

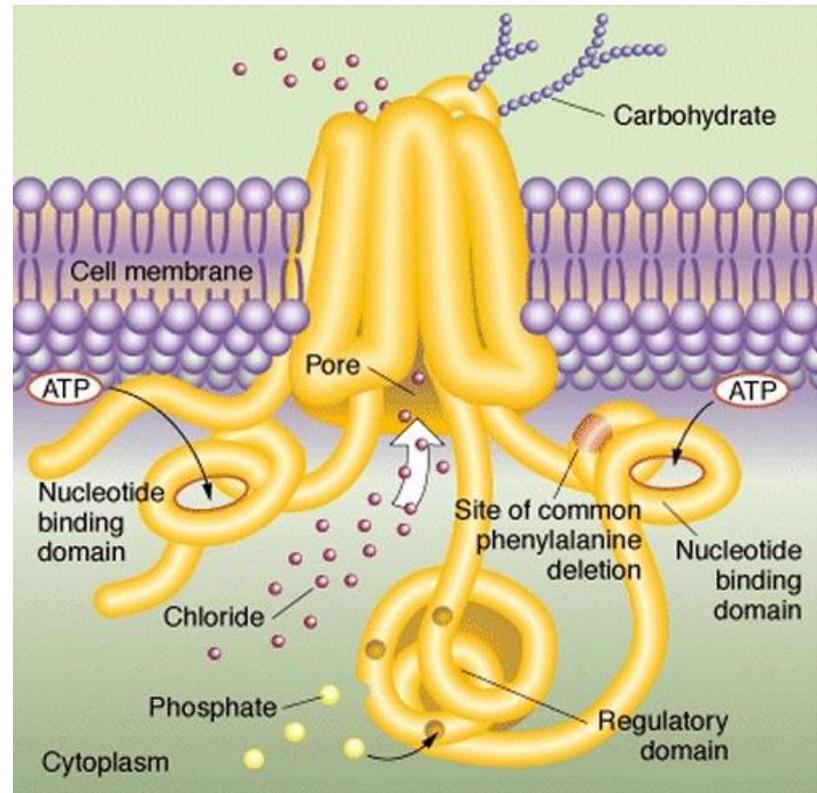
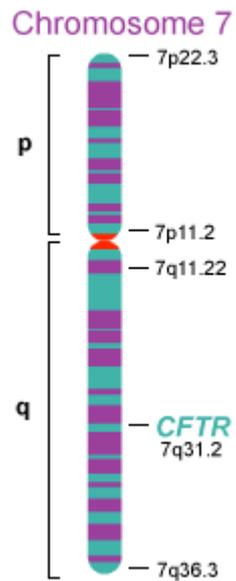
# CYSTIC FIBROSIS

