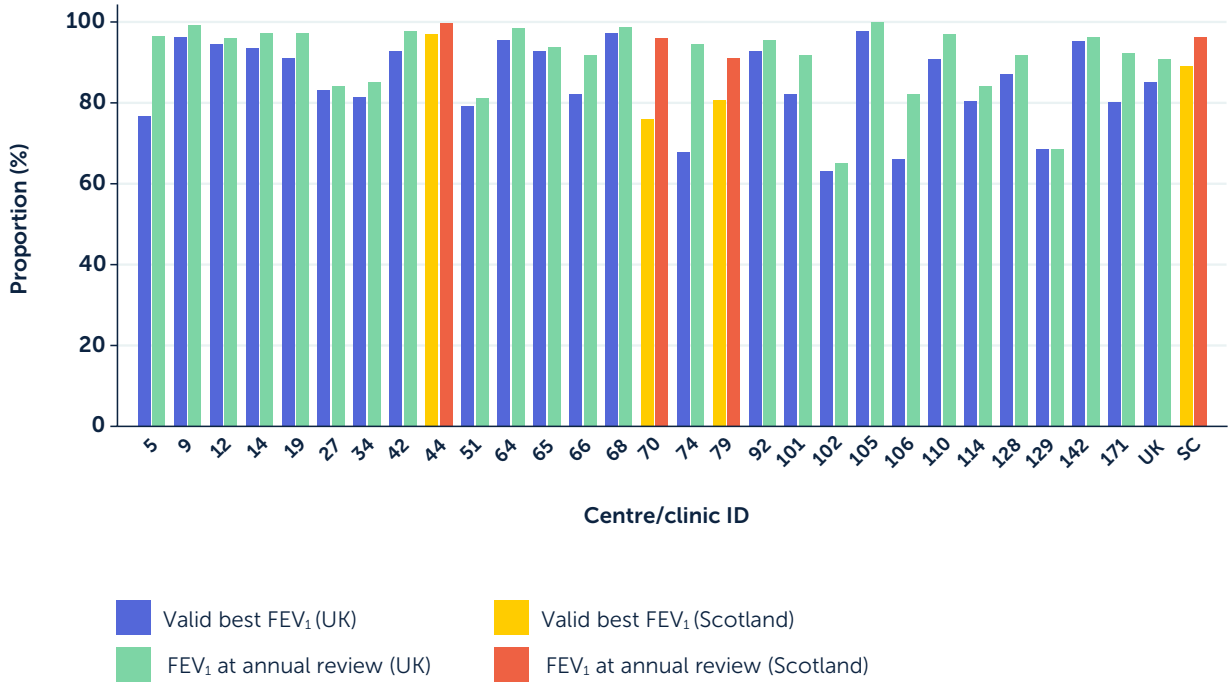
The proportion of patients attending adult services in Scotland with chronic *P. aeruginosa* is 6.3%.

