Online only supplementary material

METHODS

Ethics

In most participating countries, research laws do not require obtaining patients' informed consent for retrospectively collected observational data that are anonymised. However, even in these countries, PIs received ethics approval and informed consent for collecting patient data for local research use or national registries. In countries where informed consent is required even for anonymised observational data, PIs are responsible for obtaining ethics approval and informed consent in their country for the contribution of their anonymised data to the iPCD Cohort for research purposes. All centres are required to get informed consent for contribution of prospective data. In Switzerland, we received permission from the Bern Cantonal Ethics Committee for a national PCD registry and contributing pseudonymised data to international studies.

PCD diagnosis

PCD diagnostics have evolved quickly [1]. Initially, diagnosis was based on the Kartagener triad [2], and on transmission electron microscopy findings (EM). Then light microscopy and, later, high frequency video microscopy (VM) were introduced into the diagnostic algorithm. Current recommendations include combining EM, VM, nasal nitric oxide (nNO), and genetic testing [3], but availability of tests differs between countries [1], and not all PCD patients have been diagnosed according to current standards. Patients diagnosed years ago and patients who live in countries with limited resources are least likely to have been diagnosed according to these recommendations. Because the iPCD Cohort includes patients diagnosed since 1964, we divided patients into three diagnostic subgroups based on the results of the tests available. The first subgroup included patients with definite PCD, which was defined, based on recent guidelines of the ERS PCD Diagnostics Task Force [3], by hallmark EM findings, and/or identified biallelic PCD genetic mutation. The second subgroup, probable PCD, included patients with abnormal VM findings and/or low nNO (we used a cut-off of 77nl/min [4]). The third subgroup includes patients with clinical PCD diagnosis; these were patients for whom the PCD diagnostic algorithm had not yet been completed, or whose test results were negative or ambiguous and other possible common diseases such as cystic fibrosis had been excluded. Patients in this third diagnostic group were followed up and treated as PCD patients at the collaborating centres based on a combination of several features: situs anomalies, persistent cough, persistent rhinitis, chronic or recurrent upper or lower respiratory infections, and history of neonatal respiratory symptoms in term infants [3].

Height and BMI

We checked data quality to identify outliers and implausible values, and contacted data contributors to resolve any issues that arose. We calculated age- and sex-adjusted height and BMI z-scores separately based on international reference values from the World Health Organisation (WHO) [5], and national reference values [6-20]. We identified national growth references by contacting collaborating centres and searching the literature. For each centre, we chose one of the following methods to calculate z-scores based on national references: an LMS approach using tables that contained L, M, and S parameters needed to generate exact z-scores [21]; direct calculation via online national z-score or percentile growth calculators; or interpolating exact z-scores from plotted percentile boundaries on growth curves.

Determinants of height and BMI

We categorised patients into six age groups (0-9, 10-19, 20-29, 30-39, 40-49, and ≥50 years old). In an analysis that included only paediatric patients, we created 5-year age groups. We received date of diagnosis from the centres or derived it from the dates of diagnostic test results. In patients without positive test results, we defined date of clinical diagnosis as the date of the first clinical follow up. We categorised organ laterality in three groups (situs solitus totalis, situs inversus, and heterotaxia). We excluded one centre where only pathological situs was recorded from organ laterality models; few data were missing from other centres. For lung function, we used the Global Lung Function Initiative (GLI) reference values to calculate age, sex, ethnicity, and height-adjusted z-scores of forced expiratory volume in one second (FEV1) and forced vital capacity (FVC) [22]. Lung function data were not provided by all countries, and some patients were too young to perform spirometry.