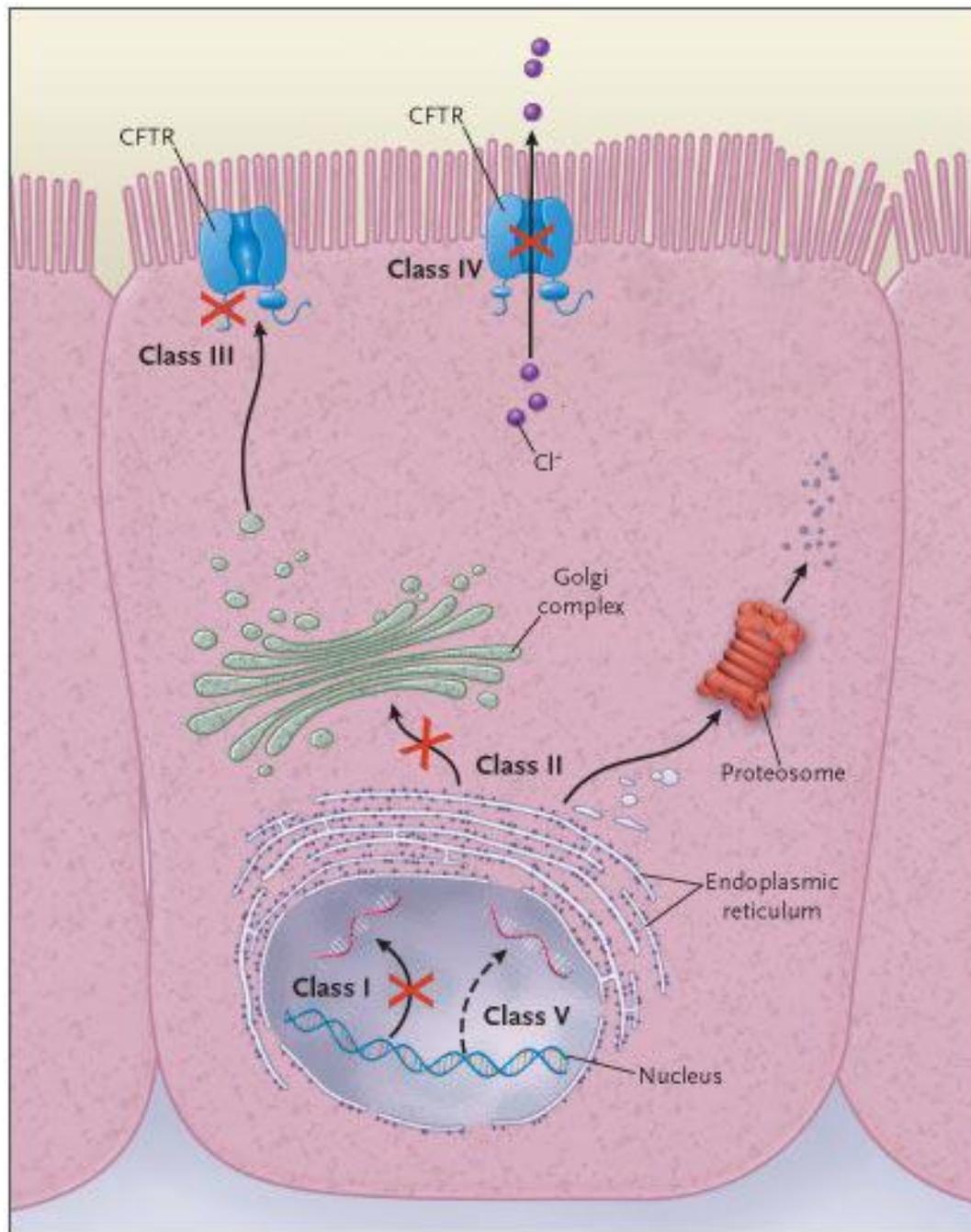
- 20 april 2017

- Terminologie:
  - Cystic Fibrosis
  - Mucoviscidosis
  - Taaislijm ziekte



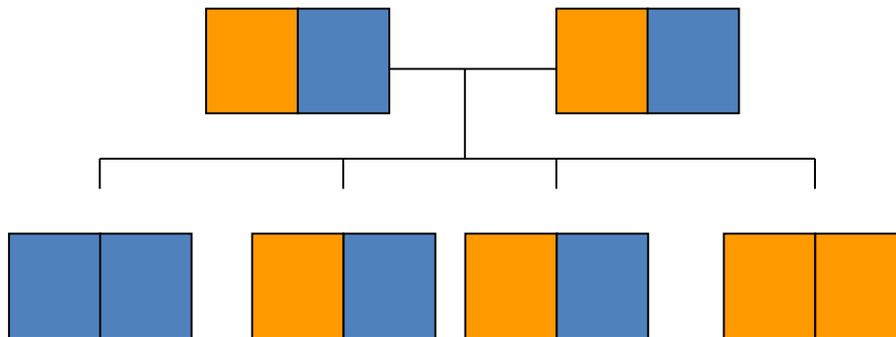
- mucus
- viscidum = mistletoe





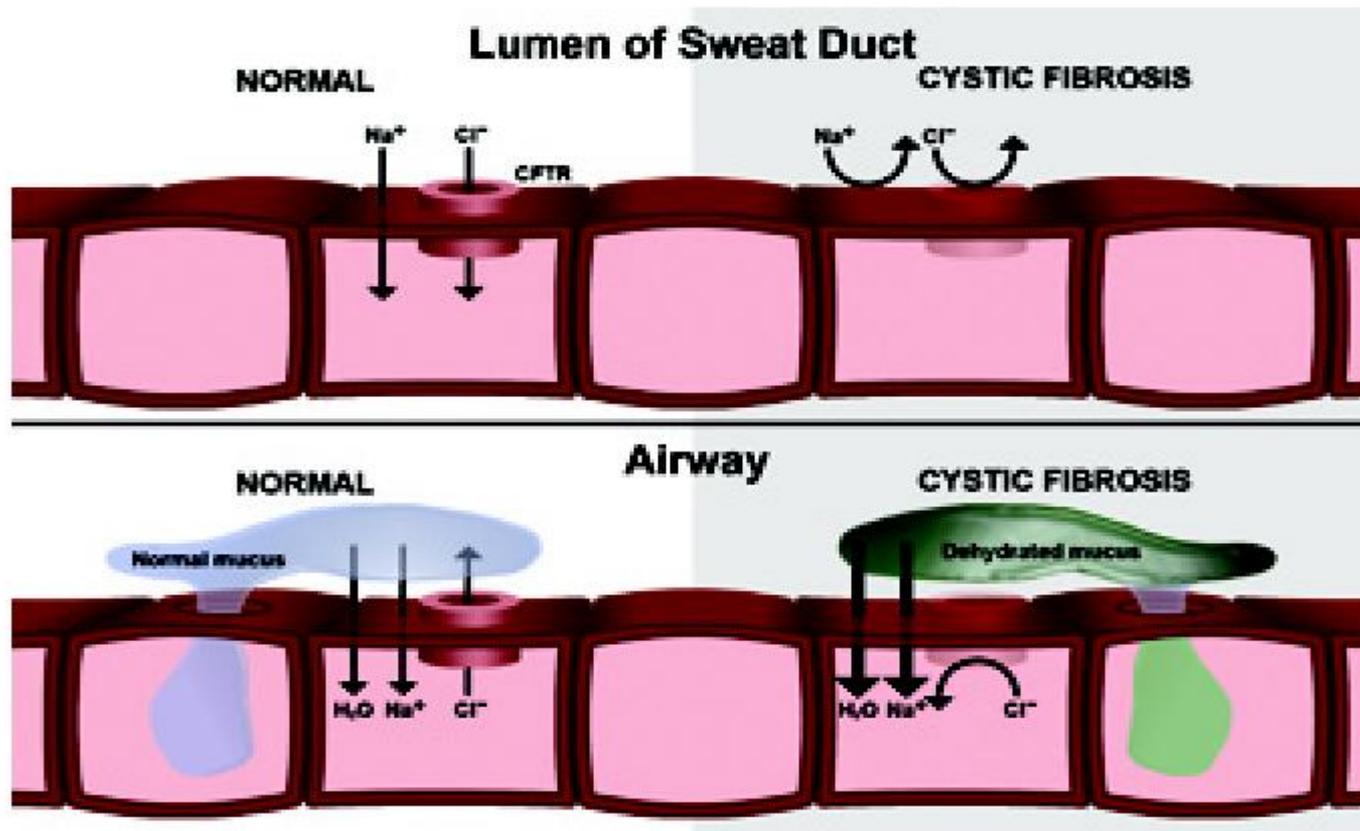
- dF508 73,6%
- A455E 3,5%
- G542X 2,0%
- R117H 2%
- 1717-1A>G 1,8%
- S1251N 1,2%
- N1303K 1,1%
- R553X 1,1%
- R1162X 1,1%

- White race: 1 : 2.000 - 3.000
- Netherlands: 1 : 4750
- Yellow race: 1 : 100.000
- Black race: 1 : 20.000

