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Physical activity, respiratory physiotherapy practices, and nutrition among people with primary ciliary dyskinesia in Switzerland – a cross-sectional survey

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Summary

AIMS OF THE STUDY: We know little about the level of physical activity, respiratory physiotherapy practices and nutritional status of people with primary ciliary dyskinesia (PCD), although these are important aspects of patients with chronic respiratory disease. We assessed physical activity, respiratory physiotherapy practices and nutritional status among people with primary ciliary dyskinesia in Switzerland, investigated how these vary by age and identified factors associated with regular physical activity.

METHODS: We sent a postal questionnaire survey to people with primary ciliary dyskinesia enrolled in the Swiss PCD registry (CH-PCD), based on the standardised FOLLOW-PCD patient questionnaire. We collected information about physical activity, physiotherapy, respiratory symptoms and nutritional status. We calculated the metabolic equivalent (MET) to better reflect the intensity of the reported physical activities. To assess nutritional status, we extracted information from CH-PCD and calculated participants' body mass index (BMI).

RESULTS: Of the 86 questionnaires we sent, 74 (86% response rate) were returned from 24 children and 50 adults. The median age at survey completion was 23 years (IQR [interquartile range] 15–51), and 51% were female. Among all 74 participants, 48 (65%) performed sports regularly. Children were vigorously active (median MET 9.1; IQR 7.9–9.6) and adults were moderately active (median MET 5.5; IQR 4.3—6.9). Fifty-nine participants (80%) reported performing some type of respiratory physiotherapy. However, only 30% of adults saw a professional physiotherapist, compared with 75% of children. Half of the participants had normal BMI; one child (4%) and two adults (4%) were underweight. People who were regularly physically active reported seeing a physiotherapist more often.

CONCLUSIONS: Our study is the first to provide patient-reported data about physical activity, respiratory physiotherapy and nutrition among people with primary ciliary dyskinesia. Our results highlight that professional respiratory physiotherapy, exercise recommendations and nutritional advice are often not implemented in the care of people with primary ciliary dyskinesia in Switzerland. Multidisciplinary care in specialised centres by teams including physiotherapists and nutrition consultants could improve the quality of life of people with primary ciliary dyskinesia.

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Introduction

Due to increased respiratory workload, chronic respiratory diseases affect exercise capacity and nutritional status. These diseases are often complicated by recurrent infections that contribute further to increased metabolic demand [1–4]. Promoting a good level of physical activity and monitoring nutritional status are crucial for maintaining respiratory health; thus, they are important parts of managing chronic lung diseases. In the chronic lung disease primary ciliary dyskinesia (PCD), genetic mutations cause structural and functional changes of motile cilia, which reduce mucociliary clearance [5]. Chronic respiratory symptoms and recurrent infections of the upper and lower airways are common in primary ciliary dyskinesia [6]. Therefore, among people with primary ciliary dyskinesia physical activity is recommended for improving airway clearance, in addition to specific respiratory physiotherapy practices [7–9]. We know from other chronic lung diseases, such as cystic fibrosis, that physical activity is associated with improved sputum expectoration. Particularly

beneficial are higher intensity activities followed by respiratory physiotherapy [10, 11].

Yet we know little about patients with primary ciliary dyskinesia and their levels of physical activity, respiratory physiotherapy practices and nutritional status. So far, the few studies involving physical exercise among people with primary ciliary dyskinesia focused on muscle strength measurements and aerobic cardiorespiratory performance [12–15]. No studies examined the level of physical activity of people with primary ciliary dyskinesia in their everyday lives. Regarding nutritional status, no studies assessed appetite and nutritional advice given to people with primary ciliary dyskinesia. A large study from the international primary ciliary dyskinesia (iPCD) cohort reported that children younger than 9 years had lower body mass index (BMI) z-scores than healthy peers [16]. A single centre study of 43 children in the United Kingdom studied nutrition using impedance spectroscopy [17]. Both studies suggested patients with primary ciliary dyskinesia should receive nutritional advice to improve growth and delay lung disease progression [17, 18]. In Switzerland, where there is no centralised care for people with primary ciliary dyskinesia, we have no information about supportive care. Our study described physical activity, respiratory physiotherapy practices and nutritional status among people with primary ciliary dyskinesia in Switzerland, investigated how these vary by age and identified factors associated with regular physical activity.

Methods

Study design and population

Our national cross-sectional questionnaire survey was nested in the Swiss PCD registry (CH-PCD). CH-PCD is a population-based patient registry (www.clinicaltrials.gov; identifier NCT03606200) enrolling all people with confirmed or clinical diagnoses of primary ciliary dyskinesia in Switzerland [19]. Patients with a clinical primary ciliary dyskinesia diagnosis have a strong clinical suspicion, such as situs anomalies, persistent cough, persistent rhinitis, chronic or recurrent upper or lower respiratory infections and history of neonatal respiratory symptoms as term infants, but have not completed the diagnostic algorithm and have negative or ambiguous results for the tests performed so far [19–21].

Our reporting conforms with the Strengthening the Reporting of Observational studies in Epidemiology (STROBE) statement [22]. Detailed information about the study design is published elsewhere [23]. CH-PCD received ethical approval from the Cantonal Ethics Committee of Bern in 2015 (KEK-BE: 060/2015). We obtained written informed consent from participants or parents of participants younger than 14 years.

In February 2020, we sent a postal questionnaire to all people with primary ciliary dyskinesia enrolled in the CH-PCD. After we mailed the questionnaire by post, we distributed the questionnaire at the primary ciliary dyskinesia outpatient clinic in Bern to an additional five people newly enrolled in CH-PCD (fig. 1). After 2–3 weeks, we sent a reminder by post to everyone who had not yet returned questionnaires.