Table S1. National references used for calculation of height and BMI z-scores

| Country | Growth reference source Year of p | ublication |
|-----------------------|---|------------|
| Australia | Centre for Disease Control and Prevention [13] | 2000 |
| Belgium | Flemish growth study (Roelants et al.) [15] | 2009 |
| Cyprus | Growth curves for Greek children 0-5 years (Papadimitriou et al.) [14 | 1] 2000 |
| | Growth curves for Cypriot children 6-17 years (Savva et al.) [17] | 2001 |
| Denmark | Danish growth references (Tinggaard et al.) [20] | 2014 |
| France | French references for Height (Sempé et al.) [19] | 1979 |
| | French references for BMI (Rolland-Cachera et al.) [16] | 1991 |
| Germany | KiGGS study [11] | 2006 |
| Israel | Centre for Disease Control and Prevention [13] | 2000 |
| Italy | Centre for Disease Control and Prevention [13] | 2000 |
| Netherlands | Fifth Dutch Growth Study [18] | 2009 |
| Norway | Growth charts for Norwegian children (Júlíusson et al.) [9] | 2009 |
| Poland | Growth references (Kulaga et al.) [10] | 2010 |
| Serbia | Not available* | - |
| Switzerland | Swiss growth curves (Braegger et al.) [8] | 2011 |
| Turkey | Growth references for Turkish children (Neyzi et al.) [12] | 2006 |
| United Kingdom | Royal College of Paediatrics and Child Health [6,7] | 1990 |
| USA/Canada | Centre for Disease Control and Prevention [13] | 2000 |

^{*} No national growth references were available in Serbia, for which the WHO references are used instead. These patients were therefore included only in analyses using WHO references.

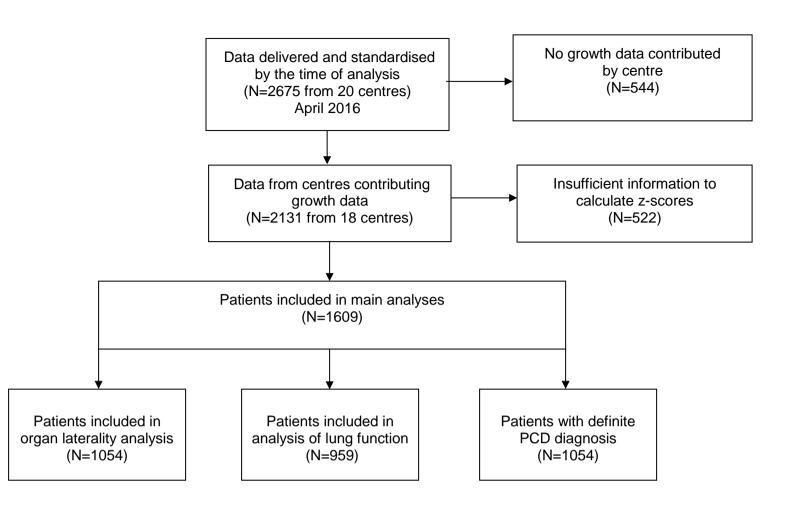


Fig S1. Flow chart showing the patients included for the different analyses

Table S2. Characteristics of PCD patients included in this study compared to those who were excluded

| Characteristic | Study participants | Insufficient information | Comparison with | No growth data | Comparison with |
|------------------------|--------------------|--------------------------|--------------------------------------|------------------------|--------------------------|
| | (n=1609) | on growth (n=522) | participants p-value [¶] | contributed (n=544) | participants p-value¶ |
| | n (%) | ` n (%) | | ` n (%) | , |
| Sex | | | 0.159 | | 0.198 |
| Male | 780 (49) | 266 (51) | | 226 (42) | |
| Female | 829 (51) | 245 (47) | | 209 (38) | |
| Missing | 0 (0.0) | 11 (2) | | 109 (20) | |
| Country of residence* | | | < 0.001 | | < 0.001 |
| Australia | 55 (3) | 52 (10) | | 0 (0) | |
| Northern Europe | 390 (24) | 118 (23) | | 0 (0) | |
| Western Europe | 405 (25) | 292 (56) | | 443 (81) | |
| Eastern Europe | 97 (6) | 1 (0) | | 0 (0) | |
| Southern Europe | 46 (3) | 3 (1) | | 0 (0) | |
| Western Asia | 202 (13) | 56 (11) | | 0 (0) | |
| North America | 414 (26) | 0 (0) | | 0 (0) | |
| South America | 0 (0) | 0 (0) | | 101 (19) | |
| Current age# | | | < 0.001 | | < 0.001 |
| 0-9 years | 195 (12) | 139 (27) | | 131 (24) | |
| 10-19 years | 689 (43) | 154 (30) | | 192 (35) | |
| 20-29 years | 326 (20) | 109 (21) | | 61 (11) | |
| 30-39 years | 151 (9) | 63 (12) | | 46 (9) | |
| 40-49 years | 100(6) | 20 (4) | | 54 (10) | |
| >50 years | 148 (9) | 37 (7) | | 39 (7) | |
| Missing | 0 (0) | 0 (0) | | 21 (4) | |
| Available measurements | | | | | |
| Height | 1609 (100) | 0 (0) | | 0 (0) | |
| BMI | 1494 (93) | 0 (0) | | 0 (0) | |

