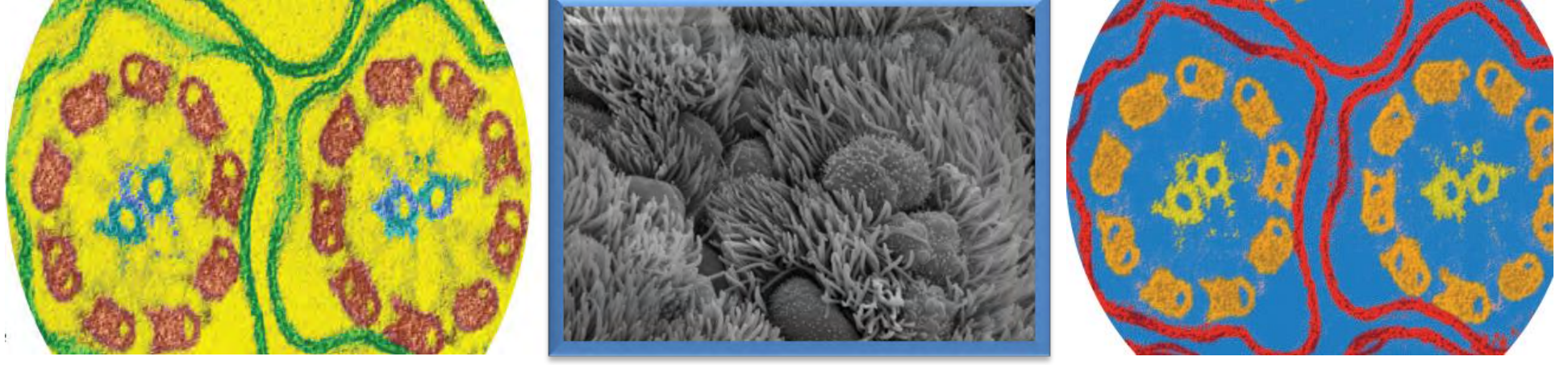


Primary Ciliary Dyskinesia in children

Dr Biju Thomas MD, FRCPCH
Senior Consultant in Paediatric Respiratory Medicine
KK Women's and Children's Hospital
Singapore

KK Women's and Children's Hospital
SingHealth

Image courtesy Nature 2007

Structure of the talk

- Historical perspectives
- Cilia in humans
- Disorders of Motile Cilia – Primary Ciliary Dyskinesia
- Clinical features of PCD
- Diagnosis of PCD in the molecular age
- Management of PCD

History

1683: Anton de Hide – first description of ciliary movement

History

1834

- **Purkinje and Valentin: discovered mammalian cilia**

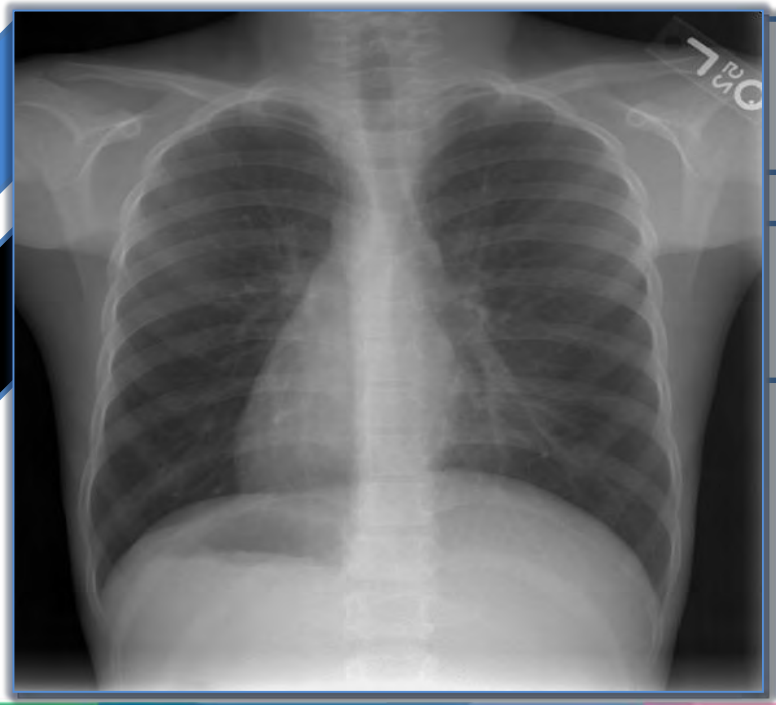
History

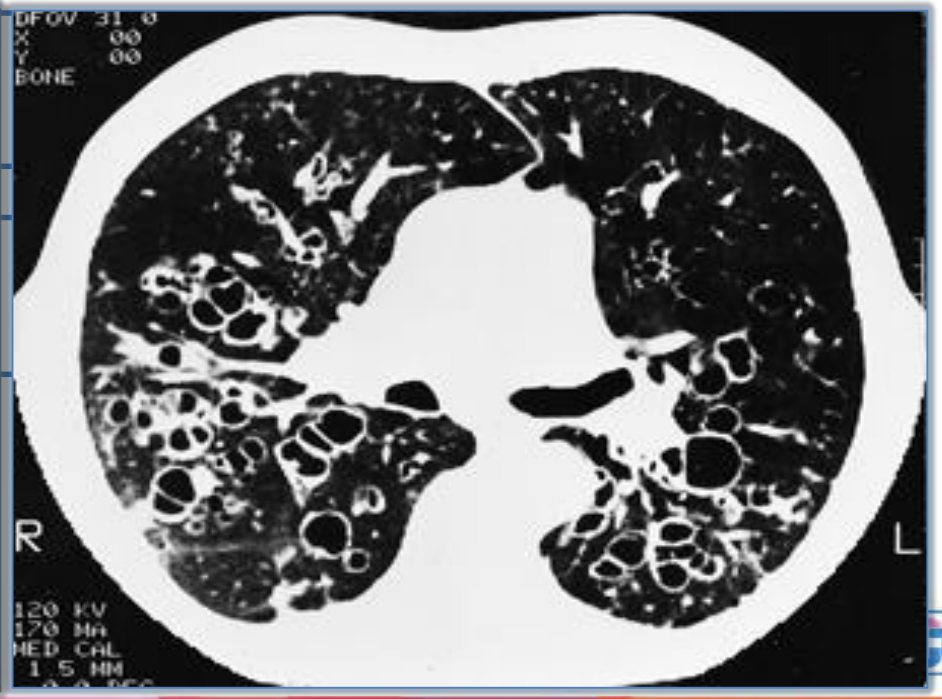
1834

- **Purkinje and Valentin: discovered mammalian cilia**

1904

- **Siewert**
- **Situs inversus and Bronchiectasis**





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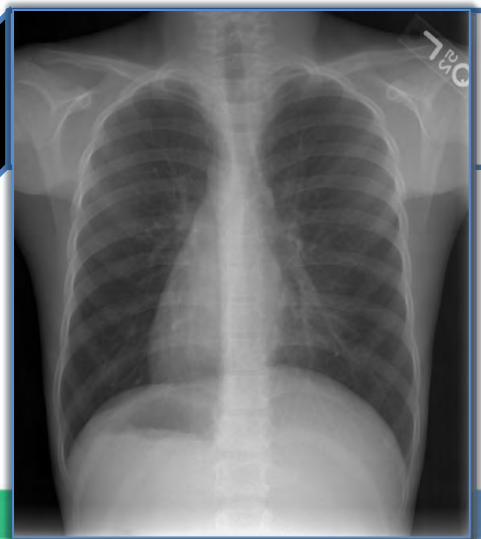
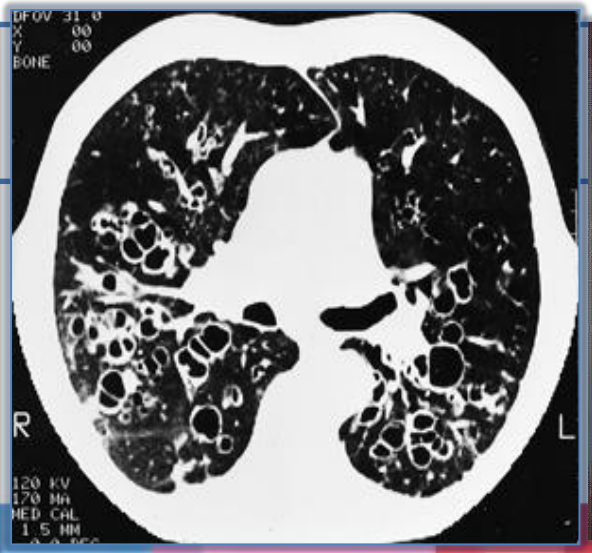
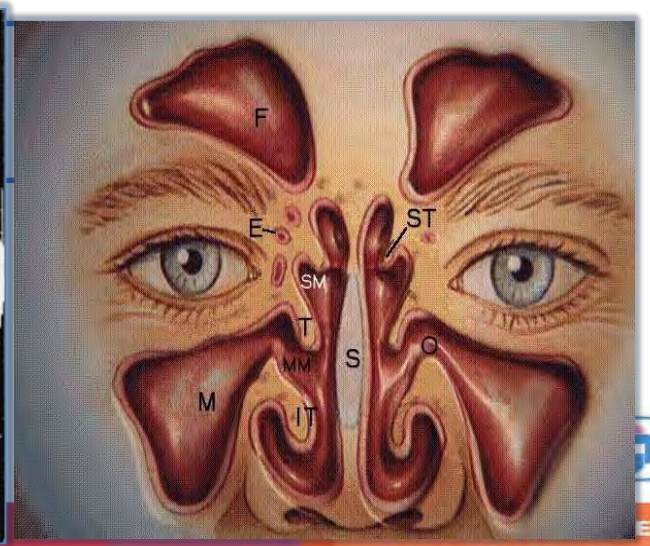
1933
- Kartagener
- Situs inversus, Bronchiectasis, Sinusitis







History

1834
- Purkinje and Valentin: discovered mammalian cilia

1904
- Siewert
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1933
- Kartagener
- Situs inversus, Bronchiectasis, Sinusitis

1976
- Afzelius and Pederson
- Abnormal ciliary movement as the cause of the disorder

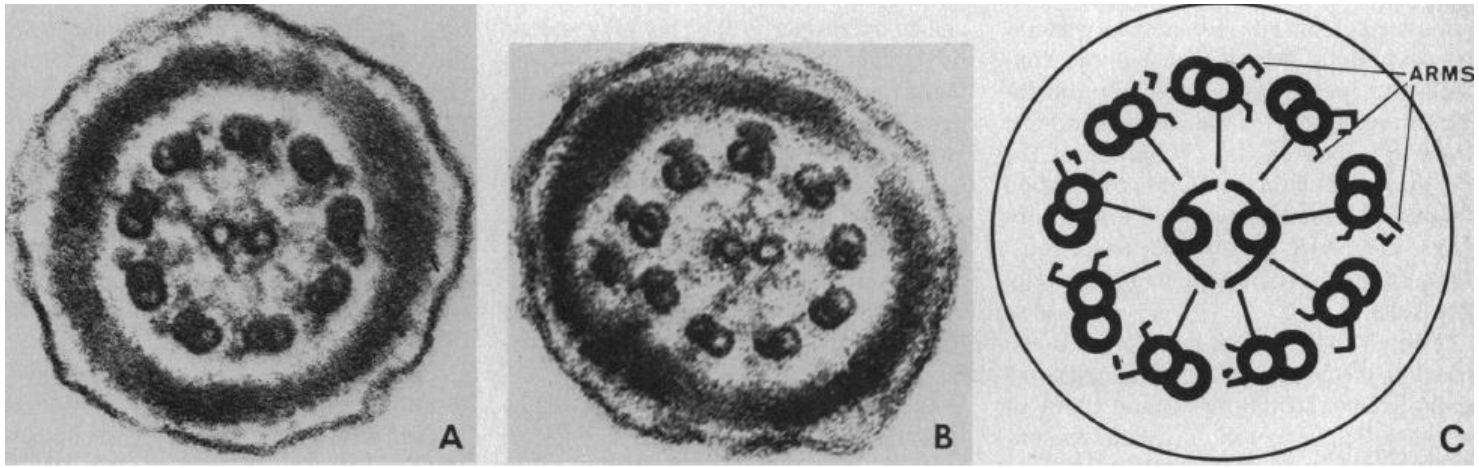
Afzelius BA. Science 1976; 193: 317-9
Pedersen & Mygind. Nature 1976; 262: 494-5

A Human Syndrome Caused by Immotile Cilia

Abstract. *Four subjects who produced immotile sperm were studied. In three of the subjects, who had frequent bronchitis and sinusitis, there was no mucociliary transport, as measured by tracheobronchial clearance. Electron microscopy indicated that cilia from cells of these patients lack dynein arms.*

BJÖRN A. AFZELIUS

Wenner-Gren Institute,
S-113 45 Stockholm, Sweden



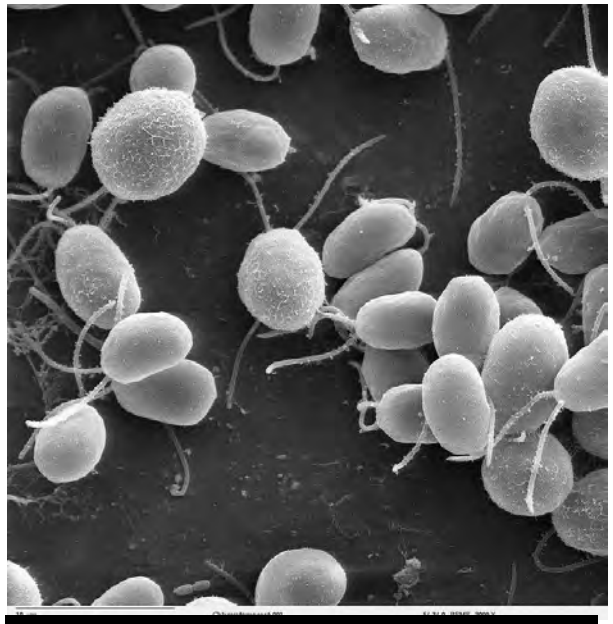
Nature Vol. 262 August 5 1976

nature

Absence of axonemal arms in nasal mucosa cilia in Kartagener's syndrome

HENNING PEDERSEN

Understanding cilia biology and genetics



Chlamydomonas Reinhardtii

Frog (Xenopus) larvae

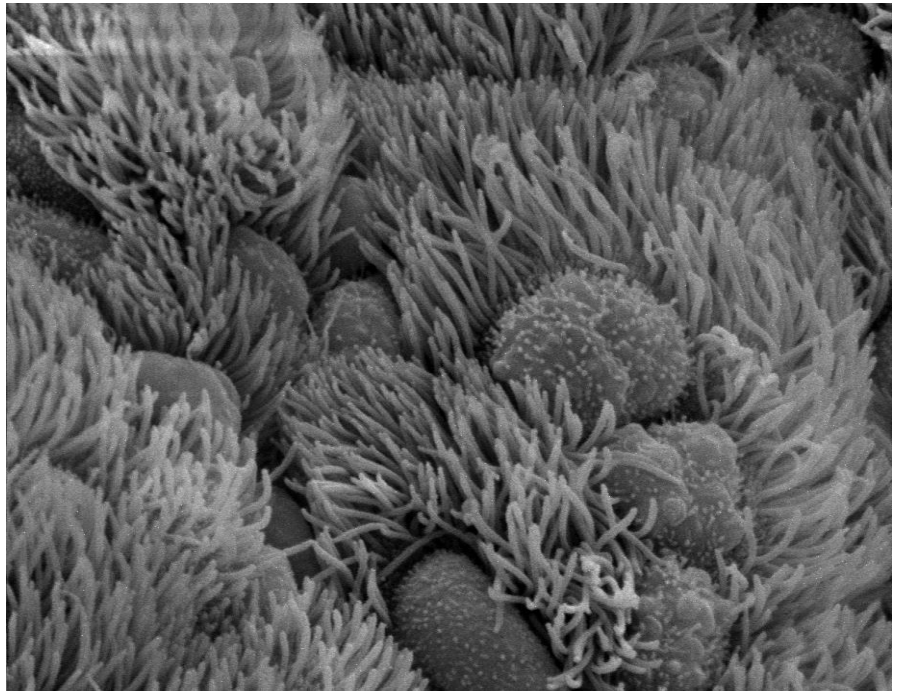
Drosophilia melanogaster

Zebra fish embryo

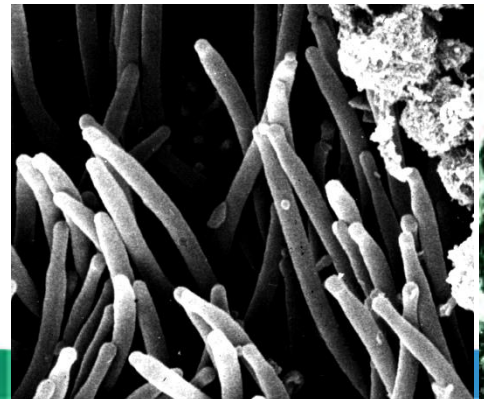
Image courtesy J Cell Biology 1996; Ninghui Shi