Questionnaire

The questionnaire was based on the FOLLOW-PCD questionnaire (version 1.0), a PCD-specific standardised questionnaire, and part of a follow-up form developed by an international PCD expert group [24]. The FOLLOW-PCD questionnaire's main domains included chronic respiratory symptoms from the past 3 months and health-related behaviours, such as physical activity during the past 12 months [23]. Our study used all original questions from the FOLLOW-PCD questionnaire and included additional questions about respiratory physiotherapy and nutritional intake. We added a section on respiratory physiotherapy based on the relevant clinical module from the FOLLOW-PCD form [24]. We reviewed the physiotherapy-related questions with respiratory physiotherapists in Switzerland to ensure they would be understandable by participants and relevant to local techniques. Lastly, we added more questions about nutritional advice in Switzerland based on expert suggestions.

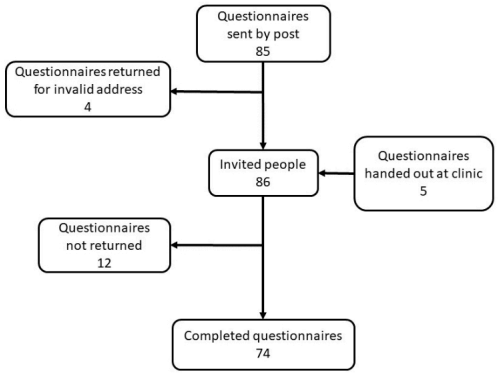
We developed new questions in German, then a native French speaker with knowledge about primary ciliary dyskinesia translated them into French. Questionnaires were age-specific for adults (ages ≥18 years), adolescents (ages 14–17 years), and parents of children (ages 0–13 years) with primary ciliary dyskinesia, available in German and French [23].

Physical activity

We asked if participants performed sports, what type of sports and how many hours per week. For school-age children, we asked if they played school or any additional sports. We classified physical activity into five categories: aerobic activity, such as endurance or cardio activity; muscle strengthening; bone strengthening; balance activity; and flexibility activity [25]. If multiple types of physical activity were listed, we reported them all.

Since types of physical activity differ in intensity of cardiorespiratory exertion, we used metabolic equivalents (METs) to better reflect the intensity of reported activities. The MET of the adult compendium is described as the ratio of work metabolic rate to the resting metabolic rate [26]. One MET is defined as the amount of oxygen consumed sitting at rest; it is equivalent to 3.5 ml oxygen/kg

Figure 1: Flowchart of people with primary ciliary dyskinesia living in Switzerland and their parents who were invited and participated in the survey.



body weight/min. We calculated the total MET per week based on participants' activities. Since there is no standardised reference, we added a MET combination of gymnastics (3.8 MET) and children's games (5.8 MET) to the total MET per week for children who always performed school sports. For children attending school sports sometimes or usually, we added one third or two thirds of the MET combination of gymnastics and children's games, respectively. We categorised physical activity in three MET intensity categories: light (<3.0 METs), moderate (3.0–5.9 METs), and vigorous (≥6.0 METs) [27].

Nutrition

We assessed participant nutritional status by collecting information about appetite, physician advice for increasing caloric intake during the past 12 months and calculating participants' BMIs. If increasing caloric intake was recommended, we asked about specific recommendations, such as increases of energy-dense food, larger portion size, or meal frequency. Furthermore, we assessed if recommendations had been successful in increasing energy and if body weight stabilised or increased. Other questions revolved around intake of oral nutritional supplements.

To calculate BMI, we used height and weight data from the CH-PCD measured in the hospital or private practices closest to the time when participants completed the survey. We calculated BMI by dividing weight in kilograms by height in meters squared (kg/m^2). We classified BMI for adults as underweight (<18.5 kg/m^2), normal (≥18.5 to <25), overweight (≥25.0 to <30) or obese (≥30.0 to <35) by World Health Organization (WHO) standards [28]. For children and adolescents aged 5–17 years, we calculated sex and age-specific BMI z-scores based on the 2007 WHO references [29]. We defined thinness (<—2 z-scores), normal (—2 to 1 z-scores), overweight (1 to 2 z-scores), and obesity (>2 z-scores).

Respiratory physiotherapy

We collected information on respiratory physiotherapy practices during the past three months, including if participants performed any physiotherapy; whether they had seen a professional physiotherapist; and which respiratory practices they used for upper and lower airway clearance, such as nose blowing, nasal rinsing, specific airway clearance techniques and use of breathing exercise aids. We asked about the use of inhalation for the upper and lower airways, particularly with isotonic (0.9% sodium chloride) or hypertonic (>0.9% sodium chloride) saline. Lastly, we asked how difficult airway clearance was for them during this period and if they felt any improvement of their respiratory symptoms after respiratory physiotherapy.

Respiratory symptoms

The questionnaire asked about several upper and lower respiratory symptoms and their frequency during the past 3 months. We defined symptoms as frequent if they were reported daily or often. Reported frequency of all symptoms ranged from daily, often, sometimes, and rarely to never [23].

Possible factors associated with regular physical activity

We categorised physical activity into regular (once a week or more) and irregular (less than once a week). We considered the following factors as possibly associated with regular physical activity: experiencing respiratory symptoms, such as chronic nasal symptoms, cough, shortness of breath and sputum production, that might influence physical activity performance; performing any physiotherapy; and seeing professional physiotherapists.