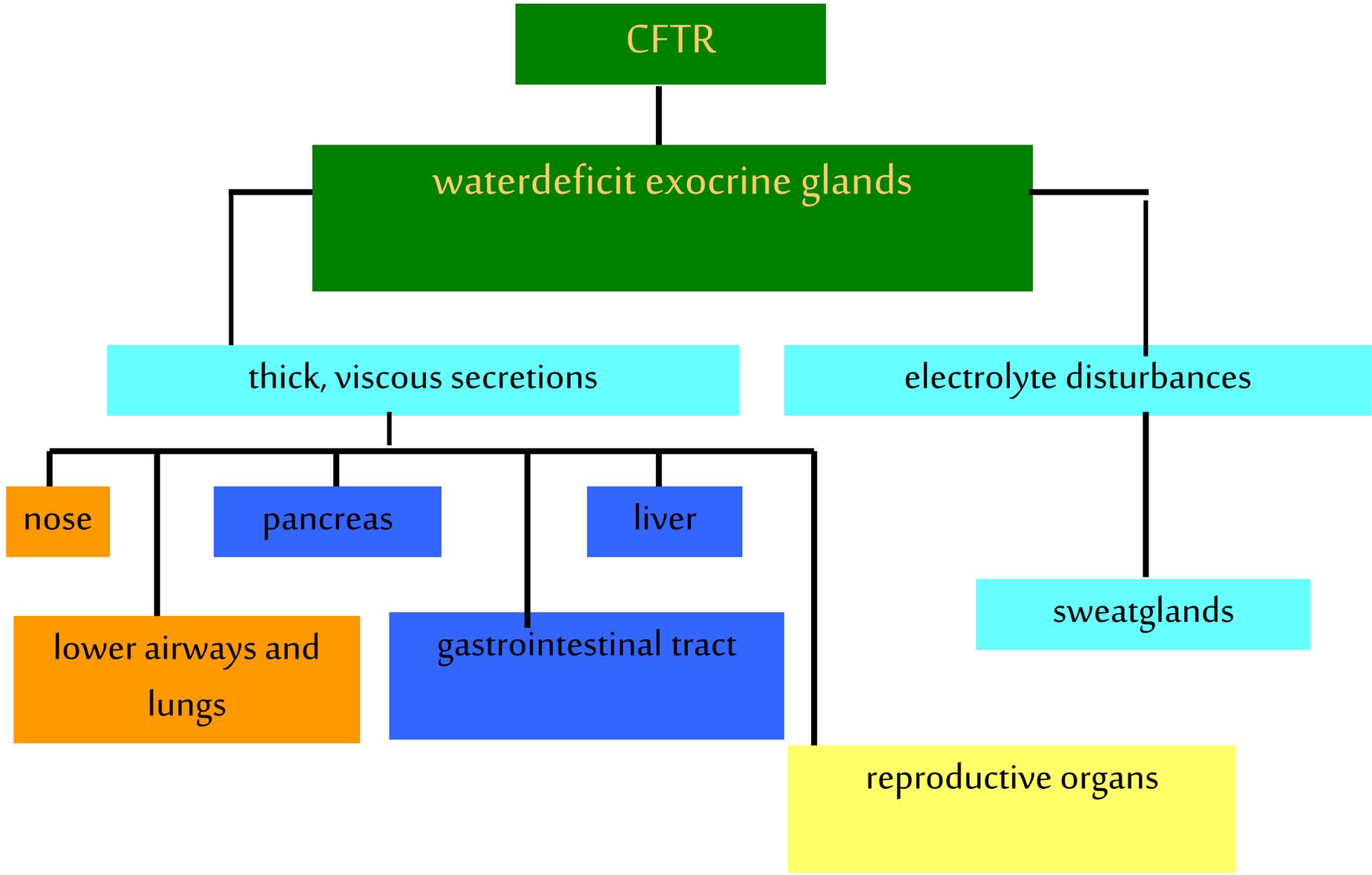


- Meer dan half miljoen mensen in Nederland zijn drager van het CF-gen
- Bijna elke week wordt in Nederland een baby met CF geboren
- Gemiddelde levensverwachting in Nederland is rond de 40 jaar (van iemand met CF)
- Er zijn op dit moment ongeveer 1530 mensen met CF in Nederland waarvan 650 kinderen

Figure A.  
CFTR channel in a normal individual and a patient with cystic fibrosis.



# CF Pathophysiology



**Lungs and sinuses**  
Infection, inflammation and obstruction

**Sweat gland**  
Elevated sweat chloride concentration

**Liver**  
Cirrhosis

**Pancreas**  
Exocrine dysfunction, diabetes and pancreatitis

**Intestine**  
Neonatal obstruction (meconium ileus) and distal intestinal obstruction

**Male reproductive tract**  
Obstructive male infertility

**Airway obstruction**  
*TGFB1*, *MBL2*, *EHF*, *APIP*, *Chr20q13.2*, *SLC9A3* and *SLC6A14*

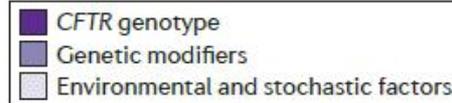
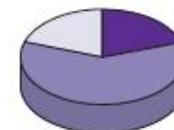
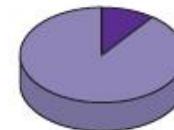
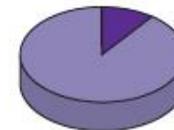
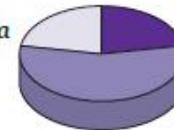
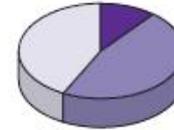
**Infection with *P. aeruginosa***  
*MBL2*, *DCTN4* and *SLC6A14*