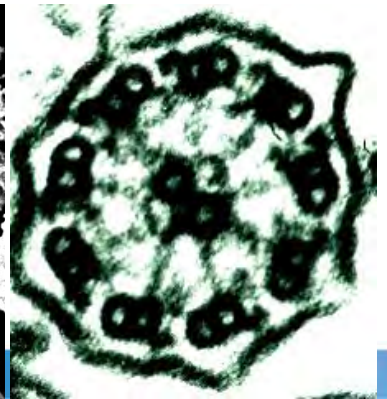
Ciliated respiratory epithelium

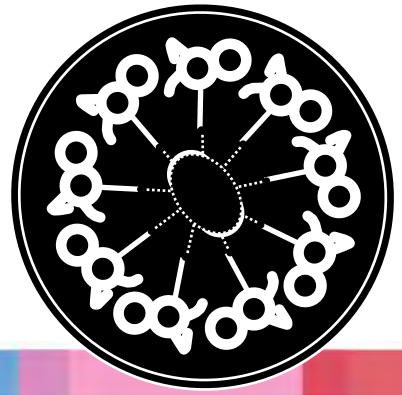
- **Human respiratory cilia**



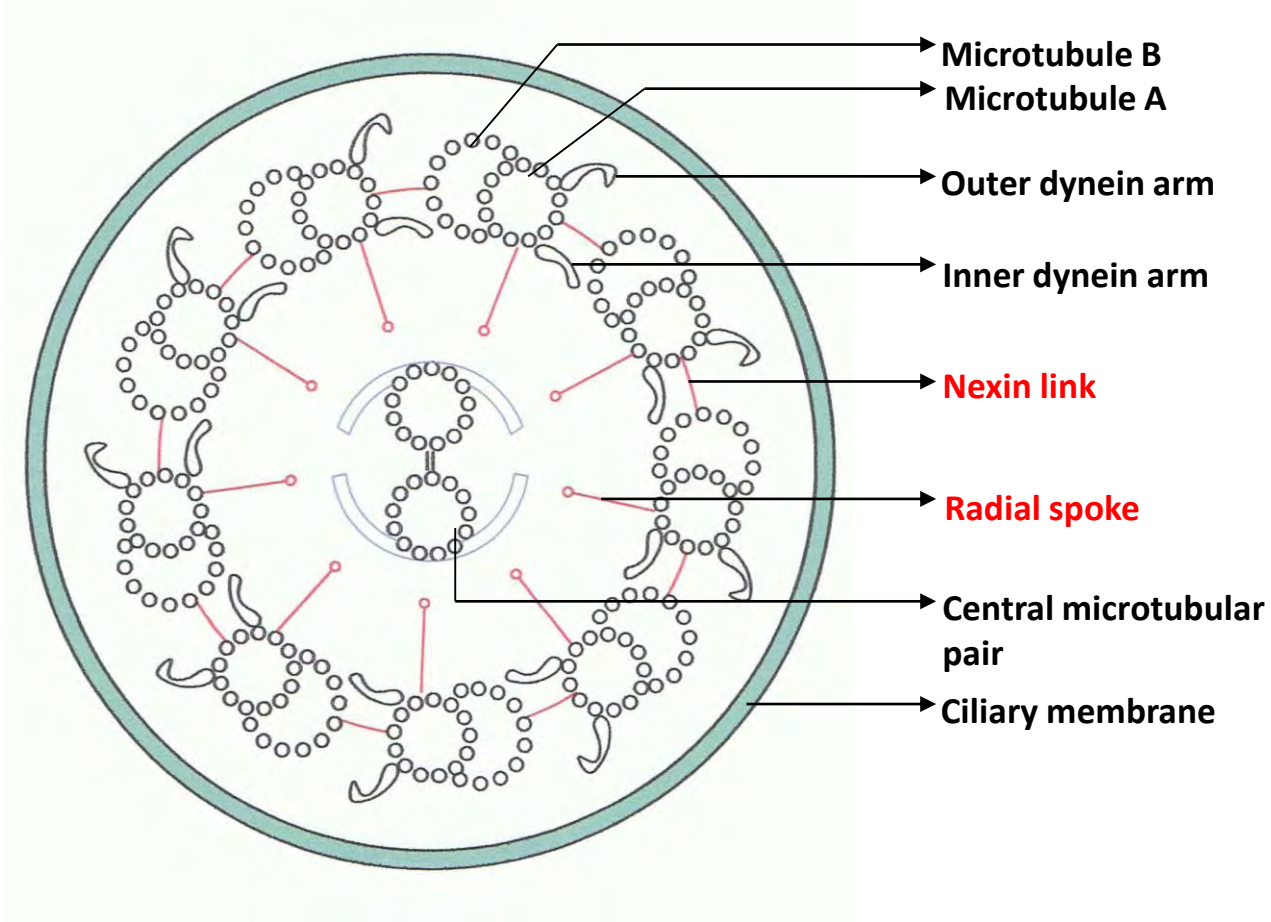
- ~ 200 cilia/cell
- 6µm long
- 0.3µm wide
- Frequency 10-14 Hz





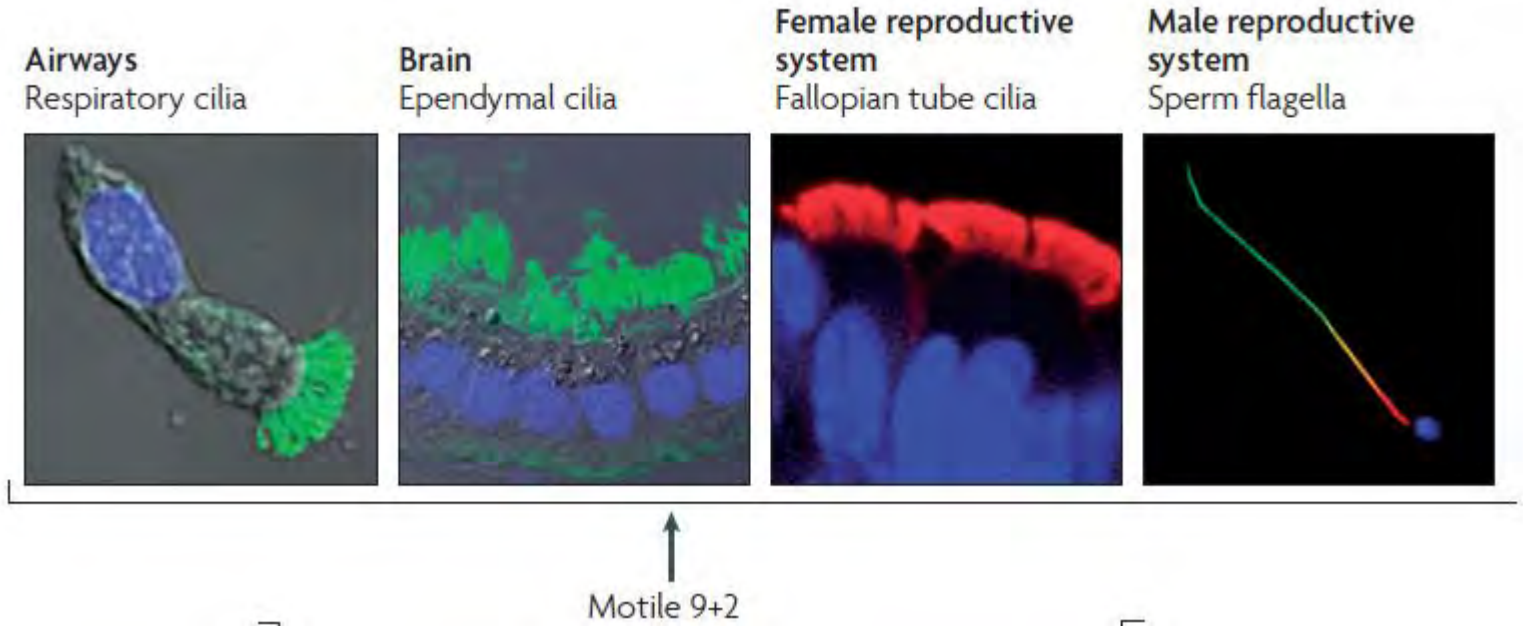


Cross section of human respiratory cilium



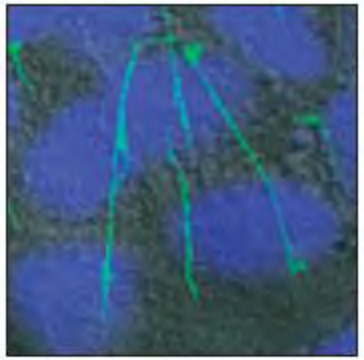
www.nature.com/reviews/molcellbio
880 | NOVEMBER 2007 | VOLUME 8

Motile Cilia in Humans (9+2)

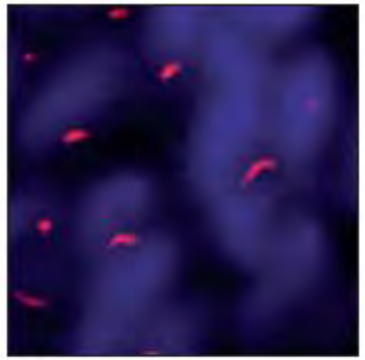


www.nature.com/reviews/molcellbio
880 | NOVEMBER 2007 | VOLUME 8

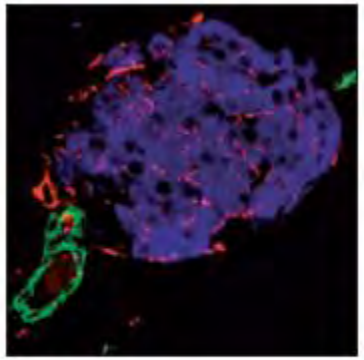
Non-motile Cilia in Humans (9+0)



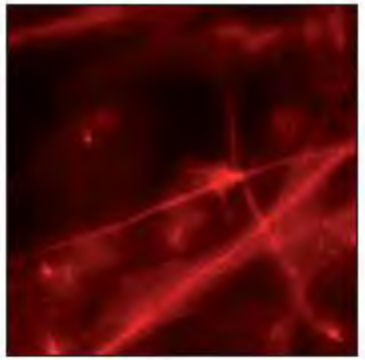
Kidney
Renal cilia



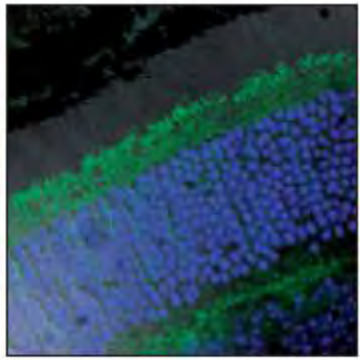
Bile duct
Cholangiocyte cilia



Pancreas
Pancreatic duct cilia

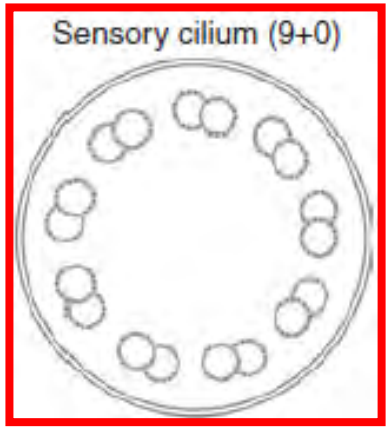


Bone/cartilage
Osteocyte/
chondrocyte cilia



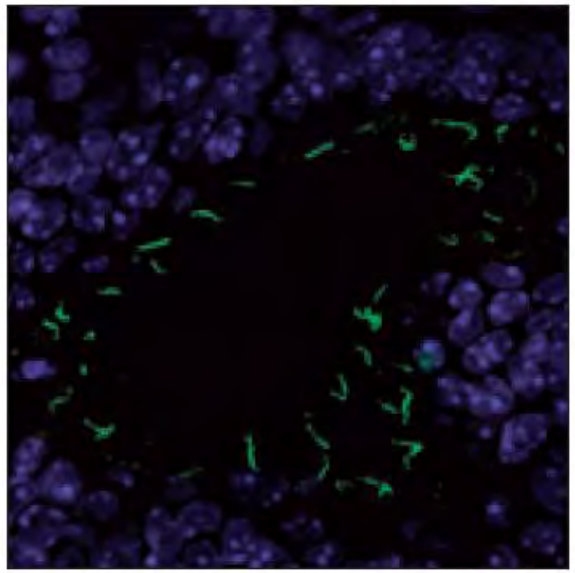
Eye
Photoreceptor
connecting cilia

Non-motile 9+0

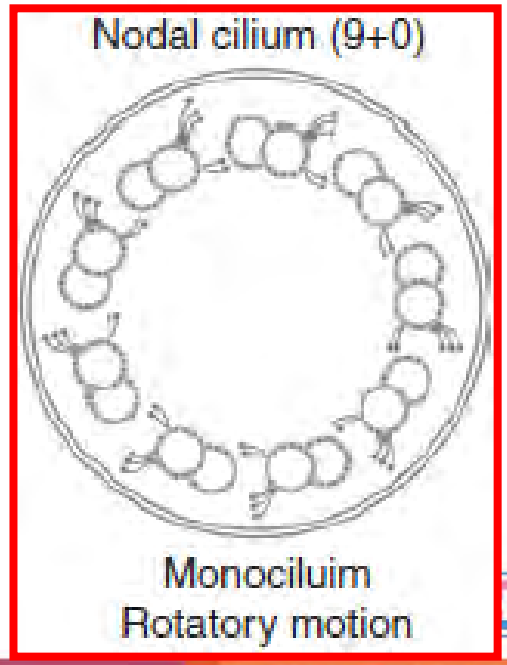


Motile Cilia in Humans (9+0)

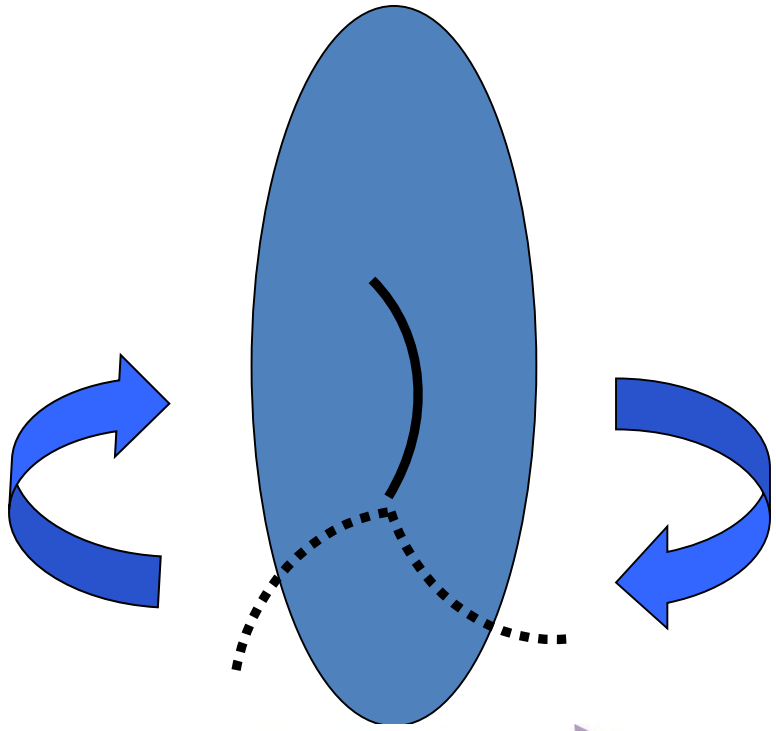
Embryo
Nodal cilia

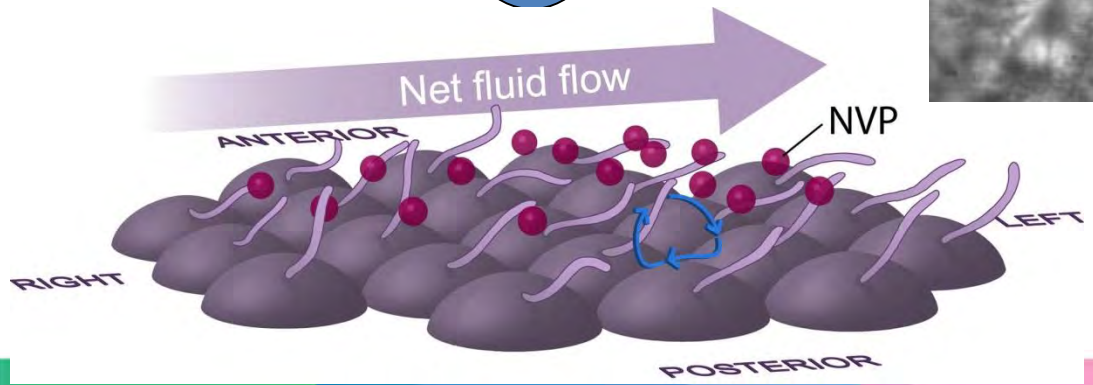


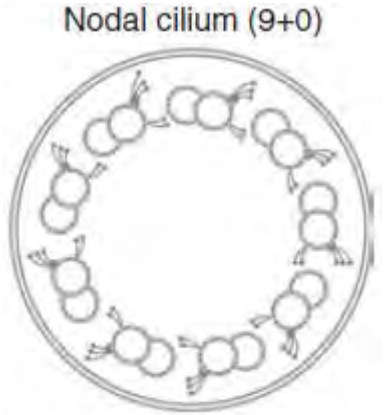
← Motile 9+0



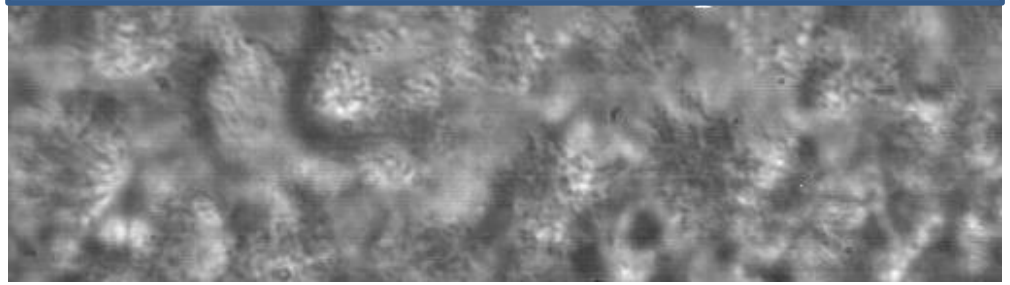
Determination of Situs: Role of Nodal cilia