Statistical analysis

We described characteristics of the population, physical activity, respiratory physiotherapy practices, and nutritional status in the total population and separately among children (<18 years) and adults with primary ciliary dyskinesia. We also described possible differences in physical activity, and respiratory physiotherapy practices by sex and nutritional status, and in nutritional status by sex. For continuous variables, we used median and interquartile range (IQR); for categorical variables, we used numbers and proportions. We used Wilcoxon rank-sum and Pearson's Chi square to study differences between children and adults and factors possibly associated with regular physical activity. We calculated Wilson 95% confidence intervals (CIs) for proportions. Missing data were reported as such and were excluded from analyses. We performed all analyses with Stata version 16 (StataCorp LLC, Texa, USA).

Results

We invited 86 people; 74 (86%) participants returned the questionnaire (fig. 1). The median age at survey completion was 23 years (IQR 15–51) and 38 (51%) were female. Among the participants, 24 were children or adolescents (referred as children from now onwards) of whom 17 attended school. Of the 50 adults, 42% were employed full-time (80–100%), whereas 18% were retired or on disability pension (table 1). In the past 12 months, most children and adults (64%) missed less than 1 week of school or work due to PCD-related symptoms; few adults (10%) missed more than 2 weeks. About half of participants (46%) lived in areas with little or no traffic. Age and sex did not differ between questionnaire respondents and non-respondents (supplementary table S1 in the appendix). Missing responses to individual survey questions were less than 5%.

Physical activity

Among all 74 participants, 48 (65%) performed sports regularly (table 2). All 17 schoolchildren attended school sports; 14 of them (82%) participated always. The seven children not yet in school were all physically active. Categories of sports did not differ by age. The most common type of physical activity were aerobic activities, for example, children played soccer or hockey (25%), whereas adults preferred jogging or walking (56%). We found children were vigorously active (median MET 9.1; IQR 7.9–9.6) and adults were moderately active (median MET 5.5; IQR 4.3–6.9). Adults spent more time (median 7 hours/week; IQR 3.5–12) performing light sports when compared with children who spent most of their time (me-

dian 4 hours/week; IQR 3–7) performing vigorous sports. We present the most reported physical activities in supplementary table S1 (appendix). We found no differences in physical activity by sex. Underweight and normal weight participants reported more frequently aerobic activities compared with overweight or obese individuals (supplementary table S3).

Respiratory physiotherapy

Overall, most participants (59; 80%) reported they performed some type of respiratory physiotherapy; of these, 18 children (75%) and 28 adults (56%), this was daily. Only 15 adults (30%) saw a professional physiotherapist in comparison with 18 children (75%) (p = 0.008). Regarding upper airway physiotherapy, 69% of the participants blew their noses, most daily (table 3). Over half of participants (61%) performed nasal rinsing, 8 children (33%) and 13 adults (26%) daily. Only 21 participants (28%) performed

Table 1:
Characteristics of people with primary ciliary dyskinesia in Switzerland participating in the survey, overall and by age group (n = 74).

	Total, n (%)	Children <18 y, n (%)	Adults ≥18 y, n (%)	p -value
Number of participants	74 (100)	24 (100)	50 (100)	
Age, median (IQR)	23 (15–51)	11 (6–15)	32 (23–57)	
Sex, female	38 (51)	7 (29)	31 (62)	0.008
Occupation				
Student/preschool	29 (39)	24 (100)	5 (10)	
81–100% employment	21 (28)	-	21 (42)	
61–80% employment	2 (3)	-	2 (4)	
41–60% employment	5 (7)	-	5 (10)	
≤40% employment	8 (11)	-	8 (16)	
Retired/disability pension	9 (12)	-	9 (18)	
Missing (pre)school or work days due to primary ciliary dyskinesia in the past 12 months				0.345
0–6 days	46 (62)	15 (63)	31 (63)	
1–2 weeks	8 (11)	4 (17)	4 (8)	
>2 weeks	5 (7)	0 (0)	5 (10)	
Not in school yet/ retired	12 (16)	3 (12)	9 (18)	
Not reported	3 (4)	2 (8)	1 (1)	
Area of residence				0.949
Street with dense traffic	17 (23)	6 (25)	11 (22)	
Street with moderate traffic	23 (31)	7 (29)	16 (32)	
Street with little or no traffic	34 (46)	11 (46)	23 (46)	

y: years. All characteristics are presented as n and column % with the exception of age presented as median and interquartile range (IQR).

Table 2:
Physical activity of people with primary ciliary dyskinesia in Switzerland, overall and by age group (n = 74).

	Total, n (%)	Children <18 y, n (%)	Adults ≥18 y, n (%)	p -value
Number of participants	74 (100)	24 (100)	50 (100)	
Regular sports [a]	48 (65)	13 (54)	35 (70)	0.182
School sports attendance		17 (71)	NA	
Always		14 (59)	NA	
Usually		2 (8)	NA	
Sometimes		1 (4)	NA	
Not yet in school		7 (29)	NA	
Sports categories [b]				
Aerobic activity	37 (50)	9 (38)	28 (56)	0.136
Muscle strengthening	21 (28)	5 (21)	16 (32)	0.319
Bone strengthening	4 (5)	2 (8)	2 (4)	0.440
Balance	5 (7)	2 (8)	3 (6)	0.708
Flexibility	3 (4)	1 (4)	2 (4)	0.973
Total MET per week [c]	6.8 (4.8–8.9)	9.1 (7.9–9.6)	5.5 (4.3–6.9)	<0.001
Sports hours/week by MET categories [d]				
Light (<3.0 METs)	7 (3.5–12)	0 (0)	7 (3.5–12)	
Moderate (3.0–5.9 METs)	3 (2–3)	2.5 (2–6)	3 (2–3)	0.336
Vigorous (≥6.0 METs)	4 (3–6)	4 (3–7)	5 (3–5.5)	0.198

y: years; NA: not applicable.

Characteristics are presented as n and column %, metabolic equivalent (MET = 1 kcal/kg/hour) described in median and interquartile range (IQR).

