

Meet the Patient Interview with Lenke
Interviewed by Jazzmin, Charity Officer, Myrovlytis Trust

Jazzmin: Hello my name is Jazzmin and I am a charity officer at the Myrovlytis Trust and BHD foundation. We are delighted today to have Lenke all the way from Belgium to come and talk to us about her BHD experience with a focus on pregnancy and women's health.

Thank you so much Lenke for joining me today and it's a pleasure to have you here. I thought we should start at the very beginning, you were 14 when you had your first symptom of BHD, although at the time you didn't know what that was, and pneumothoraxes are rare in younger individuals but we know they do happen. Could you tell me about that experience?

Lenke: I was in puberty and none of my family members ever had anything similar. I was actually ill. I was coughing a lot and I had a pretty bad cold and it was actually after a couple of weeks that I noticed that my general physique hadn't really improved. When I rode the bike to school, I was tired and I had to take a break and yeah at a certain point in time my teacher at school said well isn't it time that you just go and see a doctor and just see what's wrong. So I went to a doctor and he said well that's funny if I listen to your lungs I can't hear you breathing on one side, but everyone was kind of relaxed so he just said well go to the hospital let them take a picture and we'll see. So I went to the hospital they took a picture of my lungs and just in 10 minutes there were people coming from everywhere jumping around and saying yeah you have a collapsed lung we have to operate immediately. So it was quite scary but eventually they let me go home for the night because it was already late and they said the doctor who performs that kind of procedures isn't here at the moment and as you have no real issues just go home relax and come back in the morning. We went back in the morning and they didn't perform surgery but they put in a chest drain and I stayed in the hospital for five days and they really didn't think about what had caused it. Actually the doctor said because you were sick and you were coughing a lot it's possible that you've coughed so hard that it made a little hole in your lung and it caused your lung to collapse.

A few months later, I think that's pretty common for people who have had collapsed lungs, my lung spontaneously collapsed again. Because, I knew the symptoms and I knew what to look for I woke up and I said okay this isn't good. We went to the hospital and they put in the chest drain again but then the next day they performed surgery. So I got pleurodesis but as it was normal that if you have had a collapsed lung it collapses again a couple of months later nobody really asked any questions as to why this had happened so by the time I was 14 15 I had a collapsed lung but no one really did a lot of research. It was just bad luck.

Jazzmin: You were really quite young, and it must have been quite a shock. Had you ever been in hospital before anything?

Lenke: I had a car accident the year before. But nobody really knew how bad it would hurt. The procedure (pleurodesis) is pretty painful and my mom and dad, as moms and dads do, said it won't be that bad you'll be asleep you'll have a lot of pain medication, but it was bad. So for the people who have experienced it it's really painful it's not much fun the recovery is also quite tough. So that wasn't really a good time or a good experience, but it was fixed, and we thought everything had passed. So I was happy that my lung got glued back together and that I was safe and it was actually I think 10 years later that I got a phone call from my mother who said well guess what I'm in a hospital my lung collapsed.

And well she actually called me and she said well does it hurt and what will they do because she knew that I had experienced it and that's when doctors started asking questions like oh your daughter had one that was also classified as spontaneous and now you have one and in the meantime even my grandmother had had one. But I was the youngest and I was the first.

There was a doctor in Brussels who started asking questions and he said there's this rare syndrome it's called BHD maybe you guys have that and he wrote it on a on a little card but at a time it was very costly to have the investigation done, it was like 5000 euro just to know whether you have this condition. My mom decided not to have the test and we would just assume that we have this condition without really knowing. And time passed so my mom had a collapsed lung and she had the same experience as I did so they did the chest drain first and then a few months later the lung collapsed again and then they did the surgery and my grandmother just had the chest drain once and afterwards she had no problems at all.

Jazzmin: You were 14 the first time how old was your mum and your grandma when they had theirs roughly?

Lenke: I think my mom was late 30s early 40s something like that. My grandmother was in her 50s but the funny thing was that I was the first so they never had any issues with lungs or with their kidneys. Because I was the first we already knew what was coming and I already told them how bad it was going to hurt so they apologized later on saying sorry that we said that it wouldn't hurt, it hurts like hell. My mom always kept a card from the doctor that said BHD. So we did some research but because it also said that you had to have kidney problems and other kinds of things we always thought well maybe it's just one big coincidence.