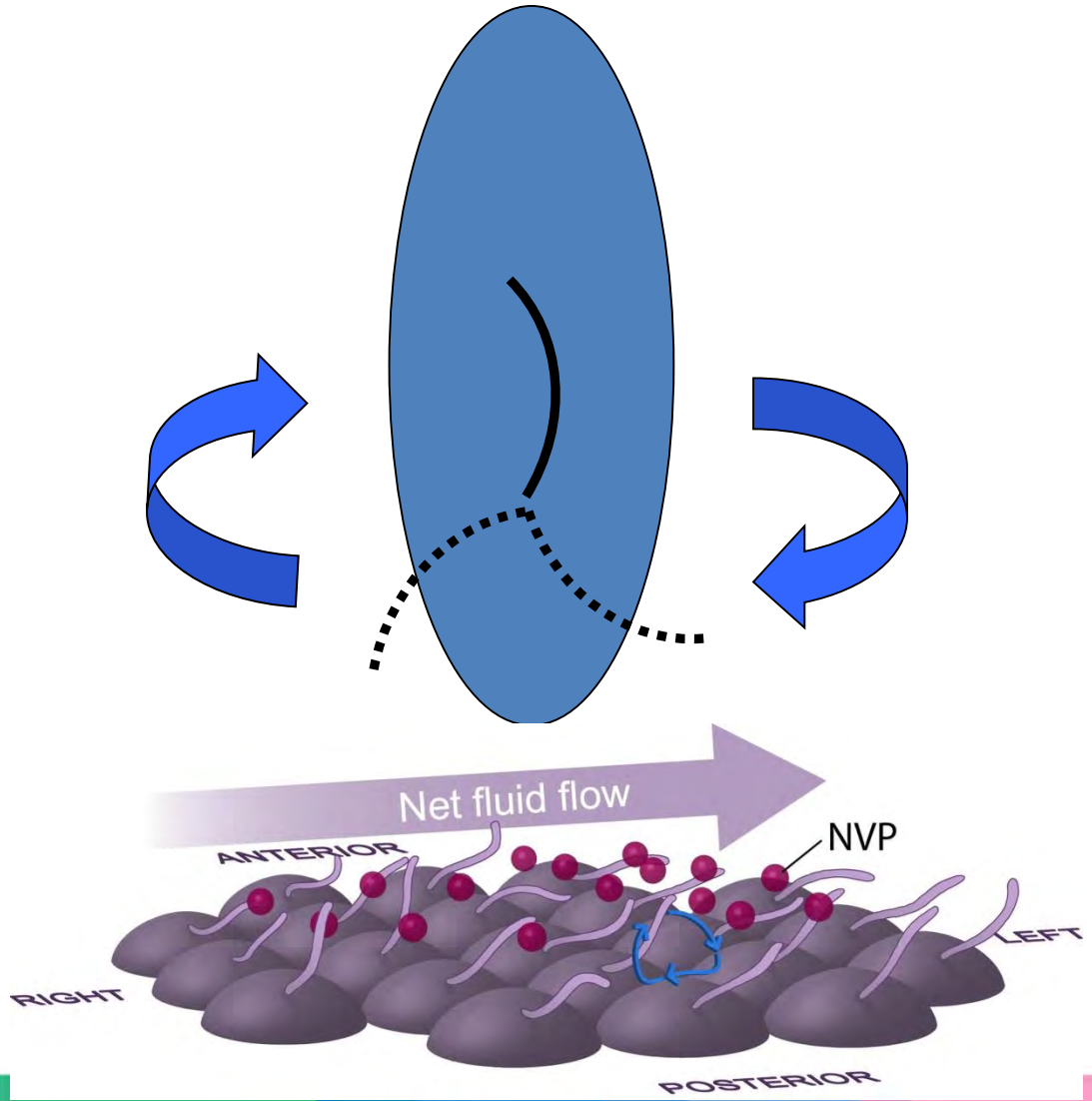


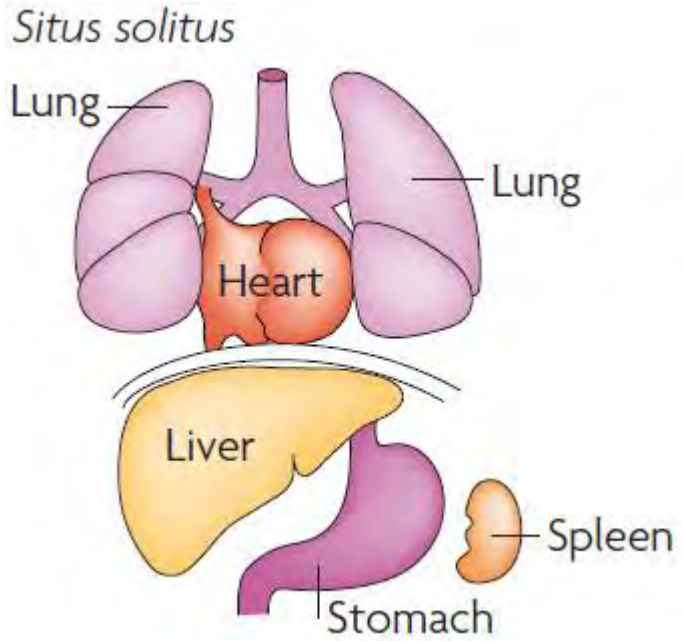
Circular/Rotatory motion of cilia



Nonaka Nature:2002

Determination of Situs: Role of Nodal cilia

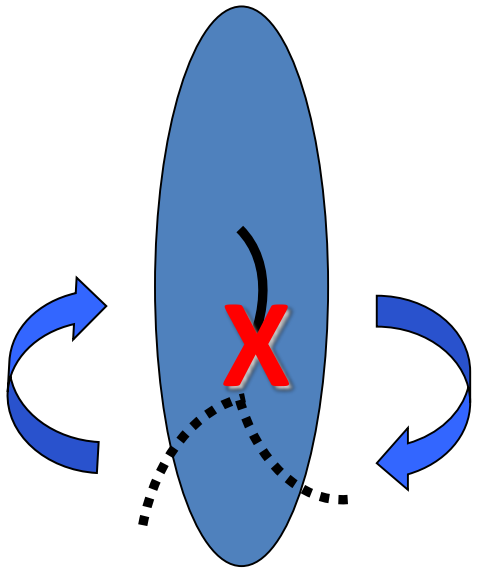


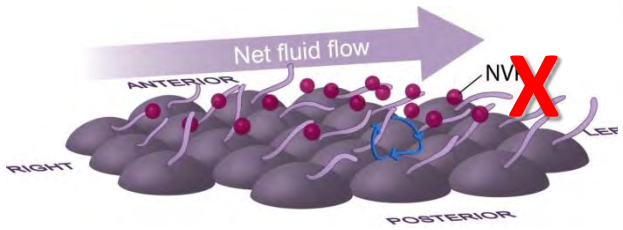


Fluid current → molecular gradients & Activation of mechanosensors

Nonaka Nature:2002

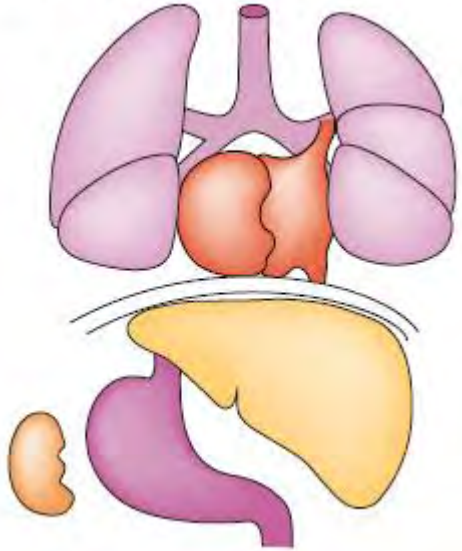
Situs Inversus: Nodal Hypothesis





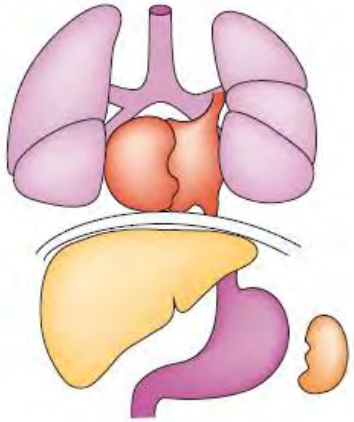
Nonaka Nature:2002

Situs inversus totalis

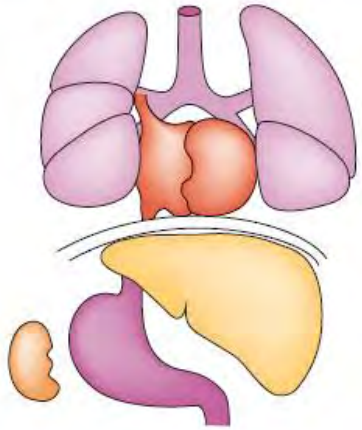


Left isomerism (polysplenia)

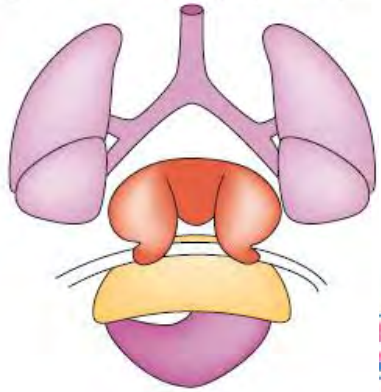
Situs inversus thoracalis



Situs inversus abdominalis



Right isomerism (asplenia)



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SingHealth

Disorders of Cilia

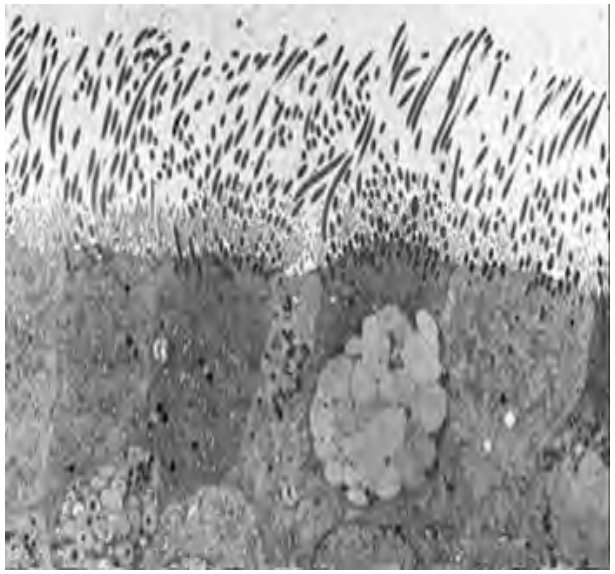
Disorders of Primary Cilia

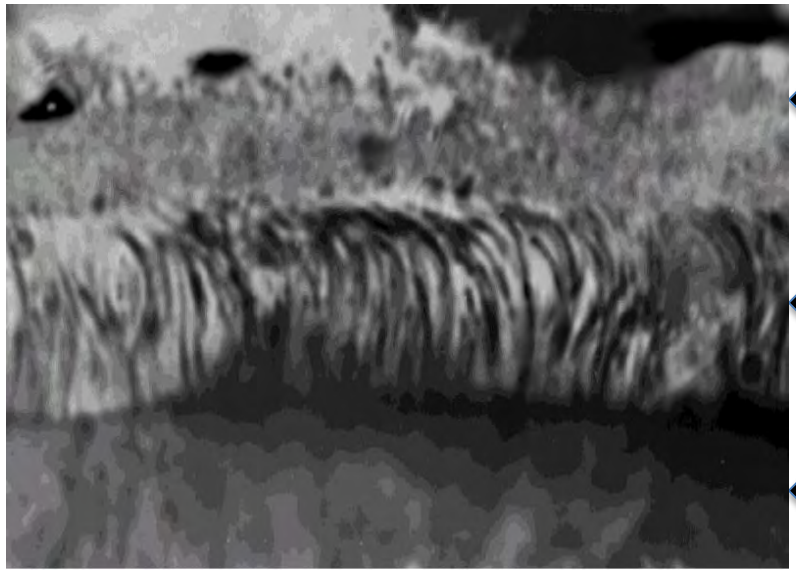
"Ciliopathies"

Disorders of Motile Cilia:

PCD

Normal mucociliary clearance





Normal mucociliary clearance

Normal mucociliary clearance

Ciliary Dyskinesia

- **Primary**
- **Secondary**

Primary Ciliary Dyskinesia: Epidemiology

- Prevalence: 1:15,000 (Caucasian)
  Higher in South Asians* (1: 2200) – UK study
  Higher with parental consanguinity

- Autosomal recessive, Rarely X-linked or Dominant

- Significantly under diagnosed

**C O'Callaghan. Ped Pulmonology 2004*

Clinical Presentation of PCD

- Infancy – late adulthood
- Manifestations vary with age
- Symptoms overlap with common respiratory diseases
- Late diagnosis is common, mean age at diagnosis: >4yr
- 30% have established bronchiectasis at diagnosis

Coren ME. Acta Paediatr 2002; 91: 667-669
Bush A. ERJ 1998; 12: 982-988, Noone PG. AJRCCM 2004; 169: 459-467
Hossain T. J Perinatology 2003; 23: 684-687

Age-related prevalence of clinical features in PCD

PCD clinical feature	Youngest age when feature present in >50% of PCD	Youngest age when feature present in >80% of PCD
Neonatal respiratory distress	12hr of life	24hr of life
Organ laterality defects	Neonatal	
Recurrent otitis media with effusion	Infancy	Infancy
Year-round, daily wet cough	Infancy	Infancy
Year-round, daily nasal congestion	Infancy	Infancy
Recurrent LRTI	Infancy	Preschool
Chronic pansinusitis	Preschool	School age
Bronchiectasis	School age	Adult
Male infertility	-	Adults

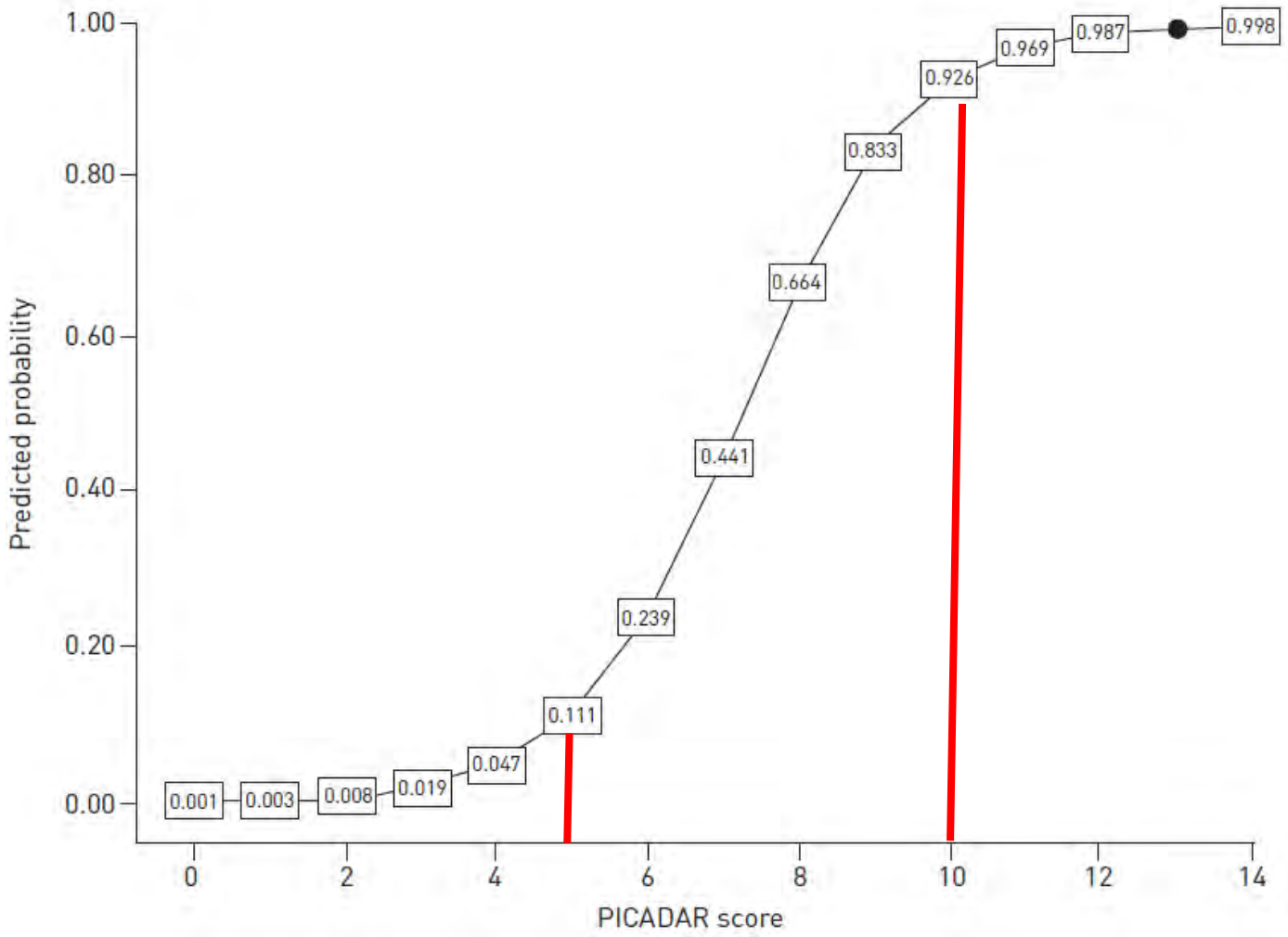
Knowles MR, AJRCCM 2013

PrImary CiliaAry DyskinesiA Rule

PICADAR		
Does the patient have a daily wet cough that started in early childhood?	**Yes** – complete PICADAR **No – STOP**. PICADAR is not designed for patients without a wet cough	
1. Was the patient born pre-term or full term?	Term	2
2. Did the patient experience chest symptoms in the neonatal period (*e.g.* tachypnoea, cough, pneumonia)?	Yes	2
3. Was the patient admitted to a neonatal unit?	Yes	2
4. Does the patient have a situs abnormality (situs inversus or heterotaxy)?	Yes	4
5. Does the patient have a congenital heart defect?	Yes	2
6. Does the patient have persistent perennial rhinitis?	Yes	1
7. Does the patient experience chronic ear or hearing symptoms (*e.g.* glue ear, serous otitis media, hearing loss, ear perforation)?	Yes	1
	Total score =	