[a] For children, this refers to additional sports besides school sports.

[b] Sports categories were not mutually exclusive

[c] Average sports intensity per week in MET (median, IQR) including school sports for schoolchildren

[d] Reported hours per week participants spent performing sports by MET categories (median and IQR)

inhalations for the upper airways, eight children (33%) and five adults (10%) daily. Among all participants, 11 (17%) used saline for upper airway inhalation, 9 of them hypertonic (>0.9%) saline. We found no differences in upper airway physiotherapy practices between children and adults apart from nose blowing and nasal rinsing, which were more common in children.

Regarding lower airway physiotherapy, 49 participants (67%) applied some airway clearance technique, such as autogenic drainage, 21 participants (28%) daily (table 4). The most common aid used for airway clearance was a flutter (19%) or positive expiratory pressure combined with flutter (7%). Overall, 65% performed inhalations for lower airways, 63% of children and 42% of adults daily, mainly with hypertonic saline. Nineteen participants (26%) reported difficulty clearing their lower airways; 13 children (54%) and 18 adults (36%) reported their lower respiratory symptoms improved after physiotherapy. Lower airway physiotherapy practices did not differ between children and adults.

We found no differences in upper and lower respiratory physiotherapy practices between males and females, as well as between underweight/normal weight and overweight/obese participants (supplement tables S4 and S5).

Nutrition

Weight and height information from clinical visits near survey completion (within 12 months, usually a few months apart) was available for 53 (72%) participants: 16 children and 37 adults. Median BMI was 22.3 kg/m^2 (IQR 20.4–25.0) for adults and median BMI z-score was 1.0 (IQR 1.0–1.0) for children (table 5). Half of the participants had a normal BMI; only one child (4%) and two adults (4%) were underweight/thin. There was no difference between children and adults or between males and females in BMI categories (table 5 and supplementary table S6). Overall, seven people reported decreased appetite (9%). Physicians recommended increased caloric intake to three children (13%) and three adults (6%); three people (4%) ate larger meal portions, two increased their meal frequency, and one child received energy-dense food. Five adult participants reported ingesting hypercaloric drinks, four only during periods of increased physical activity or colds. Increased caloric intake and hypercaloric drinks helped four patients stabilise or gain weight. One child and two adults reported these measures increased their energy.

Factors associated with regular physical activity

We found no difference in frequency of reported nasal symptoms, cough, shortness of breath and sputum production between participants who were regularly or less physically active (fig. 2). Respiratory physiotherapy was not associated with regular physical activity. However, regularly physically active people were less likely to see professional physiotherapists (p = 0.008).

Discussion

Our study is the first to provide patient-reported data on physical activity, respiratory physiotherapy and nutrition for people with primary ciliary dyskinesia. Our results suggest people with primary ciliary dyskinesia in Switzerland

Table 3:
Upper airway physiotherapy practices of people with primary ciliary dyskinesia in Switzerland, overall and by age group (n = 74).

	Total, n (%)	Children <18 y,n (%)	Adults ≥18 y, n (%)	p -value
Number of participants	74 (100)	24 (100)	50 (100)	
Nose blowing				0.021
Yes, daily	37 (50)	10 (42)	27 (54)	
Yes, often	8 (11)	5 (21)	3 (6)	
Yes, sometimes	3 (4)	2 (8)	1 (2)	
Yes, rarely	0 (0)	0 (0)	0 (0)	
Yes, only during colds	3 (4)	3 (12)	0 (0)	
No / not reported	23 (31)	4 (17)	19 (38)	
Nasal rinsing				
Yes, daily	21 (28)	8 (33)	13 (26)	0.021
Yes, often	11 (15)	4 (17)	7 (14)	
Yes, sometimes	6 (8)	5 (21)	1 (2)	
Yes, rarely	3 (4)	0 (0)	3 (6)	
Yes, only during colds	4 (6)	4 (17)	0 (0)	
No / not reported	29 (39)	3 (12)	26 (52)	
Inhalations for the upper airways				0.146
Yes, daily	13 (18)	8 (33)	5 (10)	
Yes, often	4 (5)	1 (4)	3 (6)	
Yes, sometimes	3 (4)	0 (0)	3 (6)	
Yes, rarely	1 (1)	0 (0)	1 (2)	
Yes, only during colds	0 (0)	0 (0)	0 (0)	
No / not reported	53 (72)	15 (63)	38 (76)	
Upper airways inhalation with saline	11 (15)	7 (29)	4 (8)	0.237
Isotonic (0.9% sodium chloride)	2 (3)	2 (8)	0 (0)	
Hypertonic (>0.9% sodium chloride)	9 (12)	5 (21)	4 (8)	
Upper airways inhalation with other medication	5 (7)	3 (13)	2 (4)	

y: years. All characteristics are presented as nN and column %.

are health-oriented, physically active and normal weight; they also perform respiratory physiotherapy.

A strength of our study includes our questionnaire. We based our survey on the PCD-specific standardised FOLLOW-PCD questionnaire [24], allowing for future comparisons with larger multicentre studies. We also developed

Table 4:
Lower airway physiotherapy practices of people with primary ciliary dyskinesia in Switzerland, overall and by age group (n = 74).

