

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

Jazzmin: Hello, my name is Jazzmin, and I am a Charity Officer at the BHD Foundation which is managed by the Myrovlytis Trust. We fund research, and raise awareness about BHD, and I am delighted today to be talking to a BHD patient from Denmark, Charlotte, thank you so much for joining us today.

Charlotte: Thank you for having me.

Jazzmin: So, I've heard a little bit about your background, but I thought, let's start from the beginning and would you tell me about the events leading up to you being diagnosed with BHD.

Charlotte: Yes, I will be happy to do that. I was 43 years old, and I woke up and my back hurt a lot and I was super puzzled about it because I didn't recall doing anything wrong. And then, actually, three weeks went by with me having back pain, and I thought I would kind of get over it, and I also had a lot of difficulties in running. I run every day or every second day, but I was having difficulties doing that and very fast got out of breath. So, I googles a lot and it says that, oh yeah you can get out of breath if you have a knot in the back, so I was so confident that that was it. So eventually, I thought, maybe I should go to the doctor. So I went there, and she immediately spotted that I had a pneumothorax, so that one lung had had a spontaneous puncture, and I simply went with that for three weeks. So after that there was a hole in the back. So, then of course I got hospitalised, and I was, I think it was four days in the hospital. And I got so many funny comments from that hospital that usually it was young males, who were athletics and very tall, so it was super fun that I fitted with none of these descriptions, not being very tall and a woman, and not being that young. So, I thought it was fine as well, I went into the hospital, it got done, and I was fit to get on with my life, I have four kids and a full-time job, so I was just like, that's okay. That was funny. That was a funny story to tell, but nothing more than that. And then half a year later my sister had a pneumothorax as well. Just like me. And we are very alike, even though we are not twins we look very alike. So that was even more fun she went to the same hospital as me, she had the same comments, as I did, and we were just like, oh that's so super strange. We are even more alike than we thought. Neither of us thought about a syndrome or having a genetic disorder at all. She actually continued having the pneumothorax, I think it was five or six instances. And then she eventually went to a hospital where there was a lung specialist, and then for the first time she told the story that every time she came in with a pneumothorax and first it was my sister and now me. Eventually there was a doctor saying, I have heard of BHD, maybe you should get yourself tested, with your sister, because you might have it. And that was leading up to us discovering that we had BHD.

We found out it was from our father, but he has not experienced any issues, either with pneumothorax, cyst, kidneys or any skin issues. And that goes for his mum and dad as well. Now we don't know which of them have BHD.

Jazzmin: Once your sister told you that they thought, potentially it might be something called BHD, how did you then go about finding a doctor to get first of all genetically tested and then to be monitored.

Charlotte: So, in Denmark we are small country. And so, there's not that many genetic experts. There is one in the Copenhagen area, and we went for an interview with her. She's the one that's talking to all the BHD patients. So we went at the same time, to have a talk with her about it before taking the test and then also after taking the test and being notified that both of us had BHD. It's a public healthcare system in Denmark. So, you are automatically enrolled into the system. I get a CT scan every second year to spot any cysts in the kidneys.

Jazzmin: So that brings us on to the kidneys. You had a pneumothorax, and then I believe you also have some cysts in your kidneys, is that correct?

Charlotte: Yes. So after being diagnosed we were scanned and everything looked quite nice. So, I think both of us were confident that, okay, we might be suffering from the pneumothorax but hopefully that's it. We were also told not everyone gets cysts on their kidneys. They went two years and then I had a scan again, and there they actually found a cyst right in the centre of the left kidney and it was just where you have all the veins and vessels, connecting, and it was three centimetres in diameter.

Jazzmin: What did they do at that point?

Charlotte: So, first they wanted to make sure that it was benign tumour. So, they took a biopsy from the kidney to check what type of tumor or what type of cyst it was, and I was told (it was not 100% confirmed, but almost 100% confirmed) that there was a chromophobe RCC (Renal Cell Carcinoma). So I was not in any doubt that I wanted it removed, even though it was not super big, but it has grown from nothing to three centimetres within the two years. That was actually just when COVID-19 started. I was scheduled March the 12th. And on March the 11th, Denmark closed down with COVID-19 that everything was cancelled. But luckily, it was just the day after at eight o'clock that I was scheduled so they did not have time to, to cancel my operation.

Jazzmin: You knew there was a risk that there could be a cyst which could be cancerous that could grow in this condition, but only about 1/3rd of patients do develop some type of tumor in their kidney. When you got told about the cyst, how did you feel and did you have any resources available to help you?