2.11 Inhaled antibiotic use for patients with chronic *Pseudomonas aeruginosa* by centre/clinic



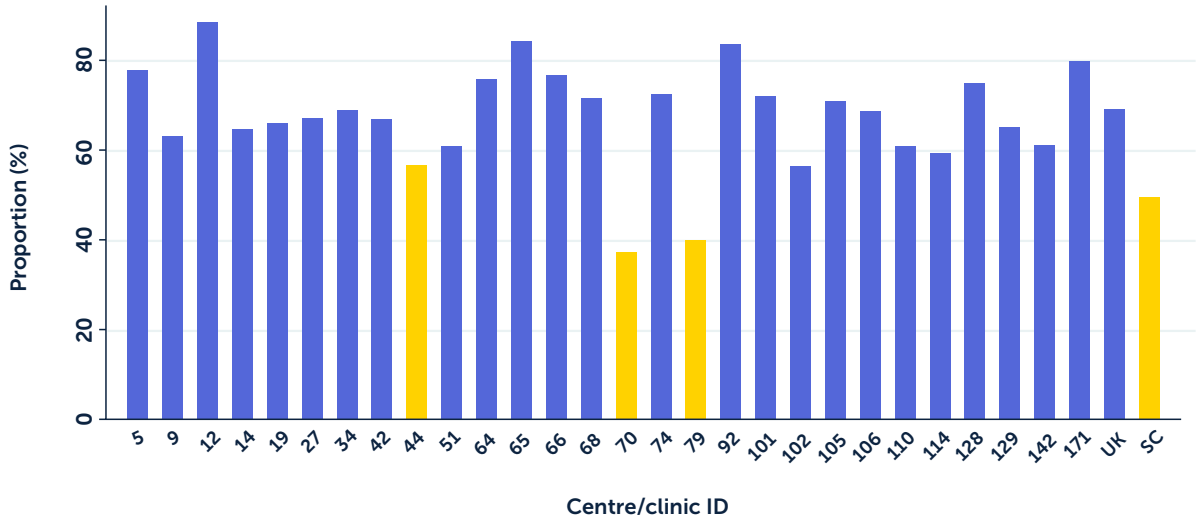
The proportion of chronic *P. aeruginosa* patients on inhaled antibiotics in Scotland is 60.8%.

2.12 Data completeness by adult centre/clinic*



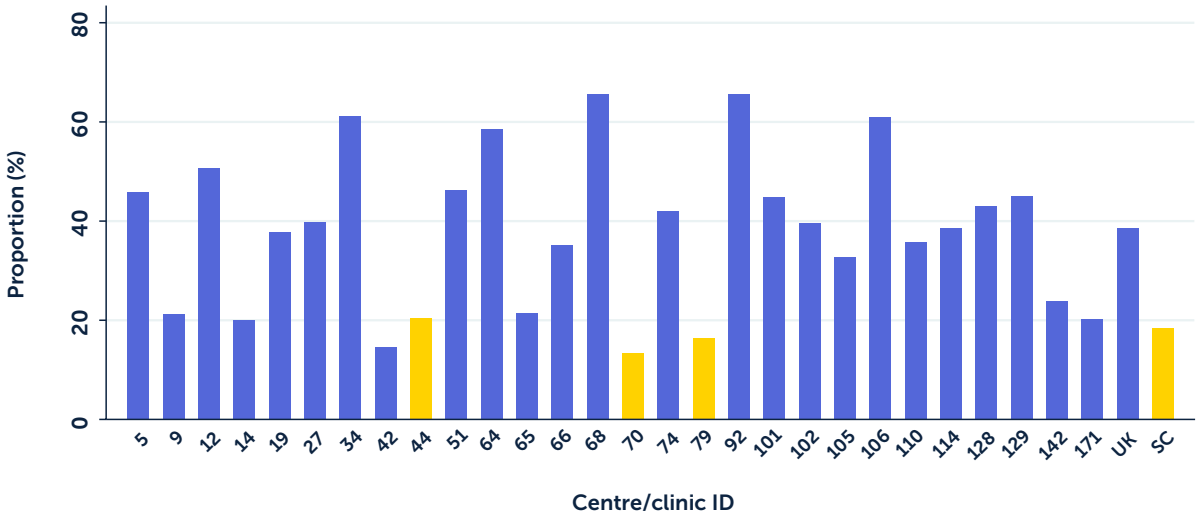
*FEV_1 was considered valid if it was not missing, and the percent predicted was not more than 0.5% lower than the annual review value. For some patients there may be medical reasons why FEV_1 could not be taken, so centres may not be able to get 100% completeness.

2.13 Proportion of patients receiving DNase treatment by adult centre/clinic



The proportion of patients attending adult services in Scotland receiving DNase treatment is 49.6%.

