Kartagener Syndrom und Primäre Ciliäre Dyskinesie e.V.
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International Primary Ciliary Dyskinesia Cohort

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PCD research

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- Rare diseases have been neglected for years
- Little funding available for research
- No routine data; difficult to identify patients (no ICD-10)
- Low awareness among physicians & public
- Complex diagnosis/ ends up to misdiagnosis or late diagnosis
- Little is known about symptoms, prognosis and treatment

Need for collaborative studies

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- Even large centres have few patients
- Many small studies, difficult to compare
- Doctors don’t always use the same “language”

Registries and cohorts:

- Increase awareness, help with patient identification
- Pool data of many countries/centres together in a standardised way
- Can be linked with other disease registries and routine statistics
- Help to identify suitable patients for studies, including trials

BESTCILIA FP7 EU funded project

- WP1: Observational trials in PCD
- WP2: International prospective PCD registry
- WP3: Introducing standardized diagnostic testing for PCD in European countries, where this is currently not available
- WP4: PCD-specific Health-Related Quality of Life Questionnaires (HRQOLQ)
- WP5: Randomized controlled clinical trial on the use of azithromycin

WP1: Observational trials in PCD

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We aimed to:

- Identify existing PCD datasets
- Pool them in standardised cohort
- Describe the disease: clinical symptoms, lung function, growth, disease severity, prognosis and treatment effects
- Use the results to inform future research

Building up the iPCD cohort

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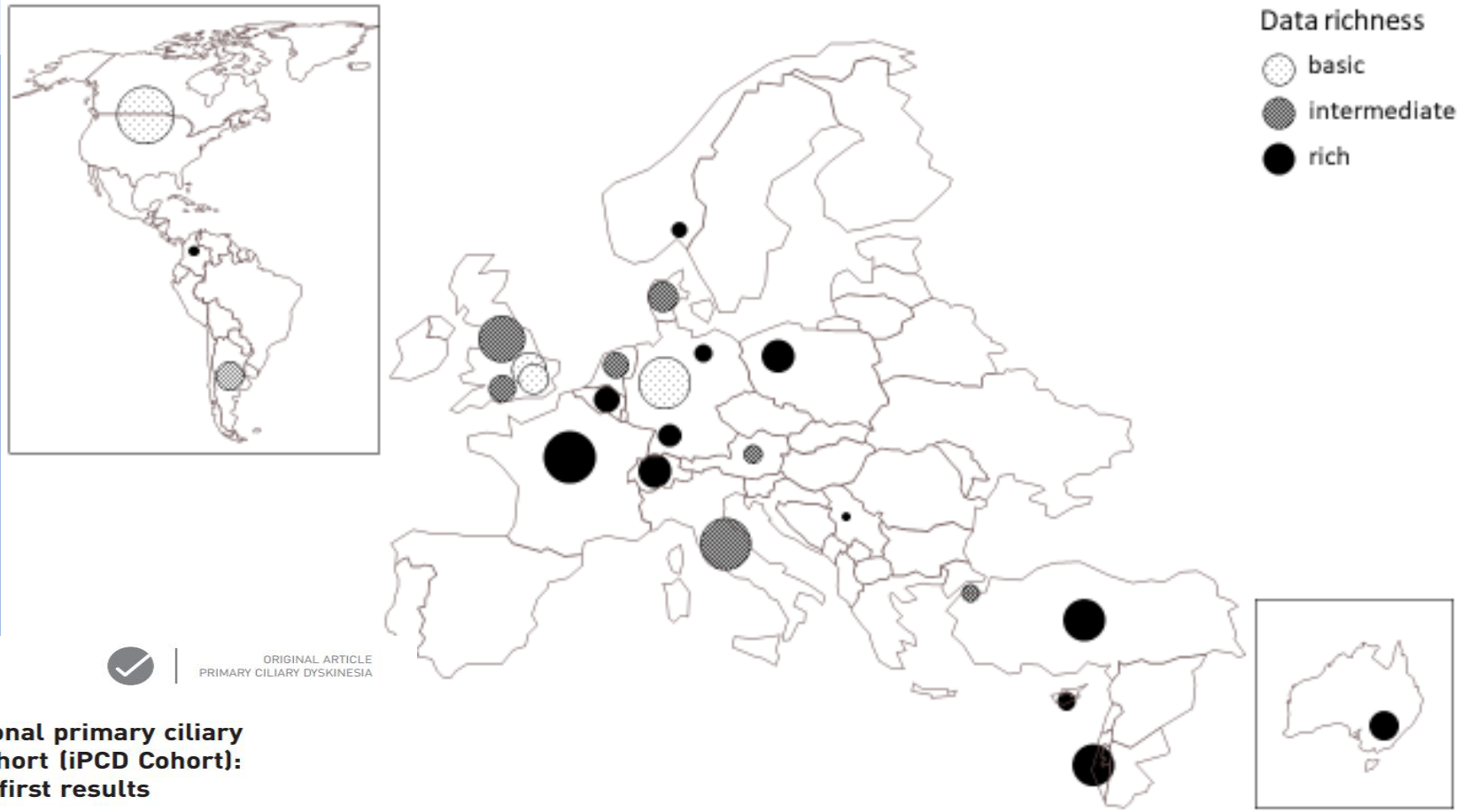
Identify all available datasets on PCD patients:

- Published and unpublished studies and case series
- Registries
- Personal contacts (2009 ERS PCD taskforce survey)
- www.clinical-trials.gov database

iPCD Cohort



ORIGINAL ARTICLE
PRIMARY CILIARY DYSKINESIA

The international primary ciliary dyskinesia cohort (iPCD Cohort): methods and first results

Myrofora Goutaki[1,2], Elisabeth Maurer[1], Florian S. Halbeisen[1], Israel Amirav[3], Angelo Barbato[4] on behalf of the PCD Italian Consortium, Laura Behan[5], Mieke Boon[6], Carmen Casaulta[2] on behalf of the Swiss PCD Group, Annick Clement[7] on behalf of the French Reference Centre for Rare Lung Diseases, Suzanne Crowley[8], Eric Haarman[9], Claire Hogg[10], Bulent Karadag[11], Cordula Koerner-Rettberg[12], Margaret W. Leigh[13] on behalf of the Genetic Disorders of Mucociliary Clearance Consortium, Michael R. Loebinger[14], Henryk Mazurek[15], Lucy Morgan[16], Kim G. Nielsen[17], Heymut Omran[18], Nicolaus Schwerk[19], Sergio Scigliano[20], Claudius Werner[18], Panayiotis Yiallouros[21], Zorica Zivkovic[22,23], Jane S. Lucas[5] and Claudia E. Kuehni[1]

@ERSpublications
The iPCD Cohort offers a unique opportunity to study PCD in an international retrospective cohort of >3000 patients http://ow.ly/rn0m304Jgsu

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21 countries
26 centres or consortia

Country	P.I.	Patients (N)	Country	P.I.	Patients (N)
Argentina	S. Scigliano	101	Netherlands	E. Haarman	82
Australia	L. Morgan	105	Norway	S. Crowley	27
Belgium	M. Boon	82	Poland	H. Mazurek	132
Colombia	S. Ucros	11	Serbia	Z. Zivkovic	10
Cyprus	P. Yiallouros	38	Switzerland	Swiss PCD group	132
Czech Rep.	P. Pohunek	44	Turkey	B. Karadag	37
Denmark	K. Nielsen	110	Turkey	U. Özçelik	221
France	Respirare (A. Clement/ B. Maitre)	380	UK	J. Lucas	104
			UK	C. Hogg	116
			UK	M. Loebinger	151
Germany	H. Omran	337	UK	R. Hirst	443
Germany	C. Koerner-Rettberg	64	USA/ Canada	PCD Foundation's Clinical & Research Centers Network	418
Germany	N. Schwerk	38			
Greece	K. Priftis	12			
Israel	I. Amirav	210	**iPCD Cohort**		**3736**
Italy	Italian PCD Consortium	331			