Behan L et al. ERJ 2016

PICADAR ≥ 5		
	Southampton (UK)	Brompton (UK)
Sensitivity	90%	86%
Specificity	75%	73%



PICADAR Probability Curve

Behan L et al. ERJ 2016

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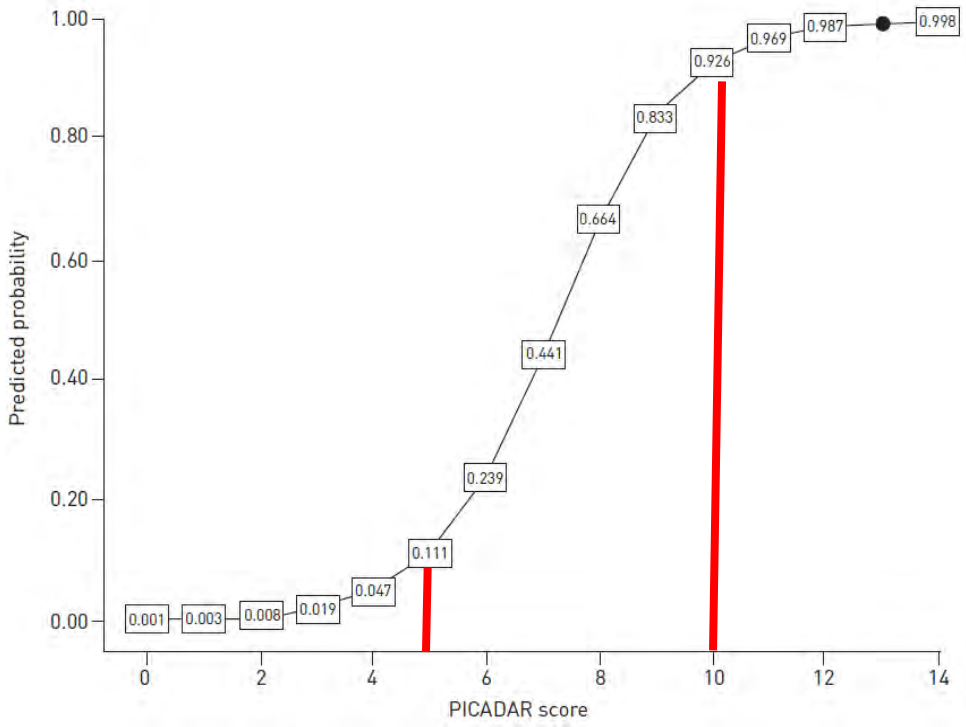

Probability of PCD	
PICADAR ≥5	PICADAR ≥10
>11%	>90%

Behan L et al. ERJ 2016

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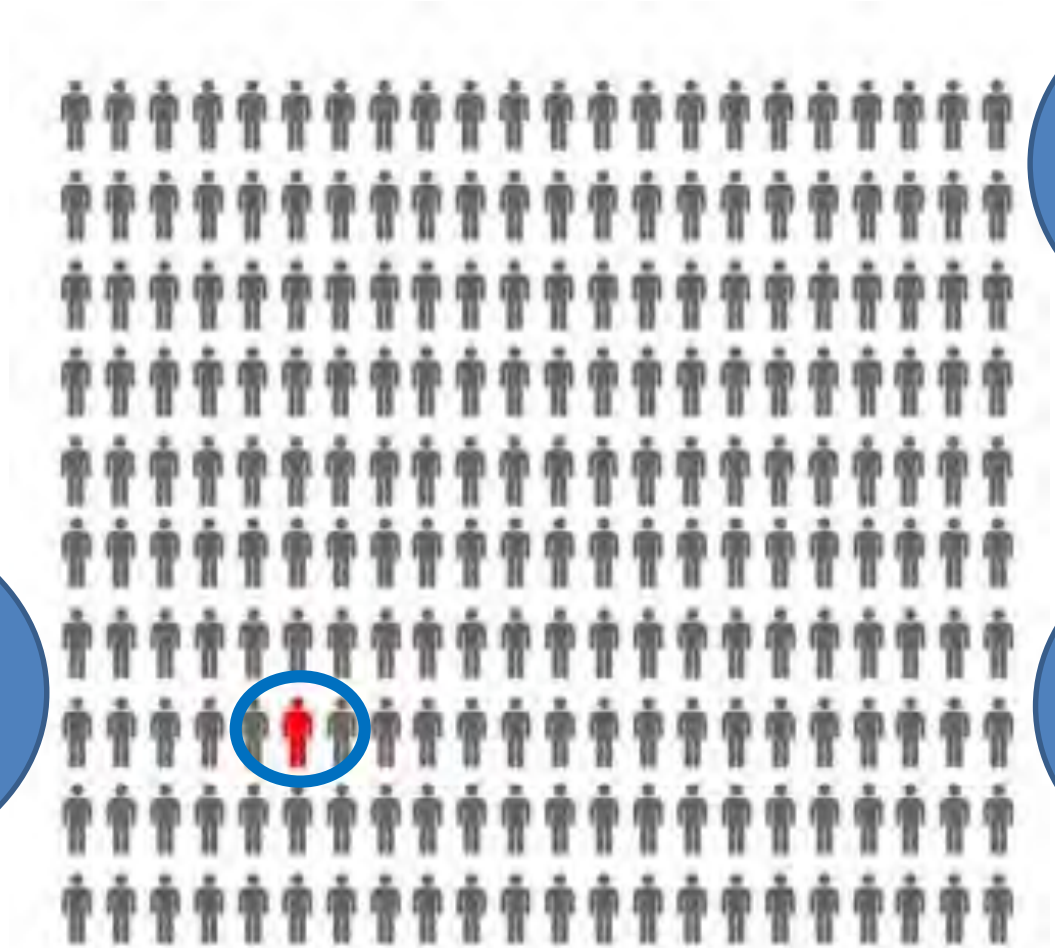
SingHealth

Diagnosis of PCD in the molecular age

MAKING A DIAGNOSIS

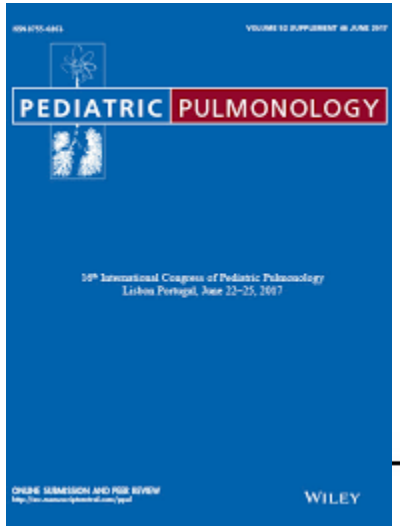
Rare

Single Symptoms - nonspecific



No Gold Standard Test

Tests
-Expensive
- Equipment
-Expertise



Pediatric Pulmonology 51:115–132 (2016)

State of the Art —

Diagnosis, Monitoring, and Treatment of Primary Ciliary Dyskinesia: PCD Foundation Consensus Recommendations Based on State of the Art Review

Adam J. Shapiro, MD,[1]* Maimoona A. Zariwala, PhD,[2] Thomas Ferkol, MD,[3] Stephanie D. Davis, MD,[4] Scott D. Sagel, MD, PhD,[5] Sharon D. Dell, MD,[6] Margaret Rosenfeld, MD,[7] Kenneth N. Olivier, MD,[8§] Carlos Milla, MD,[9] Sam J. Daniel, MD,[10] Adam J. Kimple, MD,[11] Michele Manion,[12] Michael R. Knowles, MD,[13] and Margaret W. Leigh, MD,[14] for the Genetic Disorders of Mucociliary Clearance Consortium

European Respiratory Society guidelines for the diagnosis of primary ciliary dyskinesia

Jane S. Lucas[1,2], Angelo Barbato[3], Samuel A. Collins[1,2], Myrofora Goutaki[4,5], Laura Behan[1,2], Daan Caudri[6,7], Sharon Dell[8,9], Ernst Eber[10], Estelle Escudier[11,12], Robert A. Hirst[13], Claire Hogg[14], Mark Jorissen[15], Philipp Latzin[5], Marie Legendre[11,12], Margaret W. Leigh[16], Fabio Midulla[17], Kim G. Nielsen[18], Heymut Omran[19], Jean-Francois Papon[20,21], Petr Pohunek[22], Beatrice Redfern[23], David Rigau[24], Bernhard Rindlisbacher[25], Francesca Santamaria[26], Amelia Shoemark[14], Deborah Snijders[3], Thomy Tonia[4], Andrea Titieni[19], Woolf T. Walker[1,2], Claudius Werner[19], Andrew Bush[14] and Claudia E. Kuehni[4]

Cite this article as: Lucas JS, Barbato A, Collins SA, *et al.* European Respiratory Society guidelines for the diagnosis of primary ciliary dyskinesia. *Eur Respir J* 2017; 49: 1601090 [https://doi.org/10.1183/13993003.01090-2016].

AMERICAN THORACIC SOCIETY DOCUMENTS

Diagnosis of Primary Ciliary Dyskinesia

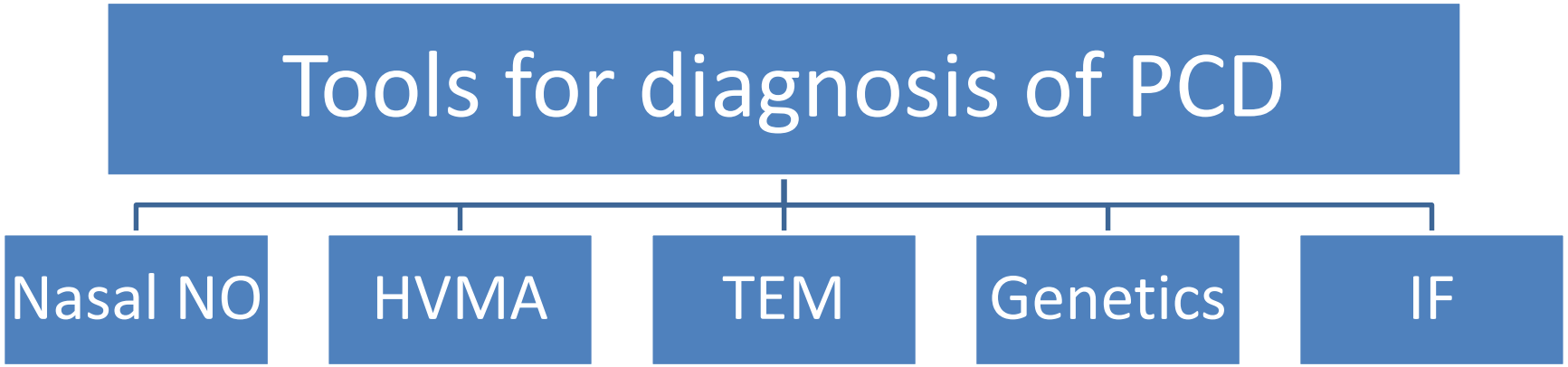
An Official American Thoracic Society Clinical Practice Guideline: Executive Summary

Adam J. Shapiro, Stephanie D. Davis, Deepika Polineni, Michele Manion, Margaret Rosenfeld, Sharon D. Dell, Mark A. Chilvers, Thomas W. Ferkol, Maimoona A. Zariwala, Scott D. Sagel, Maureen Josephson, Lucy Morgan, Ozge Yilmaz, Kenneth N. Olivier, Carlos Milla, Jessica E. Pittman, M. Leigh Anne Daniels, Marcus Herbert Jones, Ibrahim A. Janahi, Stephanie M. Ware, Sam J. Daniel, Matthew L. Cooper, Lawrence M. Nogee, Billy Anton, Tori Eastvold, Lynn Ehrne, Elena Guadagno, Michael R. Knowles, Margaret W. Leigh, and Valery Lavergne; on behalf of the American Thoracic Society Assembly on Pediatrics

THIS OFFICIAL CLINICAL PRACTICE GUIDELINE OF THE AMERICAN THORACIC SOCIETY WAS APPROVED MAY 2018

Am J Respir Crit Care Med Vol 197, Iss 12, pp 1524–1533, Jun 15, 2018

No Gold Standard Diagnostic test

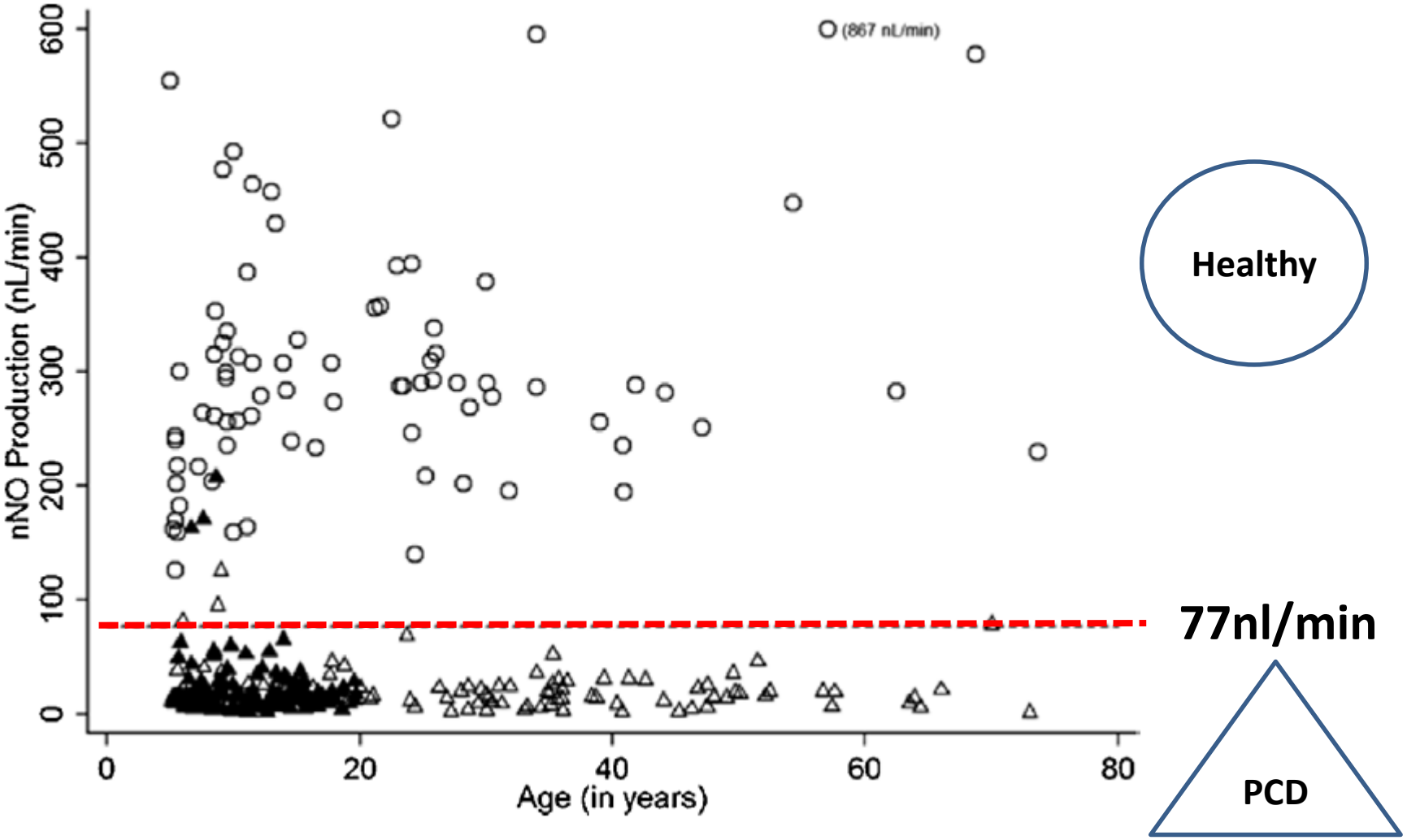


NO = Nitric Oxide
HVMA = High speed Video Microscopy Analysis
TEM = Transmission Electron Microscopy
IF = Immunofluorescence

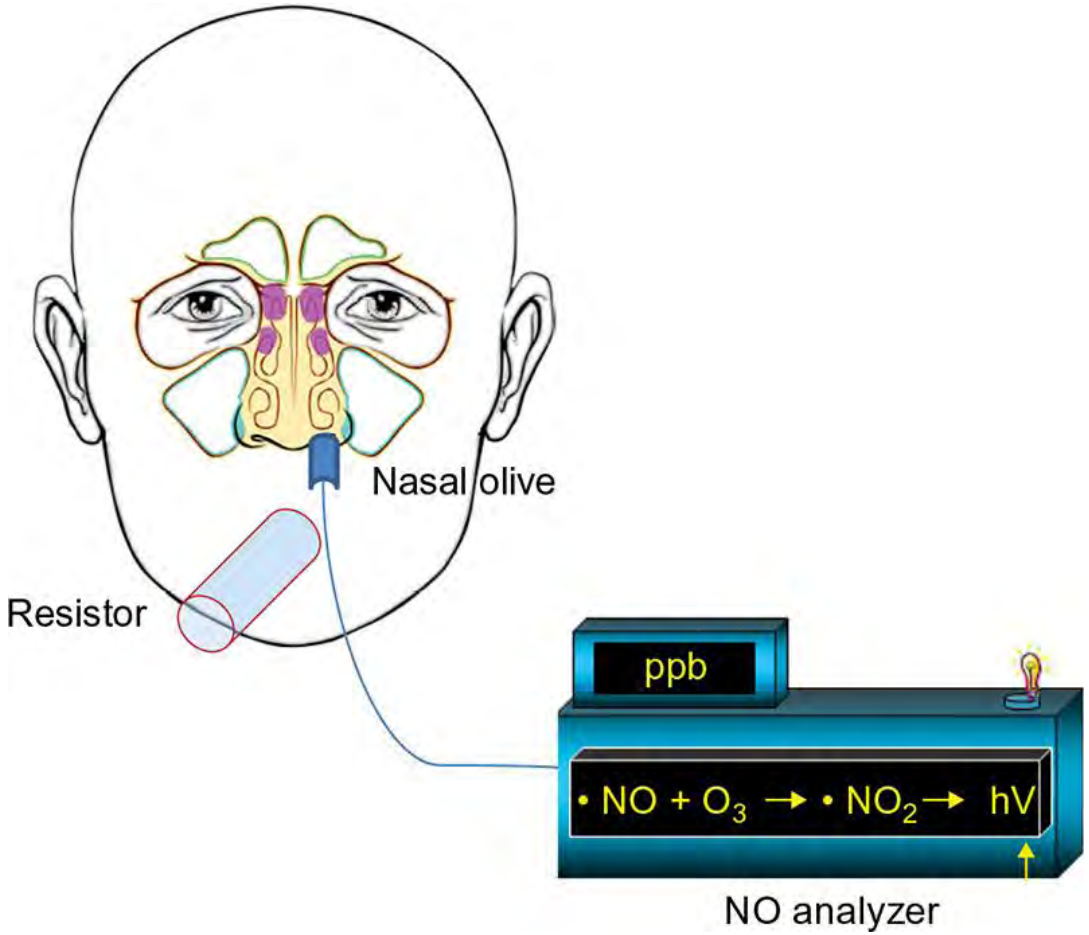
Nasal NO

nNO	Study Population	Sampling method	Threshold nl/min	Sensitivity	Specificity
Marthin 2011	117 referrals 14 PCD	Breath holding with oral exhalation	72	1.0	0.94
Leigh 2013	155 referrals 71 PCD	Oral exhalation, velum closure	77	0.99	0.75
Beydon 2015	86 referrals 49 PCD	Velum closure	82	0.91	0.86
Jackson 2015	301 referrals 34 PCD	Breath Hold, velum closure	30	0.90	0.95

