



Lungnasjúkdómar barna

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barnalungnalæknir

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Öndunarfæraeinkenni

- Hósti
- Stridor
- Mæði
- Öndunarerfiðleikar
- Blóðhósti
- Hæsi
- Ræskingar
- Nefstíflur
- Hrotur

Hósti

- Algengasta einkenni lungnasjúkdóms
- Mjög algengt einkenni hjá lögnum börnum sem leitað er með til læknis
- 10% frískra barna með kvef hóstar í meira en 2-3 vikur.

Hósti

- Bráður hósti
 - Sýkingar
 - Aðskotahlutur
 - Astmi
- Langvinnur hósti > 4 vikur
 - Astmi
 - Krónískur lungnasjúkdómur
 - Anatómísk afbrigði
 - Bakflæði
- Endurtekinn langvinnur hósti

Hósti

- Hve lengi?
- Hvernig byrjuðu veikindin?
- Áður svipuð einkenni?

Hósti

- Einkenni frá efri öndunarvegum
 - Nefrennsli/nefstíflur
 - Stridor
- Einkenni frá öðrum líffærum
 - Kvið
 - Efri öndunarfæri
- Vöxtur og þroski
- Aukahljóð frá öndunarvegi
- Mæði

Mismunandi tegundir hósta

teiknimynd

<https://www.youtube.com/watch?v=UDG6z5AiU-A&vl=en>

Croup:

<https://www.youtube.com/watch?v=xwOfOgY8Ye8> 1:46

<https://www.youtube.com/watch?v=zGWZSTkAotI&list=PLKUX6UbAKeUCY3oLIEj5thB7YojOcPozU>

RS

<https://www.youtube.com/watch?v=d1mv4VZ4HiM> 1,4 0 2 m

Kikhósti

<https://www.youtube.com/watch?v=8Ka2h5bLVjw>

Skoðun

- Almennt ástand
- Hvernig hljómar hóstinn
- Hlustun
 - Hvæsiöndun
 - Stridor
 - Brak
 - Slímhljóð
- HNE
- Clubbing
- Mettun

Series: 1001
C: 315,0, W: 981,2
S: 124



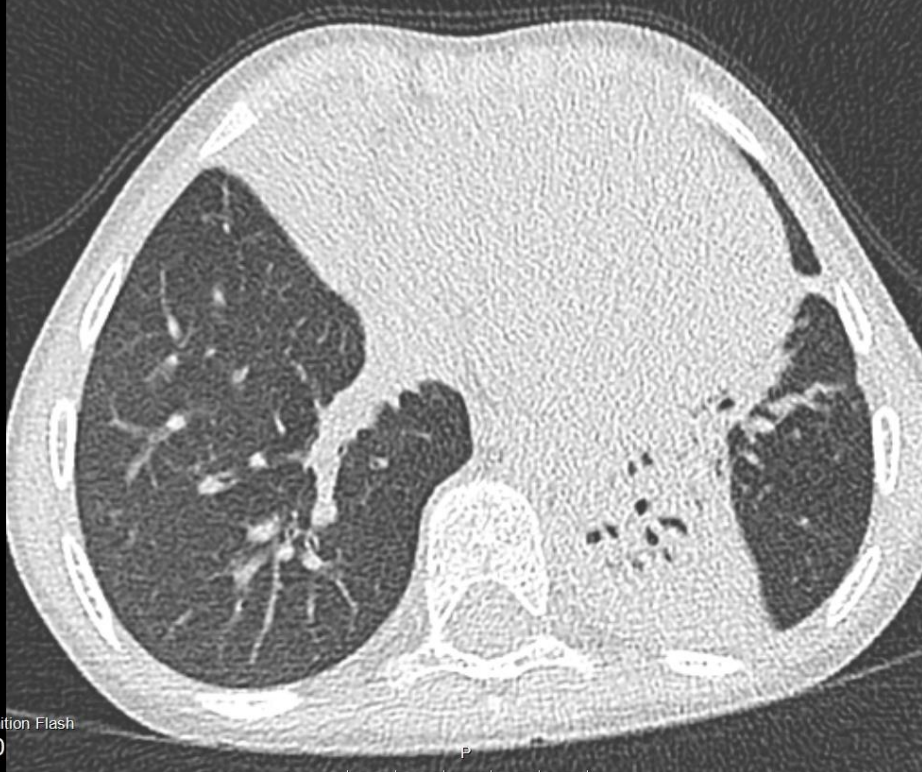
BFC USiL Lund
2009-11-11 13:03:26

LUNGA BARN LIGGANDE FRONTAL

FoV: 192 mm

Series: 2
Frame 298 av 522
Nivå: 420 mm
Snitt: 0,75 mm
Pos: FFS
C: -565,0, W: 1685,0
WINDOW1 1/13
Z
R
L

LU Röntgen 1 BFC
I70f1
Time: 285 ms
27 mAs
58 mA
100 kV
Model name SOMATOM Definition Flash
2013-01-30 15:12:20



HRCT SPIRAL 0.75 I70f 1
HRCT



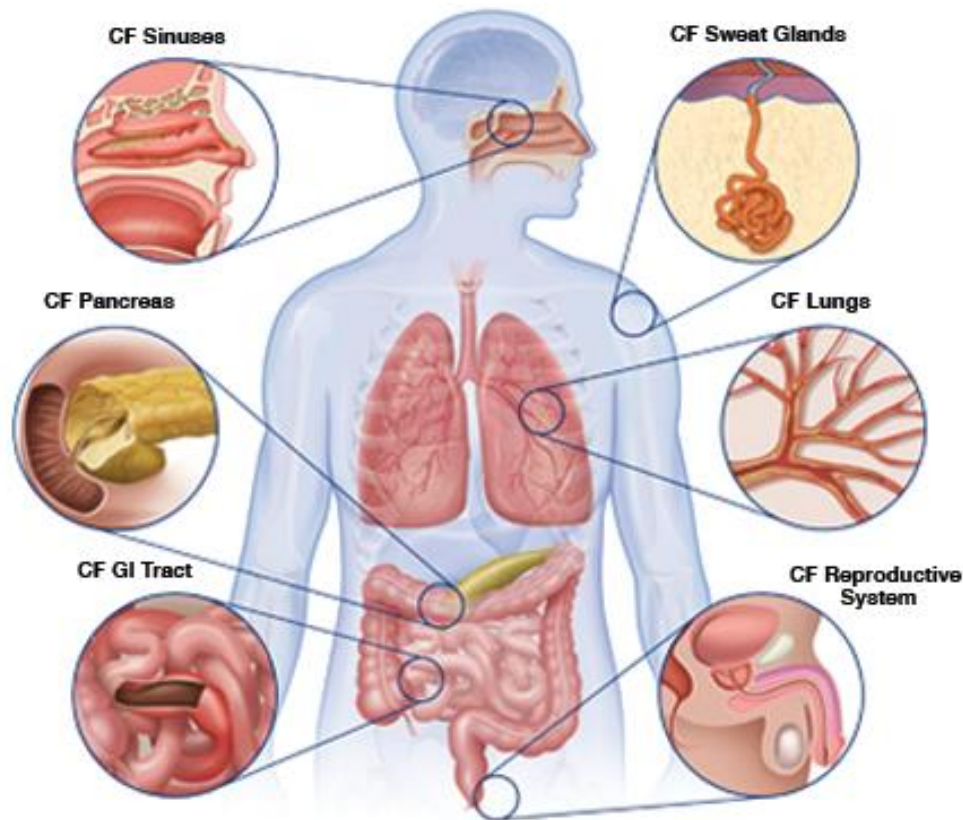
L
F

Tilfelli

- 9 mánaða drengur
- Eðlileg meðganga og fæðing
- Greindur með bronkit/astma á Hg
- Þyngdist ágætlega fyrstu mánuðina
- Gengið verr sl mánuði
- Órólegur og alltaf svangur

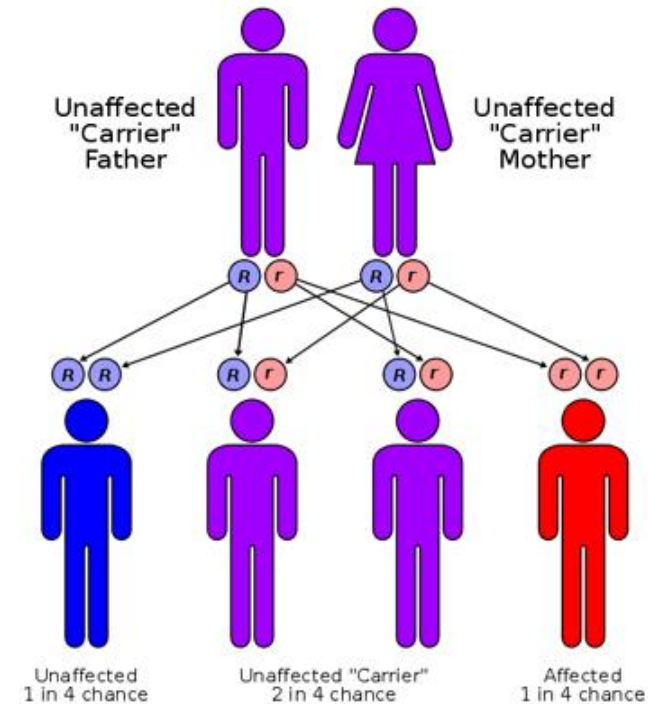
CF – Fjölkerfa sjúkdómur

- Lungu
- Bris
- Efri öndunarfæri
- Lifur
- Meltingarvegur
- Liðir og bein
- Vas deferens
- Eggjastokkar
- Svitakirtlar