**Exocrine insufficiency**

**Risk and age at onset of diabetes**  
*TCF7L2*, *CDKAL1*, *CDKN2A/B*, *IGF2BP2* and *SLC26A9*

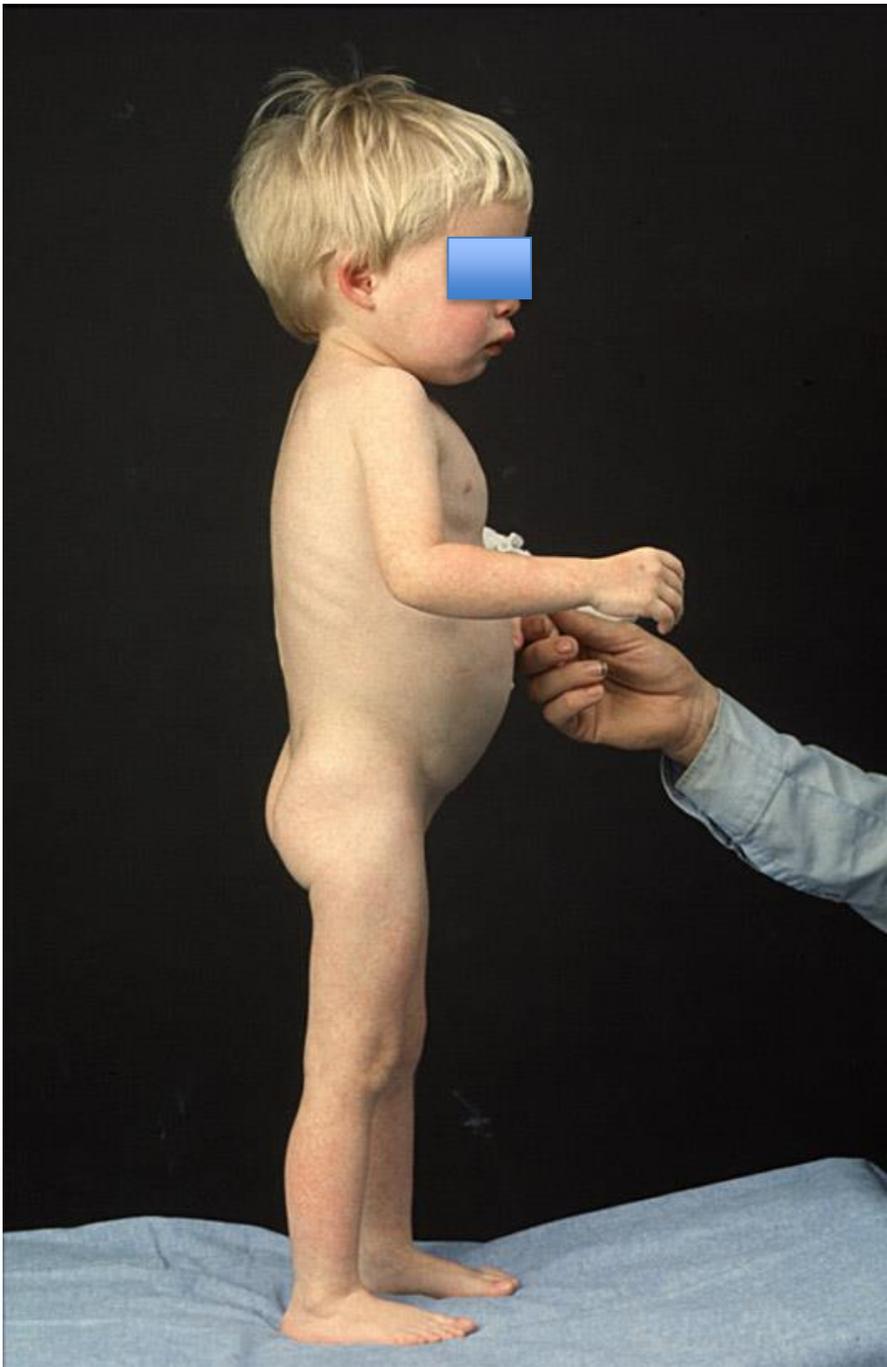
**Risk of intestinal obstruction**  
*MSRA*, *SLC6A14*, *SLC26A9* and *SLC9A3*

**BMI**  
*Chr1p36.1* and *Chr5q14*

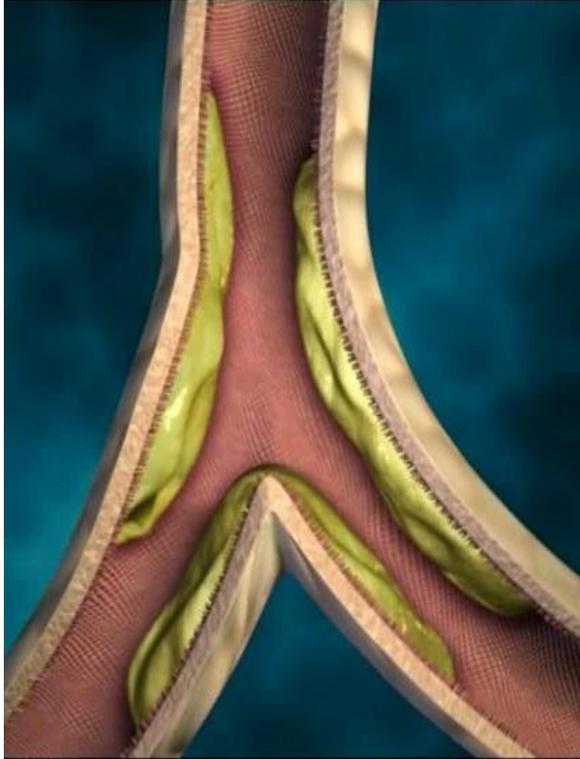


“Wow the that child which when kissed on the forehead tastes salty.  
This child is bewitched and soon must die”