Nasal NO in PCD and healthy controls



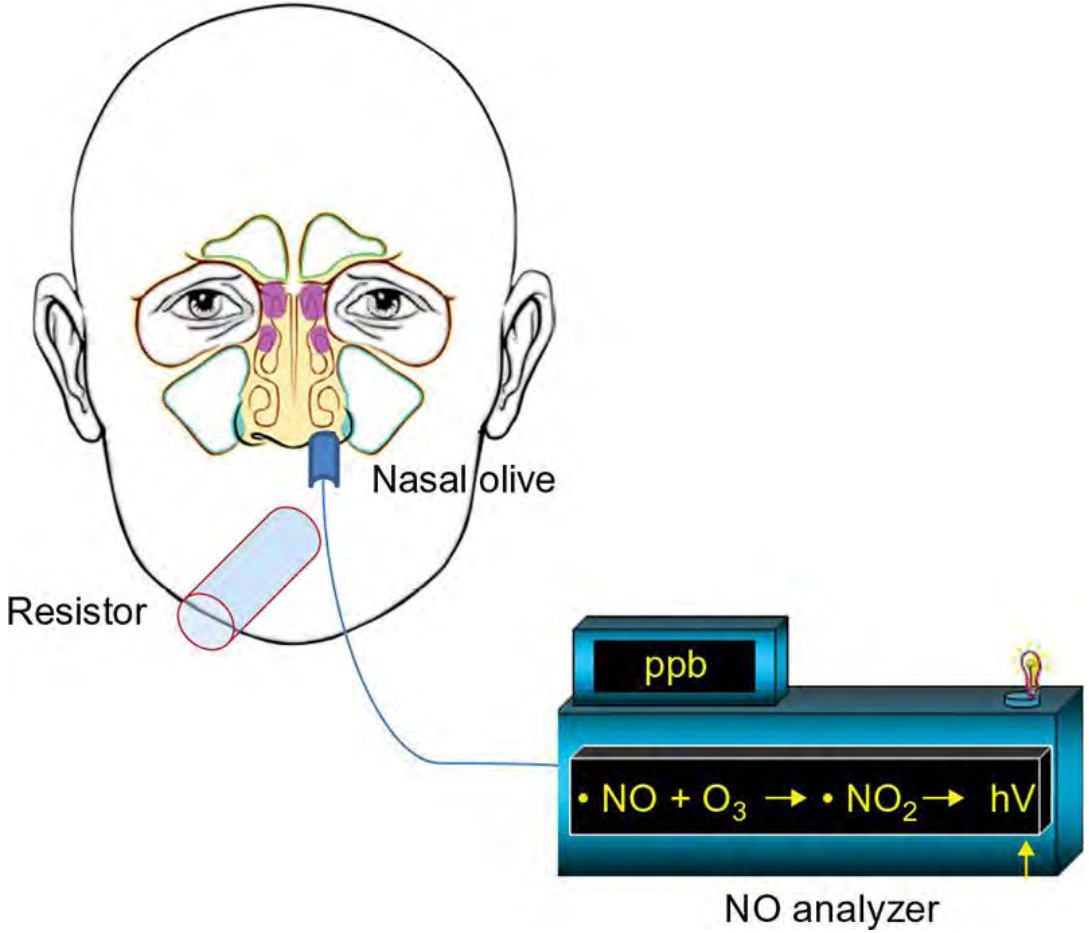
Leigh MW et al. Ann Am Thorac Soc 2013



NIOX MINO® Nasal

Chemiluminescence analyser

Electrochemical analyser

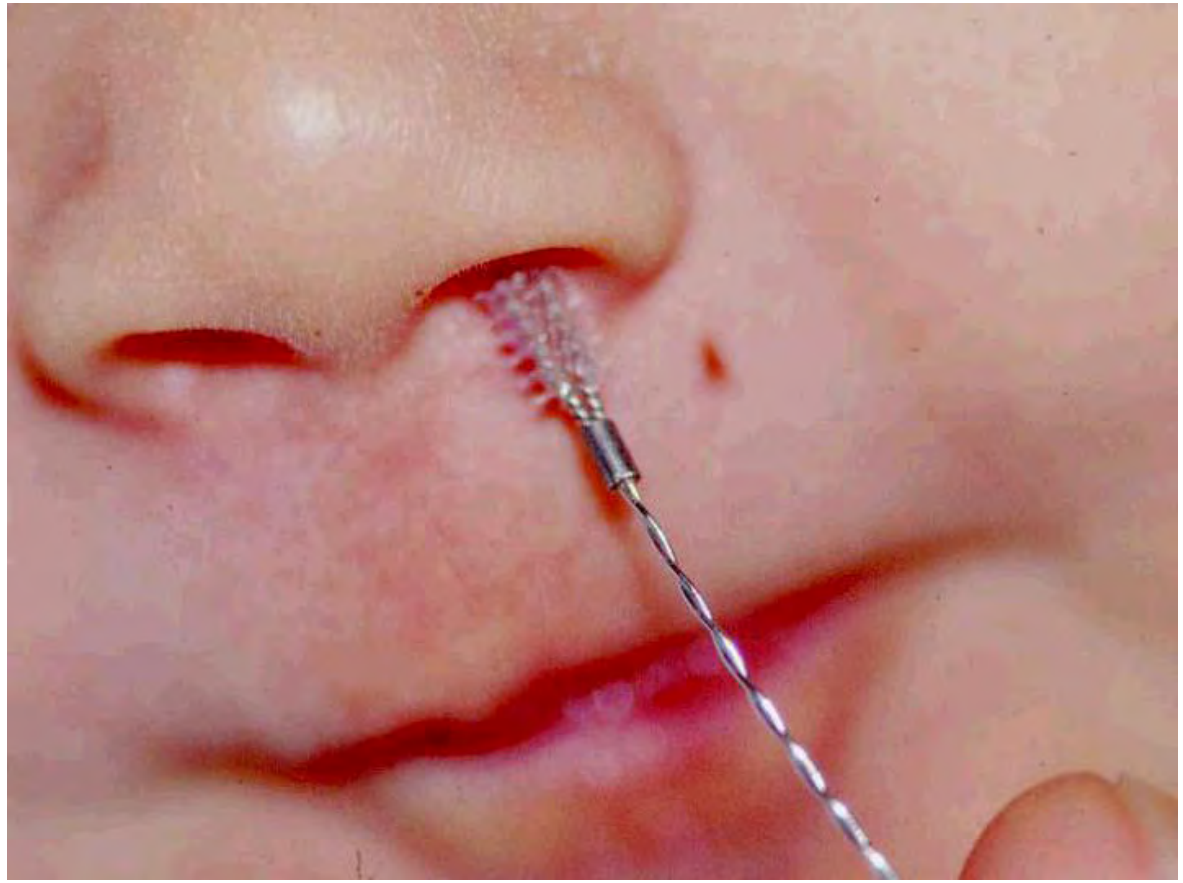


NIOX MINO® Nasal

Chemiluminescence analyser

Electrochemical analyser

Obtaining sample: Nasal brushing
For HVMA, TEM and IF



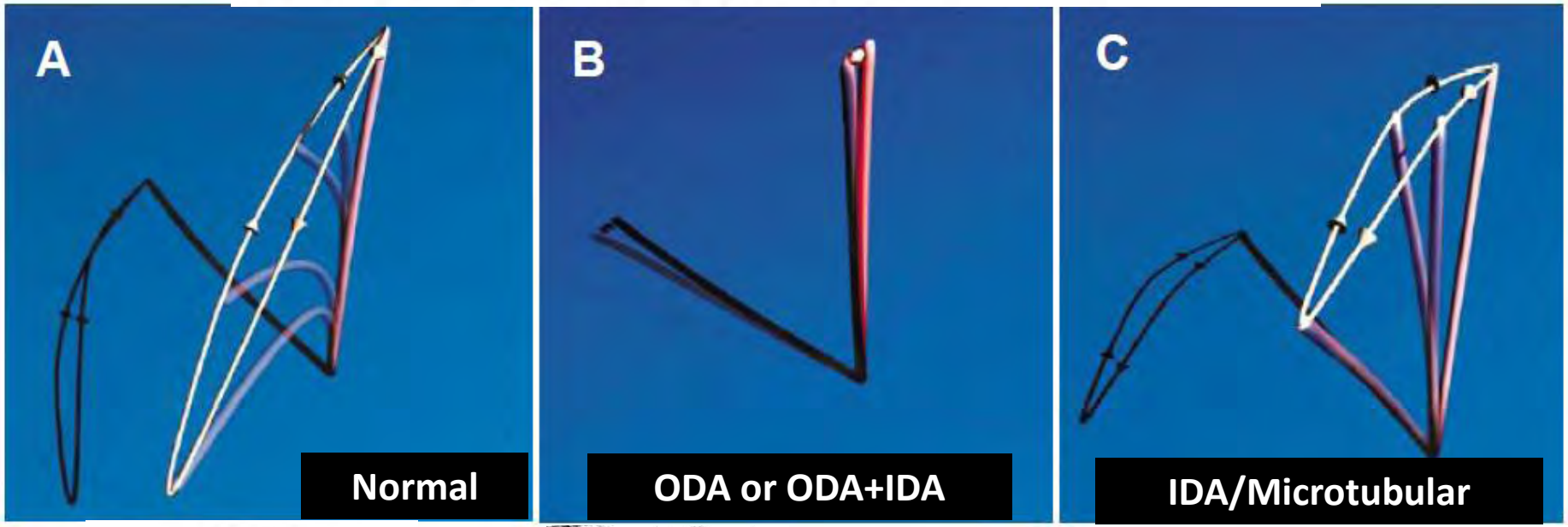
New diagnostic service for Primary Ciliary Dyskinesia at KKH

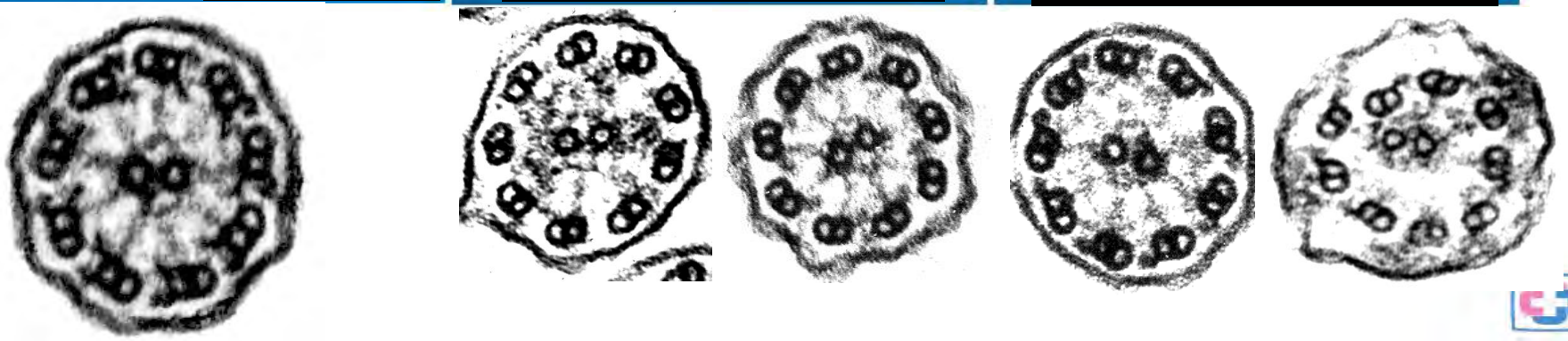
Dr Biju Thomas
Consultant, Respiratory Medicine Service, Department of Paediatrics, KK Women's and Children's Hospital

Ciliary beat pattern is associated with specific ultrastructural defects in primary ciliary dyskinesia

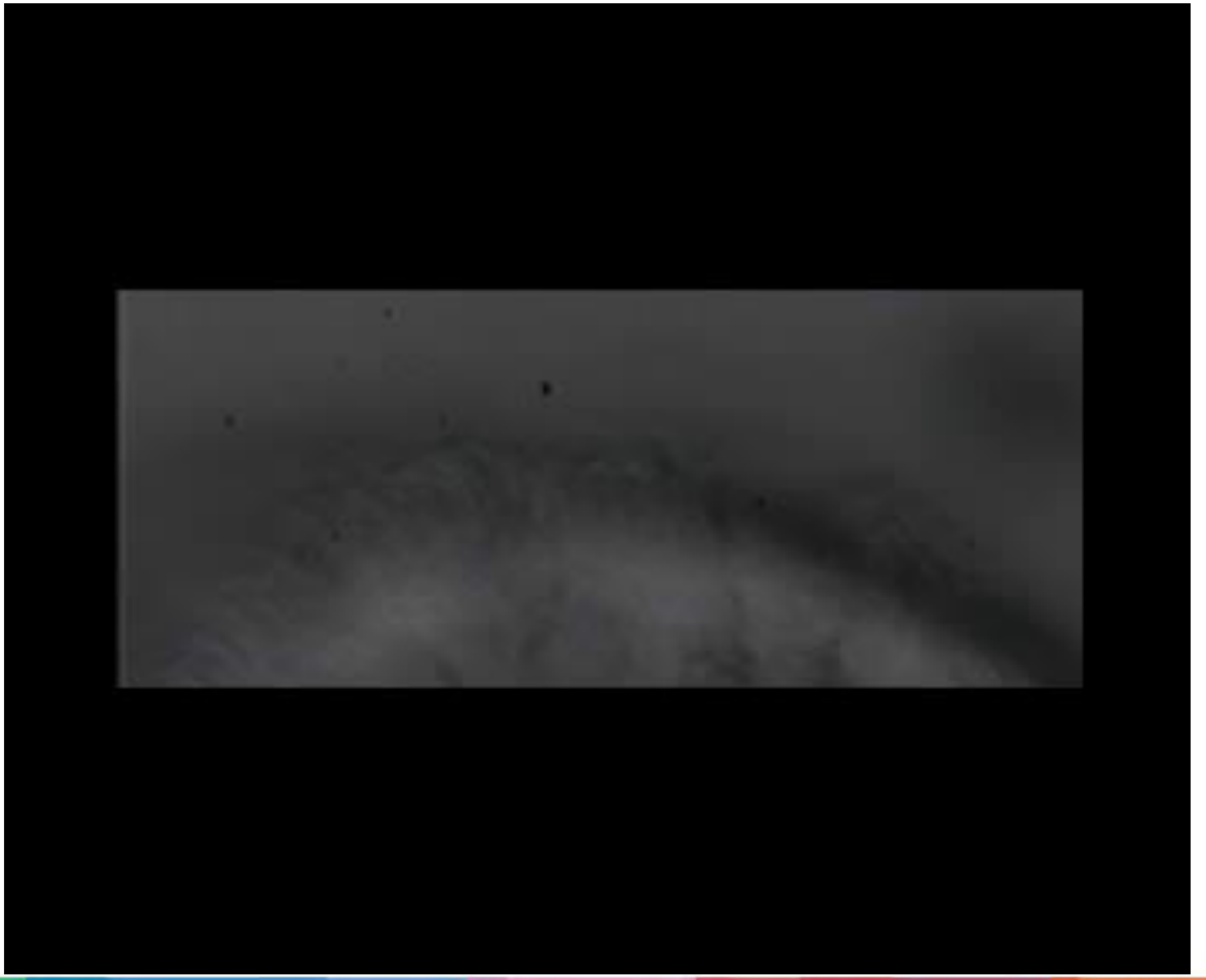
JACI 2003

Mark A. Chilvers, MRCPCH, Andrew Rutman, and Christopher O'Callaghan, FRCPCH, PhD *Leicester, United Kingdom*