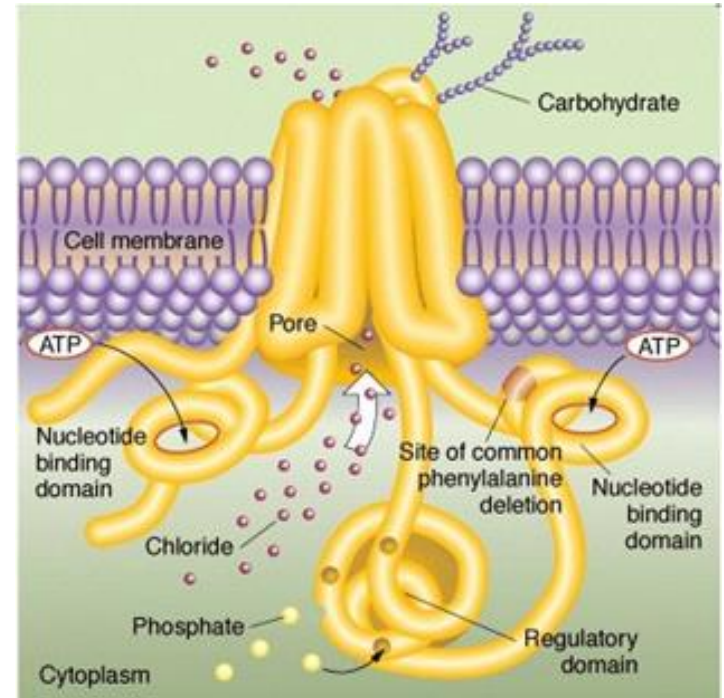
CF genið

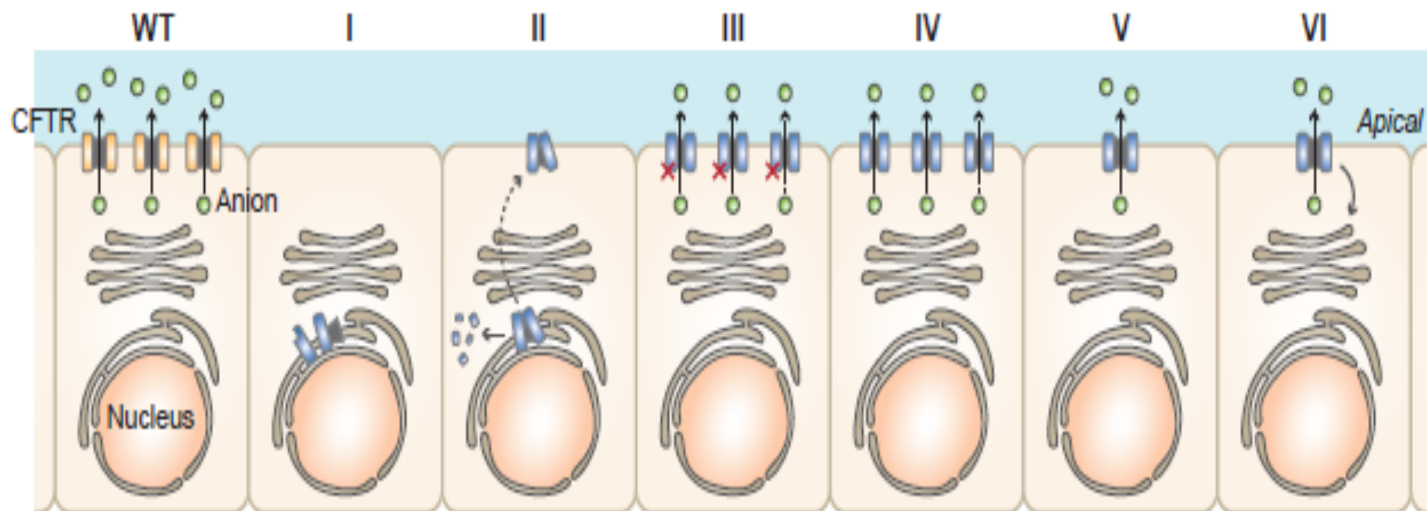
- Galli í CFTR geni á litningi 7 (7q31.2)
- Víkjandi erfðir
- Yfir 2000 þekktar genabreytur
- Delta F508 (80-90%)
- Genabreytum er skipt í flokka 1-6
- Tíðni mismunandi eftir löndum



CF próteinið

- Cystic fibrosis transmembrane conductance regulator
- Prótein í frumuhimnu
- Klór- og bikarbónatgöng



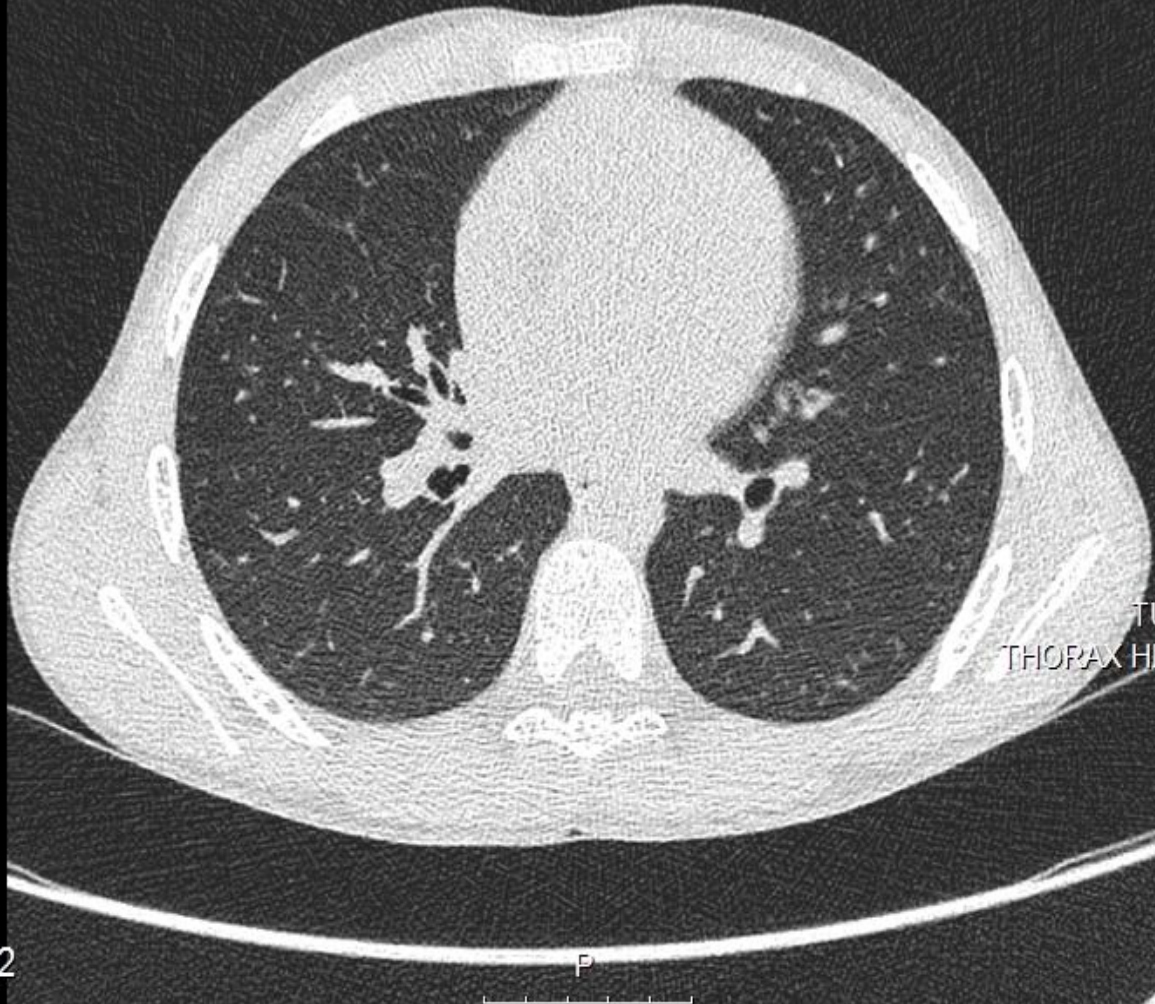


Defect	Non-functional protein synthesis	Impaired folding and trafficking	Defective regulation/gating	Impaired conductivity	Reduced levels	Reduced stability
Mutation examples	G542X W1282X N1303K 711-1G>T	F508del A455E	G551D S1251N R1349D F508del	R117H R334W L927P	TG(13)T(5) A455E	c.120del23 F508del

FoV: 278 mm

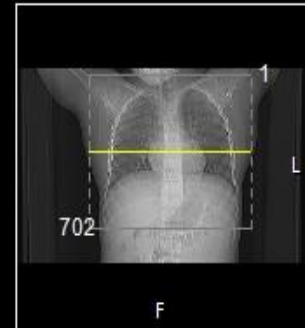
FEV₁ 115%

Series: 201
Bild 352 av 702
Nivå: -175 mm
Snitt: 0,67 mm
Pos: FFS
C: -500,0, W: 1500,0
C=-500,0, W=1500,0 1/13



TUNNA SNITT, iDose (3)
THORAX HRCT 11-50 KG /Thorax

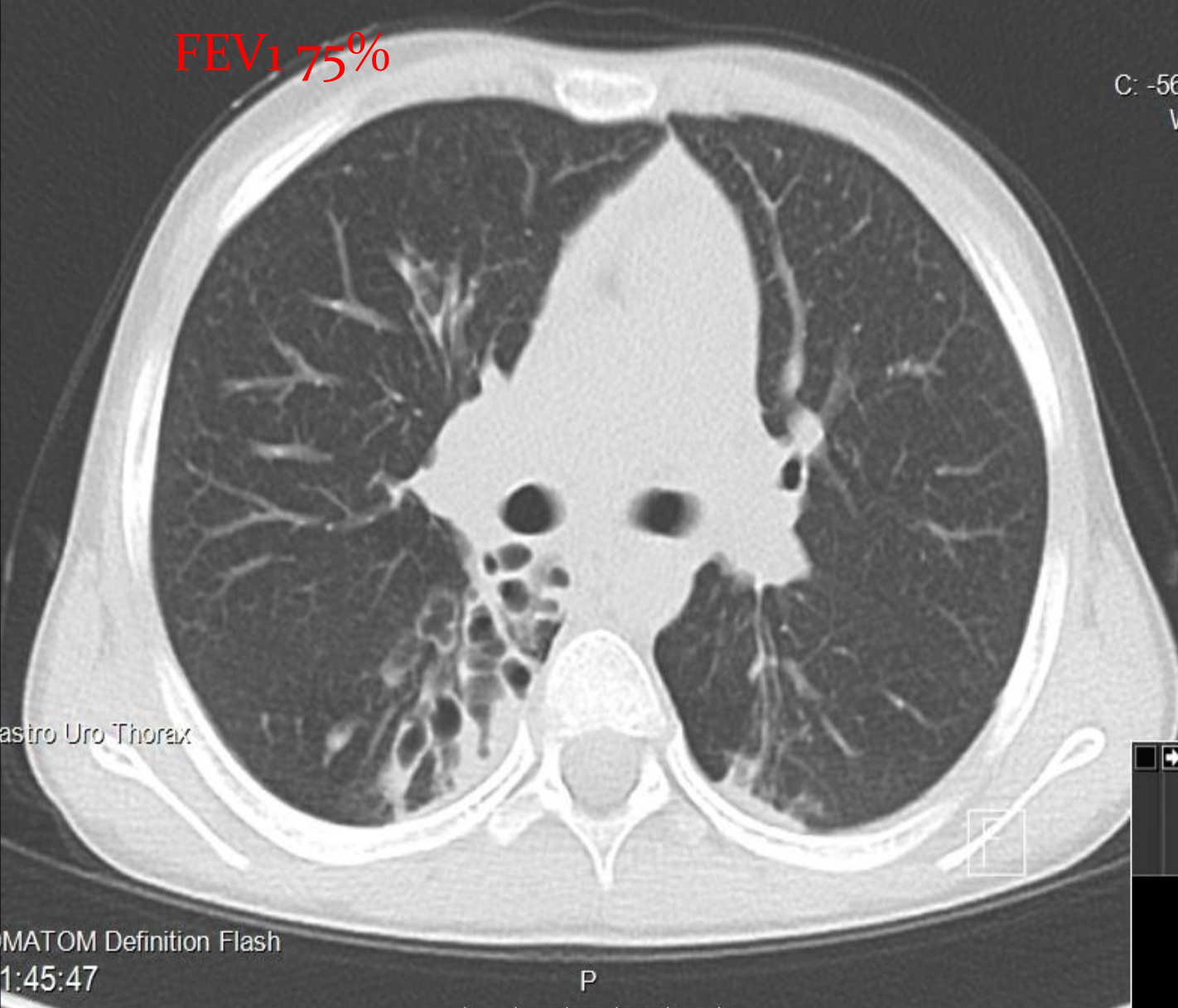
CT Lab 2 Lund
YCI
Time: 565 ms
48 mAs
85 mA
100 kV
Model name iCT 256
2016-12-01 10:27:12



