Meet the Patient Interview with Kathy
Interviewed by Katie, Charity Officer, Myrovlytis Trust

Katie: It's so nice to meet you, Kathy. My name is Katie and I'm a charity officer at the BHD foundation and Myrovlytis Trust, and it's such a pleasure to have you here today to talk about your BHD journey and experiences with us. We're so grateful for your time and thank you so much.

Kathy: Thank you.

Katie: So, I'd like to just start off with talking a bit about your diagnosis of BHD and when you first were diagnosed with BHD.

Kathy: I was first diagnosed in March of 2020, and I was also diagnosed with a very rare form of cystic fibrosis, so I have Birt-Hogg-Dubé and cystic fibrosis (CF).

Katie: Wow that's a lot to have quite a lot to handle.

Kathy: I know and my lungs started collapsing when I was 29 years old, and they said they said it happens with thin women and thin people and so time went on and I had a lot of lung issues, as I do today.

Katie: I'm not surprised with cystic fibrosis and BHD that must complicate both sides of the different conditions.

Kathy: Yes and I have to take enzymes for my pancreas. I asked the genetic counsellor if it is possible that they misdiagnosed me with CF and she said no.

Katie: Okay well there is a specific genetic test for cystic fibrosis right so they must be confident with that so that's really interesting. You got diagnosed with cystic fibrosis first because you only were diagnosed very recently with BHD so how did you get diagnosed with BHD?

Kathy: I was diagnosed at 50 years old with cystic fibrosis.

Katie: Really wow.

Kathy: Yes so I found my daughter who well she found me. I looked and looked and looked for my daughter who I gave up for adoption and I could not find her and eventually in 2011 she found me and we reunited and she was having health problems and I told her that I was diagnosed with cystic fibrosis and she went to the university of Michigan and they did all kinds of testing and they said that she had Birt-Hogg-Dubé. And they sent me a letter and said that I should get tested for it and it took me a few years and I decided to get tested for it and I am diagnosed with Birt-Hogg-Dubé.

Katie: Okay so yeah that was quite a gap from finding out your daughter had BHD to you getting diagnosed with BHD. Was that just a personal reason that it took you a while to kind of think about the consequences of getting tested or is the process just quite a long process for you?

Kathy: I just didn't want to deal with anymore you know. I've had 20 collapsed lungs yeah and then pretty difficult yes and then I had almost my whole left lung removed from collapses

until I had them glued, I've had them both glued and I have that's not helped. Last November I had another collapse.

Katie: oh no I'm sorry.

Kathy: Even though I've had my lung glue it's still partially collapsed.

Katie: Wow that sounds like a really challenging time and I'm not surprised it was a difficult decision to kind of find out you know about your BHD diagnosis.

Kathy: Right and I didn't think it was going to make any difference really.

Katie: Yeah, it's such a rare condition that so many people don't know the whole extent of the condition and the impact it could have say on your kidneys and all of those things as well.

Kathy: Yes, and I have tumours on both kidneys.

Katie: Okay and it's good that you're getting those monitored regularly then to check that needed surgery.

Kathy: I went to the NIH in November of last year and they suggest I come back in two years. But I just made an appointment with a new nephrologist to make sure that they're not growing.

Katie: Yeah, that's really important to make sure that they're not going to develop any further. So you've had a bit of an interesting journey to your BHD diagnosis, and I would like to ask you what impact it had on your family members if you have any other family members who have since been tested positive for BHD.

Kathy: I have another daughter and she will not get tested. She says she will but she has not as of yet and her grandson with bad asthma and he knows about the diagnosis he has the paperwork, but he has not got diagnosed.

Katie: Okay that's really interesting to know and would you like them to be tested or do you think it's kind of their choice.

Kathy: I think everybody has a choice in their journey in life and I think when the time comes then they will get tested.

Katie: Yeah, I think it's a personal thing. We would recommend testing if you're comfortable with it just because you know you don't want to find out that you've got severe kidney cancer down the line but as long as they're being sensible, and you know testing is something they'd be made aware of and then it's that choice.

Kathy: Yes, because my daughter has one kidney.

Katie: Oh wow one with one kidney but that's a uh something that she should probably be really considering.

Kathy: It's okay at least they're aware of it.

Katie: So we're going to maybe talk about some of your symptoms. You've mentioned some of these before like your lung symptoms, but I would just like to know a little bit more about those. Do you have any of the skin bumps or fibrofolliculomas?

Kathy: I have a few on my back okay I have one tiny one right here on my forehead but otherwise nothing.

Katie: It's so interesting that everyone has completely different manifestations of the disease and it's so interesting that you know yours seems to be very lung focused and whether that's the combination of having cystic fibrosis as well or not.

Kathy: I don't know why we're all so different.

Katie: Yeah, it's part of the complexity of the syndrome and why we're so committed to trying to fund research into it to try and find a cure or prevention.

Kathy: And is all that research basically in your part of the world.

Katie: No, we fund research worldwide. So we don't have any funded grants at the moment but we're going through this process where we're had some applications of people from around the world to do this, so we are very international and we do genuinely think that it's better if everyone works together worldwide to try and fix this problem.