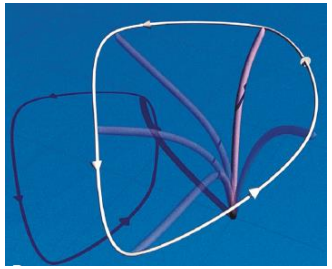


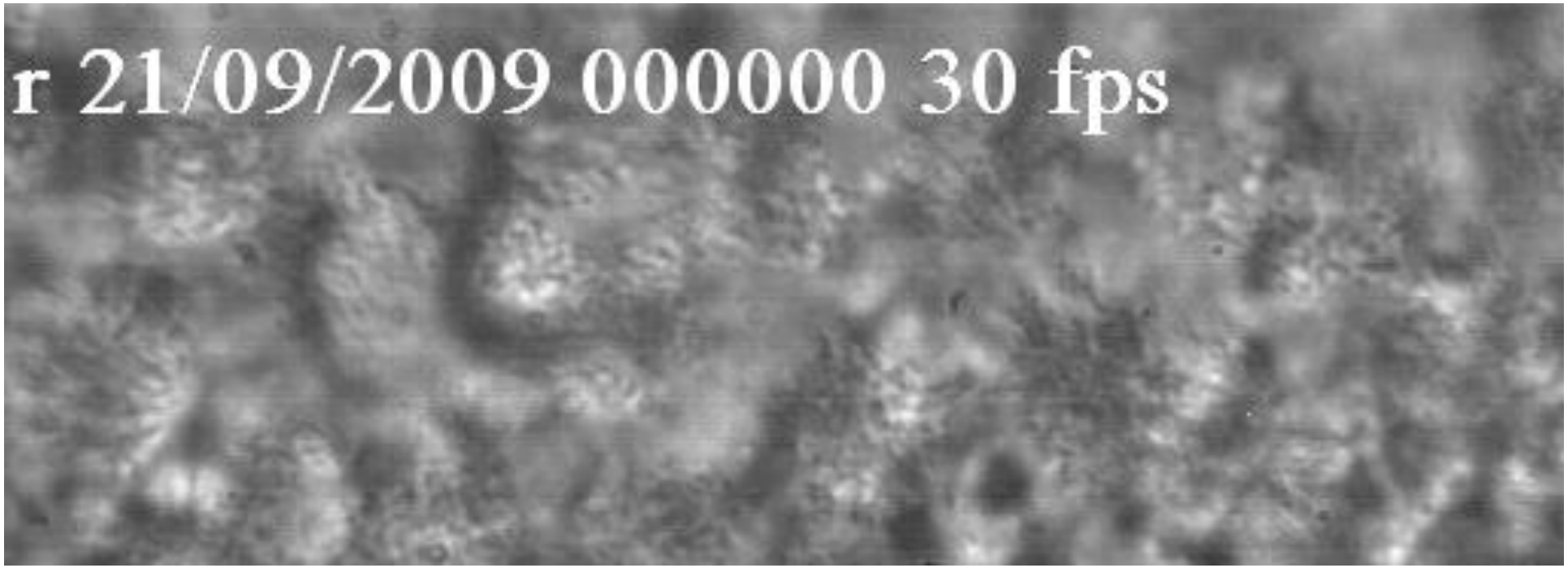
Immotile cilia



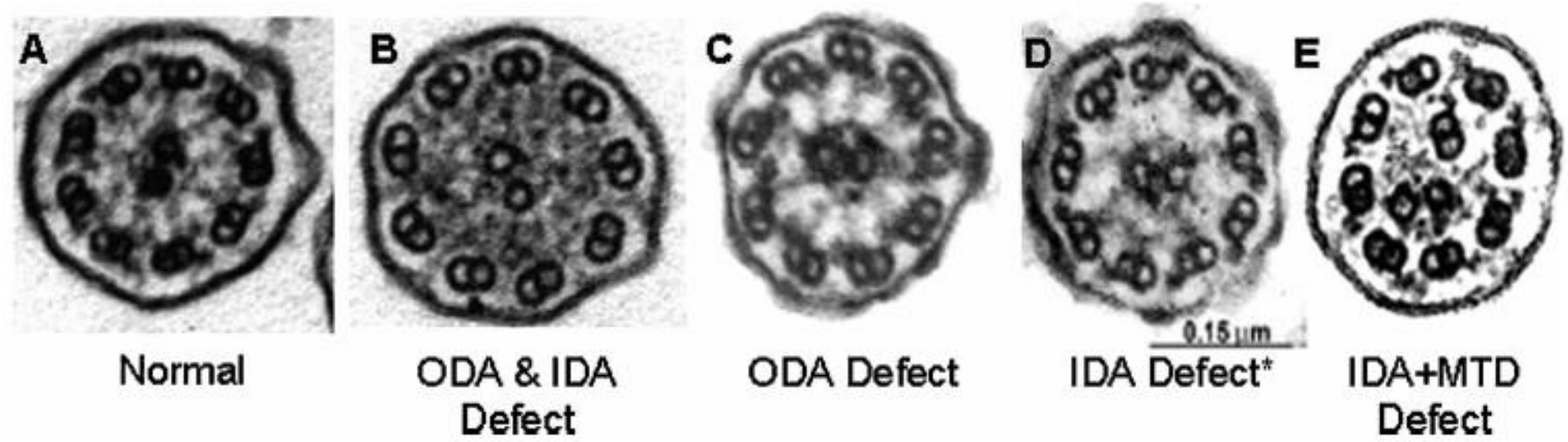
Stiff beating cilia

Circular beating cilia



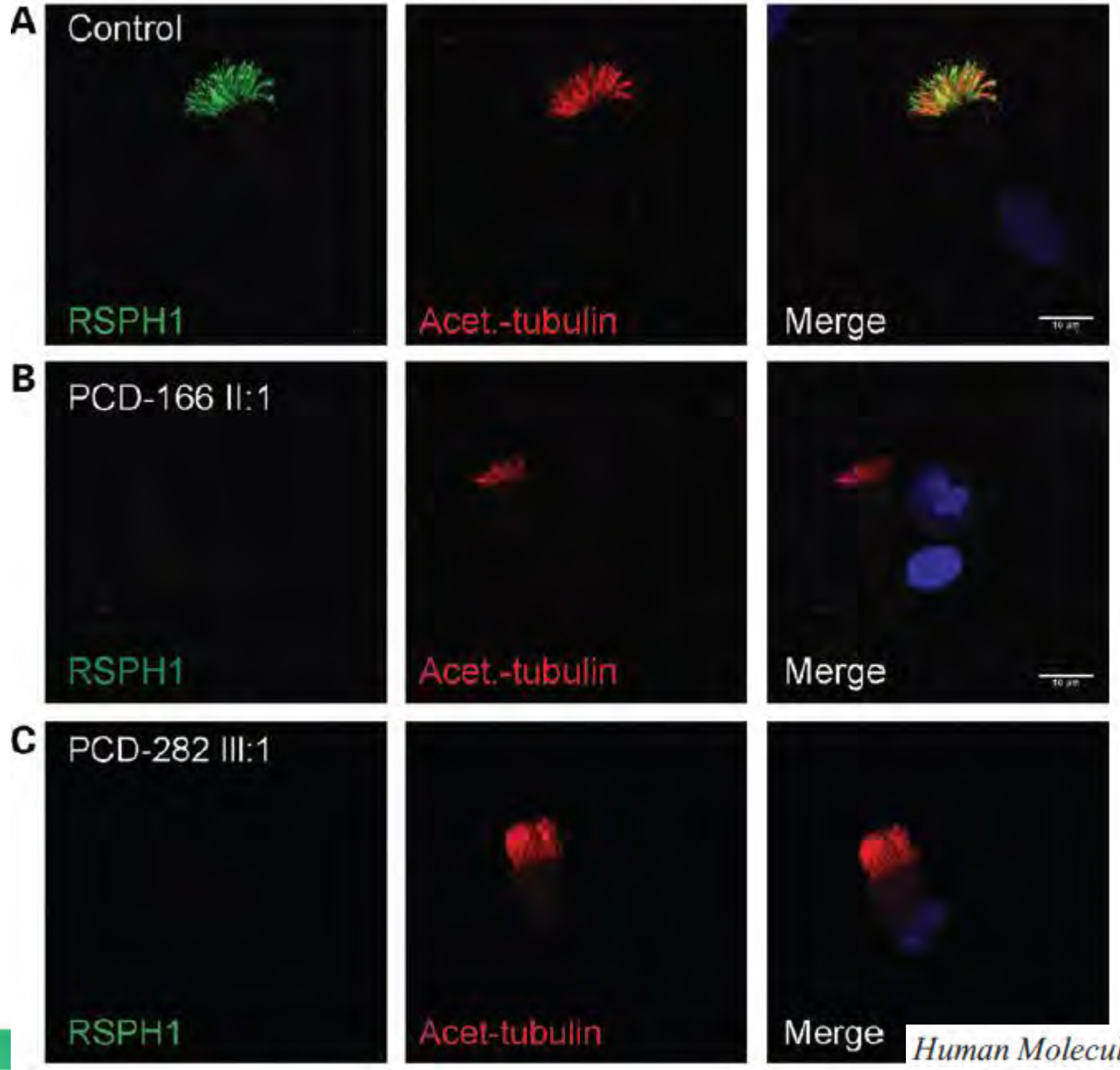


Transmission Electron Microscopy



Leigh MW et al. Genet Med 2009

Immunofluorescence



DNAH5
DNAI2
DNALI1
RSPH4A
RSPH1
RSPH 9

Human Molecular Genetics, 2014

Genetics

Genetics

- Genetically heterogeneous, NOT a single gene/locus
- Majority autosomal recessive
- Rarely, dominant or X-linked
- ~39 disease causing mutations identified
- Bi allelic mutations in one disease causing gene
- No documented cases of digenic inheritance (heterozygous mutations in two different PCD gene)

Genetic basis of about 30% patients with PCD - unknown

PCD genes	Prevalence in PCD	Ciliary structural defect	Detected on current commercial PCD NGS panels
NME8	+	Partial ODA defect	Yes
DNAH5	++++	ODA defect	Yes
DNAI1	+++	ODA defect	Yes
DNAI2	++	ODA defect	Yes
DNAL1	+	ODA defect	Yes
CCDC114	++	ODA defect	Yes
CCDC103	++	ODA ± defect	Yes
DNAAF1	++	ODA and IDA defect	Yes
DNAAF2	++	ODA and IDA defect	Yes
DNAAF3	+	ODA and IDA defect	Yes
LRRC6	++	ODA and IDA defect	Yes
HEATR2	+	ODA and IDA defect	Yes
RPGR	+	Normal	Yes
OFD1	+	Normal	Yes
DNAH11	+++	Normal	Yes
CCDC39	+++	IDA defect + MTD defect	Yes
CCDC40	+++	IDA defect + MTD defect	Yes
RSPH9	+	Central pair defect or normal	Yes
RSPH4A	++	Central pair defect or normal	Yes
RSPH1	++	Central pair defect or normal	
RSPH3	+	Central pair defect or normal	
CCNO	+	Oligocilia (residual axoneme normal)	
MCIDAS	+	Oligocilia (residual axoneme abnormal)	
DNAH8	+	Not available	
CCDC151	++	ODA defect	
ARMC4	++	ODA defect	
DYX1C1	+	ODA and IDA defect	
C21orf59	+	ODA and IDA defect	
ZMYND10	++	ODA and IDA defect	
SPAG1	++	ODA and IDA defect	
HYDIN	+	Normal	
CCDC164 (DRC1)	+	Mostly normal (N-DRC defect)	
CCDC65 (DRC2)	+	Mostly normal (N-DRC defect)	

% of all PCDs

+	: <1%
++	: 1-4%
+++	: 4-10%
++++	: >15%

Shapiro AJ. Ped Pul 2016

PCD genes	Prevalence in PCD	Ciliary structural defect	Detected on current commercial PCD NGS panels
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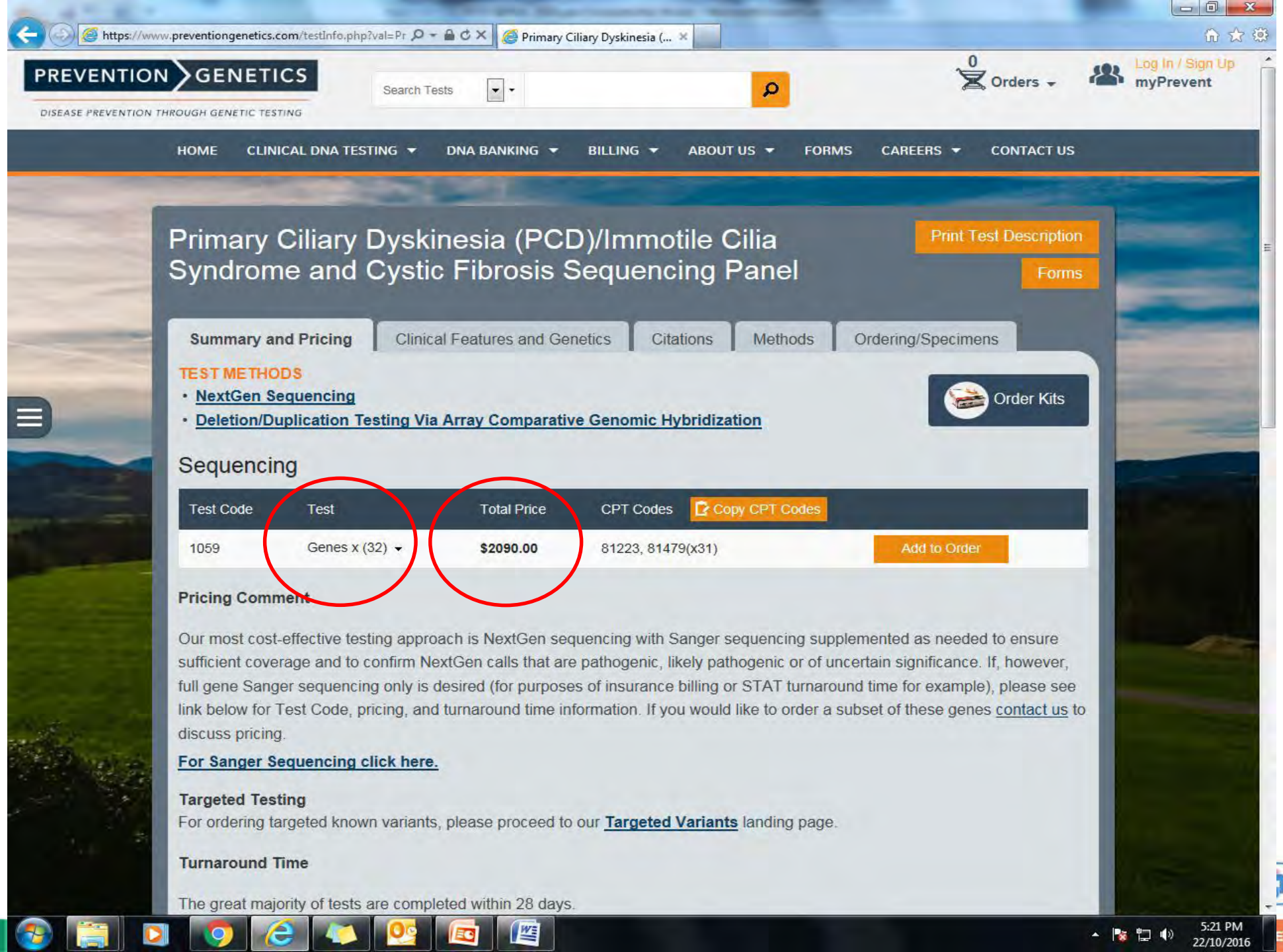
Shapiro AJ. Ped Pul 2016



PCD: Recommended Panel of Diagnostic Tests

	Potential for **false positive**	Potential for **false negative**
Nasal NO (nNO) (<77nl/min)	Low*	Low
Transmission Electron Microscopy (TEM)	Variable†	Variable‡
High speed Video Microscopy Analysis (HVMA)	Variable§	Moderate§
PCD genetic test	Low¶	Moderate¶
Immunofluorescence (IF)	Unknown	Unknown

* CF needs to be excluded. False positive: Viral infection, Epistaxis, Non atopic sinusitis.

† Infection, irritants, improper specimen handling/processing, inexperience.

‡ Several disease causing mutations can have normal TEM or only subtle changes.

§ Secondary changes, subtle changes may be missed (inexperience).

¶ 30% PCD have no identifiable mutation, may miss large insertions/deletions.