2.14 Proportion of patients receiving hypertonic saline or mannitol treatment by adult centre/clinic



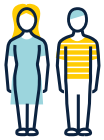
The proportion of patients attending adult services in Scotland receiving hypertonic saline or mannitol treatment is 17.6%.

Glossary

Word/Phrase	Meaning
2023	1 January 2023 – 31 December 2023.
ABPA (allergic bronchopulmonary aspergillosis)	When a person develops a respiratory allergic reaction to *Aspergillus fumigatus*.
Arthritis	A condition causing pain and inflammation in the joints.
Arthropathy	A condition causing pain in the joints.
Asthma	A respiratory condition causing reversible episodes of difficulty breathing, often associated with wheezing.
***Burkholderia cepacia* complex**	*B. cepacia* complex is a group of bacteria, some of which threaten the health of people with cystic fibrosis.
BMI (Body Mass Index)	A measure designed to show whether a person is a healthy weight for their height.
CF	Cystic fibrosis.
CFTR (cystic fibrosis transmembrane conductance regulator)	A protein at the cell surface that controls the salt and water balance across a cell. The gene that causes cystic fibrosis is the blueprint for the CFTR protein. Everyone has two copies of the gene for CFTR. To be born with cystic fibrosis, both CFTR genes must be affected by a CF-causing mutation.
Chronic	Persistent, or long-lasting.
Cirrhosis	A chronic liver disease.
CI (confidence interval)	A way of expressing how certain we are about our statistical estimates of a clinical measure (eg BMI). It gives a range of results that is likely to include the 'true' value for the population. A narrow confidence interval indicates a more precise estimate. A wide confidence interval indicates more uncertainty about the true value of the clinical measure - often because a small group of patients has been studied. The confidence interval is usually stated as '95% CI', which means that the range of values has a 95 in 100 chance of including the 'true' value.
Enzymes	Biological molecules that help complex reactions, such as digestion of food, occur in the body.
FEV_1 (forced expiratory volume in one second)	This is the amount of air that a person can blow out of the lungs in the first second of a forced exhaled breath. People with healthy lungs can blow out most of the air held in this time.
FEV_1% predicted	The FEV_1 can be converted from absolute litres of air blown out into a predicted percentage (%). A healthy range for % predicted is calculated from a very large population sample, and is normally considered to be between 80-120% predicted.
Fibrosing colonopathy	A condition causing narrowing of part of the colon.
Gall bladder	The small sac-shaped organ under the liver that stores bile after it is secreted by the liver, before it is released into the intestine.
Gastrointestinal (GI) tract	The GI tract is an organ system responsible for digesting food, absorbing nutrients and expelling waste.
Genotype	Part of the genetic makeup of a cell, organism or individual that usually controls a particular characteristic (known as a phenotype).
GORD (gastroesophageal reflux disease)	A chronic symptom of damage caused by stomach acid coming up from the stomach into the oesophagus.
GI bleed	Bleeding in the gastrointestinal tract.
GLI equations	Global Lung Initiative, the equation used for calculating FEV_1% predicted from absolute FEV_1, which takes into account age, gender, height and ethnicity.
Haemophilus influenza	*H. influenza* is a bacterium that can cause serious illness.
Haemoptysis	The coughing up of blood.
Hepatobiliary disease	A liver or biliary disorder.
Heterozygous	Everyone living with cystic fibrosis has two mutations of the gene for CFTR, one inherited from their mother and one from their father. Someone who has two different mutations is heterozygous.