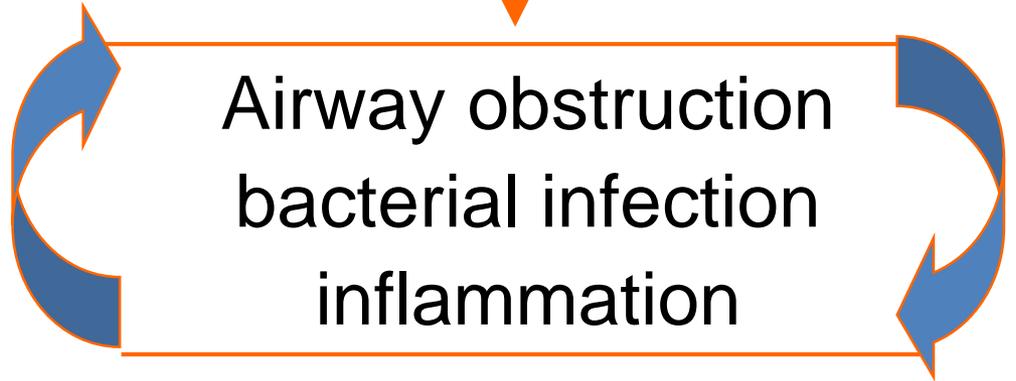
- thin
- thick belly
- deep thorax



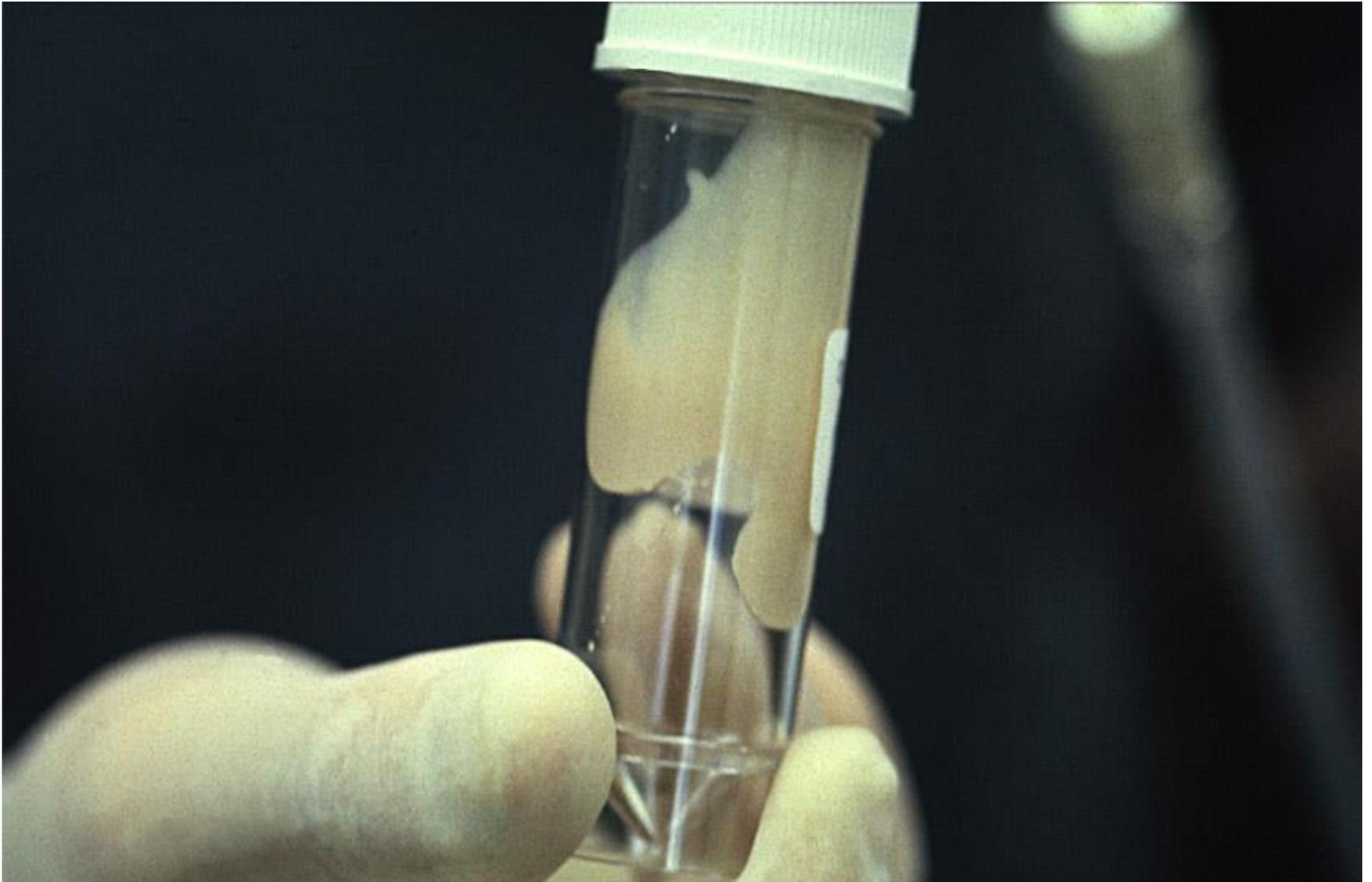
Thick, viscous  
mucus



Airway obstruction  
bacterial infection  
inflammation



# CF sputum



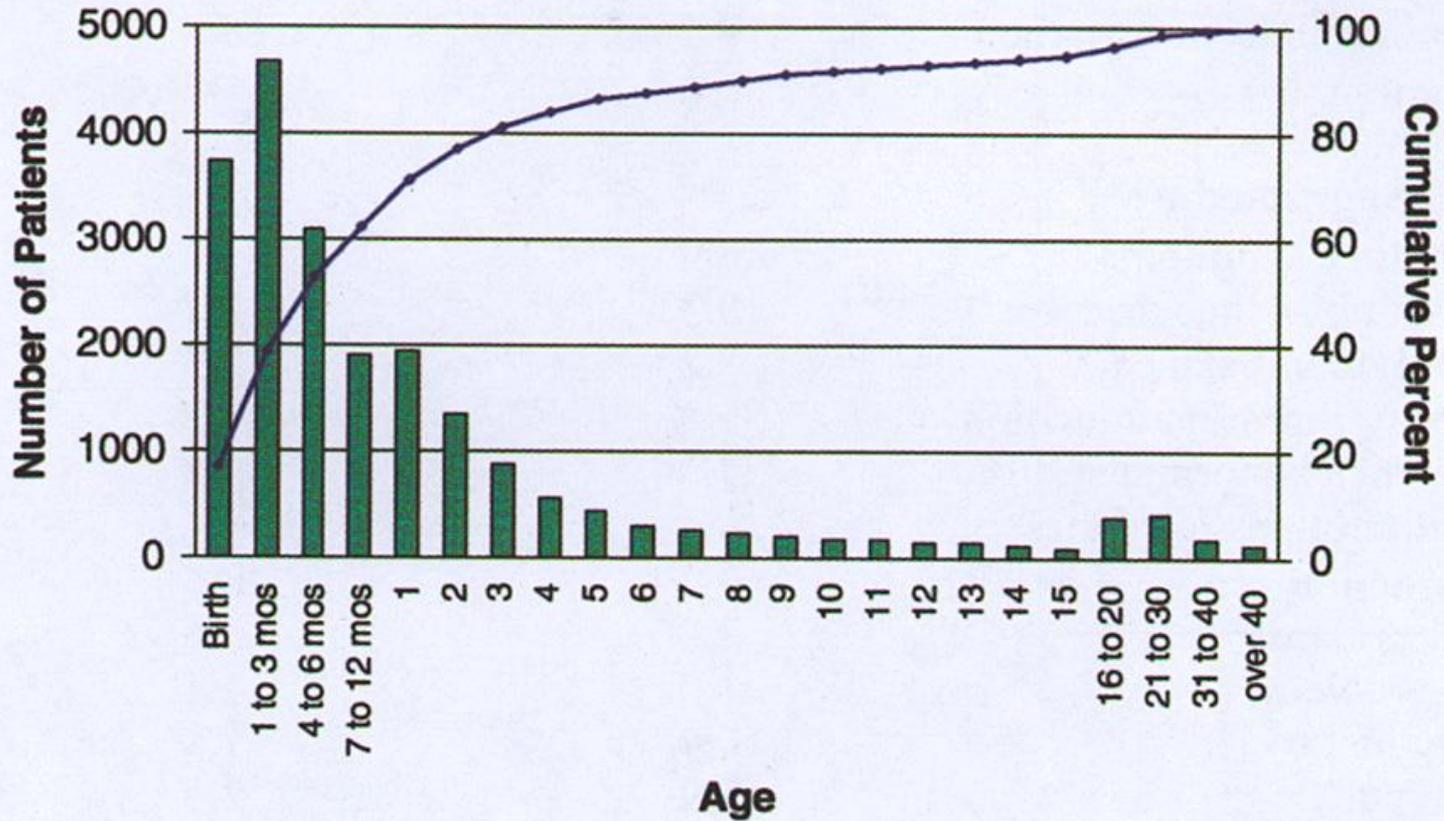
