

Cassandra's BHD Story
Interviewed by Jazzmin Huber, Charity Officer at the Myrovlytis Trust

Jazzmin: Thank you so much Cassandra for joining us today. So, my name is Jazzmin and I am a Charity Officer at the BHD foundation and Myrovlytis Trust. We are a charity dedicated to raising awareness and funding research into BHD. And we're so happy to get the opportunity to talk to a member of the BHD community. So thank you so much Cassandra for joining me today.

Cassandra: Thanks for having me.

Jazzmin: Well, I'm gonna start at the beginning. When and how did you first get diagnosed with BHD?

Cassandra: And so I was really quite fortunate in that I wasn't the first one to go. My grandmother's line is where the BHD comes from on my father's side, and so one of my grandmother's uncle's had been diagnosed with it he had gone through the big genetic tests the one that's the most expensive and they put the pieces together and then he sent a letter to everybody that he was related to, and so it filtered through all the different generations down to my generation, and I, you know, as a family we were talking back and forth about what the pros and cons of getting tested to find out if we have that same genetic mutation. And finally, it just came down for me that I wanted the information rather than being caught off guard. We got the letter from my relative in 2007, and then I was diagnosed in 2013.

And that was like a whole thing in itself, it's got to be better now because it's been several years but to get tested was super tough, because they didn't have like a kit, they would send you. I had to work with my genetic counsellor at the hospital that's near us. And she like hodgepodge this thing together using the blood department and their hospital. And then, like getting it there within a certain amount of time it was super stressful. It all worked out we got done, and I got the letter back and they said it was positive. And so I was able to just add my information to the rest of our family information.

Jazzmin: And how did you then find doctors? Getting a genetic test was hard so how did you find a doctor who knew about the BHD.

Cassandra: Well, I didn't actually I kind of just happened upon someone, and I found this very interesting because reading through all the information about BHD, it's really emphasised about how rare this diseases is. I went to the lung doctor and he just was like oh yeah BHD. I was like, did you study this disease specifically and he's like, I know it's just a lung condition so like all the lung people know about it and I'm like really, that's not what I read.

But I guess maybe it is now, I don't know but in his particular case wherever he did his training it was something that they talked about and something he was familiar with, so it didn't like scare him. I had like all these pamphlets from bhdsyndrome.org. I didn't have to go through the whole rigmarole of trying to convince them I needed all this other screening and everything, and my general practitioner doctor was also very helpful. She is not familiar with BHD but she has she read up on it, she agreed that these are all the screenings we need to send you to and said I'll hook you up with all the different specialists and so I got to a lung person, I got to a kidney person and she's been kind of like my, my home base, trying to organize things for all the stuff we've gone through so.

Jazzmin: Well, that's fantastic to hear because we still get a lot of people saying that doctors have never heard about the BHD so the fact you had a doctor who was just like, yeah, obviously. So you had that family history, so you knew it might be a possibility but knowing that you did have BHD, what impact did that have on you.

Cassandra: Um, I was kind of surprised when I was waiting for the results. Like, I would try and tell myself Okay, you've gotten a positive result. How do you feel? Okay you've got a negative result, how do you feel? and honestly I felt worse with the negative results because of the guilt, like just knowing that other people are gonna have to go through this and I wasn't. I was going to be spared.

I like having information so I don't like being surprised by things. So having the information knowing I was positive, I felt much more empowered because now I understood consistently what's going to happen if I have a lung collapse this is what happens if I have trouble with my kidneys, this is what needs to happen. And so it was really a relief to find out.

Jazzmin: Have you had any BHD symptoms?

Cassandra: I have not had anything on my kidneys. So my kidney scan right after was clear and I haven't had any issues with kidneys stuff since then.

My, main issues have been with lungs, and my first scan showed I had several blebs on my right lung, and the largest one was five centimetres, but they told me it doesn't really matter about the size, they will pop, whenever they want to pop. They don't have to be a particular size.