Charlotte: I was surprised that there was a cyst at all. But when I was told that there was a cyst, I think I was prepared that it might be something that was not benign. Yeah, maybe a chromophobe as that's the usual cyst for our syndrome. I think I was prepared for that and that it might go through an operation and get the cyst removed. You have all thoughts of being afraid of what will happen, had it already spread or was that just being lucky finding the cyst very early and having it removed. And I was hoping for the last, that it was discovered so early and many times I thought about how lucky it was that we were actually diagnosed, that my sister had five to six pneumothoraxes. If she had only had one, I wouldn't have been diagnosed my sister, either. And I would probably have had that cyst. I did not feel it. I felt perfectly healthy. So, I wouldn't have discovered that, before it was too late. In that sense I thought a lot about how lucky it was that I was actually being diagnosed. It's why the screening is so important and why we so need to recognise this condition.

Jazzmin: What surgery did you go for in the end?

Charlotte: So, I was told that it was quite a difficult place that the cyst was located in because it was in that centre of the kidney and normally it sits on the surface, and then you will do a partial nephrectomy, but they were not sure that they were able to do that. So they kind of warned me that it was going to be a quite difficult, operation, and then you go to sleep, you don't know what happens and when you wake up, you're not all awake. So, you're kind of in a sleeping mode but you can still sense and hear everything that people are talking about around you. And I could sort of sense that they did not only do a partial nephrectomy, they actually removed the entire kidney. It was so difficult for them to, to just remove the cyst, it was so much grown into the tissue of the kidneys that wouldn't have been a problem, if it was on the surface but it wasn't. It was in the middle of the kidney so they couldn't and on discovering that they took my whole kidney.

I felt quite vulnerable, I would say, because within a couple of weeks I went from feeling perfectly healthy, to having a cyst in my kidney, being afraid of what that could mean to only having one kidney. That was not what I thought I would be going through, if I looked back just four weeks.

Jazzmin: How was the recovery process and what advice would you give to someone else who has to go through the same or similar procedure to you?

Charlotte: Before that operation I did a lot in terms of just keeping exercising. For me that's been actually really important since I had my pneumothorax because it was there that I really could feel that there was something wrong with my body. So for me, taking a run every second day is important because then I know whether I have a pneumothorax or not, because if I have one, I cannot breathe, properly. And even though I knew that I had a cyst in my kidney I kept on exercising because I wanted to be fit for the recovery. And I think that went surprisingly fast. I notified my job that I would have to stay at home for six weeks, but I actually didn't, I recovered very fast. I was out of the hospital within two days. And then I just started after a couple of weeks walking and being able to run and walk longer and longer distances, so I was, I thought at that point I was fit to work. I don't have a tough work, just sitting at my computer, and I thought I was fit for doing that after a couple of weeks. So, I was surprised, actually how fast it went.

Jazzmin: And to only be in hospital for two days after the procedure as well.

Charlotte: Yeah, I think I impressed the doctors and nurses because I was doing so well. And of course, you have a scar and that hurts a bit. But, being at home, just with simple painkillers worked. And maybe it was because of COVID-19, why I was sent home but it was really nice for me.

Jazzmin: I'm glad I think recovering at home if you're safe to do so is very nice because then you can be surrounded the people you care about.

Charlotte: COVID-19 was also a good period where you could be together with the entire family. In March 2020, everyone was at home either working or home schooling. Being surrounded by the family is very important, I think, after such a procedure, and also feeling vulnerable. You just want to be close to your family as much as possible. So if you can arrange that, I would really recommend that. Also making sure that you train and exercise, before the procedure, but also being close to some family or friends after the procedure.

Jazzmin: As you said it must have initially been a bit of a shock what happened with that procedure but being able to talk and discuss it is so important and really good.

Charlotte: That's also a recommendation that helped me a lot. Having known that I have a cyst being afraid of everything, will I survive this, am I now a cancer patient, having that talk to a lot of your friends and also your family helps a lot. And you kind of feel that you have a whole team that hopes for your recovery. And that helps when you are on the other side, thinking okay, but now I'm good.

Jazzmin: What impact has BHD had on our everyday life?

Charlotte: I would say, when I got diagnosed with BHD I was told not to dive too much, go jumping out of a plane with a parachute, play the trumpets. But none of these things I wanted to do. So, in that sense I can live with that. I didn't feel that I lost anything not doing these things. I have been travelling a lot in my life, but I would say that this kidney cyst and me being aware that I have only one kidney, and I also know that I have some big cysts in my lung has actually put a limit to my eagerness for traveling. I would say, I'm quite nervous of going to a country where I will have a pneumothorax, and not being able to drive home. I have a job in an international department and it was actually the intention that I should go travel a lot, but right now I am in a state where I can travel in Europe, but I would not like to travel outside of Europe. I think that I can feel that limits me and my kids are also saying are we never going to travel anywhere else. And maybe not, maybe not with me. Maybe they can.

Jazzmin: There are so many beautiful places in Europe. There are still so many places I want to explore so that that's sensible to do what you feel you're comfortable with.