Kathy: Yes, I want to because I have a key. Each of us with Birt-Hogg-Dubé have a key and we can unlock doors.

Katie: The BHD community is so involved in research as well and that's a really nice thing to hear.

Kathy: You could share my information with anybody.

Katie: So, you've mentioned that you had pleurodesis where they stick your lungs yes?

Kathy: Yeah

Katie: So, have you had any other treatments or anything?

Kathy: Pleurodesis in both lungs, I have a small piece on my left because they removed the upper lobe and the upper lobe in the lower lobe due to blebs that were there and had to be taken off and they said I had emphysema when they did those lung surgeries. So, I didn't know what I had but I I never got better after the surgeries.

Katie: Yeah, it's a difficult one isn't it when you're balancing lung function and the health of your lungs generally and your health overall. losing a lung is never going to be easy on anyone I'm sure it was a decision that the surgeons took carefully.

Kathy: Yes

Katie: Okay so would you say that your lung symptoms have been the most challenging aspect of living with BHD?

Kathy: Yes, most definitely.

Katie: Yeah, and you've had these collapsed lungs throughout your life.

Kathy: Pretty much yes from age 29 until November of last year where my upper part collapsed. It's very painful and I hate the pain involved because they can't well Pittsburgh will not touch my lungs. I went to surgeons, and they said we can't help you and I didn't know

that that would ever be the case because they always inflated my lung with chest tubes but they said I don't have enough lung and it is too risky. They don't think I'd make it.

Katie: Oh, that's so challenging and it's so sad to hear you've kind of battled with severe lung issues for most of your life. I'm so sorry to hear that and so was there anything that you could do to kind of try and manage your symptoms? Did you know when pneumothorax was happening and I guess you maybe avoided going deep-sea diving.

Kathy: Yeahand mountain climbing, so then I just pace my day as I can and sometimes, I need pain medication for my lungs because they are partial collapse and if it's very cold they'll do it and I can feel it. If it's very wet and damp humidity is my worst enemy.

Katie: Oh, okay that's really interesting to hear. So you must be quite aware of your lungs and what's going on. It's something that the average person probably never thinks about how their lungs are feeling.

Kathy: Yes.

Katie: Yeah but it's really good that you're aware of all of these things and that you are in the right place and position to be able to kind of seek medical attention if you need to and you know exactly what's happening with you now. So, you're having had your pleurodesis, that was pretty successful up until last November when you had another collapsed lung. Is there anything else further that they can do to kind of help treat and manage your lung symptoms?

Kathy: No there's not enough it's too risky they said that's right.

Katie: And you're very careful with what you do and I'm really glad to hear that you pace your day and kind of do what you can and that's a really good kind of attitude towards it as well I think.

Kathy: Yes, because every day I could change the day if I have to just take it easy that day and just watch good movies on Netflix.

Katie: Yeah, there is too many on Netflix. Okay so you've never had any treatments for your skin, and you get your kidneys scanned regularly right.

Kathy: So, I've had things taken off my back okay yes and actually I just went to my primary care doctor and she wants to look at them again in January.

Katie: It's good to get them checked over regularly just to make sure nothing abnormal is going on right and if they are you know causing you any discomfort then there are ways to treat that.

Kathy: And I don't know any of that yet because I've only had them taken off one time maybe four years ago so I'm learning more about that.

Katie: Yeah the skin is such an interesting one where it's often overlooked by a lot of clinicians because they don't think it's really important because it doesn't really have any severe consequences, you know compared to your lung collapsing or you're getting kidney cancer, but actually it can affect people quite a lot. It's good to hear that you are going to check in with a dermatologist and and keep that up and see what happens with that.

I would like to move on to some of your thoughts and views the BHD community. What advice would you give to people who are newly diagnosed or maybe going through the genetic testing process?

Kathy: I would say that the Birt-Hogg-Dubé Foundation is a really interesting place to go, and I love the Facebook community because we all can share our experiences and we don't feel alone because we have each other. I would say if you're getting tested for me it's better to know than not to know. I can't fix anything if I don't know what it is to fix and so for me it was a gift to find out what it was so then I can better care for myself in the future.

Katie: That's such a great that's such a great message Kathy thank you so much for that advice.

Kathy: I think that's you know the best advice you could give to anyone really. And so thinking about the future of BHD and research and trying to improve the quality of life of BHD patients what would you like to see in terms of treatment for something or anything?

Kathy: I would like to see more studies, more stuff done and more people followed and I would like to see more testing for genetics done. That way we can understand numbers. When I first heard about it it was very very very rare but I don't really think it is very very rare. I really don't and and maybe I'm wrong but I don't know.

Katie: Well these are questions that we are asking as well because we don't know the prevalence of BHD you know where there are other diseases where they can say one in a hundred thousand people have the disease or something like that, we don't know what that is for BHD and that is something that we are working towards to try and understand further. We need to also understand the disease and the syndrome itself and who it affects and how it affects people. So yeah you mentioned that the BHD foundation is a great resource and as well as the Facebook patient community which is also really great. It's so nice to see so many people asking questions about their experiences and so many people giving their kind of honest and supportive answers in that but how can do you think we can better support you and patients in general?

