

The International Primary Ciliary Dyskinesia (iPCD) Cohort: methods and first results

-Online supplement

Data protection/ Ethics

In most participating countries, research laws do not require obtaining patient informed consent for observational retrospectively collected anonymised data. However, even in these countries, PIs have received already 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 a registry permission from the Bern Cantonal Ethics Committee that permits to have a national PCD registry and to contribute pseudonymised data to international studies.

Table S1. Standardised dataset structure of the iPCD cohort (REDCap)

Topic 1: General information

Variable label	Variable description	Values	Type
idpat	Unique patient identifier		Text
idfam	Identifier for family		Text
idcent	Identifier for centre	0-12	Numeric
dob	Patient's day of birth	1-31	Numeric
mob	Patient's month of birth	1-12	Numeric
yob	Patient's year of birth	1900-2014	Numeric
sex	Sex of the patient	0=female 1=male	Categorical
ethn	Patient's ethnicity	1=White/ European descent 2=Arab (for instance, Egypt, Iraq, Jordan, Lebanon) 3=West Asian (for instance, Turkey, Iran, Afghanistan) 4=Indian subcontinent 5=Pakistani/Bangladeshi 6=Southeast Asian (for instance, Malaysia, Indonesia, Vietnam) 7=East Asian (for instance, China, Japan, Korea, Taiwan) 8=African/ African American 9=Latin American/Hispanic 10=American Indian/ Alaska Native 11=Australian aboriginal 12=Native Hawaiian or other Pacific Islander 13=Caribbean 14=Jewish 15=Roma/ Irish traveler 16=Other ethnic group (not listed above) 17=Don't know 18=Prefer not to answer 19=Bangladeshi	Categorical
ethn_oth	If other please explain		Text
vislast	Date of last follow up	dd/mm/yyyy	Date
alivlast	Date last known alive	dd/mm/yyyy	Date

Table S1: Dataset structure of the iPCD cohort (REDCap)

Topic 2: Diagnostic Evaluation

Variable label	Variable description	Values	Type
pcd_status	Diagnostic status of PCD	0=PCD excluded 1=definite PCD 2=PCD probable, positive HFVM, not all testing finalised 3= PCD probable, positive EM, not all testing finalised 4= mainly clinical diagnosis of PCD	Categorical
dd	Date of definite diagnosis of PCD	dd/mm/yyyy	Date
ddd	Date that this patient was first considered as PCD	dd/mm/yyyy	Date
no	nasalNO performed	0=no 1=yes 2=unknown	Categorical
no_dat	Date of nNO testing	dd/mm/yyyy	Date
no_res	nNO result (in ppb)	Valid numbers	Numeric
no_eq	Equipment used for nNO testing	0=Ecomedics CLD 88 Exhalyzer 1= NIOX MINO 2=NIOX Flex 3=Other	Categorical
no_man	breathing maneuver	1=breath hold 2=exhalation against resist 3=humming 4=tidal breathing 5=other	Categorical
no_prrat	nNO production rate	valid numbers	Numeric
hfvm	HFVM performed	0=no 1=yes 2=unknown	Categorical
light_micr	How was the measurement performed?	1=with HFVM 2=with light microscopy 3= other	Categorical
hfvm_dat	Date of HFVM testing	dd/mm/yyyy	Date
hfvm_fr	HFVM ciliary beating frequency	0=decreased 1=increased 2=normal	Categorical
hfvm_pat	HFVM ciliary beating pattern	0=regular power stroke and recovery stroke 1=almost immotile cilia 2=extremely stiff due to reduced ciliary bending 3=stiff beating pattern due to slight reduced amplitude 4=abnormal circular beating pattern 5=other	Categorical
hfvm_con	HFVM conclusion	0=normal 1=abnormal 2=highly abnormal 3=inconclusive test	categorical
em	EM performed	0=no 1=yes 2=unknown	Categorical
em_dat	Date of EM testing	dd/mm/yyyy	Date
em_res	EM result	0=normal 1=abnormal 2=unclear	Categorical
em_oda	Is there an ODA-defect	0=no 1=yes 2=unclear	Categorical