FoV: 233 mm

FEV₁ 75%

Series: 6
Bild 15 av 44
Snitt: 5 mm
Pos: FFS
C: -565,0, W: 1685,0
WINDOW1 1/13
Z R L



L

TRA
HRCT



F

LUN Röntgen Gastro Uro Thorax
I70R1
Time: 285 ms
41 mAs
88 mA
100 kV
Model name SOMATOM Definition Flash
2015-09-03 11:45:47

P

Einkenni - börn

- Mekonium ileus
- Langdregin gula
- Slímhósti
- Sýkingar
- Lausar hægðir, oft á dag, miklar hægðir, ómeltur matur, olíukenndar
- Rektal prolaps
- Vanþrif
- Nefstíflur
- Saltbragð

Einkenni - eldri

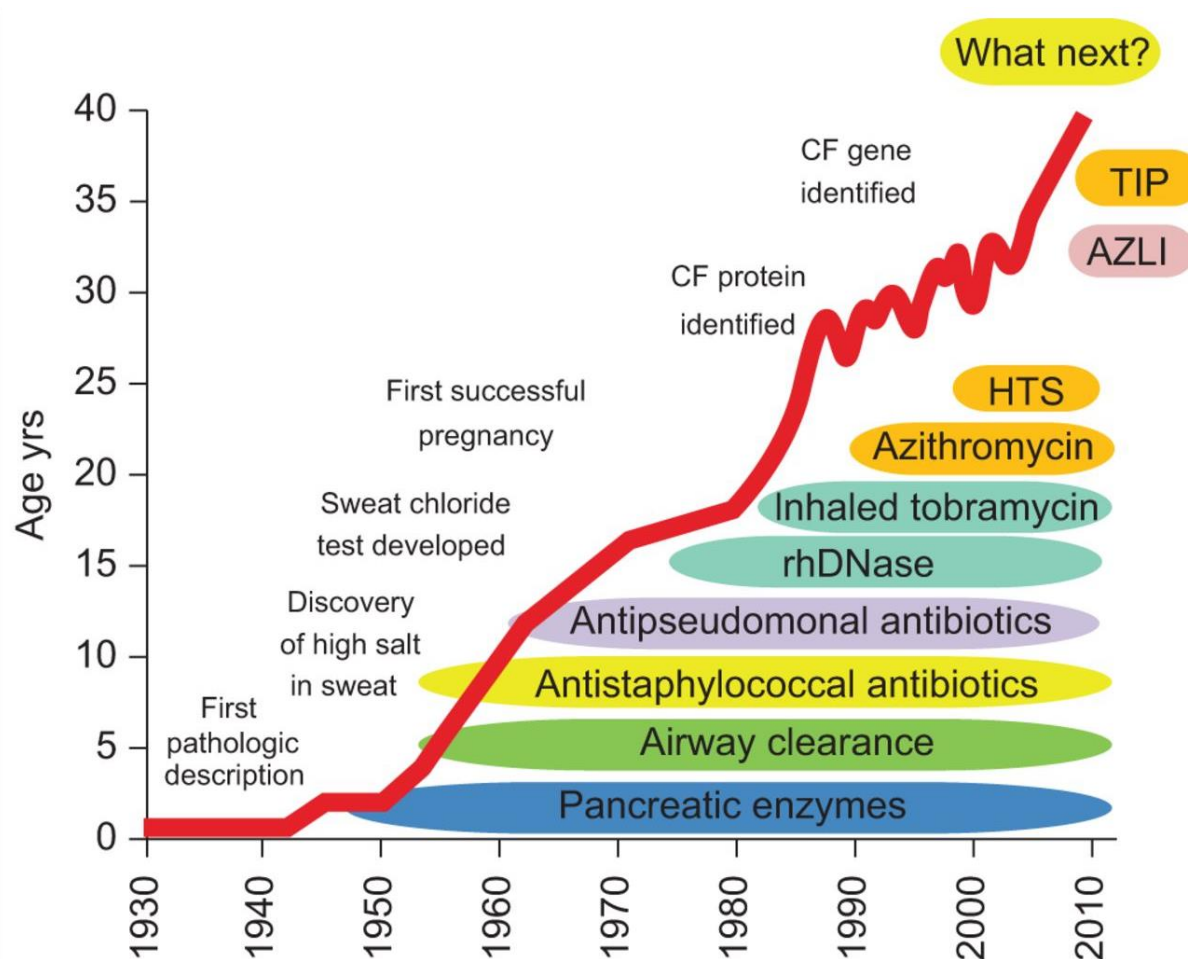
- Seinkaður kynþroski
- Brisbólga
- Kinnholubólgur
- Nefsepar
- Ófrjósemi

Cystic fibrosis í Evrópu

Outcome		Females	Males	Total
Patients registered in the ECFSPR	n (%)	21220 (47.45)	23499 (52.55)	44719
Age at follow-up (in years; patients alive on 31/12/2016)	mean	20.5	21.4	21.0
	median	18.3	19.5	19.0
Patients ≥ 18 years (patients alive on 1/12/2016)	%	50.7	53.8	52.4
Age at diagnosis*	mean (years)	4.2	4.1	4.1
	median (months)	4.2	4.0	4.1
Patients with at least one F508del allele recorded*	%	82.4	82.3	82.4
Patients living with lung transplant*	n (%)	1152 (5.7)	1162 (5.2)	2314 (5.4)
Patients living with liver transplant*	n (%)	90 (0.4)	157 (0.7)	247 (0.6)
Patients deceased in 2016**	n	254	225	479
	(%)	(1.2)	(1.0)	(1.1)
Age at death (years)**	mean	31.5	31.2	31.4
	median	29.0	30.0	30.0

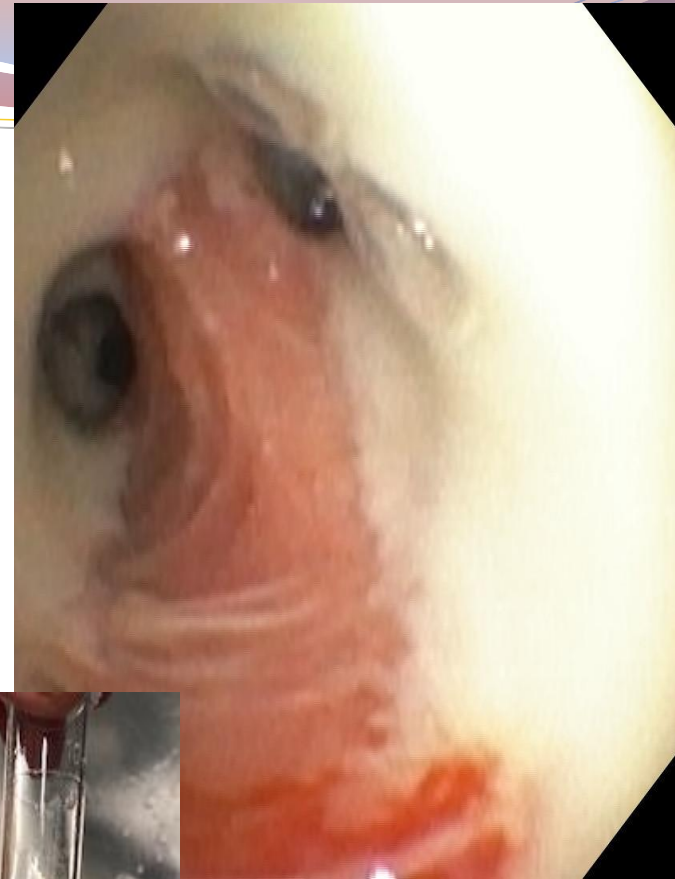
Betri meðferð-lengri lifun

CFTR
modulators



CF Lungu

- Þykkj slím
- Bólga
- Sýkingar
- Bronchiectasis
- Fibrósa
- Skert lungnastarfsemi
- Öndunarbílun
- Ígræðsla/dauði



Lungnastarfsemi

Lung function

For the evaluation of lung function all patients from 6 years without transplantation, with lung function measurement 2018 were considered. A total of 4997 data sets were available.