Shapiro AJ. Ped Pul 2016

AMERICAN THORACIC SOCIETY DOCUMENTS

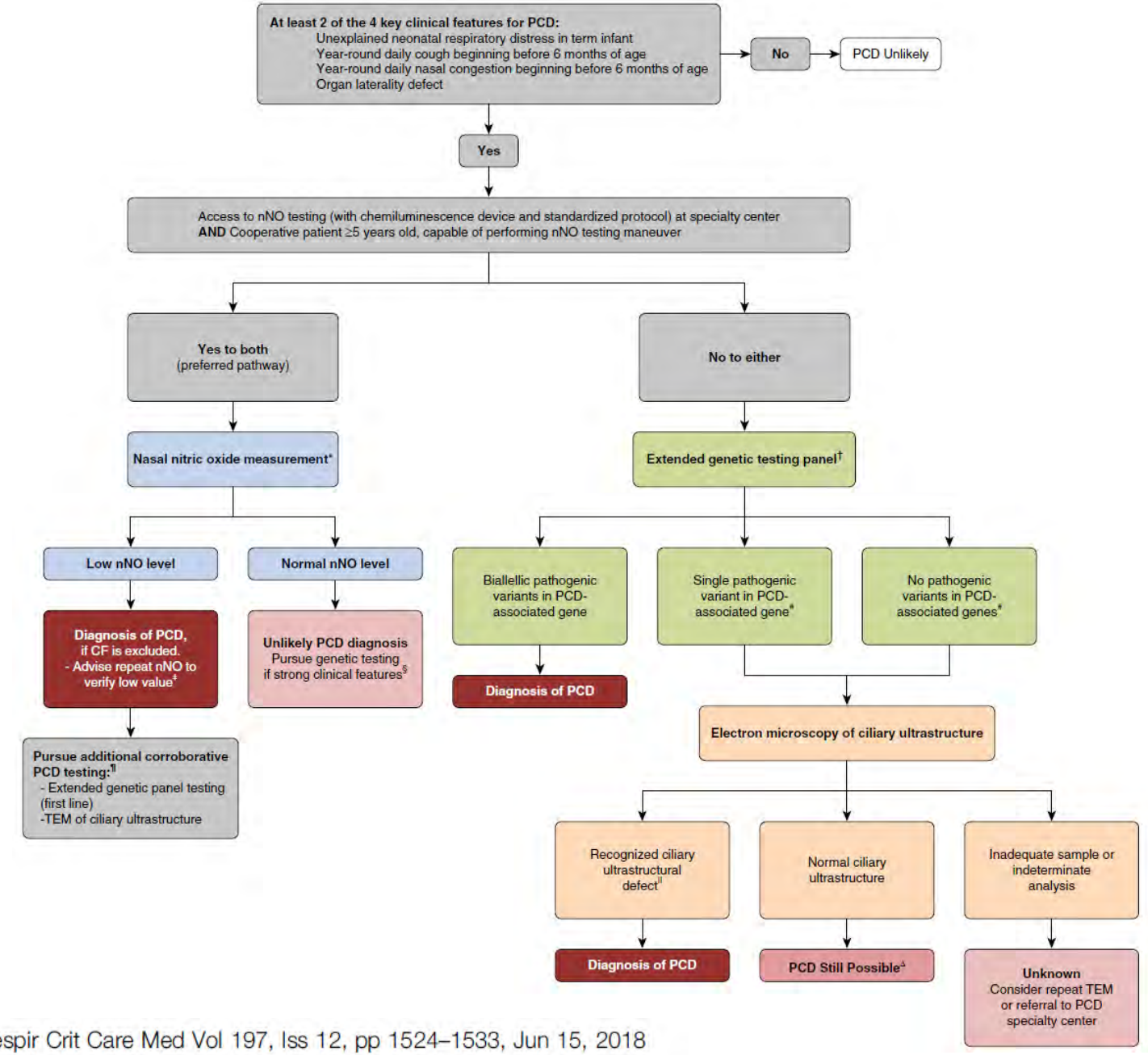
Diagnosis of Primary Ciliary Dyskinesia

An Official American Thoracic Society Clinical Practice Guideline: Executive Summary

Adam J. Shapiro, Stephanie D. Davis, Deepika Polineni, Michele Manion, Margaret Rosenfeld, Sharon D. Dell, Mark A. Chilvers, Thomas W. Ferkol, Maimoona A. Zariwala, Scott D. Sagel, Maureen Josephson, Lucy Morgan, Ozge Yilmaz, Kenneth N. Olivier, Carlos Milla, Jessica E. Pittman, M. Leigh Anne Daniels, Marcus Herbert Jones, Ibrahim A. Janahi, Stephanie M. Ware, Sam J. Daniel, Matthew L. Cooper, Lawrence M. Nogee, Billy Anton, Tori Eastvold, Lynn Ehrne, Elena Guadagno, Michael R. Knowles, Margaret W. Leigh, and Valery Lavergne; on behalf of the American Thoracic Society Assembly on Pediatrics

THIS OFFICIAL CLINICAL PRACTICE GUIDELINE OF THE AMERICAN THORACIC SOCIETY WAS APPROVED MAY 2018

Am J Respir Crit Care Med Vol 197, Iss 12, pp 1524–1533, Jun 15, 2018



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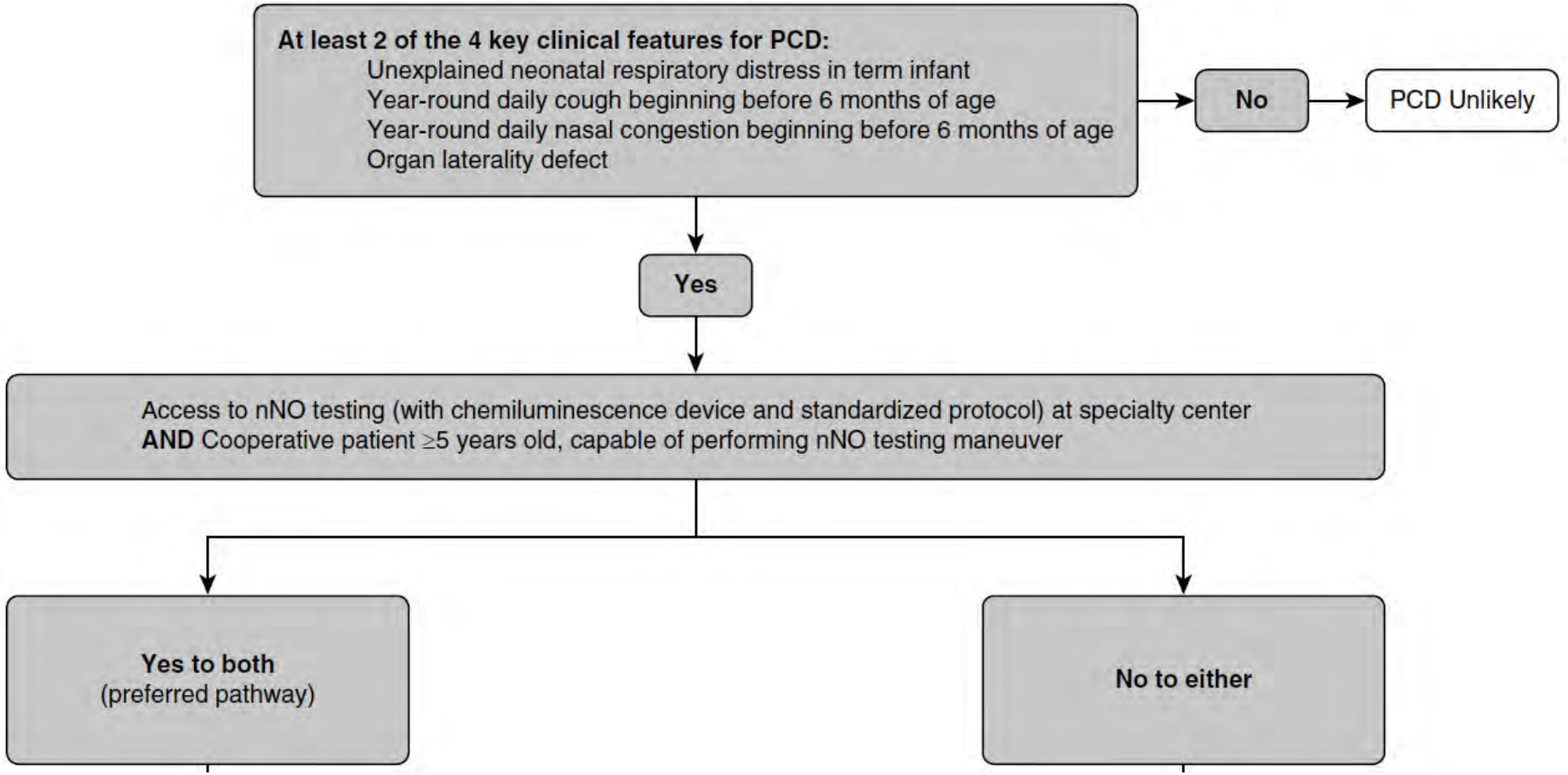
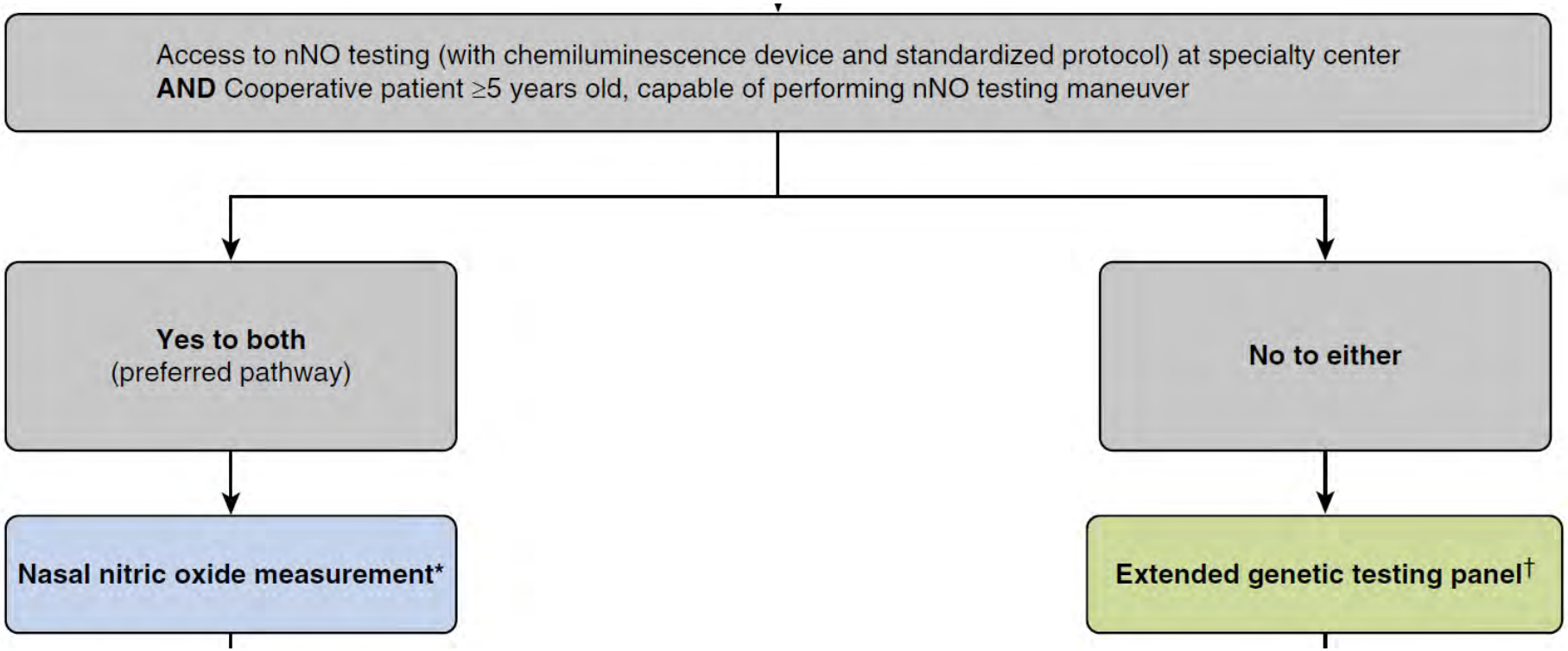
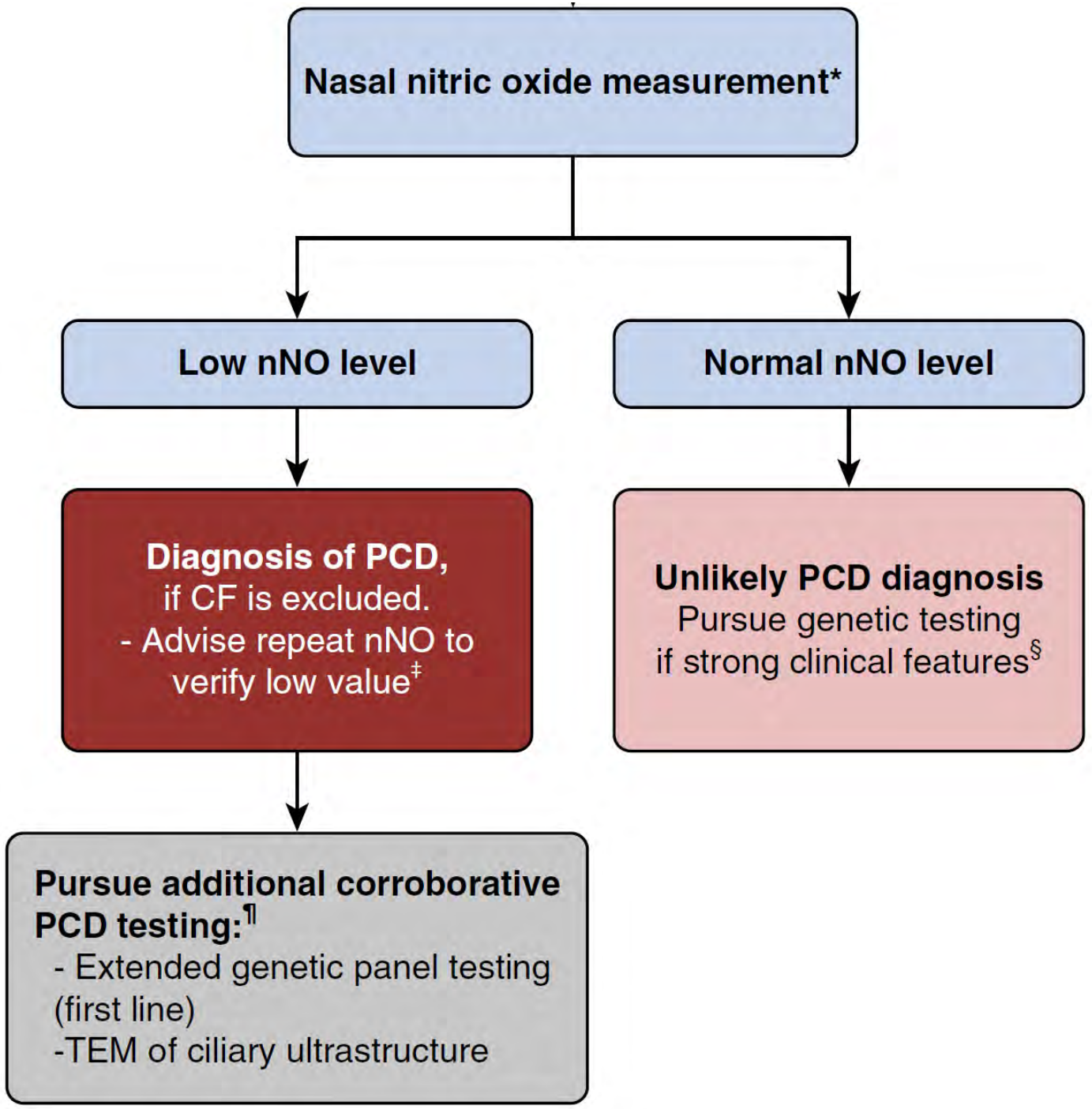
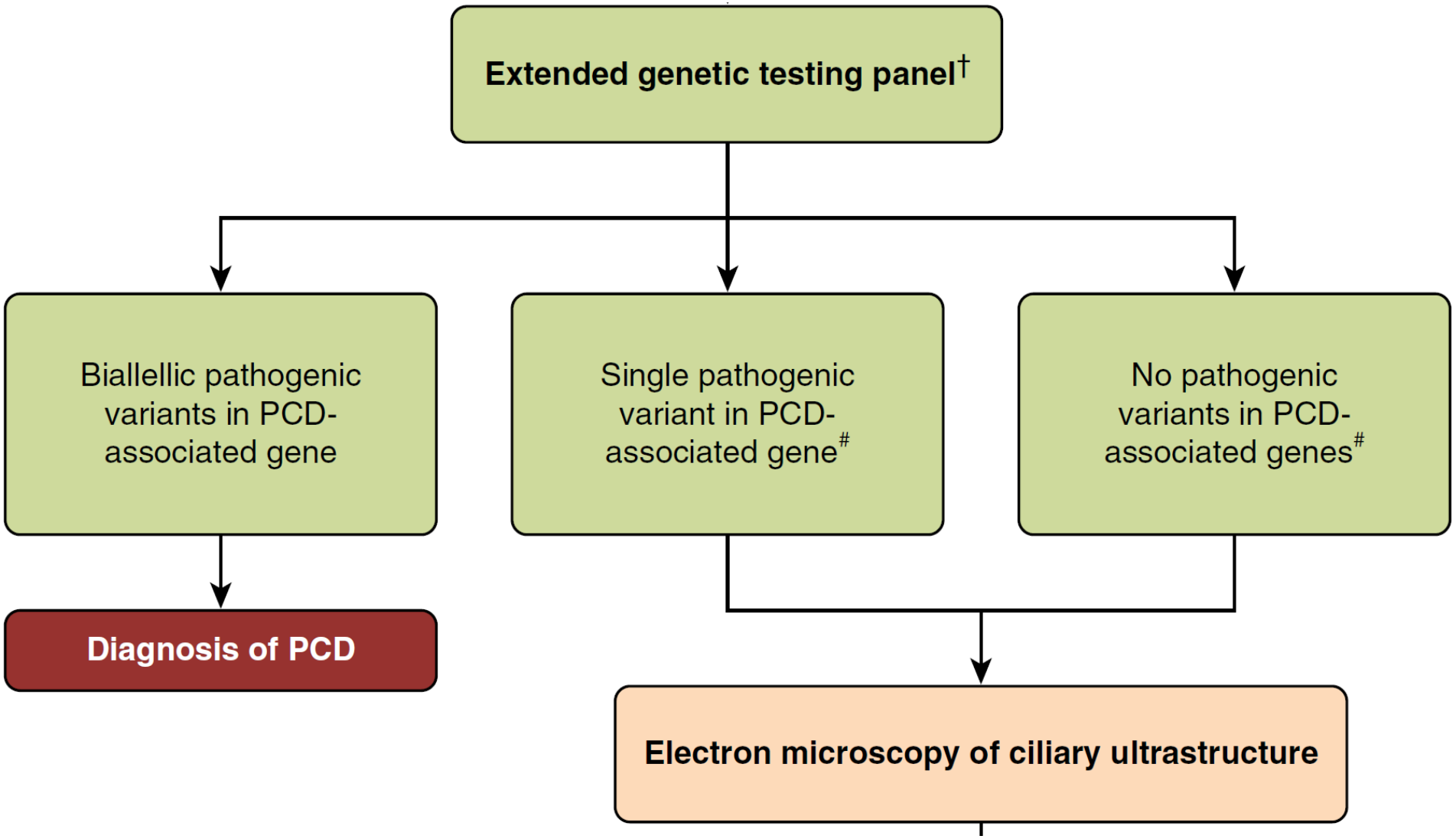
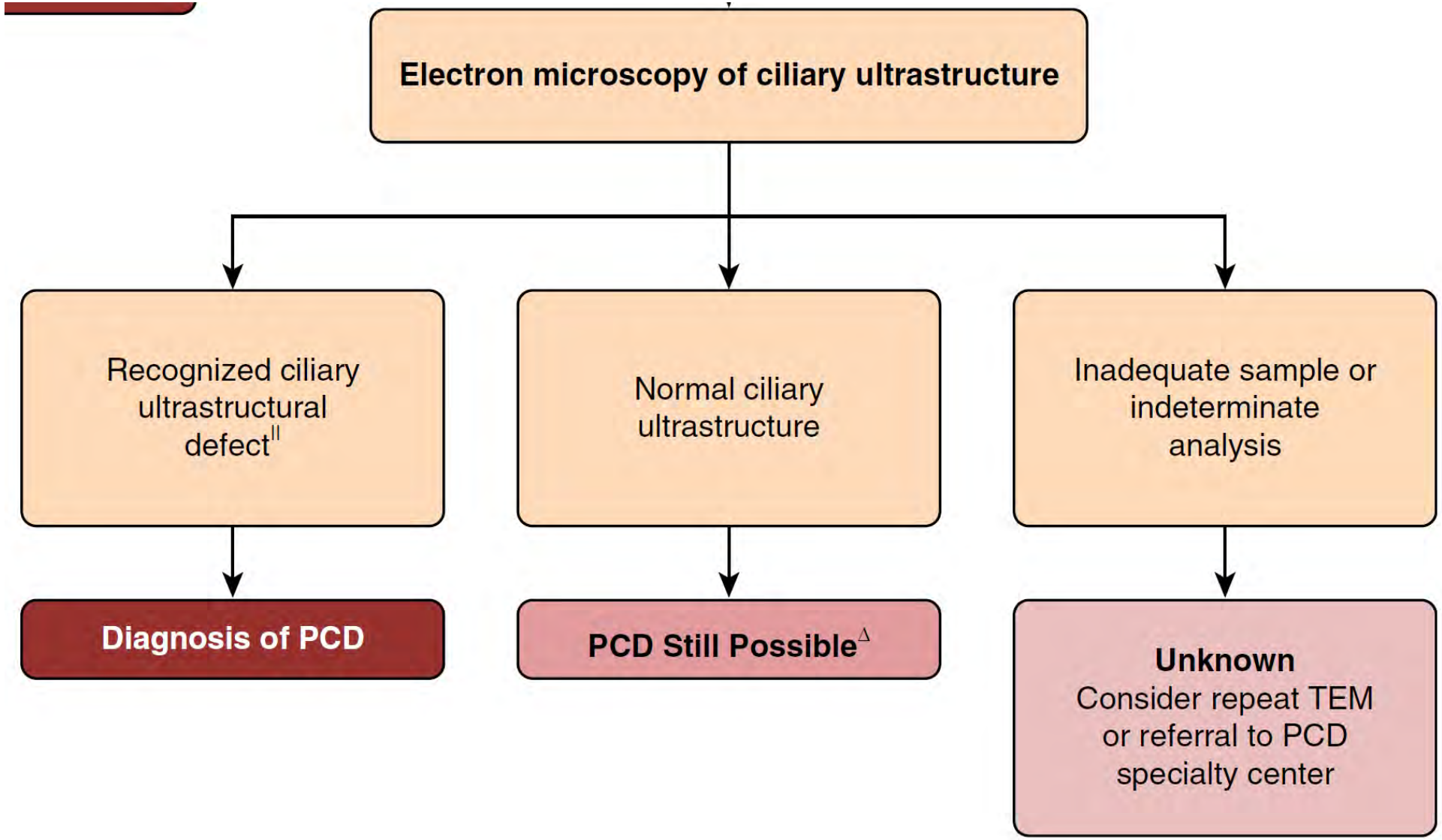