	Total, n (%)	Children <18 y, n (%)	Adults ≥18 y, n (%)	p -value
Number of participants	74 (100)	24 (100)	50 (100)	
Airway clearance techniques [a]				0.735
Yes, daily	21 (28)	8 (33)	13 (26)	
Yes, often	8 (11)	2 (8)	6 (12)	
Yes, sometimes	13 (18)	4 (17)	9 (18)	
Yes, rarely	5 (7)	3 (13)	2 (4)	
Yes, only during colds	2 (3)	1 (4)	1 (2)	
No / not reported	25 (33)	6 (25)	19 (38)	
Use of aid for airway clearance				0.565
Flutter	14 (19)	5 (21)	9 (18)	
PEP	3 (4)	2 (8)	1 (2)	
Flutter and PEP	5 (7)	2 (8)	3 (6)	
Blow in straw/nozzle	3 (4)	3 (13)	0 (0)	
Vibrating device	2 (3)	2 (8)	0 (0)	
No / not reported	47 (63)	10 (42)	37 (74)	
Inhalation for the lower airways				
Daily	36 (49)	15 (63)	21 (42)	0.233
Often	4 (5)	1 (4)	3 (6)	
Sometimes	6 (8)	0 (0)	6 (12)	
Only during colds	2 (3)	1 (4)	1 (2)	
No / not reported	26 (35)	7 (29)	19 (38)	
Lower airways inhalation with saline				0.515
Yes	35 (47)	14 (58)	21 (42)	
Isotonic (0.9% sodium chloride)	4 (5)	1 (4)	3 (6)	
Hypertonic (>0.9% sodium chloride)	31 (42)	13 (54)	18 (36)	
Lower airways inhalation with other medication	28 (38)	9 (38)	19 (38)	
Difficulty of clearing lower airways				0.581
Easy	42 (57)	12 (50)	30 (60)	
Difficult	19 (26)	8 (33)	11 (22)	
I don't know / not reported	13 (17)	4 (17)	9 (18)	

y: years; PEP: positive expiratory pressure. [a] Airway clearance techniques e.g., autogenic drainage. All characteristics are presented as n and column %.

Table 5:
Nutritional information of people with primary ciliary dyskinesia in Switzerland, overall and by age group (n = 74).

	Total, n (%)	Children <18 y, n (%)	Adults ≥18 y, n (%)	p -value
Number of participants	74 (100)	24 (100)	50 (100)	
BMI, median (IQR)			22.3 (20.4–25.0)	
BMi z-score, median (IQR)		1.0 (1.0–1.0)		
BMI categories				0.894
Thinness/underweight	3 (4)	1 (4)	2 (4)	
Normal weight	37 (50)	12 (50)	25 (50)	
Overweight	10 (14)	2 (8)	8 (16)	
Obesity	3 (4)	1 (4)	2 (4)	
Missing	21 (28)	8 (34)	13 (26)	
Physician recommendation to increase caloric intake by				
Energy-dense food	1 (1)	1 (4)	0 (0)	
Larger meal portions	3 (4)	1 (4)	2 (4)	
Increased meal frequency	2 (3)	1 (4)	1 (2)	
Use of hypercaloric drinks in the past 12 months				
Yes	5 (7)	0 (0)	5 (10)	
Weight gained/stabilised due to increased caloric intake				
Yes	3 (4)	0 (4)	3 (6)	

y: years; BMI: body mass index. All characteristics are presented as n and column % with the exception of BMI and BMI z-score presented as median and interquartile range (IQR).

BMI was available for 16 children and 37 adults. BMI categories were based on the World Health Organization (WHO) 2007 standards, using BMI in kg/m^2 for adults (underweight <18.5, normal weight ≥18.5 to <25, overweight ≥25.0 to <30, obesity ≥30.0 to <35), and calculating BMI z-scores based on the WHO references for children (thinness <—2 z-scores, normal weight —2 to 1 z-scores, overweight 1 to 2 z-scores, obesity >2 z-scores).

additional questions based on input from experts, such as physiotherapists. Further, we nested our study in the national PCD registry, which provided objectively measured BMI data. Other strengths include a remarkable response rate of 86% and a low proportion of missing survey data.

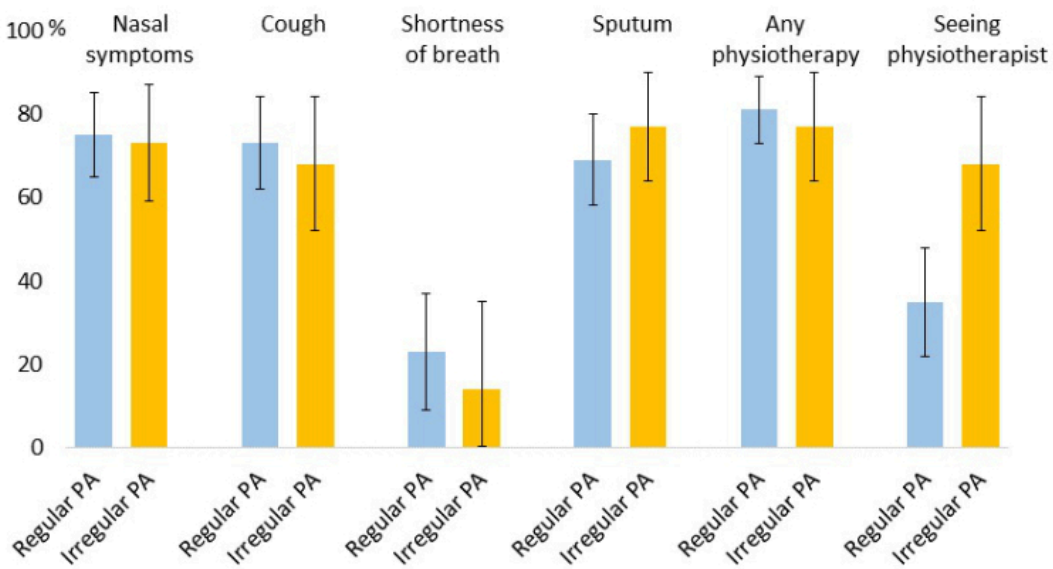
Our study has several limitations. Although we invited all people enrolled in CH-PCD, selection bias is possible because primary ciliary dyskinesia in Switzerland is underdiagnosed [19]. The COVID-19 pandemic could have affected physical activity. However, since more than 75% of the questionnaires were returned by March 2020 before lockdown in Switzerland started, we expect any effect to be small. Furthermore, most participants reported outdoor activities, which were not affected by COVID-19 restrictions. As many questions referred to the last 12 months, there is a risk of recall bias. We expect answers reflected mostly health-related behaviours closer to date of survey completion. Since FOLLOW-PCD does not include validated physical activity questions, we calculated MET for reported activities to describe exercise intensity. So far, MET is standardised for adults only; however, to describe and compare the intensity of physical activity for the whole study population, we calculated it for all participants.