^{*} Based on the United Nations Statistics Division ¶ Chi-squared tests # In August 2016

Table S3. Height of PCD patients (N=1609) of the iPCD Cohort compared to WHO height references by characteristics of the study population

| Characteristics | mean z-score | 95% | CI | p-value [¶] |
|-------------------------------------|-----------------|-------|--------|----------------------|
| Sex | | | | 0.048 |
| male | -0.07 | -0.14 | 0.01 | |
| female | -0.18 | -0.25 | -0.10 | |
| Age group | | | | < 0.001 |
| 0-9 y | -0.11 | -0.17 | -0.05 | |
| 10-19 y | -0.23 | -0.28 | -0.17 | |
| 20-29 y | 0.02 | -0.06 | 0.09 | |
| 30-39 y | 0.03 | -0.06 | 0.11 | |
| 40-49 y | 0.01 | -0.09 | 0.10 | |
| >50y | -0.0003 | -0.11 | 0.11 | |
| Country | | | | < 0.001 |
| Australia | -0.32 | -0.61 | -0.04 | |
| Belgium | -0.21 | -0.46 | 0.04 | |
| Cyprus | -0.10 | -0.48 | 0.27 | |
| Denmark | 0.31 | 0.09 | 0.53 | |
| France | -0.30 | -0.50 | -0.10 | |
| Germany | 0.09 | -0.14 | 0.31 | |
| Israel | -0.51 | -0.70 | -0.33 | |
| Italy | -0.36 | -0.70 | -0.02 | |
| Netherlands | 0.67 | 0.41 | 0.93 | |
| Norway | -0.22 | -0.66 | 0.23 | |
| Poland | 0.17 | -0.06 | 0.39 | |
| Serbia | 0.52 | -0.23 | 1.26 | |
| Switzerland | -0.03 | -0.39 | 0.27 | |
| Turkey | -0.66 | -1.07 | -0.25 | |
| UK | -0.13 | -0.26 | 0.0001 | |
| USA/Canada | -0.18 | -0.29 | -0.07 | |
| Diagnostic certainty | | | | 0.531 |
| Definite PCD diagnosis ⁺ | -0.14 | -0.21 | -0.08 | |
| Probable PCD diagnosis# | -0.10 | -0.24 | 0.04 | |
| Clinical diagnosis only | -0.06 | -0.19 | 0.07 | |

[¶] Likelihood ratio test p-value indicating if the characteristic explains differences in height within the study population

⁺ Defined as hallmark PCD electron microscopy findings and/or biallelic gene mutation identified based on the ERS PCD Diagnostics Task Force guidelines

[#] Abnormal light or high frequency video microscopy finding and/or low nasal NO value

Table S4. BMI of PCD patients (N=1549) of the iPCD Cohort compared to national BMI references by characteristics of the study population