Kathy: I just would love to have more studies going so then we can figure out not just what's going on with me with my lungs, but what's going on with you and your kidneys and tie it all together. I think it's too scattered out with so many different issues that it's it's not clear to me about you and you about me and if there was more studies done we could figure those puzzles out.

Katie: Definitely and so yeah we are going through funding rounds at the moment and we are hoping to be able to announce the research that we are funding by the end of this year. And hopefully we're going to try and make that as patient accessible and really explain it and try and connect the patients with the people who are doing the research so that we can understand each other better. Because, I think sometimes there's a disconnect between the researchers and the patients and we're really here to try and bridge that and provide a link so the patient community are made aware of the research that's going on but also the researchers are made aware of what's important to you as patients.

Kathy: And is that in UK or united states?

Katie: This will be worldwide so for example we have our virtual BHD symposium that's taking place on the 21st and 22nd of October. So we have a two afternoon session for UK time and morning for America time where researchers are going to be giving research talks and these are going to be aimed mainly at researchers and clinicians but patients are very welcome to attend. But we've also developed two patient-focused sessions which will be very driven by the patients where you're gonna kind of get the chance to share your opinions on the future of BHD, what you would like to see what's important to you. So yeah we would really encourage more patients to attend that because we're really excited.

Kathy: Great and you know what I'd like to know how come when I go to the doctor and tell them I've been diagnosed with Birt-Hogg-Dubé they don't know anything they never even heard of the word you know and I would like to see more knowledge given to primary care doctors to everybody because those are the ones that are going to see patients throughout their lives and have knowledge they never even heard the word you know. So I think that needs to change.

Katie: 100% and that was my final question as to how we can raise awareness among clinicians because we are aware that that's a thing that lots of patients say that it's difficult to find a doctor who has any knowledge and you kind of feel like you're the experts rather than the doctors who you think should be the experts. You end up being an expert on your condition and sharing your knowledge and information with the doctors so that's something we're trying to really work on and distribute information to the key clinicians. If you have any ideas we would love to hear them but we are creating leaflets for clinicians, and leaflets that clinicians can give to any new patients and we're hoping to be able to distribute these to key lung, kidney, skin doctors as well. Obviously this is quite a challenge as an international charity. So we're working we're working on it.

Kathy: That's a start. I think also trying to raise awareness of rare diseases in general among medical students is really important. So it comes from their training and it's inbuilt in them to think about rare diseases and to understand that you don't just have a you know a collapsed lung, you have a complex rare condition that needs to be you know carefully followed up and also kind of a more holistic approach to to treating it in terms of it's not just physical symptoms. You're living with a lifelong condition that you could pass on.

Katie: You have a 50% chance of passing on to your children and that affects you and it affects your family as well. So yeah definitely something we would love to try and get involved with the kind of medical student community to try and raise awareness of rare diseases.

Kathy: And if you had medical students follow one patient with Birt-Hogg-Dubé and see them then all the students in that class will be exposed to something that is so rare. And so I just think teaching hospitals.

Katie: Exactly yeah 100% these are really great ideas. These are things that we are thinking about and the best way that we can kind of connect with people that are unaware of BHD and should be made aware of it.

Kathy: I wonder if it's as rare as we think. I really don't believe it is.

Katie: Exactly I mean it's likely to be somewhat rare but how rare? We know from the literature that there's at least 600 families published worldwide but the Facebook group has like over 2 000 numbers right?

Kathy: Right. So the numbers just don't quite add up.

Katie: Right right and those 2 000 that have two or three kids that might have you know the mutation, then you're thinking it's bigger and that's why I think it's bigger too because of the numbers on the pages and the responses of everybody. Every day I see somebody else join you know.

Kathy: Yeah there's so many new people joining it's so great to see that the community is constantly expanding. Um and it's also nice to see some people who have joined because their partner or their family member has BHD but they don't and they want to find out how to kind of help them and support them. Um so that's a really lovely a lovely aspect yeah of the Facebook group. And I'm sure that they could help you in some sort of other dimension of Birt-Hogg-Dubé because of their experiences too. So getting patients involved in our work and involved in research is something that's kind of at the heart of us as a charity, and we really want to create that environment where everyone can work together towards the same goal. We need you as a community to tell us what it's like to have BHD and what's important to you and what are your biggest challenges. It's really useful to have people like you share your story with us because you have a really interesting story and it's been really nice to hear about your experiences and the just wonderful advice that you've got to give to people.

Kathy: I also want to mention energy. I think it wipes my energy out I don't have energy like I should, I just don't and maybe it's because my lungs are so involved but the autoimmune system is not working correctly with Birt-Hogg-Dubé.

Katie: Yeah I've seen a few posts recently about other kinds of autoimmune conditions some people have mentioned you know rheumatoid arthritis and fibromyalgia and other of these kind of fatigue diseases, and this is something again we'd really like to be able to explore and to see just exactly what the whole extent of symptoms are of BHD and that's something that we can only find out by talking to you and getting you know, getting you involved in research. So thank you so much for sharing your story today and sharing a really cute dog um thank you so much.