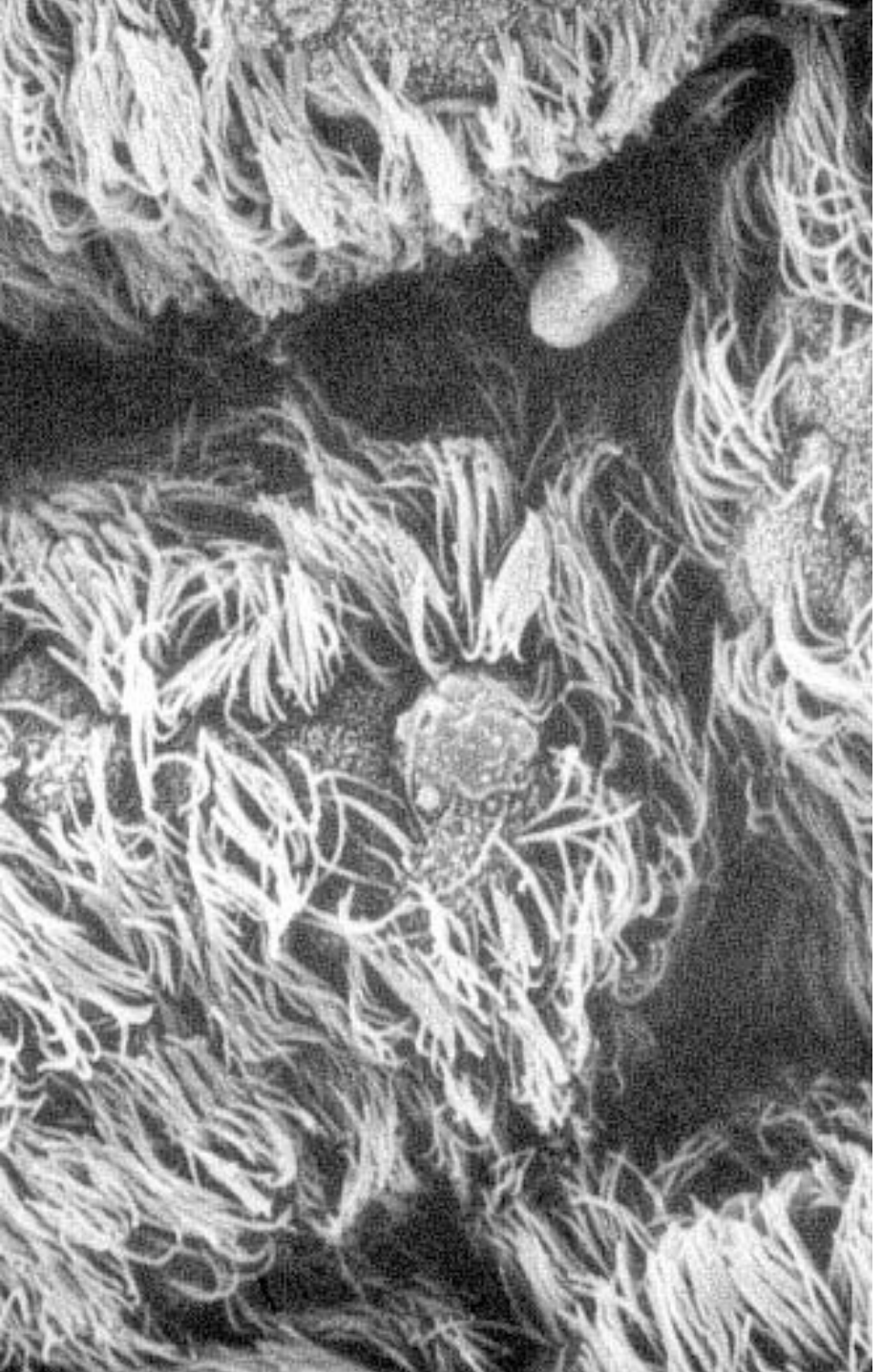
1ère rencontre autour de la DCP

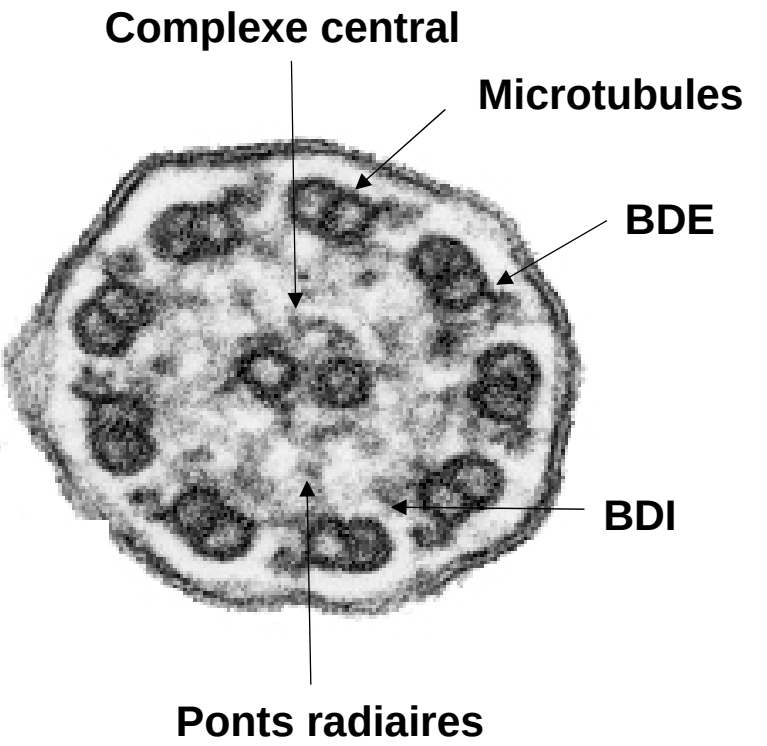
samedi 10 novembre 2007

Comment affirmer le diagnostic de DCP ?

Estelle Escudier
Service de Génétique et embryologie médicales
Hôpital Armand-Trousseau, APHP Paris
Inserm U654 et 841