Word/Phrase	Meaning
Homozygous	Everyone living with cystic fibrosis has two mutations of the gene for CFTR, one inherited from their mother and one from their father. If both mutations (or genotypes) are the same, the person is said to be homozygous.
Hypertension	High blood pressure.
Incidence	The number of people newly diagnosed with a condition in the given year.
IQR (interquartile range)	Also called the mid-spread, or middle fifty, IQR is a measure of the spread of data. It shows the difference between the upper and lower quartiles. IQR = Q3 – Q1.
Mean	A type of average, calculated by adding up all the values and dividing by the number of values.
Median	The middle number, when all numbers are arranged from smallest to largest.
Median age of death	Median age of death is based on the people with CF who died in any given year.
MRSA	Methicillin-resistant *Staphylococcus aureus* is a type of bacteria that is resistant to a number of widely used antibiotics.
Mutation	A mutation is a change in a gene. When both of a child's parents are carriers of a CF-causing mutation there is a 25% chance that the child will have cystic fibrosis. There are over 1,400 different mutations of the CFTR gene that can cause cystic fibrosis.
Nasal polyps	Small, sac-like growths of inflamed mucus membrane caused by chronic inflammation of the nasal lining.
NBS (newborn screening)	Newborn screening is part of the heel prick blood spot testing carried out on all babies at 5-7 days of age. The blood sample is tested for a number of conditions, including cystic fibrosis.
NTM (non tuberculous mycobacteria)	A mycobacterium that does not cause tuberculosis, but which can cause respiratory infection. There are several known types.
Osteopenia	A medical condition less severe than osteoporosis, where the mineral content of bone is reduced.
Osteoporosis	A condition where the bones become brittle from loss of tissue.
Pancreas	An organ in the digestive system that produces insulin and digestive enzymes.
Pancreatitis	Inflammation of the pancreas.
Peptic ulcer	An open sore that develops in the lining of the stomach, also known as a stomach ulcer.
Percentile	A percentile shows where a value stands, relative to the rest of the data. If a value is higher than 90% of the rest of the data, it is at the 90th percentile.
Pneumothorax	A collection of air in the cavity between the lungs and the chest wall causing collapse of the lung on the affected side.
Portal hypertension	High blood pressure in the portal vein system, which is the blood system of the liver.
Prenatal	Before birth, while the baby is still in the womb.
Prevalence	The overall number of people with the condition in the last 12 months.
Pseudomonas aeruginosa	A tough bacterial strain. Rarely affecting healthy people, it can cause a wide range of infections, particularly in those with a weakened immune system.
Rectal prolapse	When the rectal wall slides through the anus.
Renal	Relating to the kidneys.
Staphylococcus aureus	*S. aureus* is a bacterium that can cause disease if it enters the body.
Sinus disease	When the sinuses, which are usually filled with air, are full of thick sticky mucus.
Statistically significant	This phrase means that after careful calculations there is a definite difference between two groups, which is not simply a result of chance.

Appendix 1: Centre-level data tables

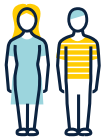


Paediatric centres/clinics providing data in 2023 – ordered alphabetically by location

Location	Name	Clinic ID	Total Active	Number with annual review
Scotland				
Aberdeen	Royal Aberdeen Children's Hospital	75	29	22
Ayr	University Hospital Crosshouse	170	19	8
Dundee	Ninewells Hospital	73	21	20
Edinburgh	Royal Hospital for Sick Children	143	124	108
Glasgow	Royal Hospital for Sick Children	56	160	99
Inverness	Raigmore Hospital	31	17	16

Paediatric centres/clinics providing data in 2023 – ordered alphabetically by location

			BMI			
Location	Name	Clinic ID	Number	Mean - unadjusted	Mean - adjusted	Median
Scotland						
Aberdeen	Royal Aberdeen Children's Hospital	75	20	59.8	59.9	58.3
Ayr	University Hospital Crosshouse	170	5	54.9	54.7	53.5
Dundee	Ninewells Hospital	73	17	53.0	53.0	54.6
Edinburgh	Royal Hospital for Sick Children	143	104	60.5	60.5	61.4
Glasgow	Royal Hospital for Sick Children	56	90	56.1	56.1	58.8
Inverness	Raigmore Hospital	31	15	53.8	53.8	56.8