Topic 2: Diagnostic Evaluation (continued)

Variable label	Variable description	Values	Type
em_ida	Is there an IDA-defect?	0=no 1=yes 2=unclear	Categorical
em_cpd	Is there a central pair defect (9+0)?	0=no 1=yes 2=unclear	Categorical
em_dis	Is there a tubulus disorganisation defect with IDA defect?	0=no 1=yes 2=unclear	Categorical
em_tutr	Is there a tubular transposition defect (8+1)?	0=no 1=yes 2=unclear	Categorical
em_nex	Is there a nexin link defect?	0=no 1=yes 2=unclear	Categorical
em_acil	Are there no cilia present (acilia)?	0=no 1=yes 2=unclear	Categorical
gene	Genetic testing performed	0=no 1=yes 2=unknown	Categorical
gene_dat	Date of genetic testing	dd/mm/yyyy	Date
gene_res	Genetic testing result	1=ARMC4 2=C21ORF59 3=CCDC103 4=CCDC114 5=CCDC164 6=CCDC39 7=CCDC40 8=CCDC65 9=CCNO 10=DNAAF3 11=DNAH11 12=DNAH5 13=DNAI1 14=DNAI2 15=DNAL1 16=DYX1C1 17=LRRCS0 18=RSPH1 19=RSPH9 20=SPAG1 21=TXNDC3 22=ZBBX 23=ZMYND10 24=other 25=Negative 26=Waiting for result	Categorical
gene_res_oth	If other, please explain		Text
diagn_comment	Add any comments relevant for PCD diagnosis <i>For example positive IF results, multiple positive HFVM tests, etc</i>		Text

Topic 3: Baseline clinical characteristics

Variable label	Variable description	Values	Type
situs	Does the patient have any situs anomalies?	0=no 1=situs inversus 2=heterotaxia 3=unknown	Categorical
chd	Does the patient have a congenital heart defect?	0=no 1=yes 2=unknown	Categorical
chd_typ	Type of congenital heart defect		Text
braincil	Did the patient at any stage had any type of brain cilia dysfunction (e.g hydrocephalus)?	0=no 1=yes 2=unknown	Categorical
braincil_typ	Type of brain cilia dysfunction		Text
retin	Does the patient have retinitis pigmentosa?	0=no 1=yes 2=unknown	Categorical
renal	Does the patient have any renal problems?	0=no 1=yes 2=unknown	Categorical
infert	Does the patient have fertility problems?	0=no 1=subfertility 2=infertility 3=unknown 4=not applicable	Categorical
infert_d	Date when fertility was evaluated	dd/mm/yyyy	Date
semen_anal	semen analysis results	0=abnormal 1=normal 2=not performed	categorical
child_n	Number of children	valid number	Numerical
assist_rep	<i>if child_n ≥ 1, were assisted reproduction methods used</i>	0=no 1=yes 2=unknown	categorical
comorb	Does the patient have any relevant comorbidities not connected with PCD?	0=no 1=yes	Categorical
comorb_com	If yes, please comment		