| Characteristics | mean | 95% (| CI | p-value [¶] |
|-------------------------------------|---------|-------|-------|----------------------|
| _ | z-score | | | |
| Sex | | | | 0.034 |
| male | 0.13 | 0.04 | 0.22 | |
| female | -0.001 | -0.09 | 0.09 | |
| Age group | | | | <0.001 |
| 0-9 y | -0.13 | -0.20 | -0.06 | |
| 10-19 y | -0.07 | -0.14 | 0.003 | |
| 20-29 y | 0.15 | 0.05 | 0.24 | |
| 30-39 y | 0.68 | 0.55 | 0.80 | |
| 40-49 y | 0.73 | 0.59 | 0.88 | |
| >50y | 0.99 | 0.82 | 1.16 | |
| Country | | | | < 0.001 |
| Australia | 0.12 | -0.19 | 0.44 | |
| Belgium | -0.06 | -0.35 | 0.23 | |
| Cyprus | -0.60 | -1.01 | -0.19 | |
| Denmark | 0.25 | 0.01 | 0.48 | |
| France | 0.24 | 0.01 | 0.46 | |
| Germany | -0.05 | -0.29 | 0.21 | |
| Israel | -0.15 | -0.36 | 0.06 | |
| Italy | 0.51 | 0.14 | 0.89 | |
| Netherlands | 0.45 | 0.16 | 0.74 | |
| Norway | -0.43 | -0.96 | 0.10 | |
| Poland | 0.13 | -0.14 | 0.41 | |
| Switzerland | -0.15 | -0.48 | 0.18 | |
| Turkey | -0.26 | -0.72 | 0.20 | |
| UK | 0.06 | -0.10 | 0.22 | |
| USA/Canada | 0.09 | -0.03 | 0.22 | |
| Diagnostic certainty | | | | 0.606 |
| Definite PCD diagnosis ⁺ | 0.04 | -0.04 | 0.18 | |
| Probable PCD diagnosis# | 0.11 | -0.05 | 0.28 | |
| Clinical diagnosis only | 0.11 | -0.04 | 0.26 | |

Abnormal light or high frequency video microscopy finding and/or low nasal NO value
Patients from Serbia were excluded from this analysis because no national references were available

[¶] Likelihood ratio test p-value indicating if the characteristic explains differences in BMI within the study population

⁺ Defined as hallmark PCD electron microscopy findings and/or biallelic gene mutation identified based on the ERS PCD Diagnostics Task Force guidelines

Table S5. BMI of PCD patients (N=1539) of the iPCD Cohort compared to WHO BMI references by characteristics of the study population

| Characteristics | maan | p-value [¶] | | |
|-------------------------------------|-----------------|----------------------|------|---------|
| Characteristics | mean z-score | 95% CI | | p-value |
| Sex | | | | 0.007 |
| male | 0.30 | 0.21 | 0.39 | |
| female | 0.12 | 0.03 | 0.21 | |
| Age group | | | | < 0.001 |
| 0-9 y | 0.03 | -0.04 | 0.11 | |
| 10-19 y | -0.03 | -0.10 | 0.05 | |
| 20-29 y | 0.36 | 0.26 | 0.45 | |
| 30-39 y | 0.99 | 0.87 | 1.11 | |
| 40-49 y | 1.07 | 0.93 | 1.22 | |
| >50y | 1.22 | 1.05 | 1.39 | |
| Country | | | | 0.004 |
| Australia | 0.36 | 0.03 | 0.69 | |
| Belgium | -0.07 | -0.36 | 0.22 | |
| Cyprus | -0.34 | -0.77 | 0.10 | |
| Denmark | 0.35 | 0.11 | 0.60 | |
| France | 0.03 | -0.20 | 0.26 | |
| Germany | 0.32 | 0.06 | 0.57 | |
| Israel | 0.06 | -0.16 | 0.28 | |
| Italy | 0.75 | 0.36 | 1.14 | |
| Netherlands | 0.24 | -0.07 | 0.54 | |
| Norway | 0.19 | -0.32 | 0.71 | |
| Poland | 0.35 | 0.09 | 0.62 | |
| Serbia | 0.70 | -0.16 | 1.55 | |
| Switzerland | -0.17 | -0.52 | 0.17 | |
| Turkey | 0.19 | -0.29 | 0.66 | |
| UK | 0.15 | -0.02 | 0.33 | |
| USA/Canada | 0.31 | 0.18 | 0.44 | |
| Diagnostic certainty | | | | 0.656 |
| Definite PCD diagnosis ⁺ | 0.21 | 0.13 | 0.29 | |
| Probable PCD diagnosis# | 0.15 | -0.02 | 0.33 | |
| Clinical diagnosis only | 0.26 | 0.11 | 0.42 | |

[¶] Likelihood ratio test p-value indicating if the characteristic explains differences in height within the study population

⁺ Defined as hallmark PCD electron microscopy findings and/or biallelic gene mutation identified based on the ERS PCD Diagnostics Task Force guidelines

[#] Abnormal light or high frequency video microscopy finding and/or low nasal NO value