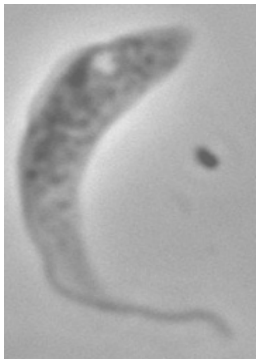


voies aériennes de conduction



épithélium respiratoire cilié

conservation des cils au cours de l'évolution



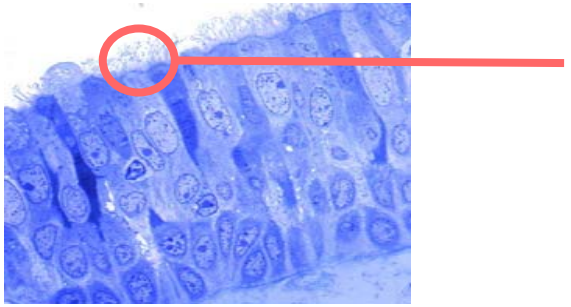
Trypanosoma brucei

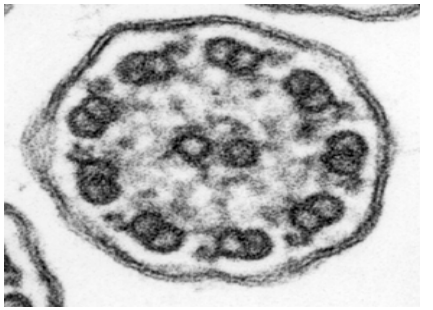
Chlamydomonas reinharditii

Oursins

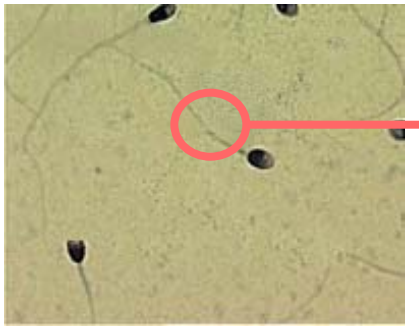
Primates

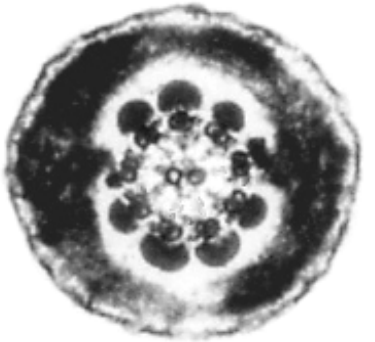
différents cils



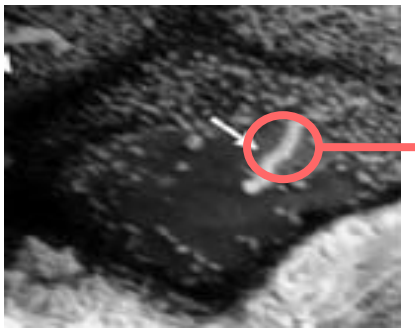


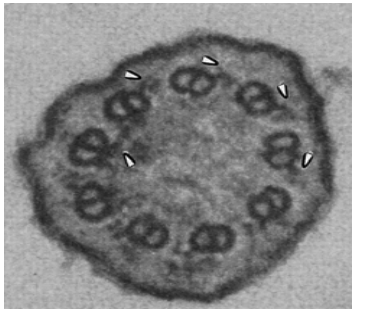
épithélium cilié





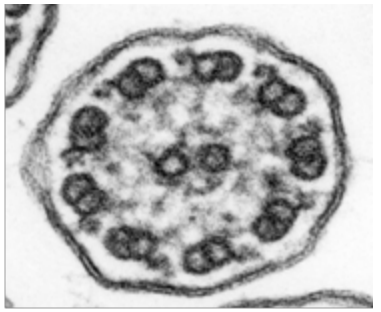
flagelle du spermatozoïde



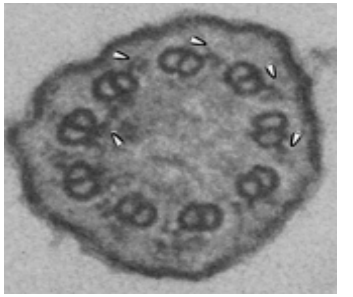


cil primaire

2 types de Cils



« 9+2 »



« 9+0 »