Growth and nutrition of PCD patients and their association with lung function

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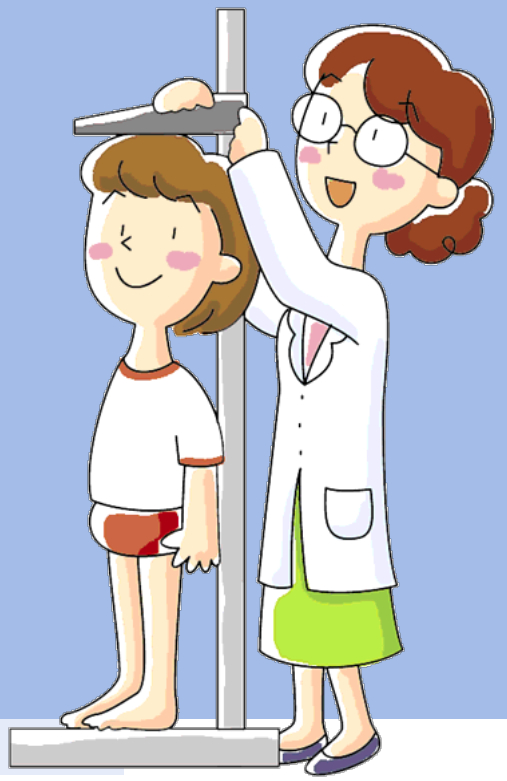
- Growth and nutrition:

a. often affected in chronic pulmonary diseases
b. associated with lung function later in life in eg. CF or bronchopulmonary dysplasia

- Few existing studies:

a. contradictory results
b. few patients
c. methodological inconsistencies



Growth and nutrition of PCD patients and their association with lung function

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We aimed to:

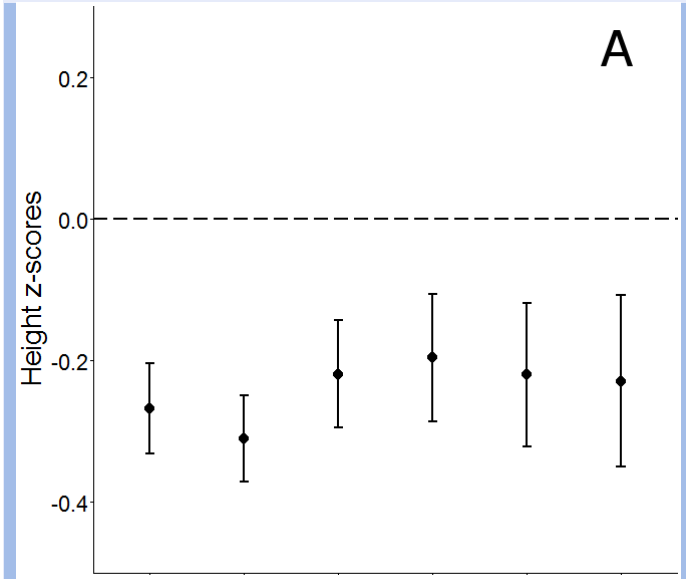
- Describe age and sex-adjusted height and BMI of PCD patients, compared to:

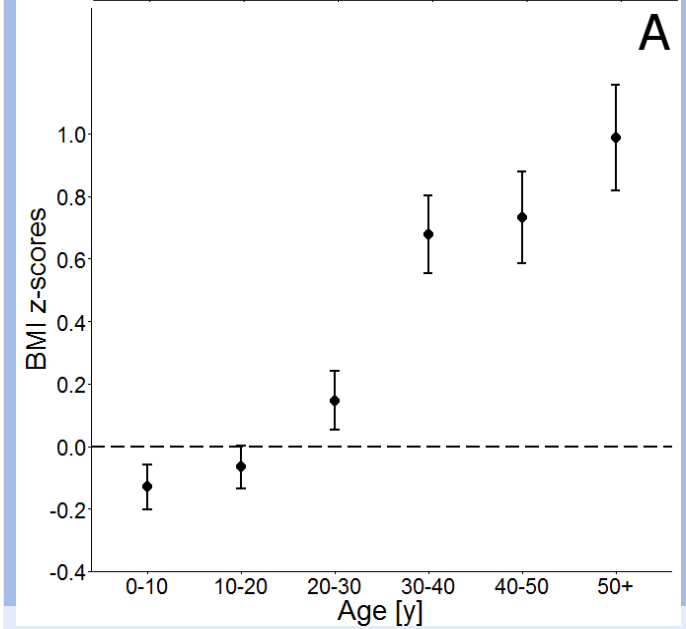
a) international reference values

b) national reference values

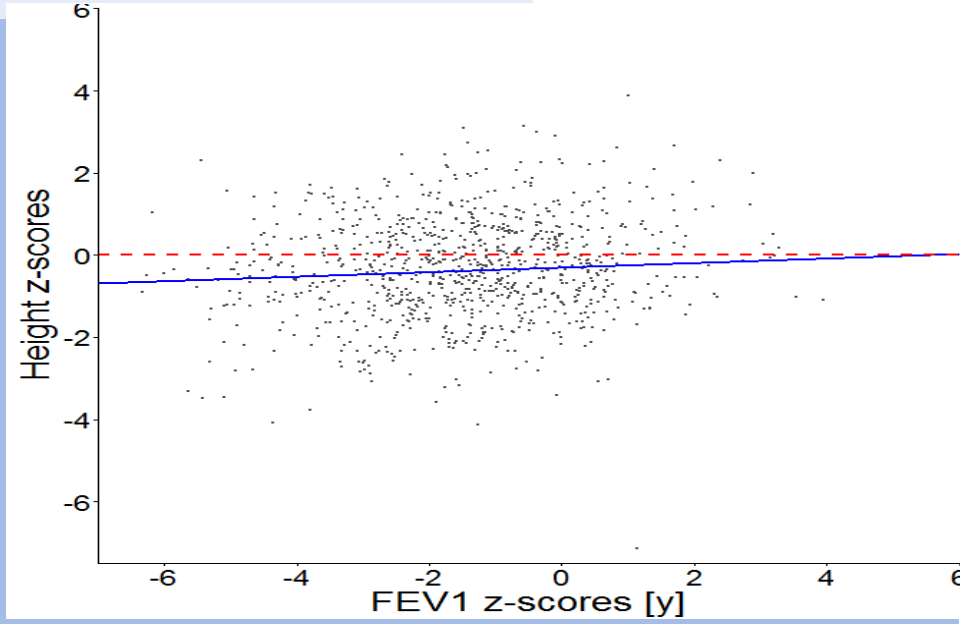
- Determine factors associated with height and BMI in PCD patients

Growth and nutrition of PCD patients and their association with lung function





1609 patients
16 countries



- Height z-scores lower in all age groups
- BMI z-scores lower in children
- Late diagnosis associated with lower height & BMI
- FEV1 positively associates with height & BMI

Growth and nutrition of PCD patients and their association with lung function

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- Early diagnosis and multidisciplinary management including nutritional interventions if needed, could delay disease progression
- Prospective longitudinal studies are needed to clarify how lung function and growth are connected in PCD

Lung function in patients with PCD

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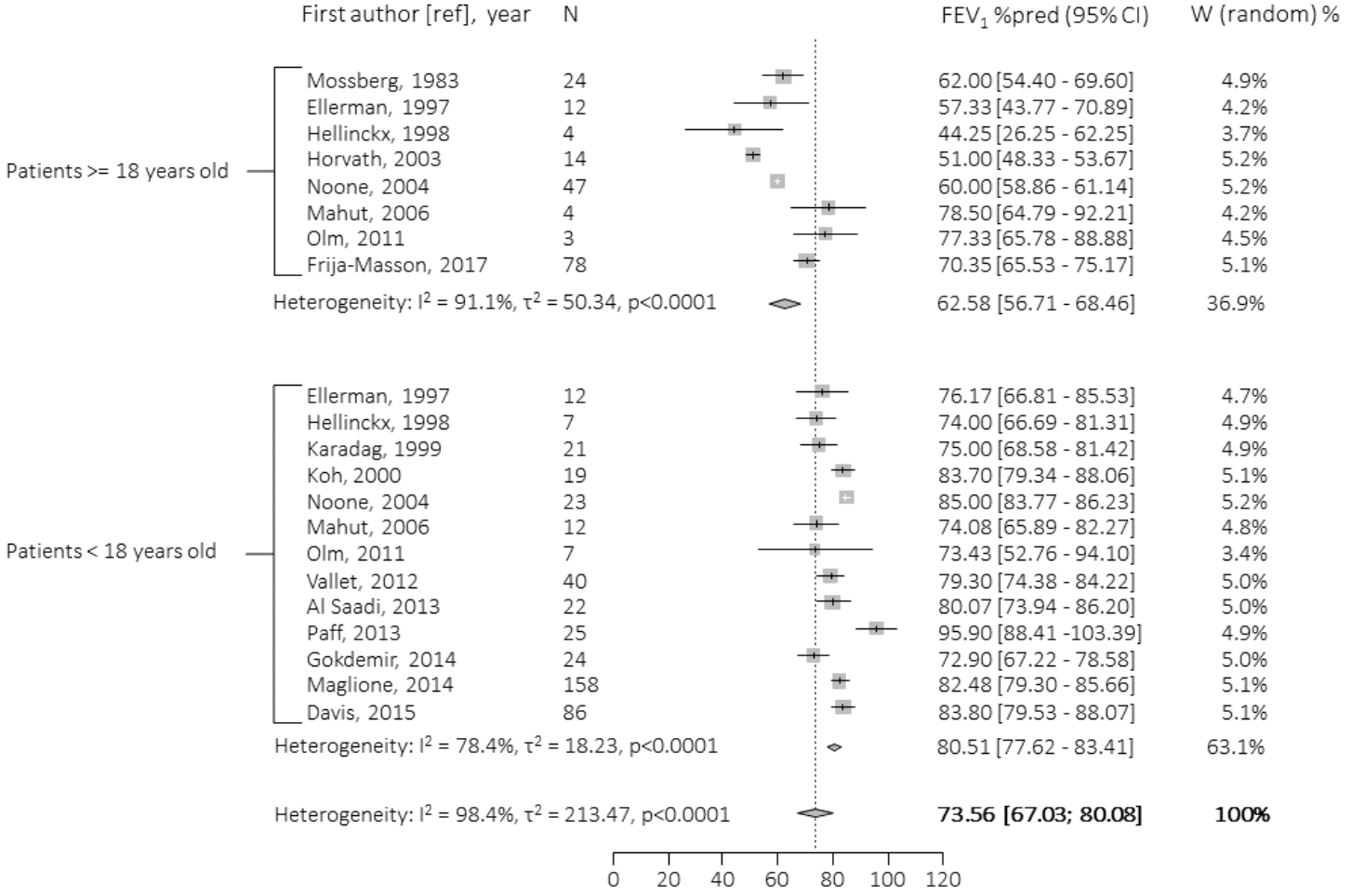
Results of published studies vary