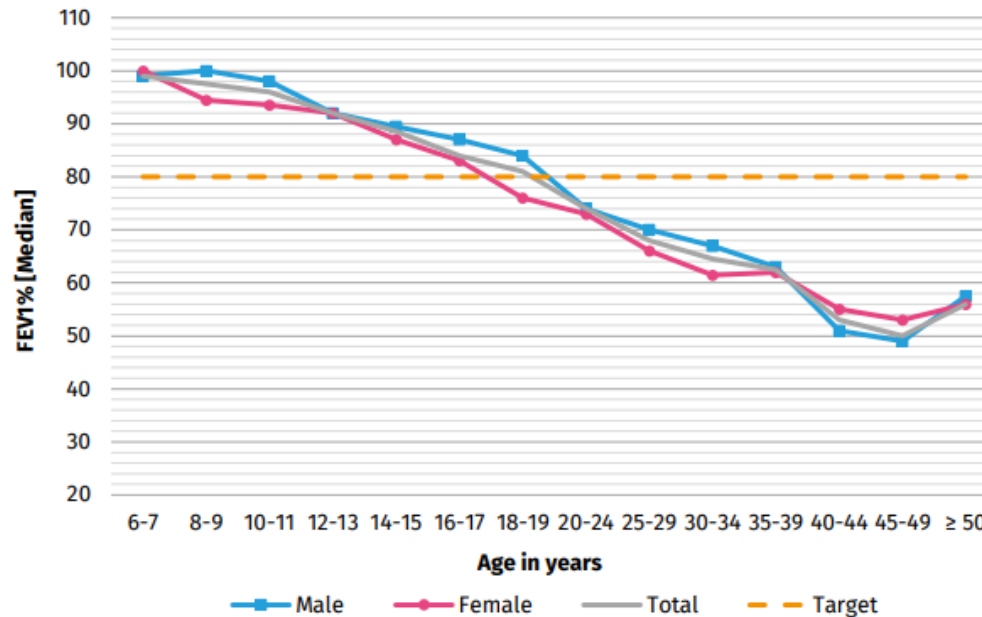
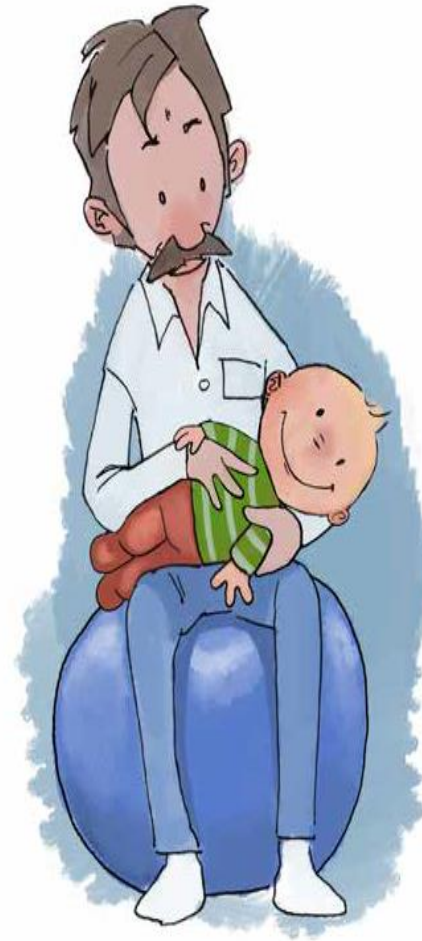


Figure 10: FEV1% value acc. Global Lung Initiative 2018

Lungu- meðferð

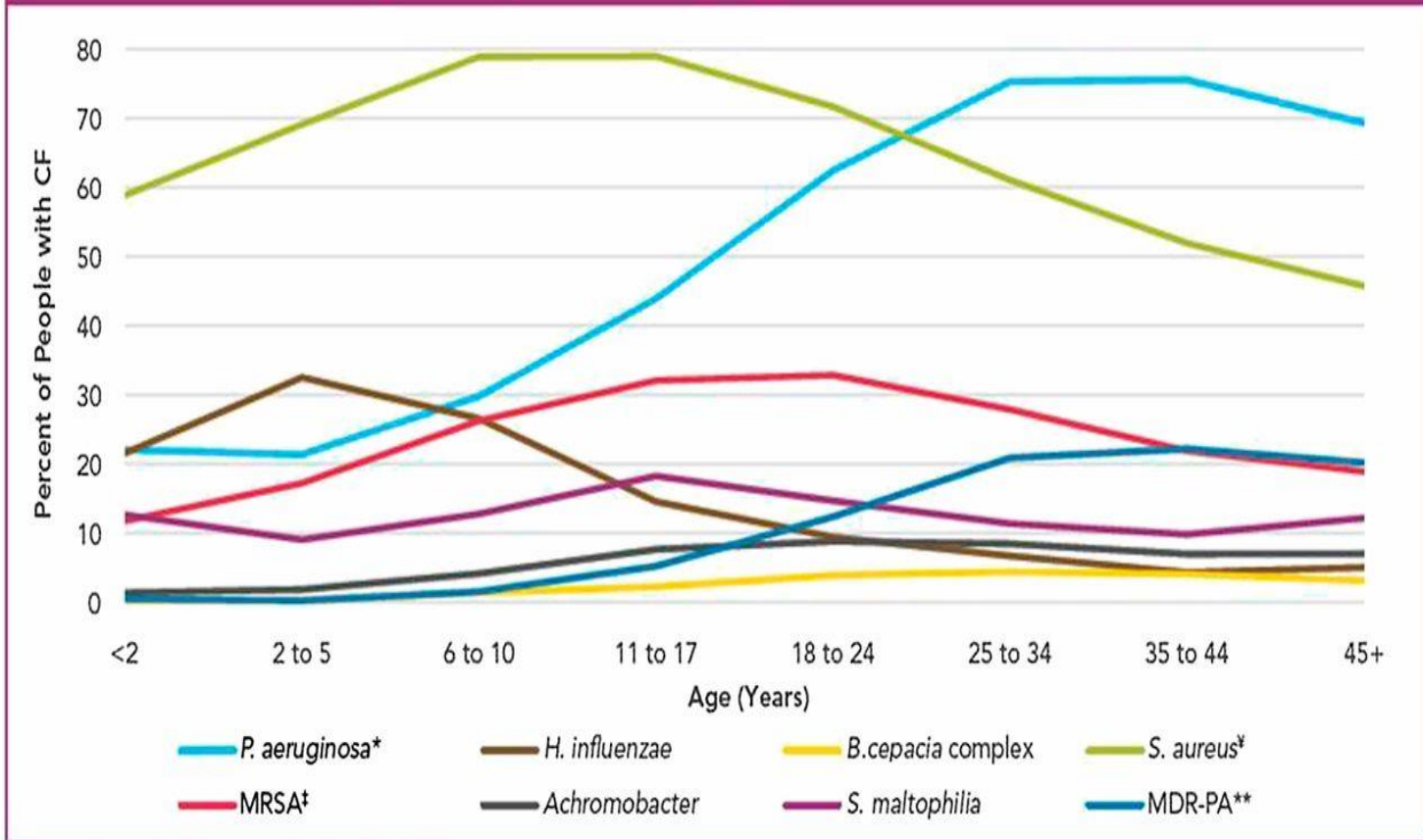
- Slímlosandi meðferð
- Öndunar sjúkraþjálfun
- Innúðalyf
 - Hyperton saltvatn
 - Pulmozyme
- Sýklalyf
 - Per os, iv, innúða
- Hreyfing!
- Berkjuvíkkandi lyf
- Bólguminnkandi lyf







Germ Found in the Lungs of People with CF by Age, 2012

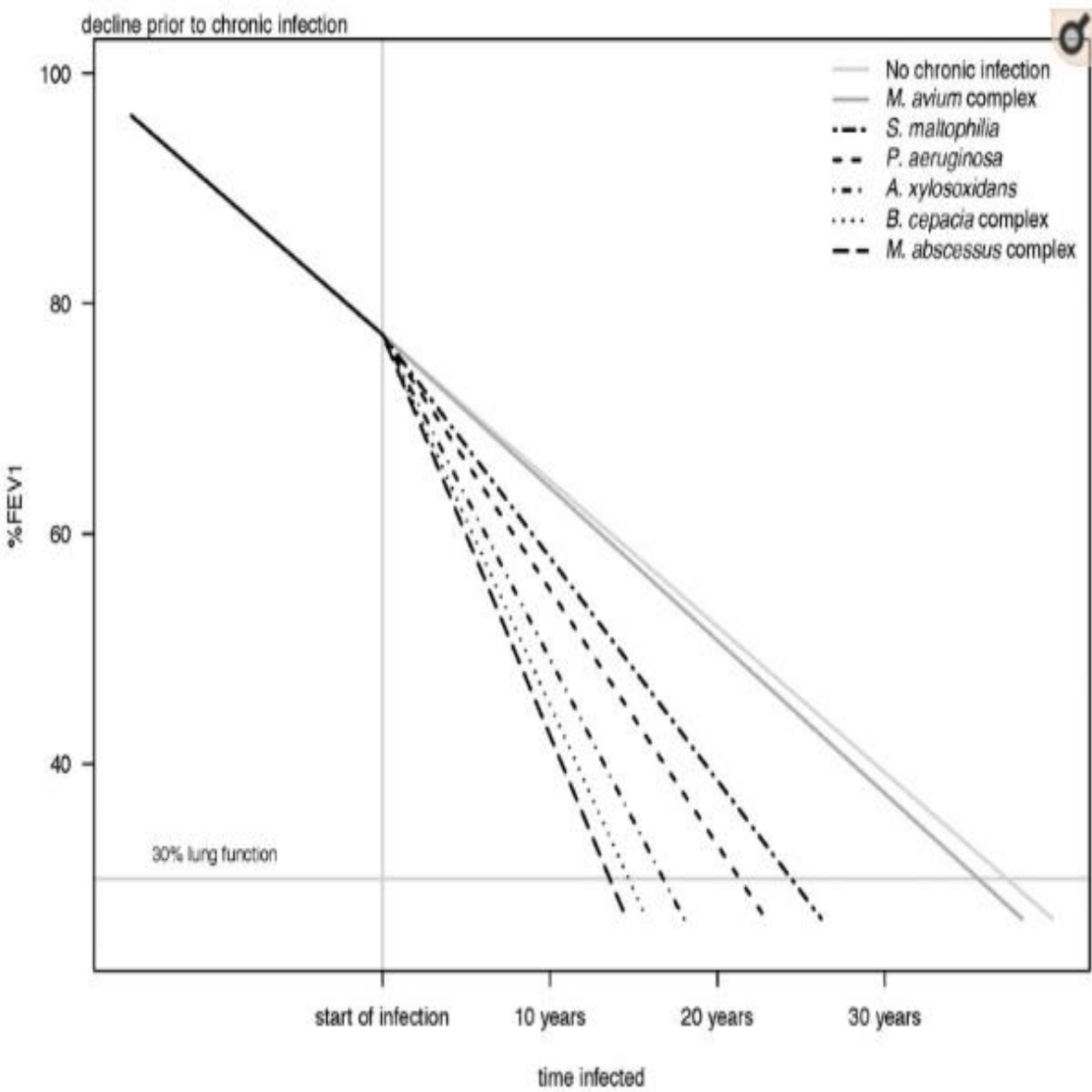
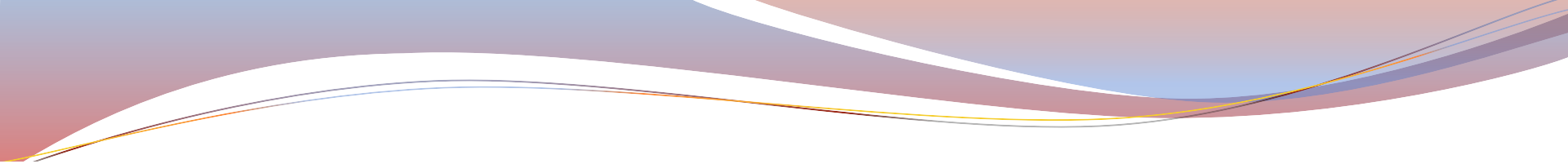


**P. aeruginosa* includes people with MDR-PA.

**MDR-PA is multi-drug resistant *Pseudomonas aeruginosa* (*P. aeruginosa*).

‡ *S. aureus* includes people with MRSA.