## CF clubbing of toes



## CF nasal polyposis



Diagnose : 50% < 6mnd, 90% < 8 jaar



Cystic Fibrosis Foundation patient registry 1999

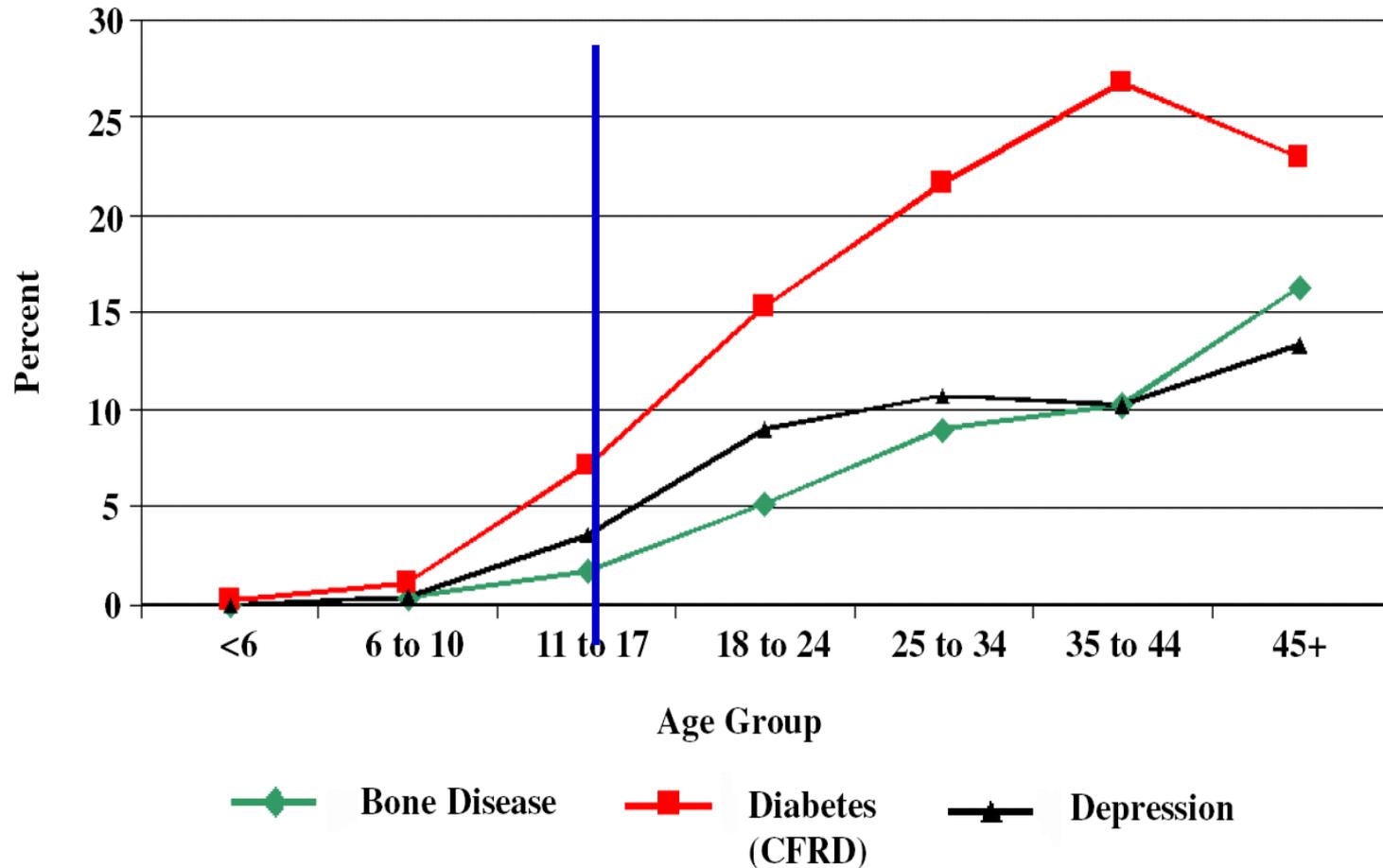
- Longen en luchtwegen:
  - Preventie infecties en behandeling infecties
  - Behandeling van ontstekingen
- Voedings status.
- Psychosociaal welbevinden