Topic 4: Lung Function & somatometric values

Variable label	Variable description	Values	Type
lufu_d	Lung function date	dd/mm/yyyy	Date
weight	Patient's weight in kg	valid numbers	Numerical
height	Patient's height in cm	valid numbers	Numerical
lufu_FVC_pre	FVC pre bronchodilation	valid numbers	Numerical
lufu_FEV1_pre	FEV1 pre bronchodilation	valid numbers	Numerical
lufu_PEFR_pre	PEFR pre bronchodilation	valid numbers	Numerical
lufu_FEF50_pre	FEF50 pre bronchodilation	valid numbers	Numerical
lufu_FEF2575_pre	FEF25-75 pre bronchodilation	valid numbers	Numerical
lufu_TLC_pre	TLC pre bronchodilation	valid numbers	Numerical
lufu_RV_pre	RV pre bronchodilation	valid numbers	Numerical
lufu_FRC_pre	FRC pre bronchodilation	valid numbers	Numerical
lufu_FVC_post	FVC post bronchodilation	valid numbers	Numerical
lufu_FEV1_post	FEV1 post bronchodilation	valid numbers	Numerical
lufu_PEFR_post	PEFR post bronchodilation	valid numbers	Numerical
lufu_FEF50_post	FEF50 post bronchodilation	valid numbers	Numerical
lufu_FEF2575_post	FEF25-75 post bronchodilation	valid numbers	Numerical
lufu_TLC_post	TLC post bronchodilation	valid numbers	Numerical
lufu_RV_post	RV post bronchodilation	valid numbers	Numerical
lufu_FRC_post	FRC post bronchodilation	valid numbers	Numerical
lufu_RV_post	RV post bronchodilation	valid numbers	Numerical
mbw	Was MBW performed?	0=no 1=yes 2=unknown	Categorical
mbw_dev	Which MBW device was used?		Text
mbw_gas	What type of trace gas was used?		Text
lufu_LCI_pre	LCI pre bronchodilation	valid numbers	Numerical
lufu_frc_mbw_pre	FRC (in MBW) pre bronchodilation in L	valid numbers	Numerical
lufu_LCI_post	LCI post bronchodilation	valid numbers	Numerical
lufu_frc_mbw_post	FRC (in MBW) pro bronchodilation in L	valid numbers	Numerical

Topic 5: Clinical manifestations (signs and symptoms)

Variable label	Variable description	Values	Type
clinic_d	Date of the clinical visit	dd/mm/yyyy	Date
cough	Did the patient have cough at visit or during the last 3 months?	0=no 1=yes 2=unknown	Categorical
ccough	Did the patient have chronic or frequent cough during the last 3 months?	0=no 1=yes 2=unknown	Categorical
ccough_def	How do you define chronic cough?		Text
sput	Did the patient have sputum at visit or during the last 3 months?	0=no 1=yes 2=unknown	Categorical
hemoptys	Did the patient have hemoptysis at visit or during the last 3 months?	0=no 1=yes 2=unknown	Categorical
wheez	Did the patient have wheezing at visit or during the last 3 months?	0=no 1=yes 2=unknown	Categorical
pneum	Does the patient have pneumonia at visit or during the last 3 months?	0=no 1=yes 2=unknown	Categorical
pneumoth	Did the patient have pneumothorax during the last 3 months?	0=no 1=yes 2=unknown	Categorical
rhin	Did the patient have rhinorrhea/rhinitis at visit or during the last 3 months?	0=no 1=yes 2=unknown	Categorical
otit	Did the patient have otitis media at visit or during the last 3 months?	0=no 1=yes 2=unknown	Categorical
sin	Did the patient have sinusitis at visit or during the last 3 months?	0=no 1=yes 2=unknown	Categorical
hearl	Did the patient have hearing impairment?	0=no 1=yes 2=unknown	Categorical
audiom	if yes what are the audiometry findings		Text
glue	Did the patient have glue ear at visit?	0=no 1=yes 2=unknown	Categorical
tymperf	Did the patient have tympanic perforation at visit?	0=no 1=yes 2=unknown	Categorical
pol	Did the patient have nasal polyps at visit?	0=no 1=yes 2=unknown	Categorical
club	Did the patient have clubbing at visit?	0=no 1=yes 2=unknown	Categorical
reflux	Did the patient have gastroesophageal reflux during the last 3 months?	0=no 1=yes 2=unknown	Categorical

Topic 5: Clinical manifestations (signs and symptoms/ continued)

Variable label	Variable description	Values	Type
exerc	Did the patient have exercise limitation during the last 3 months?	0=no 1=minor exercise limitation 2=major exercise limitation 3=unknown	Categorical
act_smok	Was the patient a smoker at the time of visit or during the last 3 months?	0=no 1=yes 2=unknown	Categorical
act_smok_cig	if yes how many cigarettes did he/she smoke per day?	Valid numbers	Numerical
pas_smok	Is the patient exposed to passive smoking in the household?	0=no 1=yes 2=unknown	Categorical
pas_smok_det	if yes who is smoking in the household?	0=mother 1=father 2=both 3=other	Categorical