‡ MRSA is methicillin-resistant *Staphylococcus aureus* (*S. aureus*).



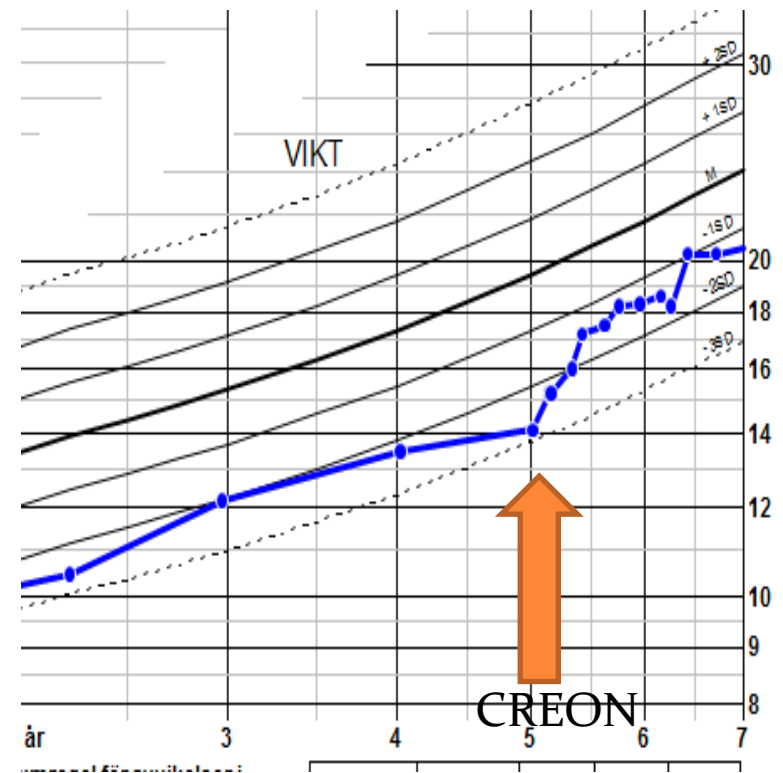
Qvist et al. J of Cystic Fibrosis 2016

Bris

- Exocrine vanstarfsemi
 - 90%
 - Malabsorption
 - Vannæring
 - Skortur á fituleysanlegum vítamínum og fitusýrum
- Endocrine vanstarfsemi
 - CF tengd sykursýki (CFRD)
 - Skimað fyrir
 - Eykst með hækkandi aldri
- Pankreatit
 - Sufficient=>insufficient

Bris - meðferð

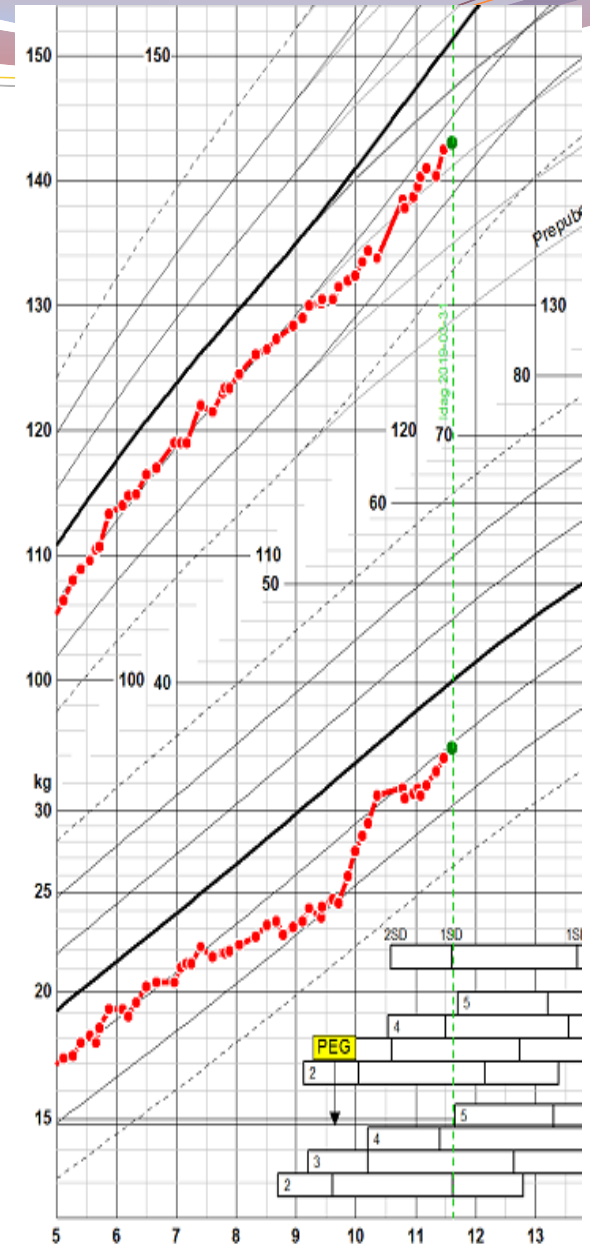
- Meltingarensím
 - Creon / Pertzye
- Vít A D E K
- Insulin við CFRD



Meltingarfæri

- Mekonium ileus
- Magabólgur og reflux
- Malabsorption
- Steatorré
- Rektal prolaps

- Eftir greiningu
 - Hægðatregða
 - DIOS
 - Lystarleysi
 - Kviðverkir



Lifur og gallvegir

- Hyperbilirubinemia /gallstasi hjá nýbura
- Gallstasi/sludge/ steinar
- Hepatic steatosis
- Focal biliary cirrhosis
- Multilobular biliary cirrhosis
- Cirrhosis=>portal hypertension=>esophageal varices
- CF lifrarsjúkdómur(CFLD)
 - 30%
 - Fyrir tvítugt
 - Skimað fyrir

Greining

- Svitapróf
 - Nanodukt aðferð <50 eðlilegt
 - Macrodukt aðferð <30 eðlilegt
 - 30-60 grátt svæði
- Feces-elasticity >200 eðlilegt
- Genarannsókn

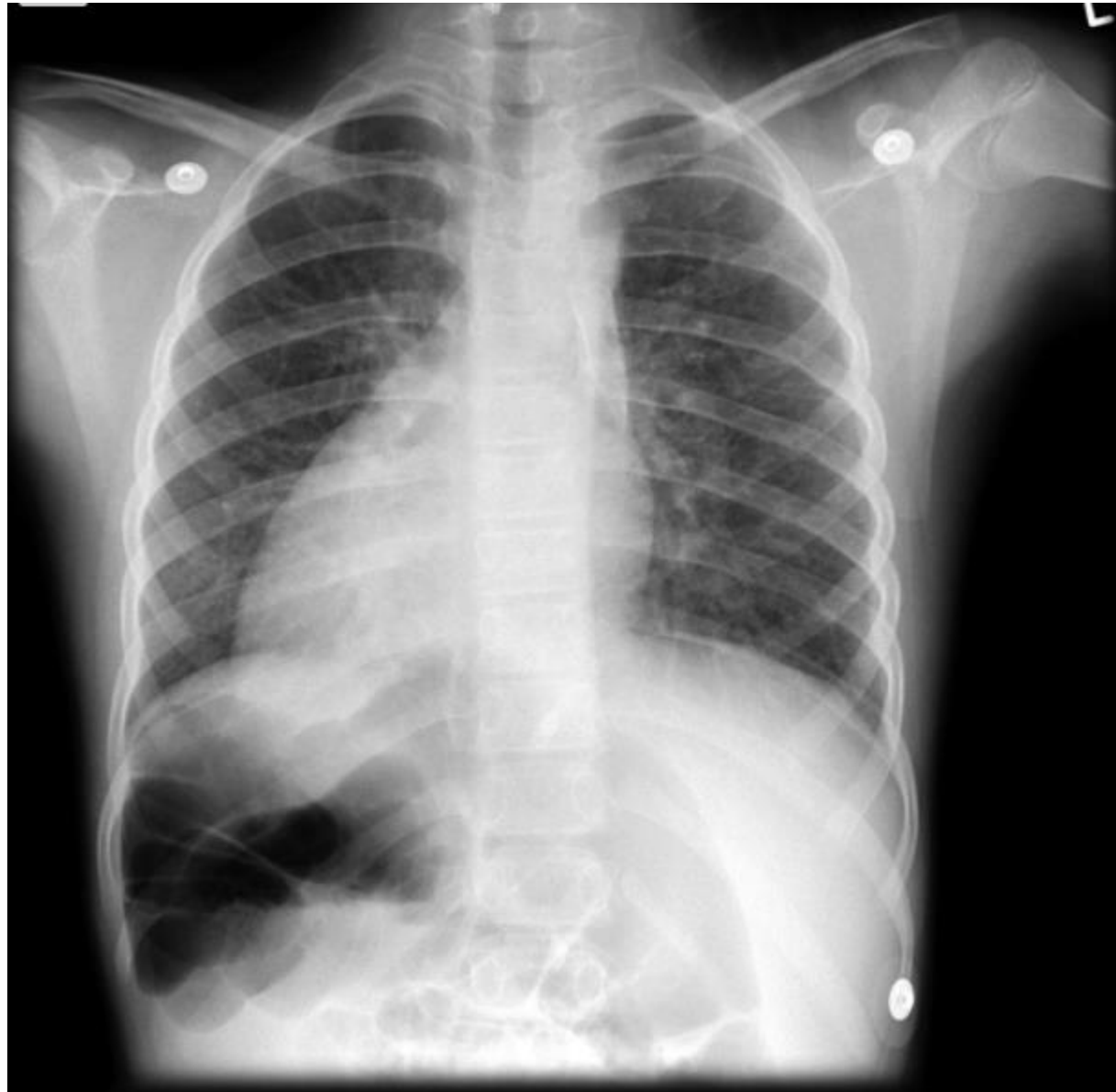


Markmið

- Viðhalda lungnastarfsemi
- Gott næringarástand
- Dagleg meðferð sem virkar fyrir barnið
- Lifa eðlilegu lífi