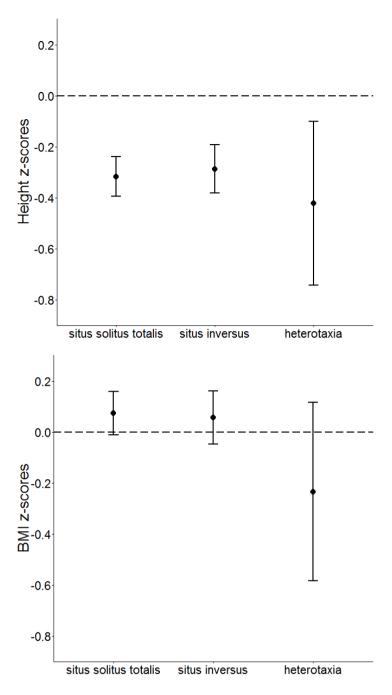


Fig S2. Height and BMI in PCD patients by situs status, compared to national references. Height and BMI are presented as z-scores (95%CI) after adjusting for sex, country, and level of diagnostic certainty.

Table S6. Height of patients with definite PCD diagnosis (N=1054) compared to national height references (sensitivity analysis) by characteristics of the study population

| 01 | | | | |
|-----------------|-----------------|-------|-------|----------------------|
| Characteristics | mean z-score | 95% (| ادَ | p-value [¶] |
| Sex | | | | 0.028 |
| male | -0.18 | -0.28 | -0.08 | |
| female | -0.33 | -0.43 | -0.24 | |
| Age group | | | | 0.313 |
| 0-9 y | -0.27 | -0.35 | -0.19 | |
| 10-19 y | -0.30 | -0.37 | -0.22 | |
| 20-29 y | -0.21 | -0.31 | -0.12 | |
| 30-39 y | -0.19 | -0.32 | -0.06 | |
| 40-49 y | -0.20 | -0.34 | -0.06 | |
| >50y | -0.19 | -0.36 | -0.02 | |
| Country | | | | < 0.001 |
| Australia | -0.35 | -0.67 | -0.03 | |
| Belgium | -0.70 | -0.97 | -0.42 | |
| Cyprus | -0.03 | -0.45 | 0.39 | |
| Denmark | -0.27 | -0.56 | 0.02 | |
| France | 0.02 | -0.25 | 0.28 | |
| Germany | -0.75 | -1.07 | -0.43 | |
| Israel | -0.72 | -1.00 | -0.44 | |
| Italy | -0.49 | -0.84 | -0.14 | |
| Netherlands | -0.27 | -0.63 | 0.09 | |
| Norway | -0.740 | -1.22 | -0.26 | |
| Poland | -0.17 | -0.52 | 0.18 | |
| Switzerland | 0.05 | -0.33 | 0.44 | |
| Turkey | -1.04 | -1.89 | -0.20 | |
| UK | -0.13 | -0.29 | 0.02 | |
| USA/Canada | -0.12 | -0.24 | 0.01 | |

Mean z-scores and 95%CI for each group after adjusting for the remaining characteristics ¶ Likelihood ratio test p-value indicating if the characteristic explains differences in height within the study population

Table S7. BMI of patients with definite PCD diagnosis (N=1019) compared to national BMI references (sensitivity analysis) by characteristics of the study population

| Characteristics | mean z-score | 95% CI | | p-value [¶] |
|-----------------|-----------------|--------|------|----------------------|
| Sex | | | | 0.009 |
| male | 0.15 | 0.04 | 0.25 | |
| female | -0.05 | -0.15 | 0.05 | |
| Age group | | | | < 0.001 |
| 0-9 y | -0.07 | -0.16 | 0.02 | |
| 10-19 y | -0.003 | -0.09 | 80.0 | |
| 20-29 y | 0.14 | 0.02 | 0.25 | |
| 30-39 y | 0.12 | -0.06 | 0.29 | |
| 40-49 y | 0.32 | 0.12 | 0.52 | |
| >50y | 0.80 | 0.58 | 1.01 | |
| Country | | | | 0.070 |
| Australia | 0.07 | -0.26 | 0.41 | |
| Belgium | 0.05 | -0.25 | 0.36 | |
| Cyprus | -0.45 | -1.00 | 0.10 | |
| Denmark | 0.16 | -0.15 | 0.46 | |
| France | 0.15 | -0.14 | 0.44 | |
| Germany | 0.04 | -0.30 | 0.38 | |
| Israel | -0.22 | -0.53 | 0.08 | |
| Italy | 0.48 | 0.11 | 0.85 | |
| Netherlands | 0.29 | -0.09 | 0.67 | |
| Norway | -0.40 | -0.84 | 0.04 | |
| Poland | 0.17 | -0.25 | 0.59 | |
| Switzerland | -0.15 | -0.56 | 0.25 | |
| Turkey | -0.45 | -1.37 | 0.46 | |
| UK | -0.06 | -0.23 | 0.12 | |
| USA/Canada | 0.12 | -0.02 | 0.25 | |

