



# Living with respiratory support

# Pompe Disease



Dr. J.C. Pompe 1901 - 1945

Spectrum disease: Severe ↔ 'Mild'

Muscles deteriorate. Respiratory support is inevitable. Muscle functions of legs, arms, neck and waist are lost, severe fatigue, muscle aches, problems with swallowing etc.

# The different stages of Pompe disease



# How did I know I had respiratory problems?

Many problems: shortness of breath, anxiety, concentration problems, night mares, emotional problems.

But I didn't know it was due to respiratory problems. This was not known to be a problem in Pompe Disease at that time.

The worst experience was that **I didn't know myself anymore!**

# What did I do?

- I went to my GP.
- I was referred to the local / regional hospital.
- I did do a sleeptest, but this failed.
- I was referred to the expert centre for respiratory care at the University Medical Centre in Utrecht.
- There I was diagnosed with respiratory problems.

# What followed?

- The diagnosis was made in January 1990.
- My problems worsened, but nevertheless I did pass my first year at university in June 1990.
- My partner and I went on holiday that Summer.
- During the holiday I had many unexplained (also to myself) emotional problems.
- When we returned home I called the hospital.

# In the hospital

- After examination it was confirmed my lungfunction deteriorated and I got a counselling with a nurse about starting ventilation.
- I was very relieved, but was send home because there was no place at the ICU.
- A couple of weeks later I pushed myself into the ICU .
- At November 1st 1990 I started home mechanical ventilation that changed my life.
- After 5 days ICU I was brought home.

# Choosing of type of ventilator

- In 1990 there were no many choices regarding ventilators. Without any discussion I received the PLV-100, a very reliable and good ventilator.
- Now the ventilators or bi-paps are still chosen by the physician as they know the specific advantages and disadvantages.
- In countries like the USA, India and the Philippines, patients must chose the ventilator themselves. They search the internet and inquire with fellow patients. Its not easy to make a well informed decission.

# Why is a certain ventilator chosen?

- What is the diagnosis?
- Does it require life support or non life support?
- When this is decided the type of ventilator is chosen: Bipap or V-pap.

# Invasive or Non-invasive ventilation

- Nowadays one starts with non invasive ventilation in almost all patients.
- When more ventilation is required the physician/nurse starts the discussion with the patient what to do next.
- I also had this discussion, but I refused a tracheostomy.
- In the Netherlands we now do have people who are 24/7 ventilated via mouth and/or nose.

# Impact of ventilation on my life

- Ventilation has of course a huge impact.
- It depends on the person, family and the environment around her/him how its experienced.
- I was relieved to have ventilation as I felt 'normal' again and could continue my study.
- Of course I am very dependent on others, I can't live without the care and support of other people.
- But when all safety measurements are met and equipment and care is well organised, one can live quite an independent life!