épithéliums ciliées

mobilisation

mucus
fluide
cellules

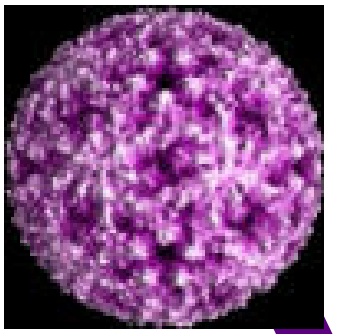
épithéliums sensoriels

perception

vision
audition
olfaction

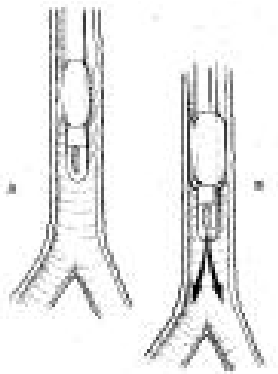
Anomalies ciliaires acquises

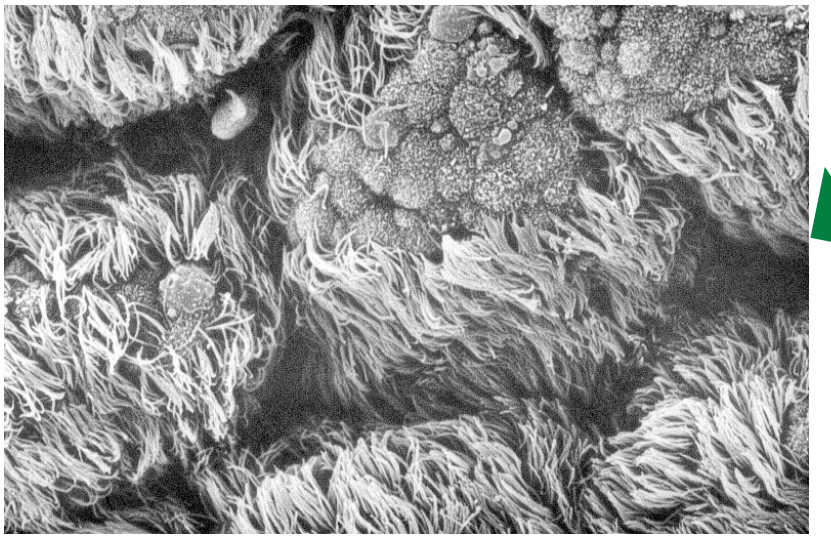
Microorganismes

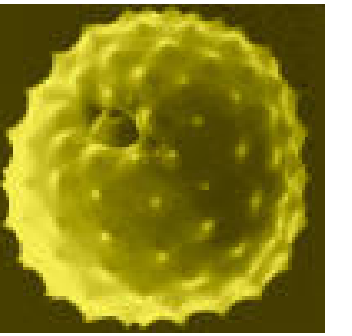


Polluants

Lésions mécaniques

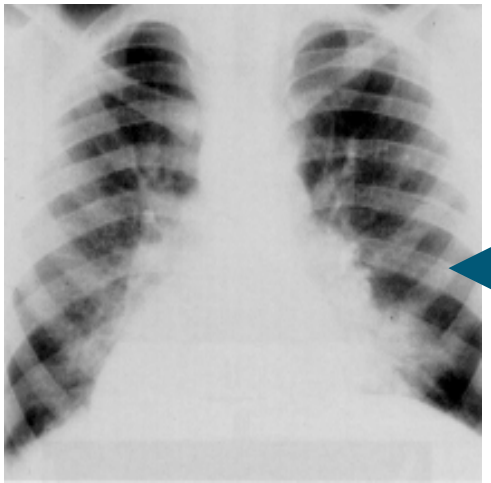




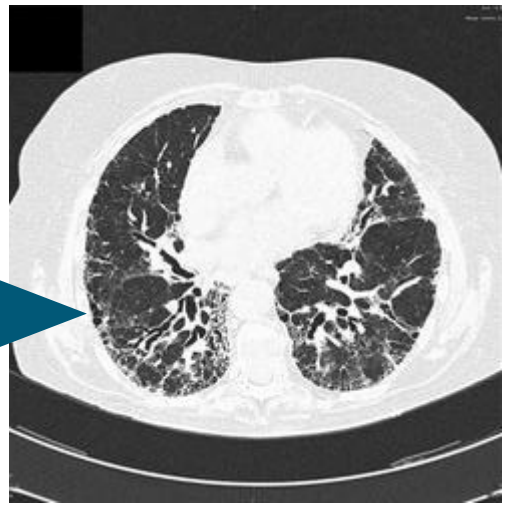


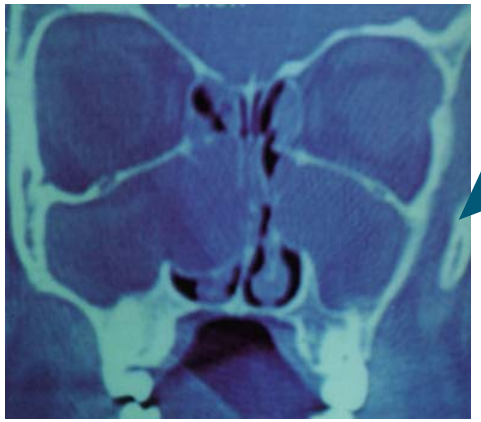
Allergènes

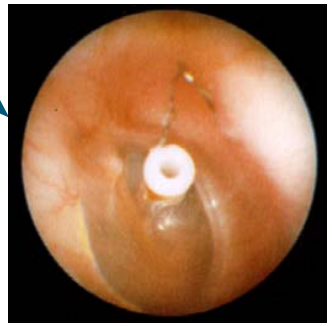
Dyskinésies ciliaires primitives



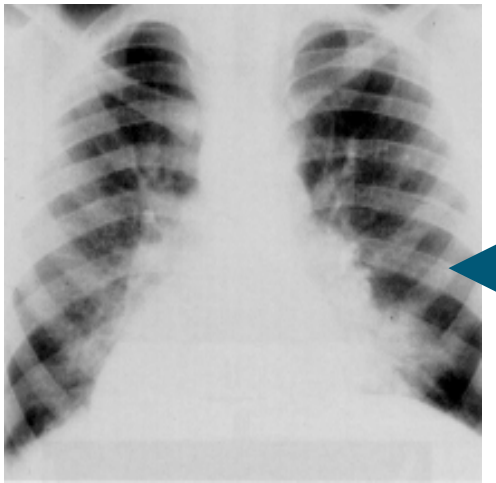
bronchiectasies
sinusites
otites séreuses
situs inversus