	Age		FEV$_1$% predicted at annual review				Best** FEV$_1$% predicted			
Clinic ID	Mean	Median	Number	Mean - unadjusted	Mean - adjusted	Median	Number*	Mean - unadjusted	Mean - adjusted	Median
75	7.4	6.5	9	109.1	108.1	108.8	12	108.2	107.1	107.5
170	9.9	12.0	5	98.6	99.3	98.9	5	101.1	102.0	98.9
73	9.6	10.8	12	97.1	96.6	95.1	14	101.1	100.5	101.4
143	9.8	10.1	80	98.7	98.4	99.0	83	101.6	101.2	101.3
56	8.9	8.8	67	97.3	96.8	95.5	69	101.0	100.3	100.4
31	9.7	9.4	13	95.5	95.1	96.3	13	100.0	99.4	99.0

	Chronic *pseudomonas*		Having at least 1 IV days		Receiving DNase treatment		Receiving hypertonic saline or mannitol treatment		Inhaled antibiotic use among patients with chronic *Pseudomonas*	
Clinic ID	Number	Proportion (%)	Number	Proportion (%)	Number	Proportion (%)	Number	Proportion (%)	Number	Proportion (%)
75	0	0.0	0	0.0	8	36.4	<5	-*	0	0.0
170	0	0.0	<5	-*	0	0.0	0	0.0	0	0.0
73	0	0.0	<5	-*	5	25.0	<5	-*	0	0.0
143	<5	-*	19	17.6	63	58.3	21	19.4	<5	-*
56	<5	-*	12	12.1	21	21.2	35	35.4	<5	-*
31	<5	-*	0	0.0	7	43.8	<5	-*	<5	-*

* Redacted to adhere to statistical disclosure guidelines.

** Where 'Best' values were missing, or lower than FEV$_1$% predicted taken at annual review, the annual review value was used.

Appendix 1: Centre-level data tables

Adult centres/clinics providing data in 2023 – ordered alphabetically by location

Location	Name	Clinic ID	Total active	Number with annual review
Scotland				
Aberdeen	Aberdeen Royal Infirmary	70	78	67
Edinburgh	Western General Hospital	44	282	268
Glasgow	Queen Elizabeth University Hospital	79	242	115

Adult centres/clinics providing data in 2023 – ordered alphabetically by location

			BMI			
Location	Name	Clinic ID	Number	Mean - unadjusted	Mean - adjusted	Median
Scotland						
Aberdeen	Aberdeen Royal Infirmary	70	66	26.1	26.0	25.0
Edinburgh	Western General Hospital	44	268	24.4	24.5	23.7
Glasgow	Queen Elizabeth University Hospital	79	115	24.6	24.6	24.0

	Age		FEV$_1$% predicted at annual review				Best** FEV$_1$% predicted			
Clinic ID	Mean	Median	Number	Mean - unadjusted	Mean - adjusted	Median	Number	Mean - unadjusted	Mean - adjusted	Median
70	35.2	33.4	60	72.0	71.8	74.3	60	75.8	75.8	77.6
44	34.3	32.0	253	74.5	73.9	77.7	254	79.3	79.0	83.1
79	33.7	31.0	109	77.3	76.9	80.1	113	79.6	79.3	83.4

	Chronic *pseudomonas*		Having at least 1 IV days		Receiving DNase treatment		Receiving hypertonic saline or mannitol treatment		Inhaled antibiotic use among patients with chronic *pseudomonas*	
Clinic ID	Number	Proportion (%)	Number	Proportion (%)	Number	Proportion (%)	Number	Proportion (%)	Number	Proportion (%)
70	0	0.0	9	13.4	20	29.9	<5	-*	0	0.0
44	20	10.9	46	17.2	136	50.7	30	11.2	14	70.0
79	12	11.3	31	27.0	36	31.3	9	7.8	6	50.0

* Redacted to adhere to statistical disclosure guidelines.

** Where 'Best' values were missing, or lower than FEV$_1$% predicted taken at annual review, the annual review value was used.