[¶] Likelihood ratio test p-value indicating if the characteristic explains differences in BMI within the study population;

⁺ Defined as hallmark PCD electron microscopy findings and/or biallelic gene mutation identified based on the ERS PCD Diagnostics Task Force guidelines

Table S8. Height of PCD patients (N=1601) of the iPCD Cohort compared to national height references by characteristics of the study population; including one measurement per patient

| Characteristics | mean z-score | 95% (| CI | p-value ¹ |
|-------------------------------------|-----------------|-------|-------|----------------------|
| Sex | | | | 0.208 |
| male | -0.26 | -0.35 | -0.17 | |
| female | -0.34 | -0.42 | -0.25 | |
| Age group | | | | 0.878 |
| 0-9 y | -0.27 | -0.36 | -0.17 | |
| 10-19 y | -0.33 | -0.44 | -0.23 | |
| 20-29 y | -0.37 | -0.58 | -0.17 | |
| 30-39 y | -0.22 | -0.47 | 0.03 | |
| 40-49 y | -0.30 | -0.58 | -0.02 | |
| >50y | -0.30 | -0.60 | 0.00 | |
| Country | | | | < 0.001 |
| Australia | -0.40 | -0.73 | -0.07 | |
| Belgium | -0.63 | -0.91 | -0.36 | |
| Cyprus | -0.10 | -0.55 | 0.34 | |
| Denmark | -0.47 | -0.72 | -0.21 | |
| France | 0.08 | -0.15 | 0.30 | |
| Germany | -0.93 | -1.18 | -0.68 | |
| Israel | -0.49 | -0.70 | -0.28 | |
| Italy | -0.45 | -0.84 | -0.05 | |
| Netherlands | -0.19 | -0.49 | 0.11 | |
| Norway | -0.61 | -1.13 | -0.09 | |
| Poland | -0.24 | -0.49 | 0.02 | |
| Switzerland | -0.15 | -0.51 | 0.20 | |
| Turkey | -1.12 | -1.58 | -0.67 | |
| UK | -0.14 | -0.29 | 0.00 | |
| USA/Canada | -0.16 | -0.28 | -0.04 | |
| Diagnostic certainty | | | | 0.593 |
| Definite PCD diagnosis ⁺ | -0.32 | -0.39 | -0.24 | |
| Probable PCD diagnosis# | -0.29 | -0.46 | -0.13 | |
| Clinical diagnosis only | -0.23 | -0.38 | -0.09 | |

Mean z-scores and 95%Cl for each group after adjusting for the remaining characteristics For each patient the earliest available measurement of height was included

[¶] Likelihood ratio test p-value indicating if the characteristic explains differences in height within the study population;

⁺ Defined as hallmark PCD electron microscopy findings and/or biallelic gene mutation identified based on the ERS PCD Diagnostics Task Force guidelines

[#] Abnormal light or high frequency video microscopy finding and/or low nasal NO value Patients from Serbia were excluded from this analysis because no national references were available