And yeah life moved on and we hadn't had any problems. I live a healthy life I work out. There were no issues until my first pregnancy. I got pregnant in 2017 in September, and I think it was October when the my other lung collapsed. I woke up in the morning and I told my husband this isn't right. So I was pretty sure that my lung had had collapsed but because I was pregnant it was much more difficult to have treatments so I went to the hospital and I had the chest drain but it didn't work so they had to perform surgery immediately. They couldn't do the pleurodesis because they said we don't know how the foetus will react. So they actually like stapled my lung to my chest so it was a similar procedure but they didn't use any type of products they used staples. It is as painful as the other surgery but I got out of the hospital I think three weeks later with a healthy baby in in my belly and I thought well that's it I had two lungs they both collapsed, I had both surgeries so I'm out of the woods. And when I was I think 32 weeks pregnant and the same lung collapsed again so we were on holiday we did like the baby mooning.

Jazzmin: Yeah you've got to do one of those!

Lenke: We were in the south of France and it was early morning and I woke up and I felt this this real ache at the top of my lung and I said this is the same feeling we have to go back. So I know my body and I trusted my body enough to do the trip back home because I said I'm not going staying in France in a hospital in a foreign language pregnant maybe having my baby here. So we packed our bags, jumped in the car, drove the whole way back and went to the hospital and actually that same night they took a picture of my lungs and the doctor said well I'm not sure whether it really collapsed let's wait a night and then during the night I felt a lot of pressure and by the morning it was clear that the staples actually had come loose and and the lung collapsed again. But, because of the pregnancy they said we'll try to keep your lung

open with the chest drain for as long as we can then deliver your baby and then have the surgery. So I was in the hospital for I think five weeks and they delivered the baby at 37 weeks and the day after I went into surgery, had the pleurodesis and recovered in the hospital together with the baby for two weeks.

Jazzmin: I'm glad you could still be together I mean it's not what you want straight after giving birth but I'm glad you could still be together during that time.

Lenke: Yeah and I had a natural births with the chest drain as well.

Jazzmin: Fantastic, so the doctors felt comfortable doing that?

Lenke: I got a green light for a natural birth with the chest drain and of course the doctors were on standby should there be anything but there was really no issue so I didn't feel different or uncomfortable because of the chest drain, my lungs were open, and I could just deliver the baby naturally. After the baby was born we were in the hospital, I recovered and I actually haven't had any problems since.

I went to the doctor because I wanted a second baby as well and I said what are the chances of that happening again and he said you have now two well normal lungs we performed surgery yes, you have the scar tissue, but actually your lungs are now more safe than the lungs of another person because they're glued together. He said I would have no issues getting pregnant and experiencing the pregnancy again. I just gave birth to my son three months ago and I had a problem-free pregnancy. I had no issues with my lungs, I exercised until I was like halfway I think, 17 weeks pregnant, and I had no issues.

Jazzmin: I understand you were offered IVF, what decision did you make?

Lenke: Well after I was diagnosed with BHD and had my annual checkup with my gynaecologist, I talked about my wish to have a 2nd child. She told me that it was an option to consider IVF. We would be able to only place non-BHD embryo's back so that I would have a 100% chance for a non-BHD baby.

As I never considered living with BHD as a burden I actually quite rapidly came to the conclusion that this was not a road that I wanted to walk on. My BHD does not limit me in any way and actually having those annual checkups gives me a lower chance of developing any kinds of diseases that would otherwise not be caught early on. I always considered having children as a natural process and I did not want to intervene in that natural process because the child has a 50% chance of that same genetic defect that I have. It has not held me back from achieving anything I want in life and if anything it has made me more aware of my health and what a precious gift it is to be healthy and to live in a society where medicine is so far advanced. My son tested negative, so he does not have BHD. At this point in time I don't know whether my daughter has BHD or not. But it won't change a thing. I know what signs to look for should she ever develop any symptoms of a pneumothorax. And by the time she's old enough to make the decision she can choose whether or not she wants the genetic testing. Should she test positive than me and my mother just add her to our annual BHD-date at the clinic.

Jazzmin: You mentioned that you kind of now know what a pneumothorax feels like. How do you know it's happening?