Appendix 2: Full list of mutations in the Scottish population

The table below shows the number of people with CF who carry at least one of each mutation.

The groups are not mutually exclusive, as people with heterozygous mutations appear twice in the table.

Nucleotide	Protein	Legacy name	N	%
c.1521_1523delCTT	p.Phe508del	F508del	876	90.1
c.1652G->A	p.Gly551Asp	G551D	95	9.8
c.350G->A	p.Arg117His	R117H	79	8.1
c.1624G->T	p.Gly542X	G542X	62	6.4
c.200C->T	p.Pro67Leu	P67L	54	5.6
c.3454G->C	p.Asp1152His	D1152H	23	2.4
c.1679G->C	p.Arg560Thr	R560T	18	1.9
c.1585-1G->A		1717-1G->A	18	1.9
c.1477C->T	p.Gln493X	Q493X	15	1.5
c.2657+5G->A		2789+5G->A	14	1.4
c.3909C->G	p.Asn1303Lys	N1303K	12	1.2
c.1135G->T	p.Glu379X	E379X	12	1.2
c.489+1G->T		621+1G->T	10	1.0
c.3717+12191C->T		3849+10kbC->T	10	1.0
c.1364C->A	p.Ala455Glu	A455E	9	0.9
c.3528delC	p.Lys1177SerfsX15	3659delC	9	0.9
c.1558G->T	p.Val520Phe	V520F	8	0.8
c.178G->T	p.Glu60X	E60X	8	0.8
c.3140-26A->G		3272-26A->G	7	0.7
c.2657+2_2657+3insA		2789+2insA	7	0.7
c.1705T->G	p.Tyr569Asp	Y569D	5	0.5
c.3846G->A	p.Trp1282X	W1282X	5	0.5
c.1519_1521delATC	p.Ile507del	I507del	5	0.5
c.1766+1G->A		1898+1G->A	5	0.5
c.1329_1330insAGAT	p.Ile444ArgfsX3	1461ins4	<5	-
c.1721C->A	p.Pro574His	P574H	<5	-
c.1657C->T	p.Arg553X	R553X	<5	-
c.948delT	p.Phe316LeufsX12	1078delT	<5	-
c.254G->A	p.Gly85Glu	G85E	<5	-
c.579+3A->G		711+3A->G	<5	-
c.2012delT	p.Leu671X	2143delT	<5	-
c.3468G->A		3600G->A	<5	-
c.429delT	p.Phe143LeufsX10	557delT	<5	-
c.1523T->G	p.Phe508Cys	F508C	<5	-
c.509G->A	p.Arg170His	R170H	<5	-
c.2490+1G->A		2622+1G->A	<5	-
c.223C->T	p.Arg75X	R75X	<5	-
c.1680A->C	p.Arg560Ser	R560S	<5	-
c.3196C->T	p.Arg1066Cys	R1066C	<5	-
c.1006_1007insG	p.Ile336SerfsX28	1138insG	<5	-
c.2988+1G->A		3120+1G->A	<5	-
c.1210-12[5](AJ574948.1:g.152T[5])		5T	<5	-