Topic 6: Therapy

Variable label	Variable description	Values	Type
ther_d	Date when medication was prescribed	dd/mm/yyyy	Date
<u>inhaled medication</u>			
nacl	Was NaCl 0.9% prescribed?	0=no 1=yes	Categorical
hyp_nacl	Was hypertonic NaCl prescribed?	0=no 1=yes	Categorical
hyp_nacl_conc	If yes in what concentration?	1=3% 2=6% 3=other	Categorical
hyp_nacl_oth	If other please specify with text		Text
b2sh_bro	Was a β 2 short-acting bronchodilator prescribed?	0=no 1=yes	Categorical
b2l_bro	Was a β 2 long-acting bronchodilator prescribed?	0=no 1=yes	Categorical
sh_musc	Was a short-acting muscarinic antagonist prescribed?	0=no 1=yes	Categorical
l_musc	Was a long-acting muscarinic antagonist prescribed?	0=no 1=yes	Categorical
ICS	Was an inhaled corticosteroid prescribed?	0=no 1=yes	Categorical
bronc_ICS	Was a bronchodilator and steroid combination prescribed?	0=no 1=yes	Categorical
inh_oth	Was any other inhaled medication prescribed?	0=no 1=yes	Categorical
<u>nasal medication</u>			
nas_ster	Was any nasal steroid prescribed?	0=no 1=yes	Categorical
nas_oth	Was any other nasal spray prescribed?	0=no 1=yes	Categorical
nas_med_typ	If yes what type of nasal medication was prescribed?	1=NaCl lavage 2=NaCl spray 3=antihistamine nasal spray 4= other	Categorical
nas_oth_typ	If other please specify with text		Text
<u>antibiotics</u>			
antib	Were any antibiotics prescribed?	0=no 1=yes	Categorical
antib_dur	If yes was it prescribed as prophylactic treatment or for an acute infection?	1=acute 2=prophylactic 3=unknown	Categorical
antib_rout	If yes give route of antibiotic administration	1=per os 2=intravenous 3=inhaled 4=intramuscular 5=other	Categorical
antib_typ	if yes which antibiotic		Text

Topic 6: Therapy (continued)

Variable label	Variable description	Values	Type
oxyg_ther	Was oxygen therapy prescribed?	0=no 1=yes	Categorical
oxyg_th_typ	Was it continuous or intermittent?	1=continuous 2=intermittent 3=unknown	Categorical
mech_vent_typ	Was it invasive or non-invasive?	1=invasive 2=non-invasive 3=unknown	Categorical
physio	Was physiotherapy prescribed?	0=no 1=yes	Categorical
physio_typ	What type of physiotherapy?	1=airway clearance therapy postural drainage and percussion 2=autogenic Drainage 3=positive expiratory pressure (PEP) 4=Active-cycle of breathing technique (ACBT) 5=Ocsillatory PEP (OPEP) 6=high-frequency chest compression (HFCC) 7=exercise training 8=unknown	Categorical

Topic 7: Microbiological findings

Variable label	Variable description	Values	Type
micr_d	Date of microbiology examination		Date
micr_sampl	Sample taken for microbiology analysis	1=sputum 2=throat swab 3=cough swab 4=laryngeal swab 5=nasal swab 6=nasal lavage 7=BAL 8=ear swab	Categorical
micr_res	First microorganism isolated	0=Aspergillus Fumigatus 1=Candida species 2=Branhamella/M.catarrhalis 3=Burkholderia species 4=Enterobacter species 5=Enterococcus species 6=E.coli 7=H.Influenza 8=Klebsiella species 9=Ps.aeruginosa 10=St.maltophilia 11=S.pneumoniae 12=S.pyogenes(A) 13=S.aureus (MSSA) 14=S.aureus (MRSA) 15=Other	Categorical
micr_res_oth	If other please specify with text		Text
micr_res2	Second bacterium isolated	see values right above	Categorical
micr_res2_oth	If other please specify with text		Text