Lenke: It feels like I'm being stabbed in my lungs so it feels really like a stabbing pain and if I breathe it gets worse but I can tell pretty easily like which side it is and even how much it collapsed. So I can go into the hospital and say it's that side and I can perfectly tell it's that top part or the bottom part. It really feels like being stabbed and an ache and when I breathe it gets worse. When my lung collapse it always goes gradually so I never had the experience where in five seconds you're like I can't breathe, so it always went gradually so that's probably why the first time when I was younger I didn't notice it. I was sick, I was coughing my nose was blocked so it's normal to have like restrictions when you breathe and it's because it took so long and I didn't really recover that actually my teacher said well that's not normal maybe go to the doctor because it's really taking a long time for you to recover and by that time my lung had collapsed completely. So I was living just with one lung maybe for a week or longer.

Jazzmin: It is amazing what the body can actually do though isn't it?

Lenke: Yeah, exactly we're still functioning, it was more restrictive but one lung your body can still function which is amazing.

Jazzmin: So you've now had two children is that correct?

Lenke: Yeah.

Jazzmin: Have you received any guidance on genetic testing for yourself and your family as they grow up.

Lenke: No, so actually got tested after delivering my first child. The price got lower so I think for the genetic testing I paid something like 12 euros.

Jazzmin: The difference that was a lot better.

Lenke: After giving birth I went to this genetic department at the hospital they did a family tree with all of the symptoms we had and had experienced. They performed the testing and she said because you have these spots on your nose and your face as well and because of the family history you probably will test positive. I think I gave birth in June and by October November I got the result back that I was positive and my family got tested. So my mother got tested as well but we were quite sure that that she would be positive but I have an older sister as well and she hadn't had any problems so she got tested as well and she tested negative; she doesn't have the genetic deficiency like we do. But for my daughter I got the advice as there are no symptoms most of the time before the age of 18 just let her be and if she has to have her blood drawn for one reason or another maybe then do the test but not to have a separate test just to know. And for my son I already know he's negative because I asked the doctor to draw the blood from the umbilical cord after giving birth. I had had symptoms at 14 already so they made an exception because actually they don't do it before the age of 18 but they said okay because you had had your first symptoms at 14 we will draw blood from the umbilical cord for your son. I got the result four weeks ago they didn't find the genetic defect in his blood. He's BHD free but for my daughter I don't know.

Jazzmin: The advice we very much give is it's always the parent's choice up until 18 but we often advise waiting and then letting the child once they reach adulthood making that choice. But we always say for parents to be aware of the symptoms of a pneumothorax, it's very rare but for example you are a case where it does happen.

Lenke: So since we got our positive diagnosis I have an annual check-up for my kidneys and I do that now together with my mother. We have a BHD date.

Jazzmin: Oh I love that, that's such a lovely idea.

Lenke: Yeah so now we go together so we do our kidney check together and we have a visit with a lung specialist once a year and we do the breathing exercises in this cabin where you have to blow as hard as you can and do all those kind of tiring exercises and they take a picture of our lungs as well just to I think see whether there's something weird that they can find. But that's actually all there is to it so we have a check-up once a year at the lung specialist and the kidney specialist.

Jazzmin: Is there a BHD specialist centre or in Belgium?

Lenke: No. Well actually the lung specialist that we go to he was the first who knew about the syndrome and knew what it was. The others that I asked they didn't know it and they had to do some research and I even had one doctor who explicitly said during my first pregnancy that's impossible that you have that because with BHD comes kidney problems and other kinds of stuff, I'm positive that you don't have it. I said well my mom said I had to check for this and they took the card they did some research and he said no it's impossible that you have it and eventually well the original doctor was right.

Jazzmin: What advice would you give to other BHD mums or expecting mothers or people with BHD who are thinking of having children?

Lenke: I would just say to go for it because I even asked the group online whether there were any other moms who have had the same thing and actually most of them just go through pregnancy normally they don't have any additional symptoms. Even my mom she had me she had my sister without any problems so yeah I would say go for it and just be aware of symptoms, but keep active stay healthy like you would during any other pregnancy. Also do not let that restrict you because in hindsight I really think that that amidst the BHD group that I'm a rare case who had collapsed lungs during the pregnancy.

Jazzmin: You had two very beautiful and healthy babies. Is there anything else you wanted to add or tell the BHD community while we're here?

Lenke: Just be happy that you know what you have and that now you know, you can act upon it. I never had a down feeling because I got diagnosed with BHD just the opposite actually. Now I know that I have it I'm being checked every year for my kidneys, for my lungs so if there are any problems I'm lucky probably to know well in advance and to act upon it.

Jazzmin: Thank you so much Lenke, it's been lovely talking to you.