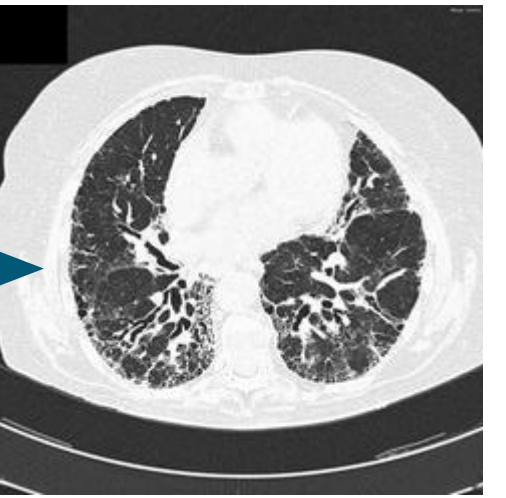
Dyskinésies ciliaires primitives

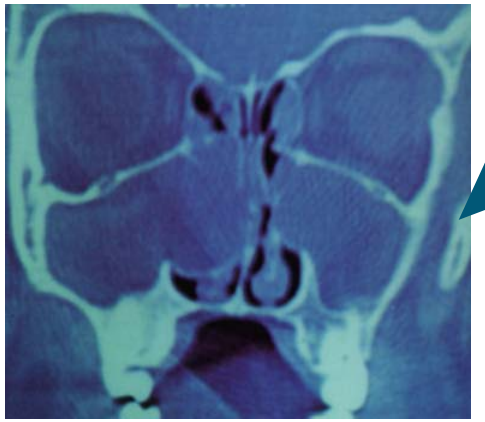


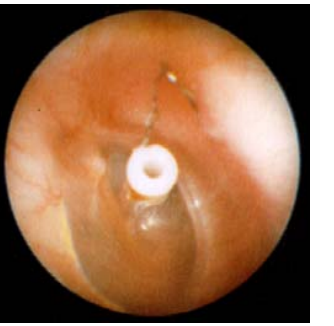
maladie récessive

1/16 000 naissances

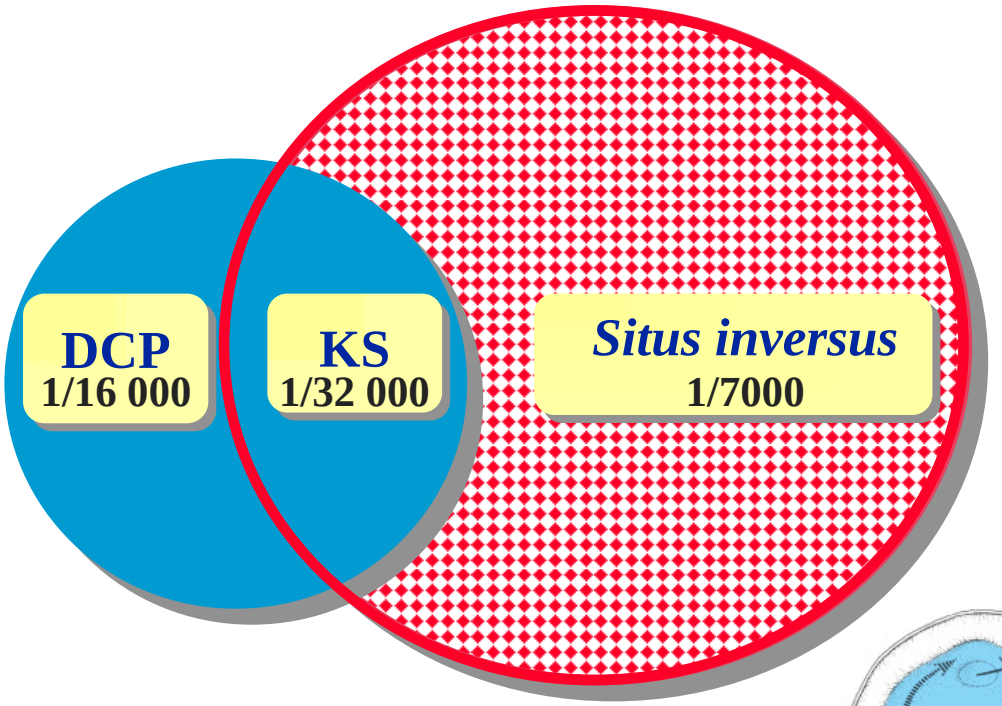
4000 cas en France

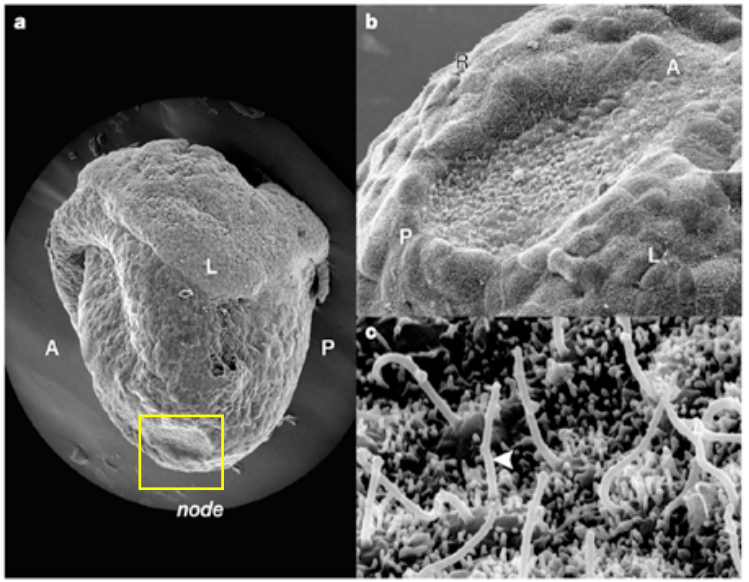


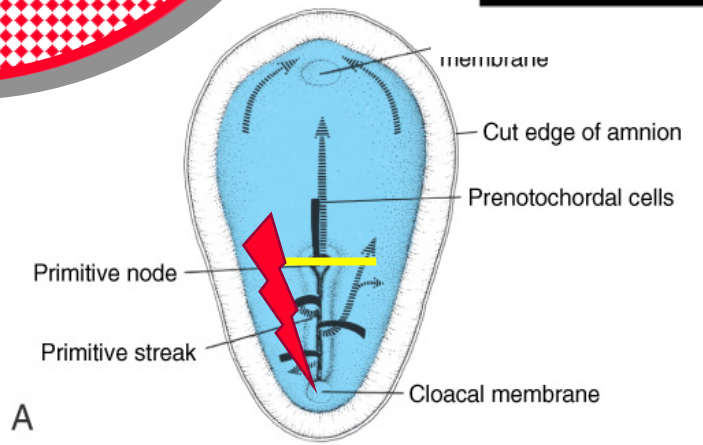


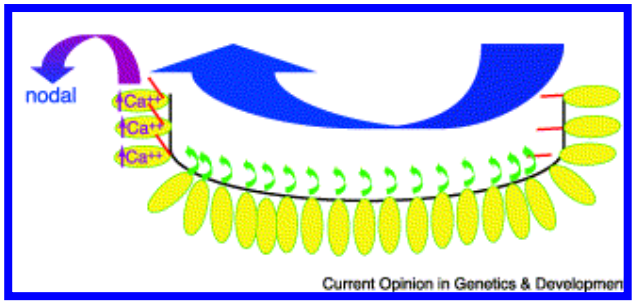


Situs Inversus et PCD









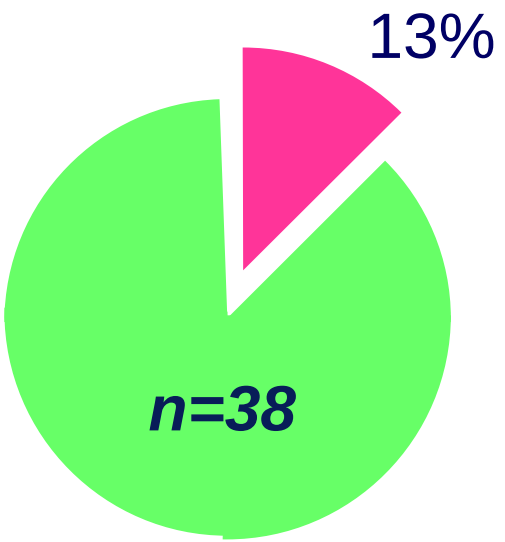
savoir y penser :

infections respiratoires récidivantes hautes et basses

- **début précoce**
- **forme familiale, consanguinité**
- **troubles sensoriels**
- **kystes des reins, hypofertilité**