Nucleotide	Protein	Legacy name	N	%
c.1367T->C	p.Val456Ala	V456A	<5	-
c.164+2T>C		296+2T->C	<5	-
c.2052delA	p.Lys684AsnfsX38	2184delA	<5	-
c.3737C->T	p.Thr1246Ile	T1246I	<5	-
c.617T->G	p.Leu206Trp	L206W	<5	-
c.3884_3885insT	p.Ser1297PhefsX5	4016insT	<5	-
c.3276C->A or c.3276C->G	p.Tyr1092X	Y1092X(C->A)	<5	-
c.292C->T	p.Gln98X	Q98X	<5	-
c.933C>G	p.Phe311Leu	F311L	<5	-
c.3475T->C	p.Ser1159Pro	S1159P	<5	-
c.274G->A	p.Glu92Lys	E92K	<5	-
c.1327G->T	p.Asp443Tyr	D443Y	<5	-
c.1986_1989delAACT	p.Thr663ArgfsX8	2118del4	<5	-
c.54-5940_273+10250del21kb	p.Ser18ArgfsX16	CFTRdele2,3	<5	-
c.2900T->C	p.Leu967Ser	L967S	<5	-
c.2859_2890delACATTCTGTTCTTC AAGCACCTATGTCAACCC	p.Leu953PhefsX11	2991del32	<5	-
c.262_263delTT	p.Leu88IlefsX22	394delTT	<5	-
c.3197G->A	p.Arg1066His	R1066H	<5	-
c.1040G->C	p.Arg347Pro	R347P	<5	-
c.2988G->A		3120G->A	<5	-
c.1585-8G->A		1717-8G->A	<5	-
c.3476C->T	p.Ser1159Phe	S1159F	<5	-
c.443T->C	p.Ile148Thr	I148T	<5	-
c.2051_2052delAAinsG	p.Lys684SerfsX38	2183AA->G or 2183delAA->G	<5	-
c.3158C->T	p.Thr1053Ile	T1053I	<5	-
c.1466C->A	p.Ser489X	S489X	<5	-
c.3208C->T	p.Arg1070Trp	R1070W	<5	-
c.349C->T	p.Arg117Cys	R117C	<5	-
c.3484C->T	p.Arg1162X	R1162X	<5	-
c.3705T->G	p.Ser1235Arg	S1235R	<5	-
c.4147_4148insA	p.Ile1383AsnfsX3	4279insA	<5	-
c.1538A->G	p.Asp513Gly	D513G	<5	-
c.2583delT	p.Phe861LeufsX3	2711delT	<5	-
c.(3873+1_3874-1)_(3963+1_3964-1) del		CFTRdele21	<5	-
c.349C->G	p.Arg117Gly	R117G	<5	-
c.1055G->A	p.Arg352Gln	R352Q	<5	-
c.273+1G->A		405+1G->A	<5	-
c.(53+1_54-1)_(489+1_490-1)del		CFTRdele2-4	<5	-
c.[1210–12[5];1210-34TG[12]]		5T;TG12	<5	-
c.1209+1G->A		1341+1G->A	<5	-
c.1753G->T	p.Glu585X	E585X	<5	-
c.[1210–12[5];1210-34TG[13]]		5T;TG13	<5	-
c.233dupT	p.Trp79LeufsX32	365-366insT	<5	-
c.1029delC	p.Cys343X	1161delC	<5	-
c.1022_1023insTC	p.Phe342HisfsX28	1154insTC	<5	-
c.1000C->T	p.Arg334Trp	R334W	<5	-

Appendix 3: Legacy names lists for modulator eligibility*

List 3a: FDA list of CFTR variants potentially responsive to elexacaftor/ tezacaftor/ ivacaftor[1]