Previous studies among people with primary ciliary dyskinesia showed reduced physical fitness in hospital settings. For example, a recent single-centre study of 27 Turkish children with primary ciliary dyskinesia reported reduced muscle strength and endurance [15]. Another single-centre study with 22 Danish children with primary ciliary dyskinesia found reduced cardiopulmonary fitness [14]. Children in our study reported a median moderate to vigorous activity of 6.5 hours/week, which is lower than the average 7.5 hours/week among 10–14-year-old children and 6.5 hours/week among 15–19-year-old adolescents who perform additional sports besides attending school sports in Switzerland [30]. However, it is comparable to the level of recreational sports (median 3.0 hours/week, IQR 1–5) of children who survived cancer in Switzerland [31]. In a multicentre service evaluation of over 300 children with primary ciliary dyskinesia in England, all children were trained and advised to perform regular airway clearance physiotherapy [32]. In our population, most people reported doing respiratory physiotherapy but only 18 children (75%) and 28 adults (56%) on a daily basis. Despite having chronic respiratory symptoms, most people with primary ciliary dyskinesia in Switzerland have a normal BMI, which is lower than the national average of three people with overweight, and one person with obesity of every ten adults in Switzerland [33]. Our study included very few children younger than age 9 so it was not possible to see whether this group had a lower BMI z-score as in the large iPCD cohort study [18].

Most participants were physically active, despite reporting frequent respiratory symptoms. This might indicate that people with primary ciliary dyskinesia are accustomed to their chronic symptoms and exercise despite them. Or they use physical activity as a method for airway clearance to relieve symptoms, explaining why fewer people who exercised regularly saw a professional physiotherapist. People might replace respiratory physiotherapy with physical activity. Exercise is a recommended method of airway clearance for patients with cystic fibrosis. However, it remains unclear whether it can replace physiotherapy or to what extent for patients with cystic fibrosis or primary ciliary dyskinesia [8, 34]. Although physical activity aids in airway clearance of larger airways, peripheral airway dysfunction persists with mucus obstruction [35].

Our study is the first to provide patient-reported data on health-related behaviours of people diagnosed with primary ciliary dyskinesia. Our results highlight the importance of implementing exercise recommendations, professional respiratory physiotherapy, and nutritional advice that are

Figure 2: Frequency and 95% confidence interval of reported respiratory symptoms and use of respiratory physiotherapy in the past three months in people with primary ciliary dyskinesia (PCD) in Switzerland who perform regular (at least once a week) physical activity (PA) compared with people who are less physically active.



currently missing in the routine care of people with primary ciliary dyskinesia in Switzerland. Multidisciplinary care in specialised centres by teams including physiotherapists and nutrition consultants could improve the daily lives of people with primary ciliary dyskinesia in Switzerland.

Availability of data and materials

The datasets used and analysed during the current study are available from the CH-PCD study team (pcd@ispm.ch) upon reasonable request.

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Author contributions: MG and CEK developed the concept and designed the survey. YTL, MG, and LH organised the survey, then cleaned and standardised the data. YTL performed the statistical analyses supervised by MG and EP. YTL, MG, and EP drafted the manuscript. All authors commented and revised the manuscript. YTL and MG take final responsibility for the content.

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Potential conflicts of interest

All authors have completed and submitted the International Committee of Medical Journal Editors form for disclosure of potential conflicts of interest. CC received advisory fees from Roche, Novartis, Boehringer, GSK, Astra Zeneca, Sanofi, Vifor, OM Pharma, CSL Behring, Grifols, Daiichi Sankyo and Mundipharma within the last 36 months. PL received grants, honorary, for participation in data safety monitoring board or advisory board from Vertex, Vifor, OM Pharma, Polyphor, Santhera, Sanofi Aventis within the last 36 months. No other potential conflict of interest was disclosed.

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Appendix: supplementary tables

Table S1:
Characteristics of people with primary ciliary dyskinesia in Switzerland, respondents and non-respondents to the survey.

	Respondents, n (%)	Non-respondents, n(%)	p -value
Number	74 (100)	12 (100)	
Age, median (IQR)	23 (15–51)	37 (20–46)	0.449
Sex, female	38 (51)	8 (67)	0.324

y: years. Characteristics are presented as n and column % with the exception of age presented as median and IQR: interquartile range.

Table S2:
Top 10 most common types of physical activity of people with primary ciliary dyskinesia in Switzerland, overall and by age group. Some people reported multiple activities.

	Total	Children <18 y	Adults ≥18 y
Number of participants	74	24	50
Training in gym	17	0	17
Walking	10	0	10
Cycling	9	1	8
Jogging	4	0	4
Ice hockey	4	1	3
Hiking	2	0	2
Horseback riding	4	2	2
Skiing/snowboarding	3	1	2
Badminton	1	0	1
Climbing	1	0	1

Table S3:
Physical activity of people with primary ciliary dyskinesia in Switzerland, overall, by sex (n = 74), and BMI categories (n = 53).