Topic 8: Imaging

Variable label	Variable description	Values	Type
imagch_d	Date when chest imaging examination was performed	dd/mm/yyyy	Date
imagch_typ	Type of chest imaging	0=x-ray 1=CT 2=MRI	Categorical
imag_bronc	Were bronchiectases present?	0=no 1=yes 2=unknown	Categorical
bronc_loc	if yes where were they located?	1=right upper lobe 2=right middle lobe 3=right lower lobe 4=left upper lobe 5=left middle lobe (in case of situs abnormalities) 6=left lower lobe 7=unknown	Categorical multiple tick list
imag_atelect	Were atelectases present?	0=no 1=yes 2=unknown	Categorical
atelect_loc	if yes where were they located?	1=right upper lobe 2=right middle lobe 3=right lower lobe 4=left upper lobe 5=left middle lobe (in case of situs abnormalities) 6=left lower lobe 7=unknown	Categorical multiple tick list
imag_infiltr	Were infiltrations present??	0=no 1=yes 2=unknown	Categorical
infiltr_loc	if yes where were they located?	1=right upper lobe 2=right middle lobe 3=right lower lobe 4=left upper lobe 5=left middle lobe (in case of situs abnormalities) 6=left lower lobe 7=unknown	Categorical multiple tick list
imag_sin	Did the patient have any sinus imaging?	0=no 1=yes 2=unknown	Categorical
imag_abdom	Did the patient have any abdominal system imaging?	0=no 1=yes 2=unknown	Categorical
imag_cns	Did the patient have any central nervous system imaging?	0=no 1=yes 2=unknown	Categorical
imag_card	Did the patient have any cardiac system imaging?	0=no 1=yes 2=unknown	Categorical

Topic 9: Surgical/Other interventions

Variable label	Variable description	Values	Type
surg_d	Date when lung surgery was performed	dd/mm/yyyy	Date
lungsurg_typ	If yes what type of lung surgery?	1=lobectomy 2=lung transplantation 3=other	Categorical
lungsurg_typ_t	If other please specify		Text
othsurg_typ	If other type of surgery/intervention relevant to PCD was performed what type of intervention was it?	1=tympanostomy tube placement (grommets) 2=adenotomy 3=sinus surgery 4=mastoidectomy 5=polypectomy 6=hearing aid placement 7=other	Categorical

Topic 10: Neonatal period (clinical manifestations and interventions during the first month of life)

Variable label	Variable description	Values	Type
neo_RDS	Did the neonate have neonatal respiratory distress syndrome?	0=no 1=yes 2=unknown	Categorical
neo_cough	Did the neonate have neonatal cough?	0=no 1=yes 2=unknown	Categorical
neo_rhin	Did the neonate have neonatal rhinitis?	0=no 1=yes 2=unknown	Categorical
neo_pneum	Did the neonate have neonatal pneumonia?	0=no 1=yes 2=unknown	Categorical
neo_hydroc	Did the neonate have neonatal hydrocephalus?	0=no 1=yes 2=unknown	Categorical
gest_ag	Gestational age	15-45	Numerical
neo_icu	Was the neonate admitted in neonatal intensive care unit?	0=no 1=yes 2=unknown	Categorical
neo_ox	Was any oxygen given to the neonate?	0=no 1=yes 2=unknown	Categorical
neo_ninv_vent	Was the neonate ventilated non-invasively?	0=no 1=yes 2=unknown	Categorical
neo_inv_vent	Was the neonate ventilated invasively?	0=no 1=yes 2=unknown	Categorical

Topic 11: Family History

Variable label	Variable description	Values	Type
sibl_n	Total number of siblings of the patient	valid numbers	Numeric
pcd_sibl_n	Number of PCD affected siblings	valid numbers	Numeric
pcd_fam_n	Number of other PCD affected family members	valid numbers	Numeric
relat_typ	What kind of relationship does the patient have with the other affected family member?	1=father 2=mother 3=cousin 4=other	Categorical
relat_typ_oth	If other, please write as text		Text
idpat_fam	Patient ID of PCD affected family member	valid numbers	Numeric