At least 2 of the 4 key clinical features for PCD:
Unexplained neonatal respiratory distress in term infant
Year-round daily cough beginning before 6 months of age
Year-round daily nasal congestion beginning before 6 months of age
Organ laterality defect
No
PCD Unlikely
Yes
Access to nNO testing (with chemiluminescence device and standardized protocol) at specialty center
AND Cooperative patient ≥5 years old, capable of performing nNO testing maneuver
Yes to both
(preferred pathway)
No to either


Access to nNO testing (with chemiluminescence device and standardized protocol) at specialty center
AND Cooperative patient ≥5 years old, capable of performing nNO testing maneuver
Yes to both
(preferred pathway)
No to either
Nasal nitric oxide measurement*
Extended genetic testing panel†


Nasal nitric oxide measurement*
Low nNO level
Normal nNO level
Diagnosis of PCD,
if CF is excluded.
- Advise repeat nNO to verify low value‡
Unlikely PCD diagnosis
Pursue genetic testing
if strong clinical features§
Pursue additional corroborative PCD testing:¶
- Extended genetic panel testing (first line)
-TEM of ciliary ultrastructure


Extended genetic testing panel†
Biallellic pathogenic variants in PCD-associated gene
Single pathogenic variant in PCD-associated gene#
No pathogenic variants in PCD-associated genes#
Diagnosis of PCD
Electron microscopy of ciliary ultrastructure


Electron microscopy of ciliary ultrastructure
Recognized ciliary ultrastructural defect‖
Normal ciliary ultrastructure
Inadequate sample or indeterminate analysis
Diagnosis of PCD
PCD Still Possible△
Unknown
Consider repeat TEM or referral to PCD specialty center

	Clinical +	≥ 1 Diagnostic Test
Newborn (0-1m)	Situs inversus totalis + Unexplained neonatal respiratory distress at term birth	TEM Genetic test HVMA
Children (1m-5 years)	≥2 major PCD clinical criteria	TEM Genetic test HVMA
Children>5 years & Adults	≥2 major PCD clinical criteria	Nasal NO TEM Genetic test HVMA

TEM	: Diagnostic abnormality
Genetic test	: Biallelic mutation in one PCD associated gene
HVMA	: Persistent and diagnostic abnormality on ≥2 occasions
Nasal NO	: <77nl/min on 2 occasions, >2m apart, with CF excluded

Major clinical criteria*

1. Unexplained neonatal respiratory distress (term) with lobar collapse ± CPAP/O_2 for >24 hr.
2. Any organ laterality defect – SIT, SA or Heterotaxy
3. Daily, year-round wet cough starting in the first year of life OR Bronchiectasis on chest CT
4. Daily, year-round nasal congestion starting in the first year of life OR pansinusitis on sinus CT

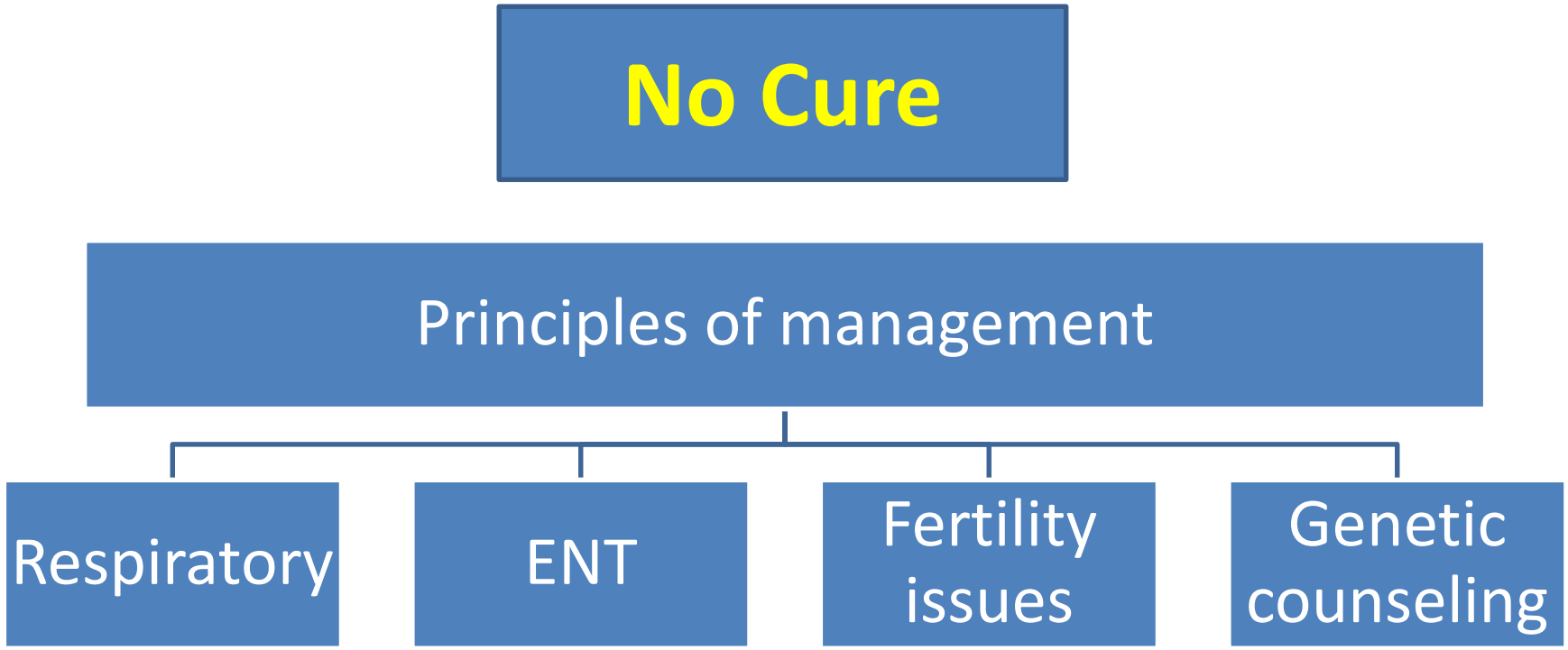
*Exclude other differentials such as CF and immune deficiencies.

Shapiro AJ. Ped Pul 2016

Choosing the test(s)

- Diagnostic test accuracy
- Confidence in the estimates of the test
- Patient values and preferences
- Costs
- Feasibility
- Acceptability

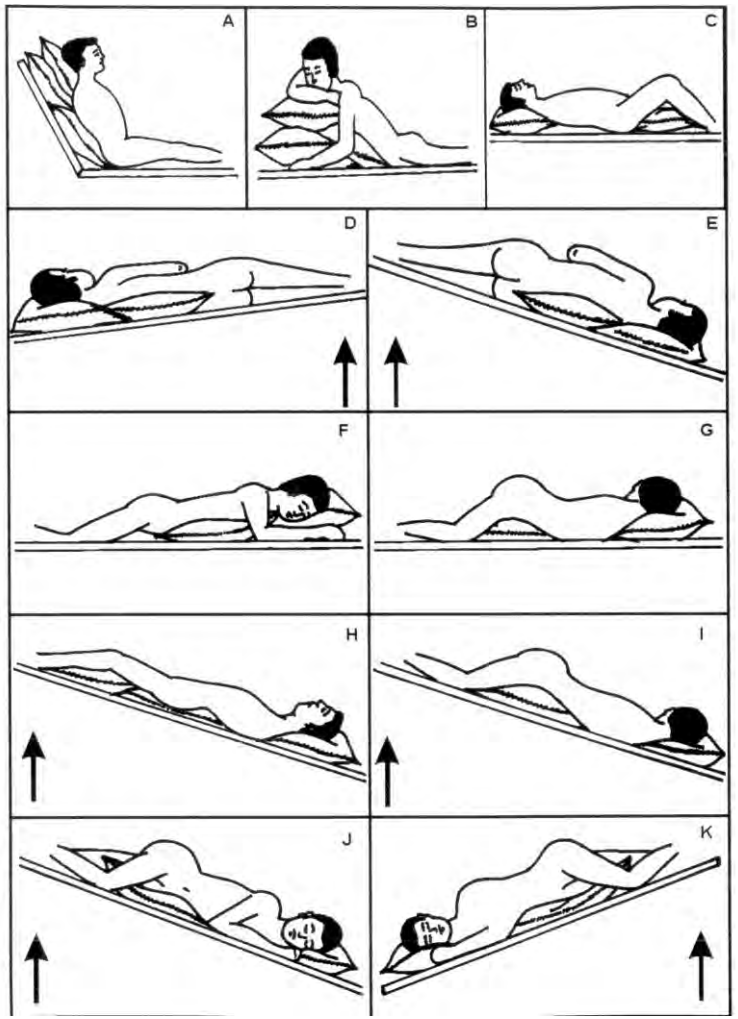
Management of patients with PCD



Summary of recommended respiratory management of children with PCD	
Clinic visit	2-4 times/year
Sputum/cough swab	At every clinic visit If respiratory exacerbations
Culture for Non Tuberculous Mycobacteria/Fungi	Every 2 years If not responding to culture-directed antibiotics Consider screening for ABPA
Spirometry	2-4 times/year
Imaging: CXR	At diagnosis, once every 2-4 years During severe exacerbations
Imaging: Chest CT	At least once (age ~ 5-7 years)
Chest Physiotherapy, exercise	Daily
Vaccinations	Routine childhood vaccines Pneumococcal vaccination Annual influenza vaccine RSV immunoprophylaxis in the first winter (case by case basis)
Antibiotics for exacerbations	Mild – Oral Severe - IV

Shapiro AJ et al. Ped Pulmonology 2016

Physiotherapy



Respiratory therapies to consider on a case by case basis in PCD		
Long term antibiotics	Oral	Co-trimoxazole Macrolide
	Nebulised	Chronic PA colonisation
Inhaled hyperosmolar agents	Hypertonic saline (3-7%)	Limited benefits in non-CF bronchiectasis No studies in PCD Proper equipment sterilization – important
	Mannitol	Limited benefits in non-CF bronchiectasis No studies in PCD
DNAse (dornase-alfa)		No trials in PCD, few case reports Adverse effects in non-CF bronchiectasis
Inhaled bronchodilators		Before nebulised hypertonic saline Reactive airway disease
ICS+LABA		Consider if associated asthma
IVIG		Consider if humoral immune deficiency present
Lobectomy		Severe localised bronchiectasis, caution
Lung transplantation		Caution - situs anomalies

Shapiro AJ et al. Ped Pulmonology 2016

Management of ENT problems

- Chronic otitis media with effusion - universal
  - grommets: controversial
  - antibiotics
  - hearing tests, hearing aids, speech therapy
  - tympanoplasty
- Chronic rhinosinusitis
  - saline lavages
  - nasal steroids
  - antibiotics
  - surgery

Involve
Paediatric ENT Surgeon &
Speech Therapist

http://www.pcdfoundation.org/ | INFOPEDIA | pcd uk - Google Search | Primary Ciliary Dyskin... | PCD Family Support Gro...

HOME | ABOUT PCD | LIVING WITH PCD | FINDING A CURE | GET INVOLVED | RESOURCES | ABOUT THE FOUNDATION | CONTACT US

PCD FOUNDATION
PRIMARY CILIARY DYSKINESIA

Forum | Clinical Trials | Volunteer | Search Site

DONATE
There are many ways to donate, find the one that's right for you!

VOLUNTEER
Support your local PCD community with a volunteer opportunity that's right for you.

ADVOCATE
Join the fight for PCD! Help raise awareness and support efforts to find a cure.

LATEST NEWS & EVENTS

- ATS Webinar on PCD 10/20/2016
- PCDF CARE Day: Boston Children's Nov. 6, 2016
- PCD On The Move 2016: Advances in Primary Ciliary Dyskinesia Research, Diagnosis & Care
- Call for Scientific Abstracts and Case Reports 2016
- PCDF Clinical Centers Network

UNMASK THE FACES OF PCD

25,000+ people

in the United States have PCD and less than 400 know it. Unmask the faces of PCD.

READ MORE...

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http://www.pcdsupport.org.uk/

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Primary Ciliary Dyskinesia
Family Support Group

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Latest News

ERS Patient Event 2016
Wednesday, October 05, 2016

Have you had your flu jab?
Wednesday, October 05, 2016

25th Anniversary Cruise Party 4th September 2016
Thursday, September 08, 2016

PCD Day Video 2016
Thursday, July 28, 2016

Lastest research news from the Royal Brompton
Thursday, July 21, 2016

PCD Day at Woburn Safari Park
Thursday, June 23, 2016

Tweets by @PCD_UK

PCD Family Suppor
UK
18 October at 08:35

Welcome

Welcome to our website which was created in 2010 with the help of a grant from Jeans for Genes.

The website provides an up to date information service about the condition, how it is diagnosed and how to live with it on a daily basis. Please look at the video case studies with real life people affected by PCD telling their stories.

Primary Ciliary Dyskinesia (PCD) is an inherited, relatively rare condition associated with the abnormality of cilia (microscopic hairs that beat in the airways, sweeping secretions out of the respiratory tract). PCD may affect the lungs, nose, sinuses, ears and fertility.

The condition involves recurrent infections in the nose, ears, sinuses and lungs. If left untreated can lead to a form of lung damage known as 'bronchiectasis'.

Up to 50% of patients with PCD also have dextrocardia (heart on the right side) and situs inversus (internal organs on opposite side to normal).

The mainstay of treatment is chest physiotherapy and targeted antibiotics enabling individuals to lead normal lives. Any problems resulting from PCD vary from person to person.

We hope you find the site useful and welcome any comments or suggestions about it.

Supported by

GENETIC

What is PCD?

Find out what PCD is, its cause and how it can be treated, plus much more.

Need to talk?

Visit our Online Community to chat to people who have experience of PCD in their lives.

Wish to donate?

All donations are gratefully received, alternatively find out how you can fundraise.

Contact us

Please contact us if you need any further information from our adult or children's contact.

4:38 PM
22/10/2016

Better Experimental Screening and Treatment for Primary Ciliary Dyskinesia

http://bestcilia.eu/

Conclusions

- PCD is a rare disease
- Symptoms overlap with common respiratory diseases
- Awareness and high index of suspicion are crucial
- There is no single gold standard diagnostic test
- Know when to refer to a specialised PCD centre

- Early diagnosis and management may improve outcome

Thank you

biju.thomas@singhealth.com.sg