Charlotte: Yeah, I don't feel comfortable going outside of Europe.

Jazzmin: In terms of having one kidney did the doctors tell you to monitor or to take any medications?

Charlotte: No actually not at all. So I think I made a fast recovery, so I have some painkillers but nothing else, and very luckily the tumour and the cyst did not grow into any other organs. So we were lucky to find it in such an early state. In terms of follow up, every year I get a scan, from my throat and down to my legs to make sure that everything is in order.

Jazzmin: Do you know if it's a CT scan or an MRI scan?

Charlotte: A CT scan with contrast fluid.

Jazzmin: And for people who might not have started having scans yet, can you briefly describe what it's like to go through a CT scanner?

Charlotte: Yeah, you get a contrast fluid, and it kind of feels like you pee in your pants.

Jazzmin: I have heard about this!

Charlotte: So yeah, so you feel like you have metal in your mouth and then you feel sort of a warm fluid going through your body, and therefore, it feels like you're peeing your pants.

Jazzmin: But you're not.

Charlotte: No, you are not at all. But it really feels like that. They're really good at telling you that before the procedure and before the scan. So, you are aware of it. But the first time I thought, Okay, this is how it feels like it's really true. I have to check afterwards.

Jazzmin: So, considering what you know now about BHD now, what would you tell your younger self who'd just been diagnosed.

Charlotte: I think I was super optimistic given the diagnosis. And what I know now, was I was so sure that I would not have a kidney cyst. I think if I would go back, I would just shut up, because having a couple of years, with just being happy go lucky, I wouldn't spoil that by my older self-telling my younger self that you will have a kidney cyst, that would have worried me a lot. I think starting off being positive and thinking this will not happen to me, was okay.

Jazzmin: Having that optimism but at the same time still going for the screening so you've got that safety net there.

Charlotte: And I think I was leaning a lot on my family because apparently, they have BHD as well and none of them knew about it.

Jazzmin: You mentioned you have four children, is that right? Have you talked about genetic testing with them in the future at all?

Charlotte: Yeah, so they're all four aware of this, and we as a family have been told that we should wait until the children are 18 and let them decide themselves to do the genetic testing, and I think that's the good way to do it because then they can decide on their own. I think it's part of their knowledge now that they might have the syndrome, so I think all of them are quite aware that they do not want to go deep diving, before getting tested. So, all of them are quite aware of symptoms and has also followed both me and my sister's journey. My oldest daughter, she's above 18. She's been tested positive for BHD. You think it won't happen to you and I think she had the same thoughts that she wouldn't have BHD. But she has. I think that was a shock, but we had a good conversation with the genetic expert because my daughter was very interested in hearing about what about her kids and whether she should do some forms of sorting eggs.

Jazzmin: IVF kind of thing.

Charlotte: Yes, exactly. I think they were really good at reassuring us and saying most people have some kind of a syndrome. So we know ours now. But most people have some kind of a syndrome and we can actually live with BHD. We can live very normal lives I will say perfectly normal lives. Until I was 43, I thought there was nothing wrong with me, I was never ill. So you can have a really good life, and I know people from my family having BHD who have also lived very normal.

Jazzmin: That fantastic to hear, especially helpful for those who are newly diagnosed. Thank you for sharing that.

Charlotte: And I'm really happy that I discovered very late in my life because in that sense you look back and think, well, if I had a healthy, good life up until now, of course I can continue to. So maybe that could be different if you're very young and being diagnosed.

Jazzmin: So, one thing we've touched upon throughout this interview is that not many clinicians and doctors know about BHD. In an ideal world where the sky's the limit, what

would you do to raise awareness about BHD, or what do you want to see the BHD foundation doing to raise awareness about BHD?

Charlotte: I think genetic disorders are undiagnosed. Other parts of our family also had the BHD. I think it could have been discovered, very early on for me and my sister, and it just didn't. It was pure coincidence that it was discovered. And I guess there's a lot of others like us that have pneumothorax, and everyone just has a funny story because you're not young and tall and male. And just having those doctors aware that having pneumothorax, could also be a symptom of BHD, could really raise the awareness. I haven't met any doctors from the lung department that was aware of it.

Jazzmin: And maybe it's also those at the front line because pneumothorax is something you're going to be brought in with. So, those are the people we need to target. The respiratory doctors and maybe even the A&E doctors who see someone come in with a history of pneumothorax.

Do you have anything else you wanted to add to at all?

Charlotte: I think it should be underlined that this is not something that will handicap you in most cases. I know that there might be severe cases, then of course it is a handicap, but in many cases and with the people I know now, it has not been a handicap, and I think that's really important to say that you can live quite a normal life just like anyone else with BHD.

Jazzmin: Thank you so much Charlotte.