Tilfelli

- 6 ára barn
- Alltaf með kvef
- Oft með slímhósta
- Fengið eyrnabólgur amk 10 sinnum
 - Fengið rör
- Engin meltingarfæraeinkenni
- Vex eðlilega



Primary ciliary dyskinesia PCD

- Víkjandi erfðir
- Prevalens 1:2200-40.000
- Mörg mismunandi gen (28+)
- Margar mismunandi genabreytur
- Syndromal genetic PCD variants
 - X-linked td Joubert sx
- Situs inversus 50%

PCD

- Nefstíflur
 - Frá nýburaskeiði
- Eyrnabólgur
 - Eiga ekki að fá rör
- Slímhósti
- Burstapróf í rafeindasmásjá
- Nasal NO
- Genapróf
- Virknipróf
- Immunofluorescence

Cilia

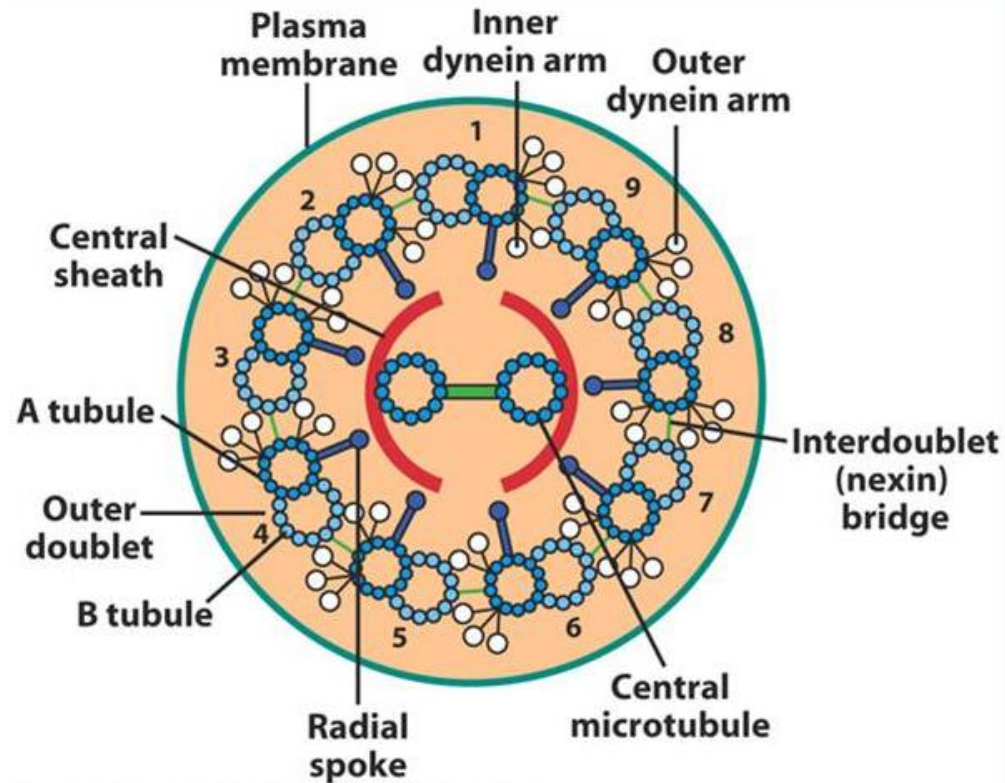


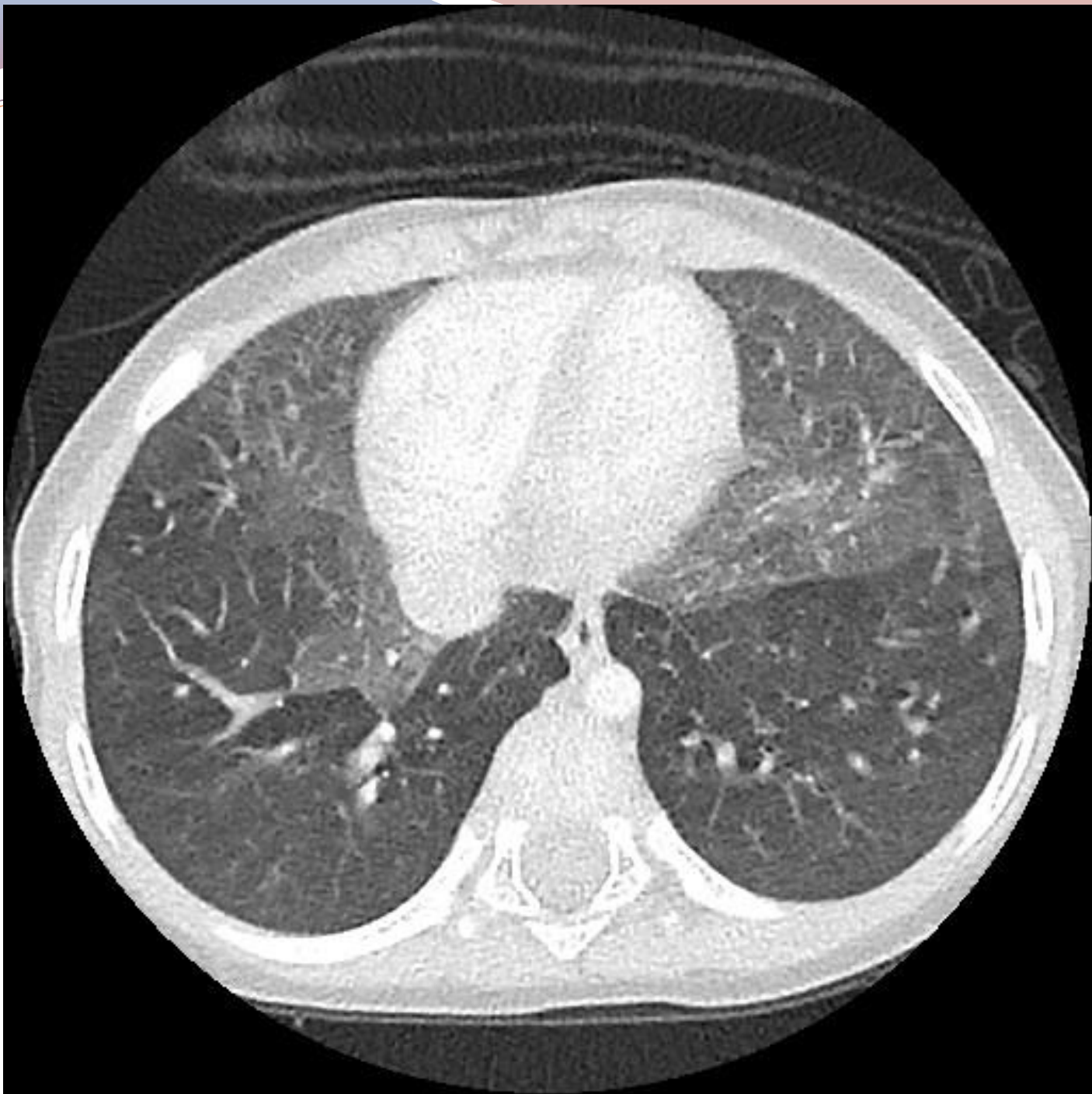
Figure 9-30b Cell and Molecular Biology, 4/e (© 2005 John Wiley & Sons)

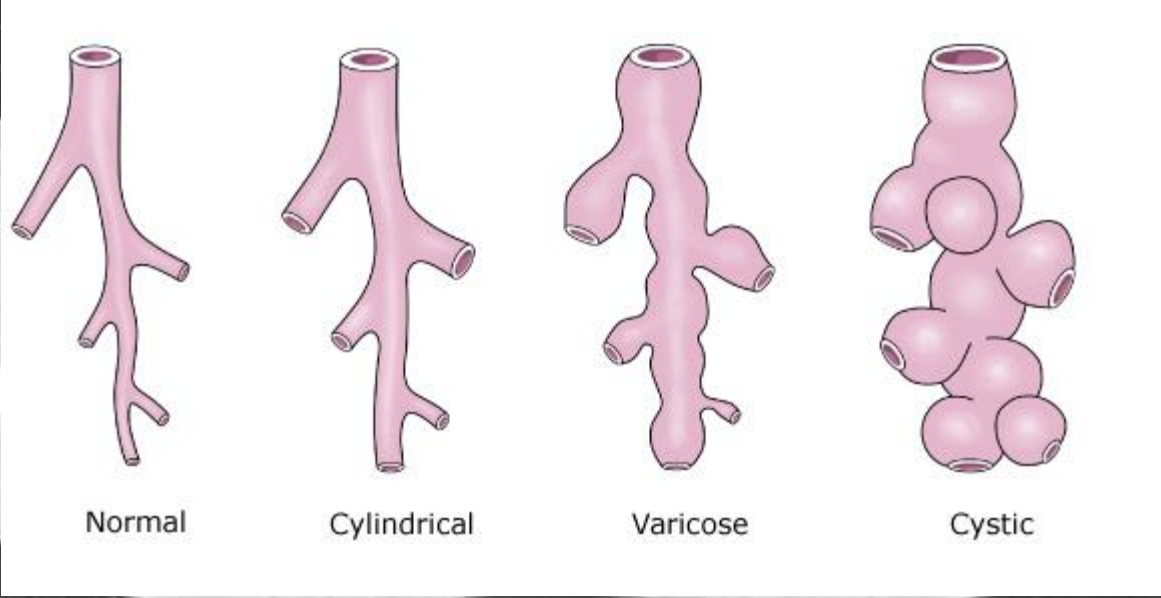
CF vs PCD

	CF	PCD
Hósti/slím	++++/þykkt og seigt	++++/laust og þynnra
Efri öndunarfæri	++	++++
Eyrnabólgur	+	++++
Nefsepar	++	+
Bronchiectasis	Byrja oftast apikalt	Byrja í lobus med/lingula
Bakteríur	S.aur, psa	H. influenza, pneumokokkar
Öndunarfæraeinkenn i nýbura	+	+++
Ófrjósemi kk	+++	+++
Metingarfæraeinkenn i	+++	-

Eitt og annað

- Meðfæddir anatómískir gallar
- Langvinnur lungnasjúkdómur fyrirbura
- Astmi
- Ónæmisgallar
- Bronkiektasíur
- Interstitial lungnasjúkdómar