3141del9	E822K	G1244E	L997F	R117P	S945L
546insCTA	F191V	G1249R	L1077P	R170H	S977F
A46D	F311del	G1349D	L1324P	R258G	S1159F
A120T	F311L	H139R	L1335P	R334L	S1159P
A234D	F508C	H199Y	L1480P	R334Q	S1251N
A349V	F508C;S1251N	H939R	M152V	R347H	S1255P
A455E	F575Y	H1054D	M265R	R347L	T338I
A554E	F1016S	H1085P	M952I	R347P	T1036N
A1006E	F1052V	H1085R	M952T	R352Q	T1053I
A1067T	F1074L	H1375P	M1101K	R352W	V201M
D110E	F1099L	I148T	P5L	R553Q	V232D
D110H	G27R	I175V	P67L	R668C	V456A
D192G	G85E	I336K	P205S	R751L	V456F
D443Y	G126D	I502T	P574H	R792G	V562I
D443Y;G576A;R668C	G178E	I601F	Q98R	R933G	V754M
D579G	G178R	I618T	Q237E	R1066H	V1153E
D614G	G194R	I807M	Q237H	R1070Q	V1240G
D836Y	G194V	I980K	Q359R	R1070W	V1293G
D924N	G314E	I1027T	Q1291R	R1162L	W361R
D979V	G463V	I1139V	R31L	R1283M	W1098C
D1152H	G480C	I1269N	R74Q	R1283S	W1282R
D1270N	G551D	I1366N	R74W	S13F	Y109N
E56K	G551S	K1060T	R74W;D1270N	S341P	Y161D
E60K	G576A	L15P	R74W;V201M	S364P	Y161S
E92K	G576A;R668C	L165S	R74W;V201M;D1270N	S492F	Y563N
E116K	G622D	L206W	R75Q	S549N	Y1014C
E193K	G628R	L320V	R117C	S549R	Y1032C
E403D	G970D	L346P	R117G	S589N	
E474K	G1061R	L453S	R117H	S737F	
E588V	G1069R	L967S	R117L	S912L	

[1] https://pi.vrtx.com/files/uspi_elexacaftor_tezacaftor_ivacaftor.pdf

List 3b: potentially responsive CFTR variants according to French Compassionate Use Programme[2]

R334W	3849+10kbC->T
R1066C	3272-26A->G
2789+5G->A	3041-15T->G
	N1303K

[2] Burgel PR et al, Eur Respir J. Feb. 2023

* Please see graphs 1.35b and 1.36 in the report which reference these lists

List 3c: CFTR variants considered suitable for tezacaftor / ivacaftor use[3]

Named variants	E56K	P67L	D110H
	R117C	E193K	R347H
	L206W	R352Q	A455E
	711+3A->G	E831X	S945L
	K1060T	A1067T	2789+5G->A
	3272-26A->G	3849+10kbC->T	
Variants with "varying clinical consequence" (VCC)	R74W	D110E	D579G
	S977F	F1052V	R1070W
	F1074L	D1152H	D1270N

[3] https://www.england.nhs.uk/wp-content/uploads/2020/08/Urgent-policy-statement-CFTR-off-label-rarer-mutations.pdf

List 3d: CFTR variants considered suitable for ivacaftor use[3]

Named variants	E56K	P67L	D110H
	R117C	E193K	R347H
	L206W	R352Q	A455E
	711+3A->G	E831X	S945L
	K1060T	A1067T	2789+5G->A
	3272-26A->G	3849+10kbC->T	
Variants with "varying clinical consequence" (VCC)	R74W	D110E	D579G
	S977F	F1052V	G1069R
	R1070Q	R1070W	F1074L
	D1152H	D1270N	

[3] https://www.england.nhs.uk/wp-content/uploads/2020/08/Urgent-policy-statement-CFTR-off-label-rarer-mutations.pdf

Table 3e: CFTR modulator use in people aged six years and older by genotype group[4]

	Genotype Group				
	Group 1	**Group 2**	**Group 3**	**Group 4**	**Group 5**
ETI*	261 (94.6%)	260 (82.8%)	22 (56.4%)	0	-**
Tezacaftor/ivacaftor	<5	<5	<5	<5	<5
Lumacaftor/ivacaftor	<5	0	<5	0	0
Ivacaftor	0	-**	8 (20.5%)	<5	0
Never used	**13 (4.7%)**	**43 (13.7%)**	**9 (23.1%)**	**<5**	**13 (65%)**
Total	**276 (100%)**	**314 (100%)**	**39 (100%)**	**7 (100%)**	**20 (100%)**

[4] Fewer than five patients were excluded because their last recorded CFTRm treatment was as part of a drug trial and specific drug was unknown

* ETI is Elexacaftor/tezacaftor/ivacaftor

** Redacted to adhere to statistical guidelines

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