	Total, n (%)	Female, n (%)	Male, n(%)	p -value	Normal weight / underweight, n (%) [a]	Overweight/obese,n (%) [a]	p -value
Number of participants	74 (100)	38 (100)	36 (100)		40 (100)	13 (100)	
Regular sports [b]	48 (65)	24 (63)	24 (67)	0.752	25 (63)	11 (85)	0.100
School sports attendance [c]				0.660			0.110
Always	14 (59)	4 (11)	10 (28)		12 (30)	2 (15)	
Usually	2 (8)	1 (3)	1 (3)		0 (0)	1 (8)	
Sometimes	1 (4)	0 (0)	1 (3)		1 (3)	0 (0)	
Not yet in school	7 (29)	2 (5)	5 (14)		4 (10)	0 (0)	
Sports categories [d]							
Aerobic activity	37 (50)	19 (50)	18 (50)	1.000	18 (45)	10 (77)	0.045
Muscle strengthening	21 (28)	8 (21)	13 (36)	0.151	12 (30)	4 (31)	0.958
Bone strengthening	4 (5)	3 (8)	1 (3)	0.331	2 (5)	1 (8)	0.715
Balance	5 (7)	4 (11)	1 (3)	0.184	2 (5)	2 (15)	0.218
Flexibility	3 (4)	1 (3)	2 (6)	0.524	2 (0)	0 (0)	0.411
Total MET per week [e]	6.8(4.8–8.9)	6(4.3–8.5)	7(4.8–9)	0.464	7.9(4.8–9.5)	6.6(4.8–9)	0.449
Sports hours/week by MET categories [f]							
Light (<3.0 METs)	7(3.5–12)	7(3.5–14)	3.5(2.5–10)	0.367	7(4.8–13.5)	8.8(3.5–14)	0.199
Moderate (3.0–5.9 METs)	3 (2–3)	2.8 (2–3)	3 (2–4)	0.336	3 (2.5–5)	2.5 (2–3)	0.611
Vigorous (≥6.0 METs)	4 (3–6)	3 (3–5)	5 (3–7.5)	0.198	4.5 (3–7)	5 (4–5.5)	0.230

Characteristics are presented as n and column %, metabolic equivalent (MET = 1 kcal/kg/hour) described in median and IQR: interquartile range.

[a] BMI was available for 53 participants. BMI categories were based on the World Health Organization (WHO) 2007 standards, using BMI in kg/m2 for adults (underweight <18.5, normal weight ≥18.5 to <25, overweight ≥25.0 to <30, obesity ≥30.0 to <35), and calculating BMI z-scores based on the WHO references for children (thinness <-2 z-scores, normal weight -2–1 z-scores, overweight 1–2 z-scores, obesity >2 z-scores).

[b] For children, this refers to additional sports besides school sports. .

[c] Sports categories were not mutually exclusive

[d] Average sports intensity per week in MET (median, IQR) including school sports for schoolchildren

[e] Reported hours per week participants spent performing sports by MET categories (median and IQR).

Table S4:
Upper airway physiotherapy practices of people with primary ciliary dyskinesia in Switzerland, overall, by sex (n = 74), and BMI categories (n = 53).

	Total, n (%)	**Female, n (%)**	**Male , n (%)**	**p -value**	**Normal weight / underweight, (%)** [a]	**Overweight/obese, n (%)** [a]	**p -value**
Number of participants	74 (100)	38 (100)	36 (100)		40 (100)	13 (100)	
Nose blowing				0.214			0.336
Yes, daily	37 (50)	18 (48)	19 (53)		23 (57)	7 (54)	
Yes, often	8 (11)	2 (5)	6 (17)		3 (7)	3 (23)	
Yes, sometimes	3 (4)	0 (0)	3 (8)		1 (3)	0 (0)	
Yes, rarely	0 (0)	0 (0)	0 (0)		0 (0)	0 (0)	
Yes, only during colds	3 (4)	2 (5)	1 (3)		1 (3)	0 (0)	
No / not reported	23 (31)	16 (42)	7 (19)		12 (30)	3 (23)	
Nasal rinsing				0.719			0.664
Yes, daily	21 (28)	8 (21)	13 (36)		12 (30)	4 (31)	
Yes, often	11 (15)	5 (13)	6 (17)		6 (15)	2 (15)	
Yes, sometimes	6 (8)	4 (11)	2 (6)		5 (12)	0 (0)	
Yes, rarely	3 (4)	2 (5)	1 (3)		1 (3)	1 (8)	
Yes, only during colds	4 (6)	2 (5)	2 (6)		2 (5)	1 (8)	
No / not reported	29 (39)	17 (45)	12 (32)		14 (35)	5 (38)	
Inhalations for the upper airways				0.374			0.146
Yes, daily	13 (18)	3 (8)	10 (28)		6 (15)	0 (0)	
Yes, often	4 (5)	2 (5)	2 (6)		3 (7)	0 (0)	
Yes, sometimes	3 (4)	2 (5)	1 (3)		2 (5)	1 (8)	
Yes, rarely	1 (1)	0 (0)	1 (3)		1 (3)	0 (0)	
Yes, only during colds	0 (0)	0 (0)	0 (0)		0 (0)	0 (0)	
No / not reported	53 (72)	31 (82)	22 (60)		28 (70)	12 (92)	
Upper airways inhalation with saline	11 (15)	2 (5)	9 (25)	0.461			-
Isotonic (0.9% sodium chloride)	2 (3)	0 (0)	2 (6)		1 (3)	0 (0)	
Hypertonic (>0.9% sodium chloride)	9 (12)	2 (5)	7 (19)		6 (15)	0 (0)	
Upper airways inhalation with other medication	5 (7)	3 (8)	2 (6)		4 (10)	1 (8)	

[a] BMI was available for 53 participants. BMI categories were based on the World Health Organization (WHO) 2007 standards, using BMI in kg/m^2 for adults (underweight <18.5, normal weight ≥18.5 to <25, overweight ≥25.0 to <30, obesity ≥30.0 to <35), and calculating BMI z-scores based on the WHO references for children (thinness <–2 z-scores, normal weight –2–1 z-scores, overweight 1–2 z-scores, obesity >2 z-scores).