Normal

Cylindrical

Varicose

Cystic

Dæmigerð uppvinnsla

- Myndrannsóknir
 - Rtg pulm
 - HRCT/CT með skuggaefni
- Lungnarannsókn
 - Spirometria
 - Body box, DLCO, IOS, MBW
- Öndunarfæraræktanir
- Ónæmisfræðilegar rannsóknir
- Status, D-vitamin, lifrarpróf, Na

Dæmigerð uppvinnsla

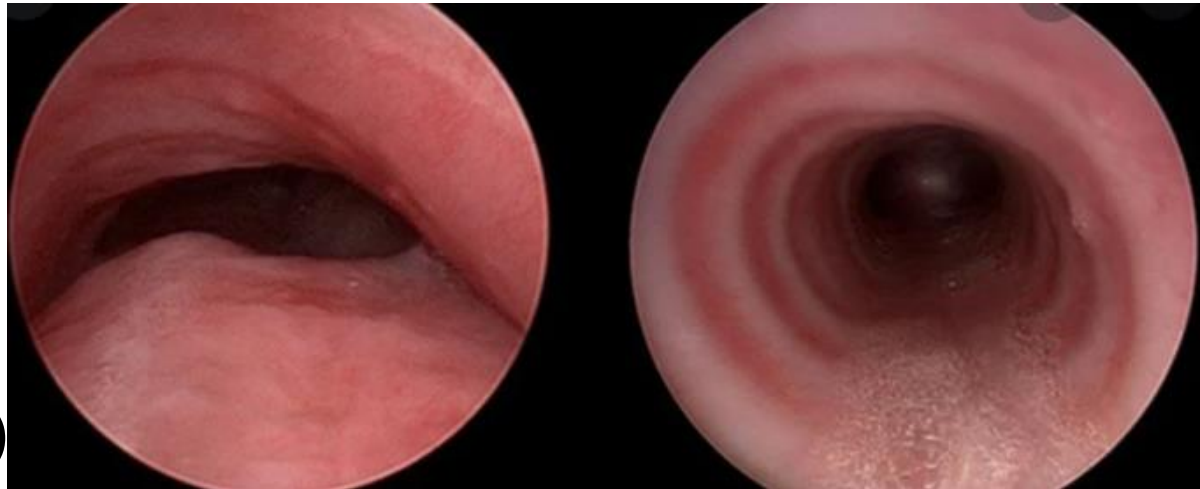
- Skoðun hjá HNE lækni
- Berkjuspeglun
- Svitapróf
- Elastasi í hægðum
- Genarannsóknir
- Cilia rannsóknir

Stridor - nýburar

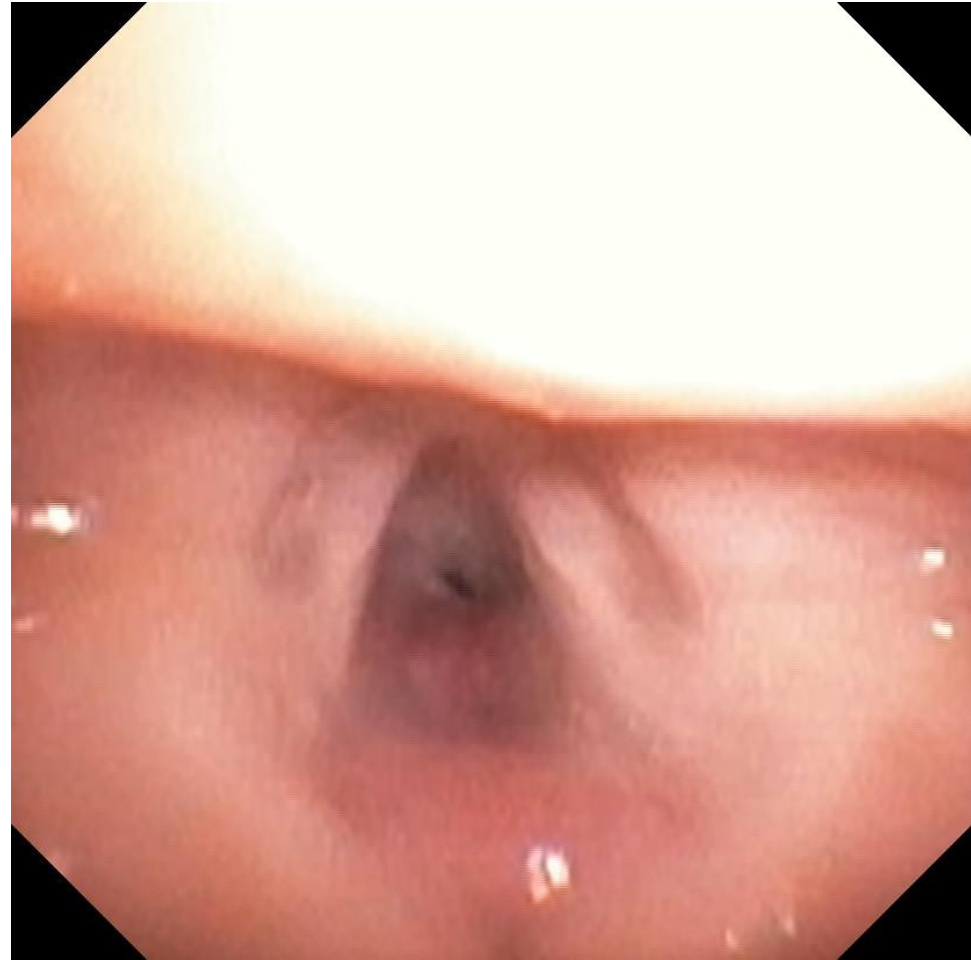
- Laryngomalacia
- Laryngeal malformation
- Raddbandalömun
- Tracheomalacia
- Æðahringur
- Stenosur
- Hemangioma
- Papilloma

Meðfædd anatómísk afbrigði

- Laryngomalacia
- Tracheomalacia
- Bronchomalacia
- Æðahringur
- Þrengsli(stenósusur)



