

WELCOME!

Louise Bouman
The Netherlands
Diagnosed with HPAH 2008



(Pre)diagnosis

- After a lot of examinations one diagnose left: PH
- Next day: delivery (using contraction inducers)
- RHC in expertcenter
- Functional class IV
- Epoprostenol per IV





Genetic counseling?

- Pulmonologist informed me after diagnosis
- Surviving was my main goal the first months
- During follow up consultation I've asked about it
- Brief information by pulmonologist
- Blood collection



Genetic counseling?

- Switched to other PH expertise center
- Genetic counseling
- BMPR2 mutation
- Family Tree research
- Family letter
- Weight on my shoulders



Family?

- Mother, father, brother received genetic counseling
- No BMPR2 gene mutation
- What about or two kids?



Choices, choices...

- No gene testing until children are able to decide
- Youngest son got some complaints at age 13
- Screening at pediatric cardiologist
- Afterwards they both decided they wanted to know



Frightening period

- Genetic counseling through Zoom
- Blood collection at local lab
- Waiting.....and being nervous
- Results through Zoom call (COVID period)
- Both are NOT carriers of BMPR2 gene mutation



Take home messages

- Always ask for genetic counseling
- Think about the pro's and con's
- Talk to family or friends
- Talk to PH team
- Take your time, don't rush