- Fysiotherapie
- Antibiotica
- Mucolytica (rhDNase)
- Anti-inflammatoire drugs (steroids)
- Optimale voeding, Voldoende energierijke voeding (voedings supplementen) : 110-150%
- pancreas enzymen, (Creon ed)
- Vetoplosbare vitamines (A, D, E, K)
- Preventie van chronische lage luchtweg infecties (vernevelen)



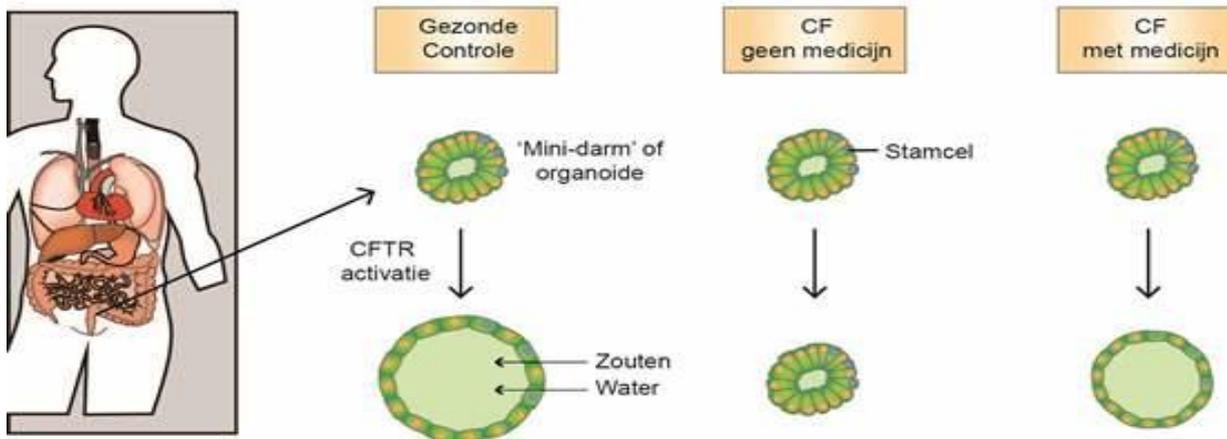
# Common Complications vs. Age



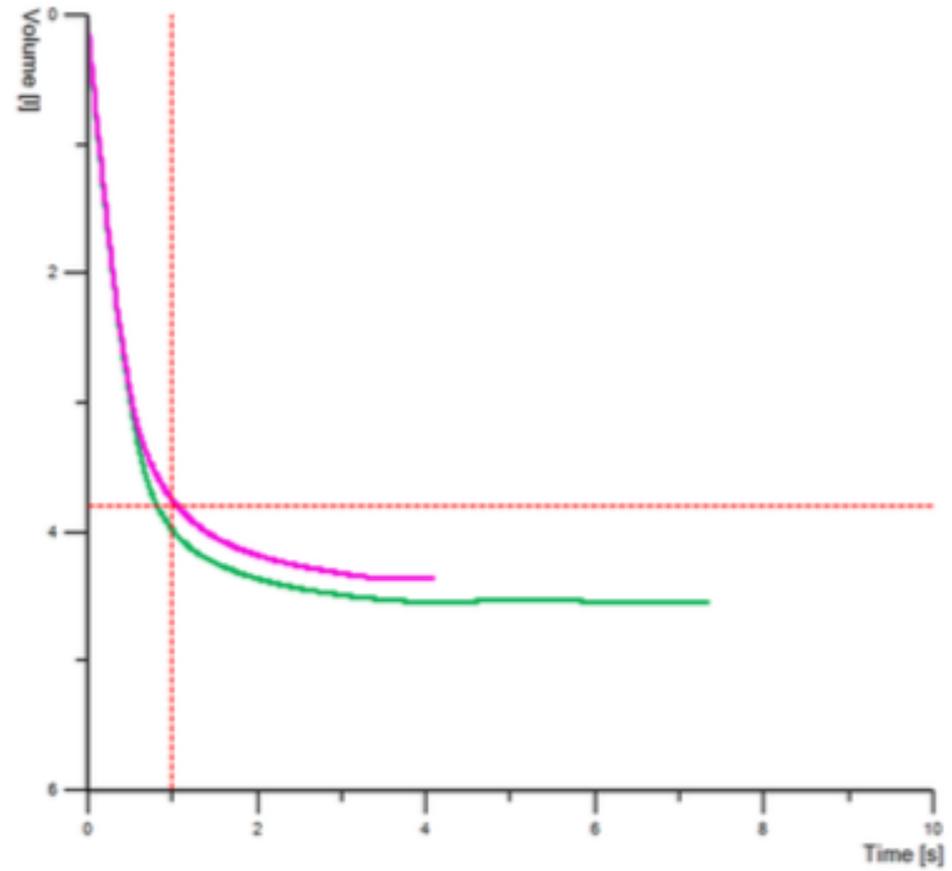
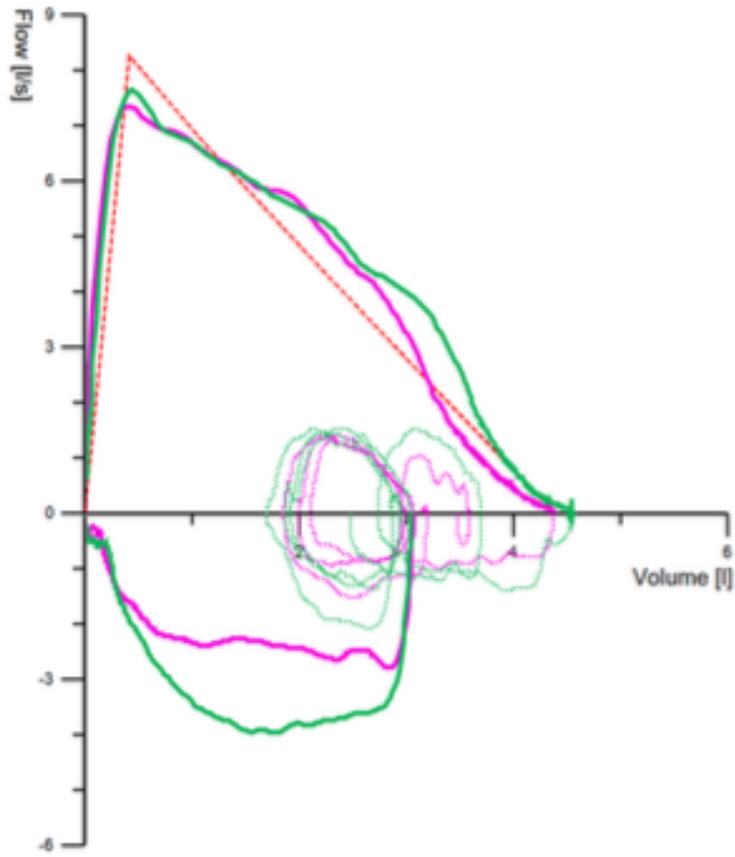
2003 CFF Registry Data