- <https://share.icloud.com/photos/oyWqGgJEcbOyqyHuiWSlqMgog>

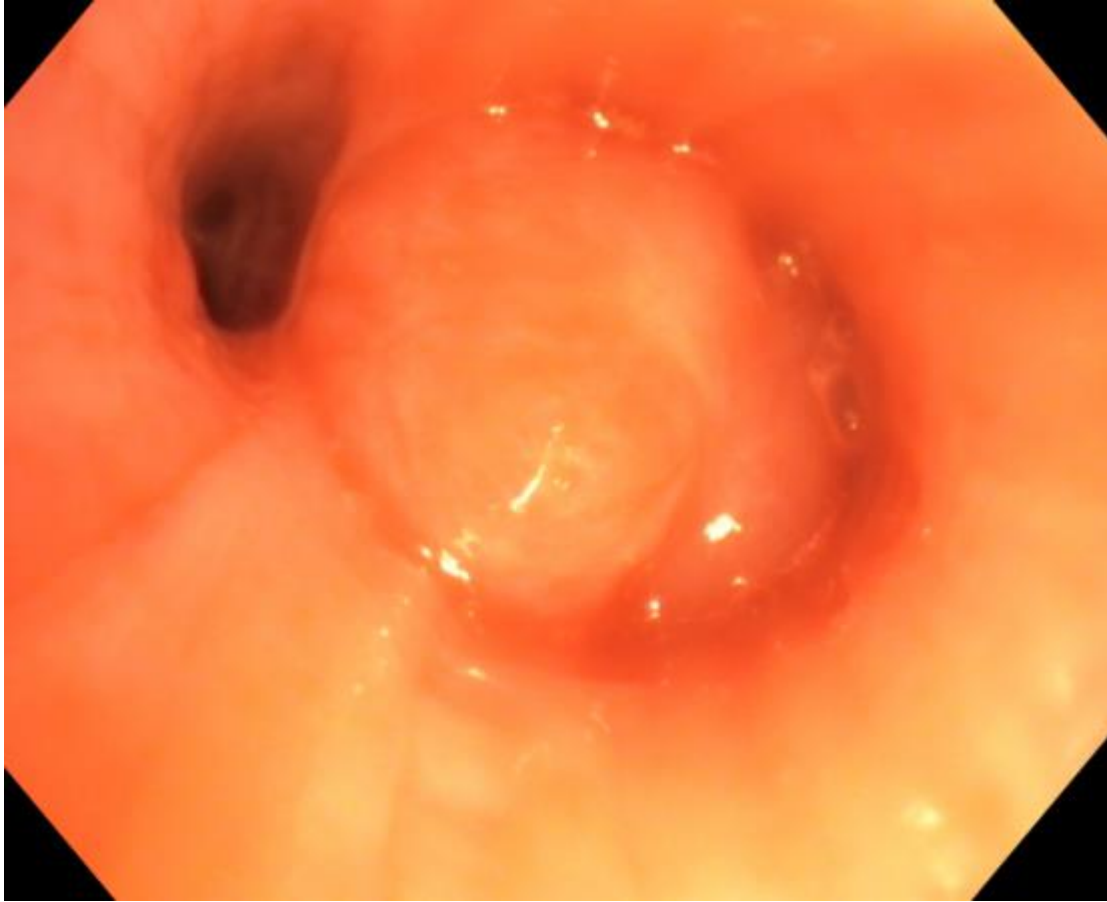


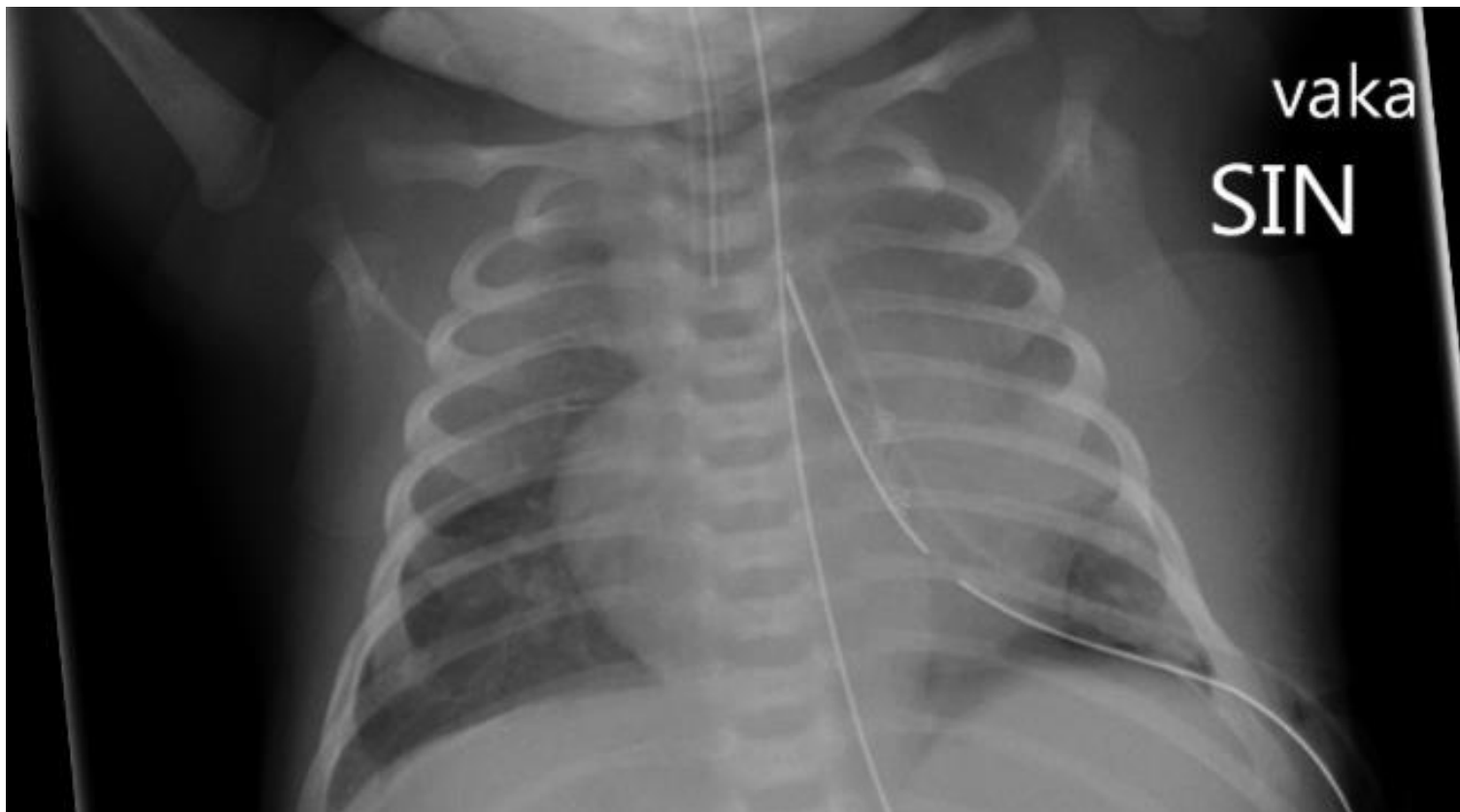
Stridor- brátt

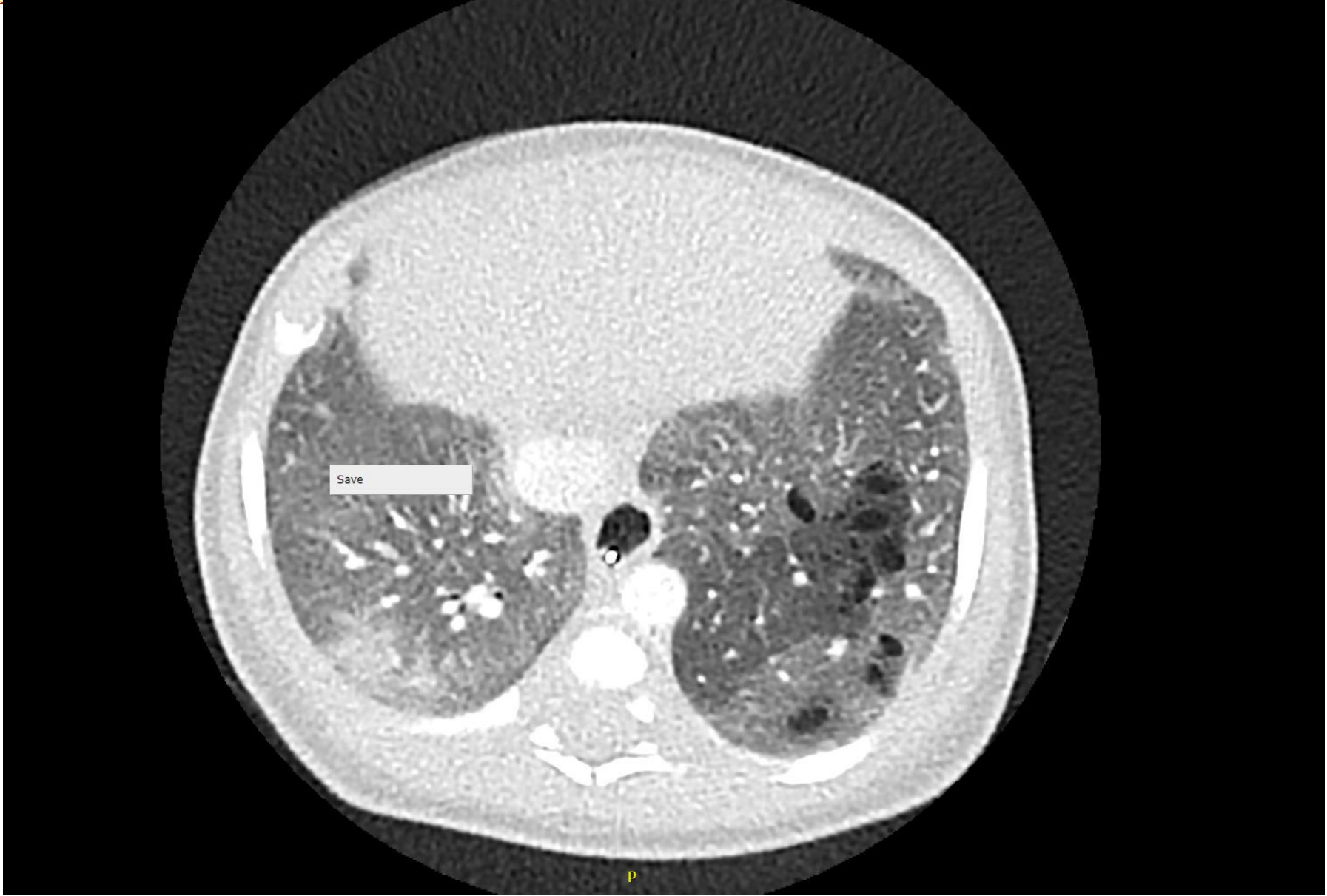
- Croup
- Epiglottit
- Tracheit
- Aðskotahlutur
- Abscess
- Anafylaxi

Stridor – eldri börn

- Vocal cord dysfunction
- Raddbandalömun
- Exercized induced laryngal obstruction
- Subglottic steonsa
- Tumor
- Tracheal web
- Hypocalcemii laryngeal spasm







Save



Meðfædd anatómísk afbrigði

- CCAM
- Pulmonary sequestration

Fleiri meðfæddir gallar

- Congenital diaphragma hernia
 - Lungnahypoplasia
 - Skert lungnastarfsemi
- Esophageal atresia með fistli
 - Tracheomlacaia
 - Hvæsiöndun
 - Endurteknar sýkingar
 - Hypersekretions vandamál

Svefntruflanir og öndun

- Kæfisvefn
 - Anatómískar orsakir, tons og ad
 - Craniofacial syndromes
 - Downs syndrome ofl
 - Offita
- Hypoventilation
 - Vöðva/taugasjúkdómar
- Trufluð central öndunarstjórn
 - CCHS
 - Chiari
 - Heilastofns lesionir

Einkenni svefntruflana

- Hrotur/öndunarhlé
- Dagþreyta
- Skert athygli, úthald og einbeiting
- Ofvirkni/Vanvirkni
- Undirmiga
- Pirringur
- Aukin/minnkuð matarlyst

Áhrif svefnháðra öndunartruflana

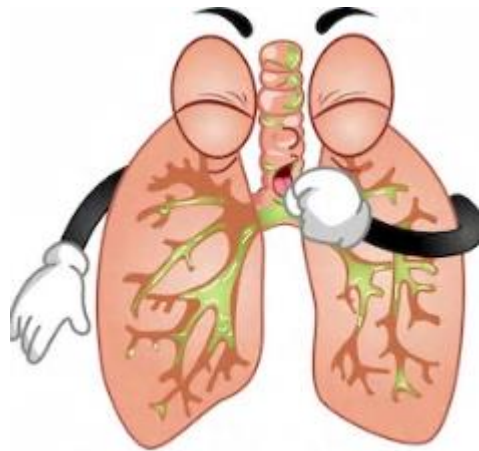
- Vöxtur
- Vitsmunafroski
- Hjarta og æðakerfi
- Lífsgæði

Hugsanleg uppvinnsla

- HNE skoðun
- Ofnæmi
- Hjarta
- Bakflæði
- Síriti

Svefnrannsóknir





- [Anne B Chang, Andrew Bush, Keith Grimwood, Bronchiectasis in children: diagnosis and treatment, The Lancet, Volume 392, Issue 10150, 2018, Pages 866-879, ISSN 0140-6736, https://doi.org/10.1016/S0140-6736\(18\)31554-X. \(https://www.sciencedirect.com/science/article/pii/S014067361831554X\)](https://doi.org/10.1016/S0140-6736(18)31554-X)
- https://www.uptodate.com/contents/bronchiectasis-in-children-pathophysiology-and-causes?search=primary%20ciliary%20dyskinesia%20children&source=search_result&selectedTitle=2~31&usage_type=default&display_rank=2
- De Boeck, K. Cystic fibrosis in the year 2020: A disease with a new face. *Acta Paediatr.* 2020; 109: 893– 899. <https://doi.org/10.1111/apa.15155>
- Shapiro AJ, et al. Genetic Disorders of Mucociliary Clearance Consortium. Diagnosis, monitoring, and treatment of primary ciliary dyskinesia: PCD foundation consensus recommendations based on state of the art review. *Pediatr Pulmonol.* 2016 Feb;51(2):115-32. doi: 10.1002/ppul.23304. Epub 2015 Sep 29. PMID: 26418604; PMCID: PMC4912005. <https://onlinelibrary.wiley.com/doi/epdf/10.1002/ppul.23304>
- Erik B. Hysinger, Laryngomalacia, Tracheomalacia and Bronchomalacia, *Current Problems in Pediatric and Adolescent Health Care*, Volume 48, Issue 4, 2018, Pages 113-118, ISSN 1538 5442, <https://doi.org/10.1016/j.cppeds.2018.03.002>. (https://www.sciencedirect.com/science/article/pii/S1538544218300294)