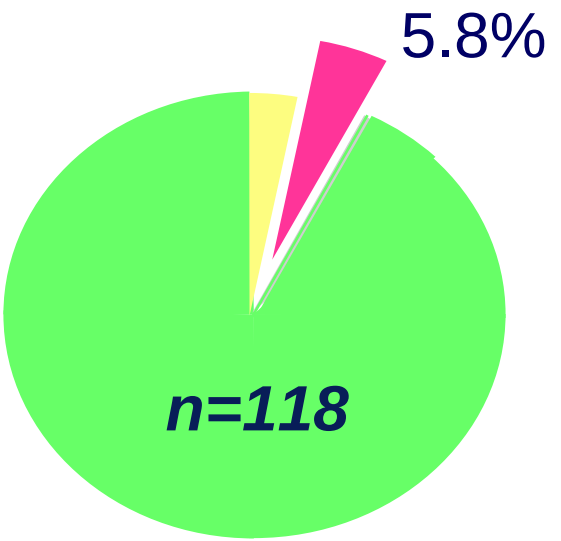
Fréquence des DCP

DDB adulte



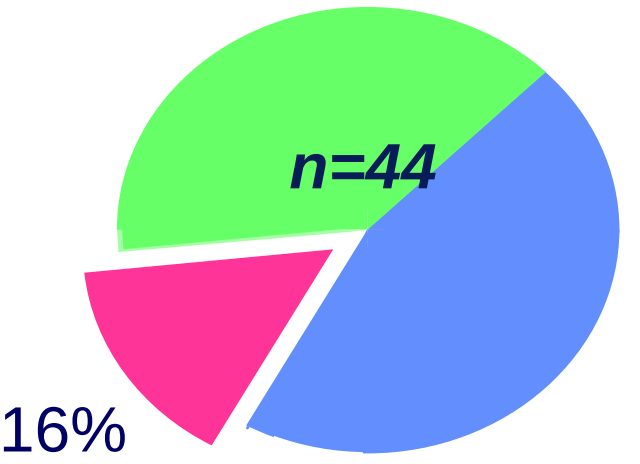
Verra et al.,1991
Eur Resp J

IRR enfant



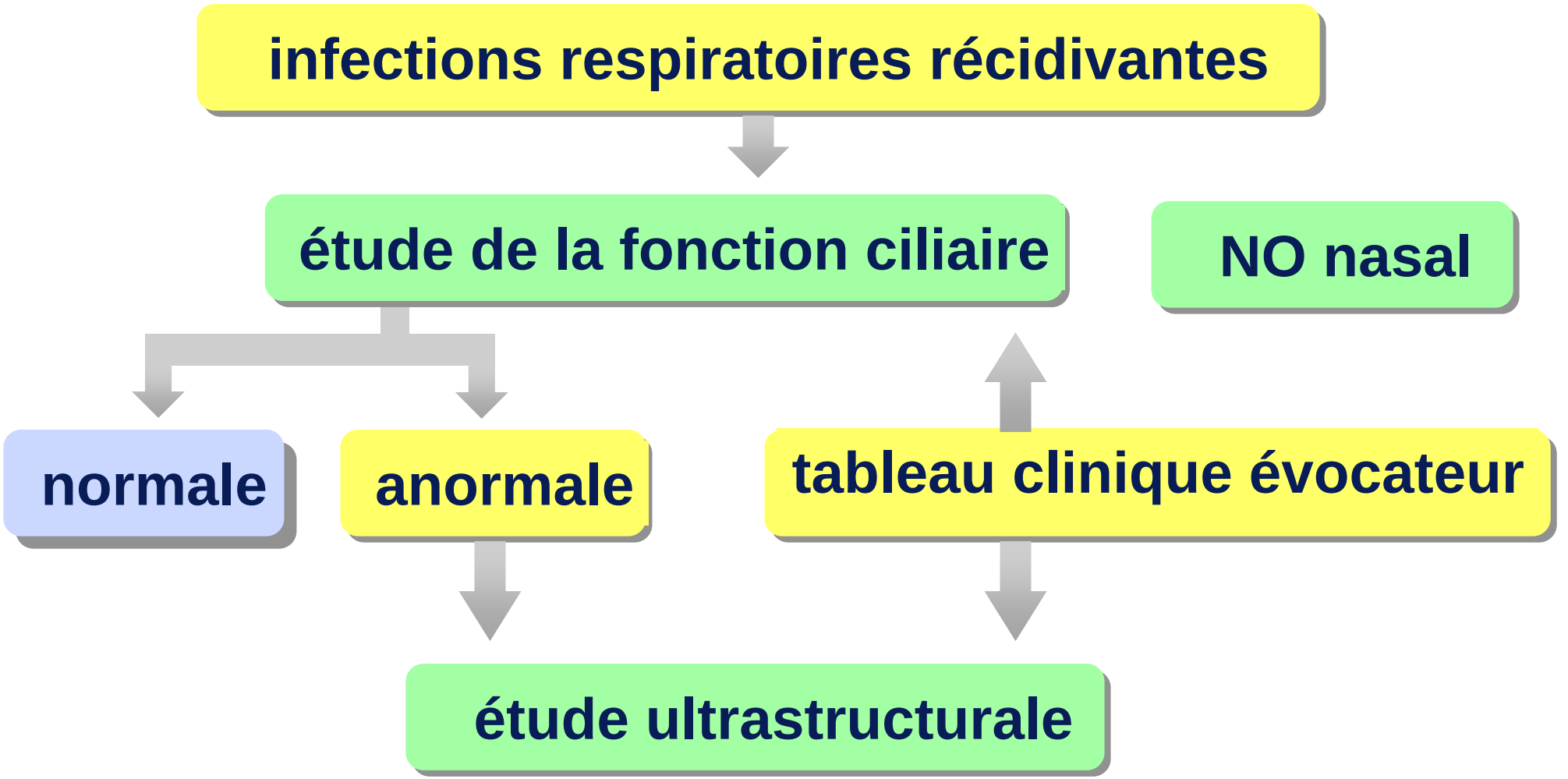
Chapelin et al.,1997
Ann Otol Laryngol

PNS atypique

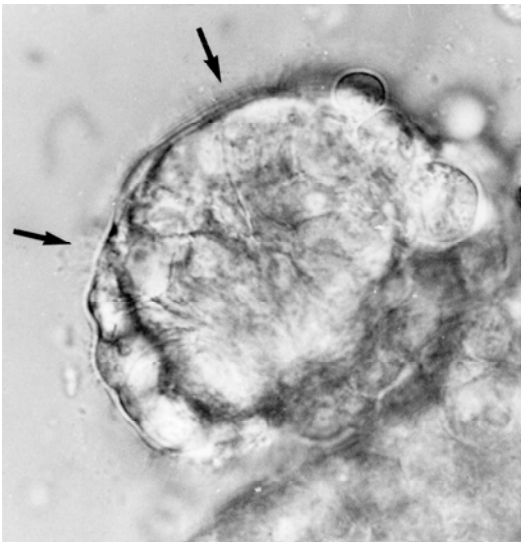


Coste et al., 2004
Laryngoscope

Démarche diagnostique



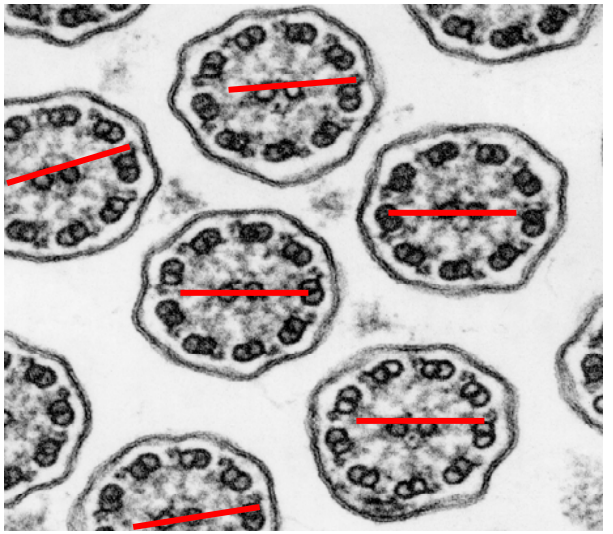
Etude du battement ciliaire



- strobotachymétrie
- caméra à haute vitesse
- photooscillométrie

10 à 15 Hz

Analyse de l'ultrastructure



- **quantitative (%)**
- **anomalie dominante**
- **orientation, extrêmité**

>30 coupes de cils dans > 10 cellules ciliées

normal : < 10% cils anormaux

anormale : >20 % cils anormaux

DCP : tous les cils portent la même anomalie

Anomalies ciliaires des DCP

absence ou défaut des bras de dynéine internes et externes

absence des bras externes

bras externes courts

absence des bras internes

absence des bras internes et des liens de nexine

absence des ponts radiaires et du complexe central

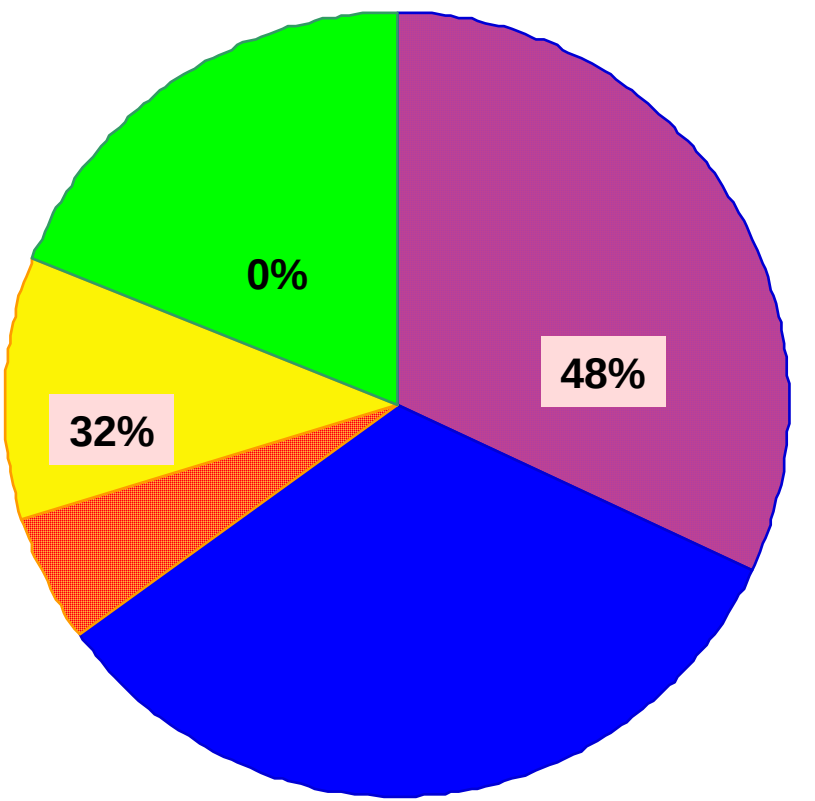
absence d’un ou des deux microtubules centraux