Table S5:
Lower airway physiotherapy practices of people with primary ciliary dyskinesia in Switzerland, overall, by sex (n = 74), and BMI categories (n = 53).

	Total, n (%)	Female. n (%)	Male, n (%)	p -value	Normal weight / underweight, (%) [a]	Overweight/obese, n (%) [a]	p -value
Number of participants	74 (100)	38 (100)	36 (100)		40 (100)	13 (100)	
Airway clearance techniques				0.130			0.119
Yes, daily	21 (28)	8 (21)	13 (36)		13 (32)	2 (16)	
Yes, often	8 (11)	1 (3)	7 (19)		2 (5)	3 (23)	
Yes, sometimes	13 (18)	7 (18)	6 (17)		7 (17)	5 (38)	
Yes, rarely	5 (7)	3 (8)	2 (6)		4 (10)	0 (0)	
Yes, only during colds	2 (3)	2 (5)	0 (0)		1 (3)	0 (0)	
No / not reported	25 (33)	17 (45)	8 (22)		13 (33)	3 (23)	
Use of aid for airway clearance				0.423			0.202
Flutter	14 (19)	7 (21)	7 (18)		5 (12)	2 (15)	
PEP	3 (4)	2 (8)	1 (2)		4 (10)	0 (0)	
Flutter and PEP	5 (7)	4 (8)	1 (6)		4 (10)	1 (8)	
Blow in straw/nozzle	3 (4)	2 (5)	1 (0)		0 (0)	1 (8)	
Vibrating device	2 (3)	2 (8)	0 (0)		2 (5)	0 (0)	
No / not reported	47 (63)	21 (55)	26 (72)		25 (63)	9 (69)	
Inhalation for the lower airways							
Daily	36 (49)	17 (45)	19 (53)	0.550	21 (53)	8 (62)	0.643
Often	4 (5)	2 (5)	2 (6)		3 (7)	0 (0)	
Sometimes	6 (8)	3 (8)	3 (8)		5 (12)	1 (8)	
Only during colds	2 (3)	2 (5)	0 (0)		1 (3)	0 (0)	
No / not reported	26 (35)	14 (37)	12 (33)		10 (25)	4 (30)	
Lower airways inhalation with saline				0.316			0.393
Yes	35 (47)	17 (58)	18 (42)		23 (58)	5 (38)	
Isotonic (0.9% sodium chloride)	4 (5)	1 (4)	3 (6)		3 (8)	0 (0)	
Hypertonic (>0.9% sodium chloride)	31 (42)	16 (54)	15 (36)		20 (50)	5 (38)	
Lower airways inhalation with other medication	28 (38)	15 (39)	13 (36)		15 (38)	8 (62)	
Difficulty of clearing lower airways				0.188			0.181
Easy	42 (57)	20 (53)	22 (61)		21 (53)	9 (70)	
Difficult	19 (26)	8 (21)	11 (31)		14 (35)	2 (15)	
I don't know / not reported	13 (17)	10 (26)	3 (8)		5 (12)	2 (15)	

[a] BMI was available for 53 participants. BMI categories were based on the World Health Organization (WHO) 2007 standards, using BMI in kg/m^2 for adults (underweight <18.5, normal weight ≥18.5 to <25, overweight ≥25.0 to <30, obesity ≥30.0 to <35), and calculating BMI z-scores based on the WHO references for children (thinness <–2 z-scores, normal weight –2–1 z-scores, overweight 1–2 z-scores, obesity >2 z-scores).

Table S6:
Nutritional information of people with primary ciliary dyskinesia in Switzerland, overall, and by sex (n =74).

	Total, n (%)	Female, n (%)	Male N (%)	p -value
Number of participants	74 (100)	38 (100)	36 (100)	
BMI, median (IQR)		22.1 (19.7–24.5)	21.9 (19.2–24.0)	0.389
BMI z-score, median (IQR)		1.0 (1.0–1.0)	1.0 (1.0–1.0)	
BMI categories				0.863
Thinness/underweight	3 (4)	1 (3)	2 (6)	
Normal weight	37 (50)	20 (53)	17 (47)	
Overweight	10 (14)	5 (13)	5 (14)	
Obesity	3 (4)	2 (5)	1 (3)	
Missing	21 (28)	9 (24)	6 (17)	
Physician recommendation to increase caloric intake by				
Energy-dense food	1 (1)	0 (0)	1 (3)	
Larger meal portions	3 (4)	1 (3)	2 (6)	
Increased meal frequency	2 (3)	1 (3)	1 (3)	
Use of hypercaloric drinks in the past 12 months				
Yes	5 (7)	1 (3)	4 (11)	
Weight gained/stabilised due to increased caloric intake				
Yes	3 (5)	1 (4)	2 (6)	

y: years. BMI: body mass index. All characteristics are presented as n and column % with the exception of BMI and BMI z-score presented as median and IQR: interquartile range.

BMI was available for 28 females and 25 males. BMI categories were based on the World Health Organization (WHO) 2007 standards, using BMI in kg/m^2 for adults (underweight <18.5, normal weight ≥18.5 to <25, overweight ≥25.0 to <30, obesity ≥30.0 to <35), and calculating BMI z-scores based on the WHO references for children (thinness <–2 z-scores, normal weight –2–1 z-scores, overweight 1–2 z-scores, obesity >2 z-scores).