And it wasn't very long after I was diagnosed well it was a couple of years I guess in 2015 I had my first lung collapse, and it was the best lung collapse, you could possibly have.

It was really funny, my husband and I had decided to get back on the wagon and started working out again and so we were in our home gym doing our stuff, and I got done doing some repetitions, and I was just laying on the ground trying to recover and I just couldn't catch my breath. And my heart rate was just pounding and pounding. I was like why am I having such a hard time recovering. This isn't normal. I was like, Maybe I should take it a little easy today and we both knew that, you know, I had this diagnosis and so when I still hadn't recovered by the end of our exercise routine, he's like, Yeah, we need to just keep an eye on that. So we went all day. I couldn't get my breath and my heart rate was so high and like by late afternoon I was like I think I should maybe, you know, go and see my doctors.

So, I got a same day appointment, and she's like yeah you have BHD. We did a chest X ray, it was a different doctor so I caught him off guard. He said I had a small pneumothorax, it was, I want to say it was less than two centimetres gap on the chest X ray. And he's like, it's pretty small, but I'm not real comfortable deciding if this needs a chest tube or not. So we're gonna send you off, instead of staying at a regional hospital. We're gonna send you off to the hospitals in the bigger city near us. And, and, when we got there, they didn't believe me at first, because I wasn't having any pain. I wasn't having trouble breathing like I felt like I hadn't gotten enough breath, each time I breathe but I didn't have trouble taking a breath, it wasn't painful to take a breath, my oxygen level was still at 100%. I've never had a lung collapse, where my oxygen was less than 100%, but my heart rate was high, and they're like, are you sure you're having a lung collapse, because you're not really showing any symptoms, and I'm like, I know I've got this condition, and it's really rare.

And so, yeah, I think I really do have a lung collapse and they sent me through the chest X rays again they're like okay it was a small one like it shouldn't even be bothering you really but, okay, and it doesn't bother us, if you want to just be on oxygen and observation overnight, and see if it'll heal on its own because the oxygen helps relax the lungs and so it doesn't have to work so hard to be able to get, you know, 100% percent oxygen level, and, and then it could just heal on its own so spent overnight on oxygen and the next day it had not grown. It wasn't much smaller, but they're like, it seems to have healed over. So, we're willing to send you home and then you just wait it out, so it was a really great first lung collapse, which was awesome because we didn't realise at the time that I was six weeks pregnant.

Jazzmin: Oh my goodness, so I'm really glad that you went there and you got all checked and it was okay. Have you had any lung collapses since then?

Cassandra: Yeah. Since then, I can't remember if it was three or four. I didn't have any for the rest of my pregnancy, which was amazing. But then after that I started having several in a year, and the doc, my general practitioner was just like you know maybe it's time to talk about the surgery because who knows how many times more this is gonna happen. And they were always really small and they always resolved, just with oxygen overnight observation. Finally just had them enough times in a year, they're like, you know, we should talk about the surgery, so they sent me up to a lung surgeon.

Jazzmin: It just shows how unique everybody's cases are, everyone has a slightly different story and a slightly different experience. So with the recurrent pneumothoraces how did it impact your everyday life, did it stop you doing anything that you normally would be doing?

Cassandra: It did make me double think travel. We didn't avoid any travel because of it, but we were hyper conscious of what was going on and like we would talk before we left for any, any sort of travel, you know, if you should experience these things what are we going to do when we get to the other side, you know, assuming we were in a plane or something. So it made us just a little bit more nervous and after the surgery we felt less nervous about traveling.

So, otherwise no I haven't had, I haven't because I have that empowerment, I feel like I can speak up for myself and I've had experience with it I don't feel nervous about doing other things. I'm not a scuba diver, though, so like if I had some sort of hobby that would require me to be in a dangerous situation probably more often I would probably have more to say about it.

Jazzmin: And how did the surgery go, what was your experience as to it?