Halbeisen, to be submitted

Lung function in patients with PCD

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We aimed to:

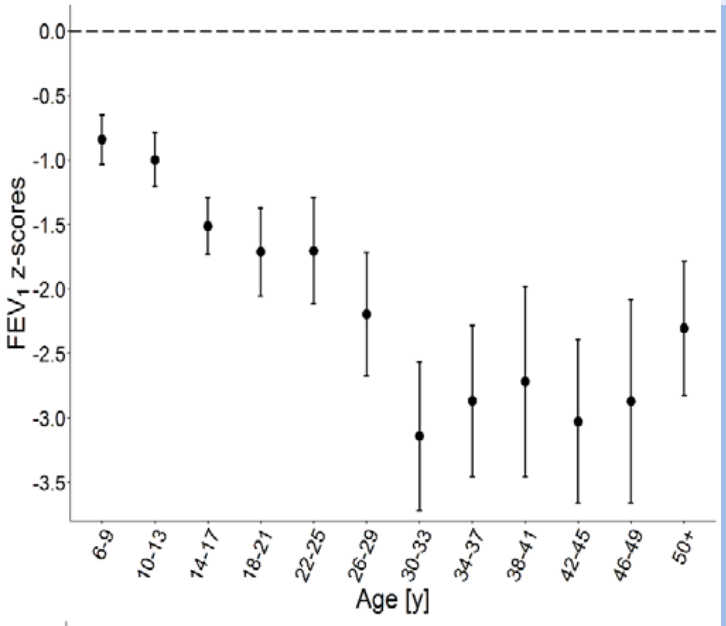
- Describe age, sex and height-adjusted FEV1 and FVC of PCD patients compared to