- Longen zijn de levensbedreigende factor.  
Bij transplantatie van longen en/of lever blijft CF in overige organen aanwezig. In 2013 : 20 volwassenen een longtransplantatie (22 volwassenen en 2 kinderen op de wachtlijst)

## HIT cf 2.0 en Regenboogproject



# F/V verslag WKZ



|          | Eenheid |      | voorspeld             | LLN  | Pre   | %Pred. | Ventolin<br>(800<br>µg) | %Pred. | %Verandering |
|----------|---------|------|-----------------------|------|-------|--------|-------------------------|--------|--------------|
| Tijd     |         |      |                       |      | 10:13 |        | 10:40                   |        |              |
| VC       | l       | (5)  | 4,07 <sup>(5)</sup>   |      | 4,12  | 101%   | 4,43                    | 109%   | +7,5%        |
| TV       | l       | (U1) | 0,87 <sup>(U1)</sup>  |      | 0,81  | 93%    | 0,62                    | 71%    | -23,6%       |
| ERV      | l       | (5)  | 1,30 <sup>(5)</sup>   |      | 1,29  | 99%    | 1,77                    | 136%   | +37,2%       |
| FVCEx    | l       | (35) | 4,40 <sup>(35)</sup>  | 3,57 | 4,37  | 99%    | 4,56                    | 104%   | +4,3%        |
| FEV1     | l       | (35) | 3,81 <sup>(35)</sup>  | 3,05 | 3,76  | 99%    | 3,99                    | 105%   | +6,0%        |
| FEV1/FVC | %       | (35) | 87 <sup>(35)</sup>    | 75   | 86    | 99%    | 87                      | 101%   | +1,7%        |
| FEV0.5   | l       | (U1) | 3,05 <sup>(U1)</sup>  | 2,84 | 2,93  | 96%    | 2,99                    | 98%    | +2,1%        |
| MEF25    | l/s     | (35) | 2,16 <sup>(35)</sup>  | 1,20 | 2,05  | 95%    | 2,89                    | 134%   | +40,9%       |
| MEF50    | l/s     | (U1) | 4,42 <sup>(U1)</sup>  |      | 5,14  | 116%   | 5,19                    | 117%   | +0,9%        |
| MEF75    | l/s     | (U1) | 6,69 <sup>(U1)</sup>  |      | 6,56  | 98%    | 6,47                    | 97%    | -1,4%        |
| MEF25-75 | l/s     | (35) | 4,29 <sup>(35)</sup>  | 2,84 | 4,56  | 106%   | 4,75                    | 111%   | +4,3%        |
| PEF      | l/s     | (U1) | 8,27 <sup>(U1)</sup>  | 8,02 | 7,36  | 89%    | 7,66                    | 93%    | +4,0%        |
| FIV1     | l       |      |                       |      | 2,75  |        | 3,02                    |        | +9,6%        |
| FVCIn    | l       | (35) | 4,40 <sup>(35)</sup>  | 3,57 | 3,07  | 70%    | 3,06                    | 69%    | -0,5%        |
| MVV      | l/min   | (5)  | 70,0 <sup>(5)</sup>   |      |       |        |                         |        |              |
| FeNO     | ppb     | (17) | 20,00 <sup>(17)</sup> | 5,00 |       |        |                         |        |              |

Casus : Jongen 16 jr  
caucasian; CF  
168 cm 60 kg BMI 21.3 kg/m<sup>2</sup>

Rustwaarden

- HF 81 bpm
- SaO<sub>2</sub> 99%
- Bloeddruk 113 mmHg
- FEV<sub>1</sub> 3.76 99% pred
- Fietsergometer verhoging van 20 w/min tot uitputting

Inspanning

- HF piek 189 bpm
- RER piek 1.26
- SaO<sub>2</sub> 99%
- VO<sub>2</sub> piek 2.62 l/min (86% pred)
- VO<sub>2</sub> piek/kg 43.6 ml/min/kg (89%)
- Wpiek 234 W (96% pred)
- Wpiek /kg 4.04 W/kg (101% pred)
- VE/VCO<sub>2</sub> piek Slope 35,3
- O<sub>2</sub> Pols 14 ml (92% pred)
- Bloeddruk 176 mmHG

Interpretatie : Kwaliteit test : goed

RER > 1.0 (1.26)

HF piek 189 (99% pred)

Pt heeft maximale inspanning geleverd.

Ventilatoire reserve