aplasie ciliaire

cils anormalement longs

absence d’anomalies

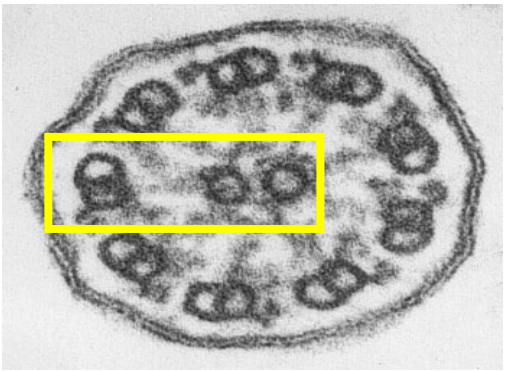
Situs inversus et anomalies ciliaires

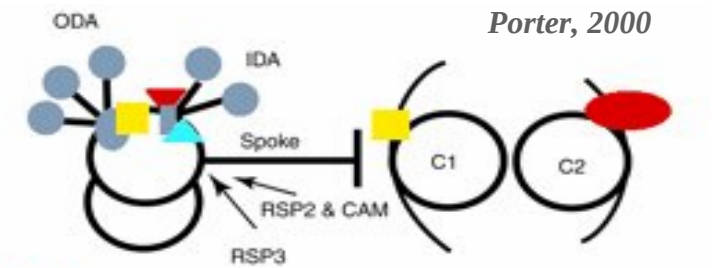


Situs inversus

BDE ± BDI
BDI seuls
CC

Syndrome de Kartagener à cils normaux (n=25)





Porter, 2000

anomalie enzymatique ?