Table S9. BMI of PCD patients (N=1601) of the iPCD Cohort compared to national BMI references by characteristics of the study population; including one

measurement per patient

| Characteristics | mean | 95% (| CI | p-value [¶] |
|-------------------------------------|---------|-------|-------|----------------------|
| | z-score | | | |
| Sex | | | | 0.033 |
| male | 0.10 | 0.00 | 0.19 | |
| female | -0.04 | -0.13 | 0.05 | |
| Age group | | | | 0.022 |
| 0-9 y | -0.005 | -0.11 | 0.10 | |
| 10-19 y | -0.04 | -0.15 | 0.07 | |
| 20-29 y | -0.05 | -0.27 | 0.18 | |
| 30-39 y | 0.18 | -0.09 | 0.44 | |
| 40-49 y | 0.35 | 0.05 | 0.65 | |
| >50y | 0.41 | 0.10 | 0.72 | |
| Country | | | | 0.0002 |
| Australia | 0.11 | -0.23 | 0.45 | |
| Belgium | 0.06 | -0.24 | 0.35 | |
| Cyprus | -0.51 | -0.99 | -0.04 | |
| Denmark | 0.08 | -0.19 | 0.36 | |
| France | 0.06 | -0.18 | 0.30 | |
| Germany | -0.25 | -0.51 | 0.02 | |
| Israel | -0.15 | -0.37 | 0.07 | |
| Italy | 0.66 | 0.24 | 1.08 | |
| Netherlands | 0.35 | 0.04 | 0.66 | |
| Norway | -0.73 | -1.36 | -0.10 | |
| Poland | 0.03 | -0.25 | 0.31 | |
| Switzerland | -0.36 | -0.73 | 0.01 | |
| Turkey | -0.39 | -0.86 | 0.09 | |
| UK | 0.06 | -0.10 | 0.22 | |
| USA/Canada | 0.12 | -0.01 | 0.25 | |
| Diagnostic certainty | | | | 0.372 |
| Definite PCD diagnosis ⁺ | -0.01 | -0.09 | 0.07 | |
| Probable PCD diagnosis [#] | 0.05 | -0.12 | 0.23 | |
| Clinical diagnosis only | 0.12 | -0.04 | 0.27 | |

Mean z-scores and 95%CI for each group after adjusting for the remaining characteristics For each patient the earliest available measurement of BMI was included

Patients from Serbia were excluded from this analysis because no national references were available

 $[\]P$ Likelihood ratio test p-value indicating if the characteristic explains differences in height within the study population;

⁺ Defined as hallmark PCD electron microscopy findings and/or biallelic gene mutation identified based on the ERS PCD Diagnostics Task Force guidelines

[#] Abnormal light or high frequency video microscopy finding and/or low nasal NO value

Table S10. Height of paediatric patients (<20 years, N=1226) of the iPCD Cohort compared to national BMI references by characteristics of the study population

| Characteristics | mean z-score | 95% | 6 CI | p-value [¶] |
|-------------------------------------|-----------------|-------|--------|----------------------|
| Sex | | | | 0.149 |
| male | -0.22 | -0.31 | -0.13 | |
| female | -0.31 | -0.41 | -0.22 | |
| Age group | | | | 0.008 |
| 0-4 y | -0.33 | -0.43 | -0.23 | |
| 5-9 y | -0.23 | -0.31 | -0.16 | |
| 10-14 y | -0.29 | -0.36 | -0.22 | |
| 15-19 y | -0.22 | -0.30 | -0.14 | |
| Country | | | | < 0.001 |
| Australia | -0.33 | -0.66 | 0.01 | |
| Belgium | -0.46 | -0.83 | -0.10 | |
| Cyprus | -0.24 | -0.75 | 0.27 | |
| Denmark | -0.30 | -0.54 | -0.05 | |
| France | 0.15 | -0.05 | 0.36 | |
| Germany | -0.69 | -0.93 | -0.44 | |
| Israel | -0.45 | -0.68 | -0.21 | |
| Italy | -0.27 | -0.64 | 0.10 | |
| Netherlands | -0.29 | -0.60 | 0.03 | |
| Norway | -0.66 | -1.12 | -0.20 | |
| Poland | -0.24 | -0.48 | -0.003 | |
| Switzerland | -0.02 | -0.39 | 0.35 | |
| Turkey | -1.18 | -1.61 | -0.75 | |
| UK | -0.10 | -0.27 | 0.07 | |
| USA/Canada | -0.22 | -0.36 | -0.07 | |
| Diagnostic certainty | | | | 0.173 |
| Definite PCD diagnosis ⁺ | -0.31 | -0.39 | -0.23 | |
| Probable PCD diagnosis# | -0.20 | -0.37 | -0.03 | |
| Clinical diagnosis only | -0.15 | -0.31 | 0.01 | |
| Age at diagnosis | | | | 0.026 |
| 0-4 y | -0.13 | -0.26 | -0.01 | |
| 5-9 y | -0.28 | -0.40 | -0.17 | |
| 10-14 y | -0.27 | -0.41 | -0.13 | |
| 15-19 y | -0.50 | -0.69 | -0.31 | |