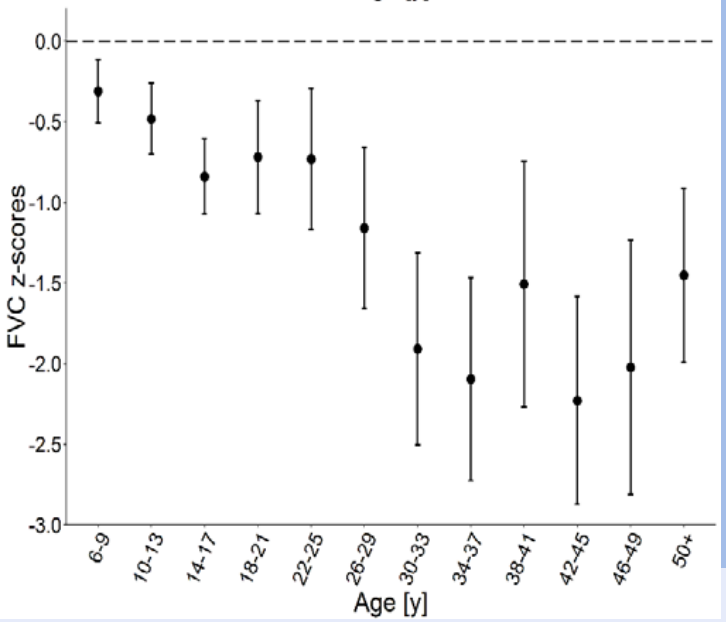
a) international GLI reference values

b) published CF lung function values

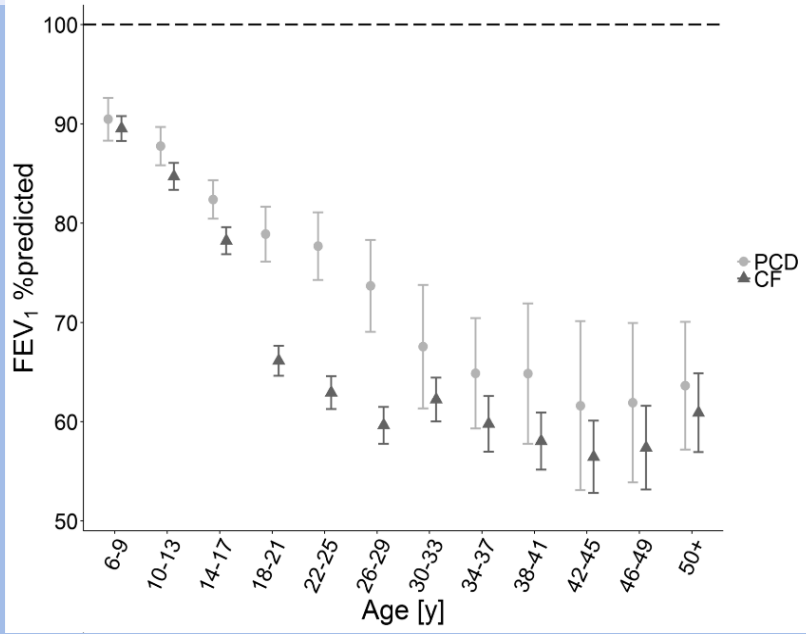
- Determine factors associated with lung function of PCD patients

Lung function in patients with PCD





991 patients
15 countries



- All age groups had lower FEV1 and FVC z-scores
- Female sex and underweight were predictors of lower lung function
- Lung function similar to CF in childhood

Halbeisen et al, in revision

Ongoing and planned studies

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- Prevalence and impact of **lung resection** in PCD
- Evolution of PCD **diagnostic testing** in Europe
- **Neonatal manifestations** in PCD

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SWISS NATIONAL SCIENCE FOUNDATION

- Describe **clinical picture and natural history** of PCD
- Distinguish **disease subgroups** (phenotypes)
- Propose simple **disease classification model**

Standardised PCD follow-up proforma

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- Need for standardised recording of symptoms
- PCD follow-up is often extrapolated from other diseases

- We aimed to **develop disease specific standardised proformas** for longitudinal data collection of PCD patients during routine care

- Interdisciplinary group (paediatric and adult pulmonologists, ENT physicians, epidemiologists, diagnosticians, nurses, physiotherapists)

Standardised PCD follow-up proforma

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Standardised PCD proforma modules	
1	Basic patient information, diagnostic evaluation and baseline medical history
2	Physical examination of lungs and heart
3	Physical examination of ear nose throat
4	Growth measurements and clinical tests (lung function, imaging, microbiology
5	Hospitalizations and treatment (surgeries, medication, physiotherapy)
6	Medical history
7	Environment and lifestyle (different for children and adults)

Very soon: **start piloting!**

Thank you for your attention

I would like to thank:

- The PCD team at ISPM Bern

Claudia Kuehni, Florian Halbeisen

- The BESTCILIA group, Heymut Omran
- BEAT-PCD network, Jane Lucas
- iPCD cohort collaborators
- Standardised PCD proforma group
- All the patients and the patient support organisations

FNSNF
Swiss National Science Foundation

BESTCILIA

BEAT-PCD
BETTER EXPERIMENTAL APPROACHES TO TREAT PCD

LUNGENLIGA BERN
LIGUE PULMONAIRE BERNOISE