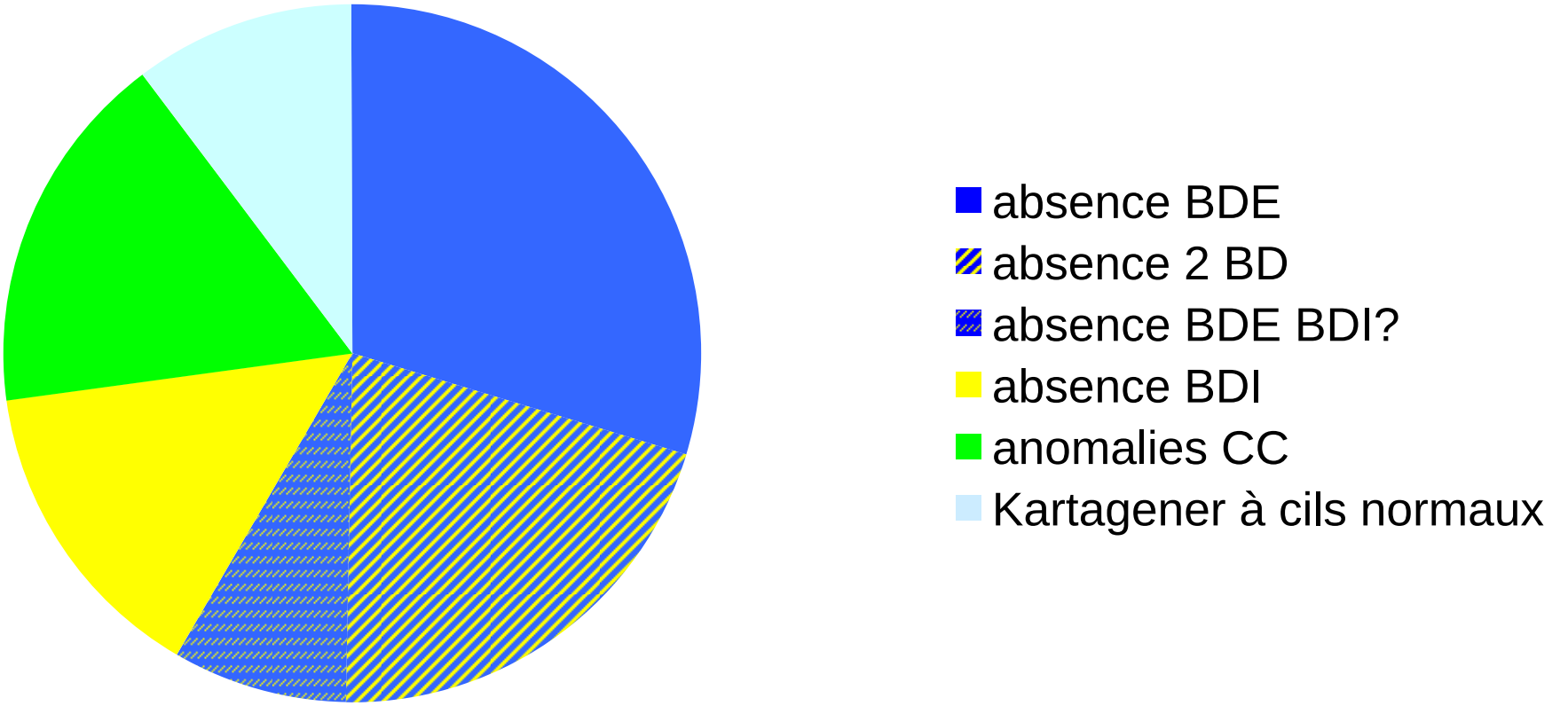
Diagnostic sur

manifestations cliniques

anomalies du battement

NO nasal effondré

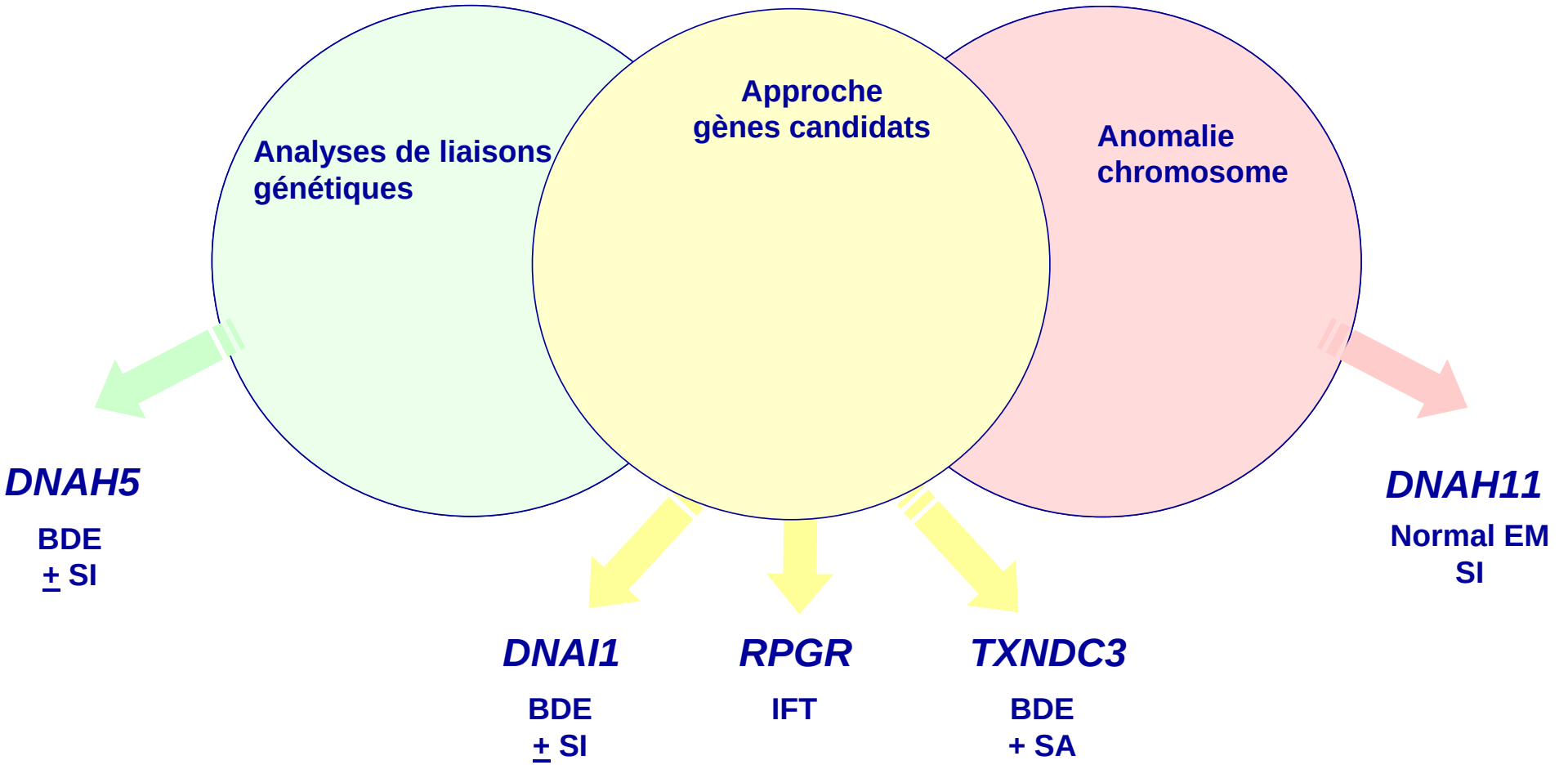
Répartition des anomalies ciliaires



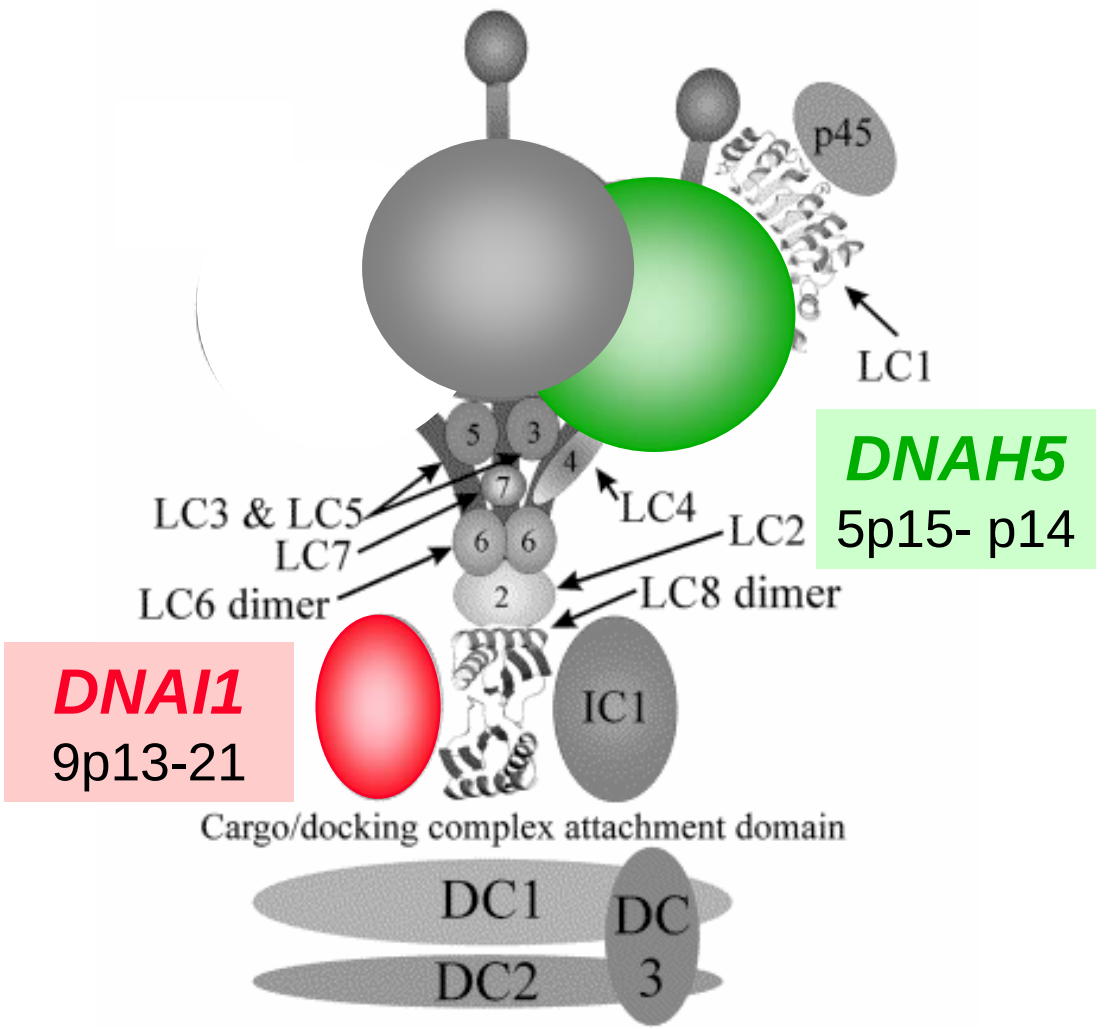
BDE : anomalie la plus fréquente

anomalie ciliaire guide les analyses génétiques

Analyse moléculaire des DCP



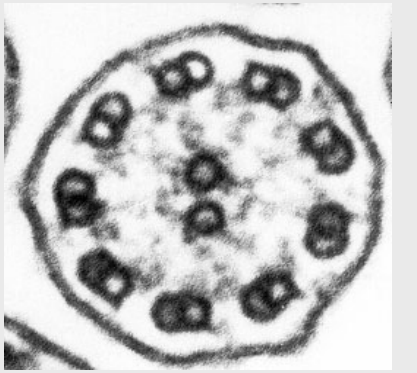
Gènes majeurs identifiés dans les DCP



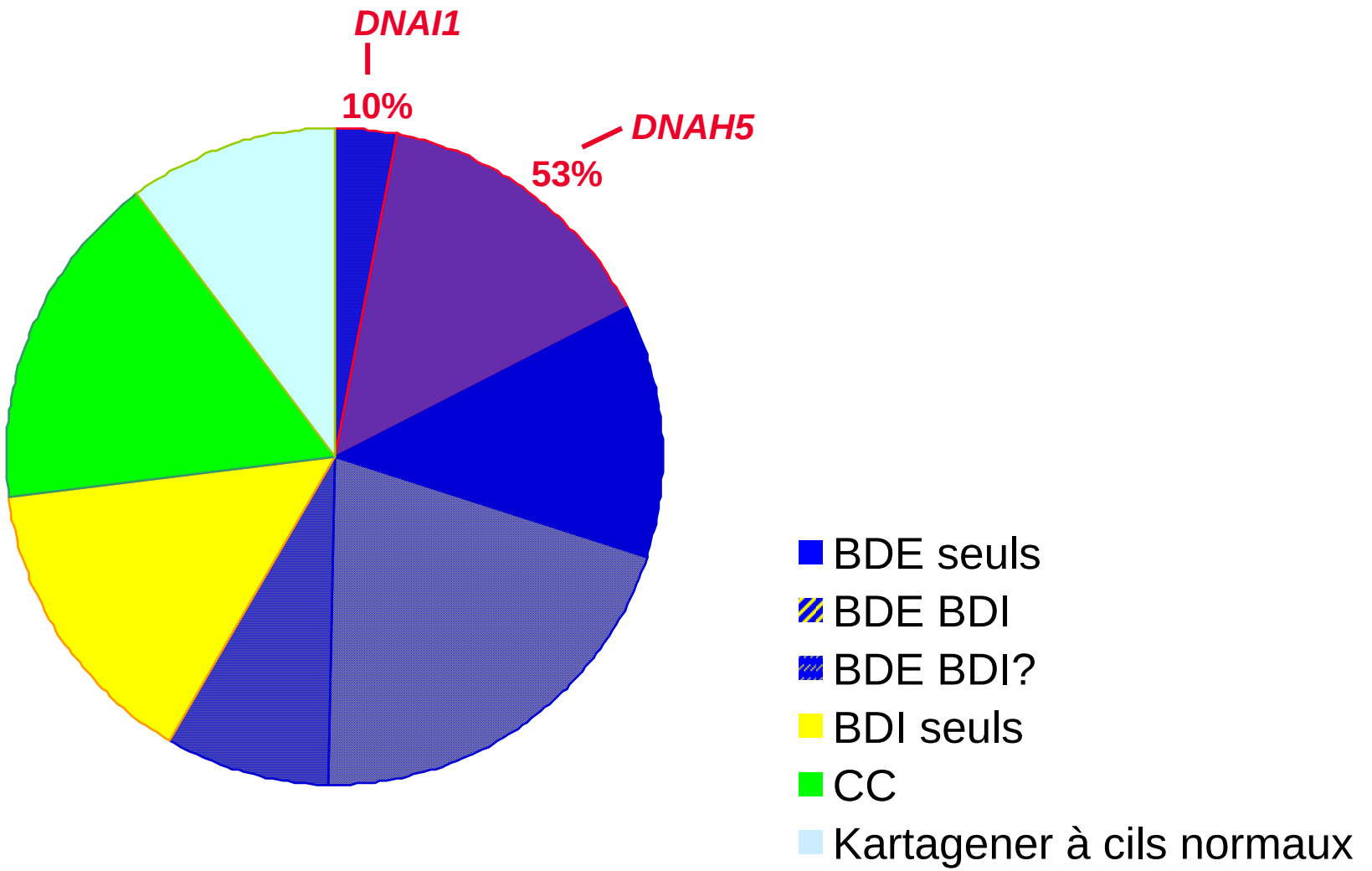
DNAH5
5p15- p14

DNAI1
9p13-21

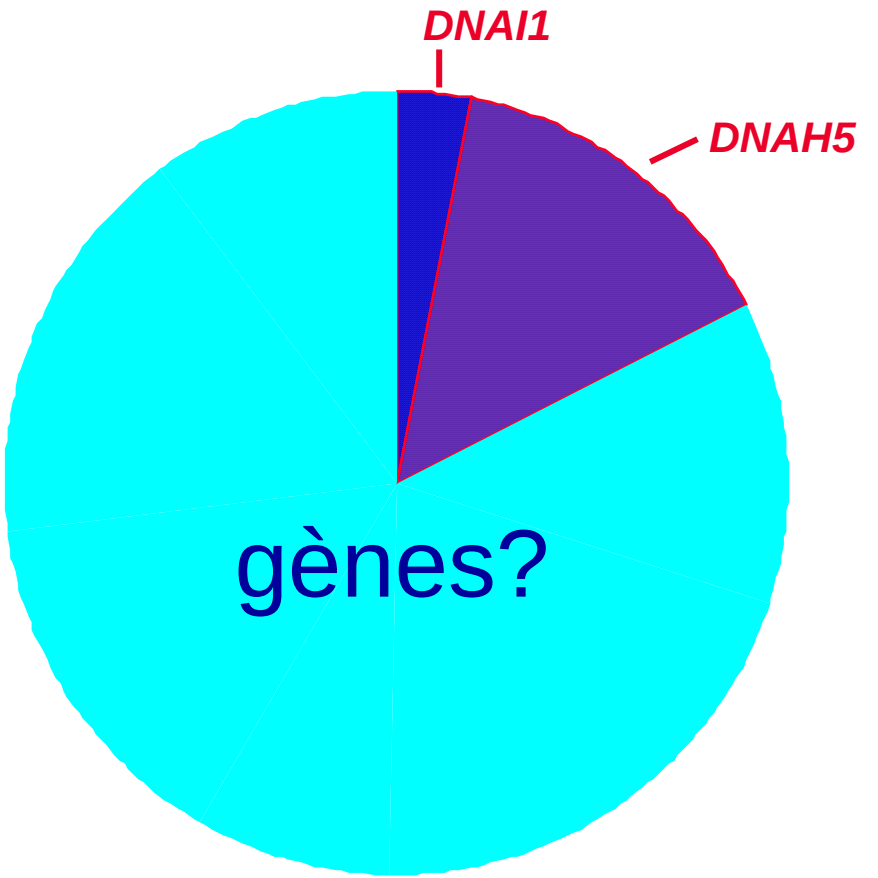
phénotype BDE

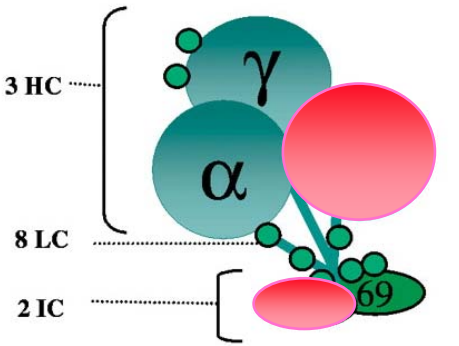


Gènes impliqués dans les DCP



Gènes impliqués dans les DCP





- BDE seuls
- BDE BDI
- BDE BDI?
- BDI seuls
- CC
- Kartagener à cils normaux

Génétique

Serge Amselem

Anne Marie Bridoux

Philippe Duquesnoy

Bénédicte Duriez

Anne Moore

Microscopie électronique

Issam Abd El Samad

Anne Marie Vojtek

ORL

Michel Boucherat

André Coste

Jean-François Papon

Roger Peynègre

Virginie Prulière Escabasse

Gilles Roger

Pneumologie Pédiatrie

Annick Clément

Jacques de Blic

Christophe Delacourt

Bruno Housset

Bruno Mahut

Aline Tamalet

Merci à tous les patients qui nous ont fait confiance et ont permis de progresser

soutenu par APHP, ANR INSERM, AFM, FRM, Legs Poix Fondation Carvajal