Cassandra: Okay anybody in the Twin Cities, Minneapolis, St Paul area of the United States. If Dr. Jacques is still available and you're thinking about it, go with him. He's amazing. Again he was not surprised by BHD either. And he was very confident that this surgery would help fix things, I mean he told us what the risks were about it. I had a mechanical thorodisis on the right side, and it was all laparoscopic, and he said there is a chance that it won't stick. And then you have to go do it again but he felt very confident that it would. And just, he seems to have a very good system like all of his nurses, all of the people that deal with him know exactly what he wants and he is really good at it. And my incisions are super small. My recovery was great. The most painful part I know I hear a lot about the chest tube being the most painful part and the chest tube was not my most painful part for some reason. Being on pain medications right away and get the stool softener right away.

He also used, massage therapy in hospital, to help me recover because the worst part of that surgery was the aches and pains in my back from the muscles re-stitching, and I tell you that massage therapists helped you so much. Just relax and helped everything heal very loosely so it wasn't too tight, but I was out of bed, walking around, I think I had that surgery in the morning, and I was walking around by that evening, and they got me up a couple more times in the night to walk around because that was his thing like he really wanted to make sure that you heal, you know with functionality.

The scar tissue on the lung is important right, but also like getting your lungs to expand and take the deep breaths and all that stuff. He was very conscious about getting that part into the recovery routine. And I was out of the hospital within 24 hours. Actually I think back, I think it was 27 hours. But still that's ridiculous. That is an amazing result, and I don't know if it's just because God is super gracious, or if it was just that good of a surgeon.

Jazzmin: Yeah, it's amazing and that's such a lovely story to hear. Do you still see a pulmonologist?

Cassandra: I do, I keep up with one, I'm trying to remember how often I see him, I think it's every couple of years I go check in with him, not had any lung troubles since then so it's really boring check ins.

Jazzmin: You mentioned you have children, is that correct?

Cassandra: we do

Jazzmin: So how do you feel about talking to her about BHD.

Cassandra: Yeah, it was really hard on her when I was recovering, because she always wants to be held and cuddled that sort of stuff and I couldn't pick her up. She could come sit on my lap but she has to be careful with mommy. So she was super aware of the surgery, and like why I had to have it. And I think that's, that's like my philosophy with everything with her, like, I want to tell her about the genetic things that are going to be possibilities for her. I don't want her to be afraid of them, but I want her to feel empowered by information, because that is how you get to be your own advocate if you don't know about it, you get caught off guard, and you can't help anybody else deal with it right. If you try to run away from it. Like with the lung, with kidney cysts and the kidney issues that can come with BHD. If you run away from it and aren't monitoring it sooner or later, that decision stops becoming yours you become less empowered by waiting to find out about it, because if it gets far enough, the decision is no longer yours, it's already been made.

And so while you know we don't get to control all things in our lives, if there's some way I can steward my body in a way that makes it possible for me to enjoy my life a little bit longer, that's the way I want to go with it. And I think that's how I'm going to try to teach her as well not to be afraid of it. But to just know what the risks are so that you can deal with it and not, not run away from it.

Jazzmin: The next question I was going to ask you was, what advice would you give someone who's just been diagnosed with BHD.

Cassandra: Yeah, I would say.

It's shocking. It takes some time to get used to, I think, like the first few days, every little ache or pain or tweak or whatever I'm thinking I'm having a lung collapse. And then, you know, you keep living and surviving days past your diagnosis and you just realize that this isn't like an end of life diagnosis, this is how to continue your life, diagnosis, and everybody does have a really different story.

Like my grandmother who, it comes through, she had no symptoms whatsoever, we all looked at her 90 something years old and we're like, why haven't you had an issue, she was like completely clear of it her whole life. And so it was surprising. But not everybody's like that So, find your community find people who can support you find good medical professionals that listen to you and will acknowledge that you feel things that maybe they don't understand about. And, yeah, building up the support is super important, so seek out the people so that you can be supported.

Jazzmin: Thank you so much, Cassandra. Those are all my questions and it was absolutely amazing talking to you. Thank you so much.