Patients from Serbia were excluded from this analysis because no national references were available

[¶] Likelihood ratio test p-value indicating if the characteristic explains differences in height within the study population

⁺ Defined as hallmark PCD electron microscopy findings and/or biallelic gene mutation identified based on the ERS PCD Diagnostics Task Force guidelines

[#] Abnormal light or high frequency video microscopy finding and/or low nasal NO value

Table S11. Height of adult patients (≥20 years, N=439) of the iPCD Cohort compared to national BMI references by characteristics of the study population

| Characteristics | mean 95% CI z-score | | CI | p-value [¶] |
|-------------------------------------|------------------------|-------|-------|----------------------|
| Sex | | | | 0.430 |
| male | -0.25 | -0.42 | -0.09 | |
| female | -0.34 | -0.47 | -0.21 | |
| Country | | | | < 0.001 |
| Australia | -0.46 | -0.96 | 0.03 | |
| Belgium | -0.88 | -1.23 | -0.53 | |
| Cyprus | -0.06 | -0.55 | 0.44 | |
| Denmark | -0.75 | -1.10 | -0.39 | |
| France | 0.36 | -0.29 | 1.02 | |
| Germany | -1.58 | -2.23 | -0.93 | |
| Israel | -0.57 | -0.91 | -0.23 | |
| Italy | -1.11 | -1.76 | -0.45 | |
| Netherlands | -0.16 | -0.69 | 0.37 | |
| Poland | -0.46 | -2.65 | 1.73 | |
| Switzerland | 0.18 | -0.35 | 0.71 | |
| UK | -0.21 | -0.45 | 0.02 | |
| USA | 0.06 | -0.14 | 0.26 | |
| Diagnostic certainty | | | | 0.381 |
| Definite PCD diagnosis ⁺ | -0.26 | -0.39 | -0.14 | |
| Probable PCD diagnosis# | -0.33 | -0.64 | -0.03 | |
| Clinical diagnosis only | -0.47 | -0.74 | -0.21 | |

[¶] Likelihood ratio test p-value indicating if the characteristic explains differences in height within the study population;

⁺ Defined as hallmark PCD electron microscopy findings and/or biallelic gene mutation identified based on the ERS PCD Diagnostics Task Force guidelines

[#] Abnormal light or high frequency video microscopy finding and/or low nasal NO value Patients from Serbia were excluded from this analysis because no national references were available

Table S12. Comparison with previously published studies on growth and/or nutritional status of PCD patients

| Author (year) | Population | Type of study | No of patients | Country | Comparison | Summary of findings |
|-------------------------------------|---------------------|---------------------|----------------|-----------------------|--|---|
| Boon et al. (2014) [23] | Children and adults | Single- centre | 168 | Belgium | Normal (national references) | Lower height than normal, no difference in BMI |
| Cohen-Cymberknoh et al. (2014) [24] | Children and adults | Single- centre | 34 | Israel | CF patients (European CF references) | Lower BMI than CF patients Trend towards a correlation between FEV1 and BMI |
| Davis et al. (2015) [25] | Children | Multi- centre | 118 | Canada and USA | Normal (national references) | Median height at the 42nd and median BMI at the 63rd percentile Lower height and BMI and FEV1 in patients with CCDC39 and CCDC40 mutations |
| Maglione et al. (2014) [26] | Children | Multi- centre | 158 | Denmark, Italy, UK | Normal (UK references) | No difference in children in height and BMI to normal No correlation between BMI and FEV1 and FVC z-scores |
| Svobodova et al. (2013) [27] | Children and adults | Single- centre | 29 | Czech Republic | Normal (national references) | Lower height in children age 7-13 years, no difference in BMI No difference in height in patients diagnosed earlier or between patients with and without impaired lung function |

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