

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

Katie: My name is Katie, and I am the charity officer for the BHD Foundation and Myrovlytis Trust. So, we are a medical research charity, raising awareness and funding research into Birt-Hogg-Dubé syndrome. I'm really delighted to get to talk to you today about one of the symptoms of BHD that I think is probably one of the most overlooked, especially from a clinical perspective, that can have a really serious impact on a person's life. And that's the skin bumps or fibrofolliculomas. But first I'd like to kind of know a little bit about your history with BHD and kind of when you were first diagnosed with BHD, and how you went about that.

Julia: Yeah, sounds great. I'm very happy to meet you. Um, I, yeah my name is Julia Thierauf, I'm a surgeon and a scientist, and I was diagnosed with Birt-Hogg-Dubé syndrome, actually, it's closely related to my work because I transitioned from being a surgeon, into a postdoc position in the United States um in molecular pathology, and I, I told my boss a little bit about my family and that my mother was recently diagnosed with bilateral oncocytoma of the parotid glands, and he saw the skin bumps um on my face and he kind of said, I think you should get genetic testing for Birt-Hogg-Dubé I think you might be. You might be a patient. That's how I got diagnosed and that is almost to this day, two years ago so very recently when I was 34.

Katie: Yeah, that is a fairly recent diagnosis, and it's great that you're a surgeon and a researcher, as well and a patient for BHD now and I know that you might be hoping to start some research on BHD and that's really exciting. It's great that we get to see all these perspectives coming from one person that's kind of one of the aims of our charity to bring together patients, clinicians and researchers and you fit that box, almost perfectly. So, you said you're, you had a family history of kind of some of the symptoms of BHD but when and what were your first symptoms?

Julia: I mean going back now that I know know more about Birt-Hogg-Dubé, I can definitely say that I got the first skin lesions on my thorax and on my face when I was when I was 20/21 Kind of. It was, it was pretty hard because you know you come right out of puberty and everyone else's skin gets flawless and you're, you develop these well lesions, and nobody really knew what they were and it was, yeah, I remember that quite vividly actually that I developed that when I was around, around 15 years ago I would say.

Katie: That's pretty like tough to deal with as a young person who, you know, just kind of finding their feet in adulthood, and then find you have got something else to deal with. Have you had any other symptoms of BHD lung, kidneys, or associated symptoms?

Julia: My kidneys were always fine and, actually, I have my annual MRI coming up tomorrow morning. Also, yeah like I said it has been two years now and we kind of um stay, stay within the timeframe of the annual screenings. So that's one exciting part. A little scary but I hope it's going to be fine. And then in terms of the lungs, I've always felt that that my lungs were not the best. I've always done a lot of sports, um, have competed in martial arts, and I definitely can say that I have a lung affliction on my left side I can feel that. However, I never got when I got diagnosed I had kind of the choice my oncologist, gave me the choice of having a CT scan of the lungs or not, because I'm also from from a medical perspective and since I'm a physician myself I um I have that rule, and I say always when when it doesn't have a clear therapeutic consequence and I, I'd rather not want to know because I'm kind of scared that it will alter my everyday behaviour. And I know that these um yeah this can be discussed.

Definitely from a medical perspective, but for me it has always felt good, not to know every single detail of what's going on in my body and I mean for the kidney, it's definitely different because that would have a clear therapeutic consequence in terms of surgery and knowing knowing cancer early enough, but I feel like for the lungs as far as I guess I can do my everyday sport program I'm fine not knowing, and obviously I don't go diving or things that could potentially potentially risk my lungs,

Katie: that's really good to hear. You're getting your regular kidney scans and fingers crossed everything looks okay. It's really interesting to hear about your perspective on the lungs cysts and I guess that's the benefit of being both a patient and a doctor so you can see it from both sides and that's really interesting to hear. So, it sounds like your kind of lungs and kidneys are okay. So, has the skin been kind of been the most challenging aspect of having BHD?

Julia: I definitely say I mean going back when I was 20, I mean I've gotten used to it. I've lived with it for a very long time now, and knowing what it is definitely helps, because it was, um, I've heard, and I don't blame anyone here because it's really, it's an orphan disease, it's so rare. I hadn't heard of it before I got diagnosed basically. Like I said I don't blame anyone but I've heard various things about my skin that my hygiene was lacking or that I should get more facials or that I could should just take better care of my skin, you know, and that those, those were just impurities that would go away or that my hormones were imbalanced and all those things that, where I was, where I was like, yeah, no I really I have tried everything it's they're there and they they're not impurities and I can see that. And I got lasers and it was really, it was, it was really tough when, when you're a young adult, I would say, because yeah like I said everyone else's skin gets kind of perfect, and out of puberty and you're left with these with these skin bumps and on camera, you can't really see them that well but definitely when the light is in, like, the wrong position and it always kind of forced me to wear makeup. When I was more of a, yeah, sports person that that would really enjoy not wearing makeup, but I always felt like I wanted to cover it a little bit so this definitely hasn't been a heavy burden, but it has definitely been something that I've been carrying around over the years and it would be. Yeah, I would be great if, if that could be tackled more in the future and if we would find a cure for that.

Katie: Definitely, it sounds like it really can affect a person's self-confidence. Kind of their sense of self-worth having all of these comments about hygiene thrown at you, and it's hard to deflect a lot, so I guess having, having a diagnosis, at least gives you like a sense of, this is what's wrong with me, kind of thing. Not that anything actually having BHD is anything wrong with you. It's a name to the symptoms that you're experiencing.

Julia: Yeah, that definitely helps accepting that you know it definitely doesn't, it doesn't leave that question mark of, yeah, everyone can see that something's wrong with my skin and a lot of people have commented on it and I mean my mom has it and my grandma had it, obviously. So now we know why. And now it makes more sense it's just gives you a little bit of peace of mind you know, and I can definitely see that's getting worse over the years, so it wasn't that bad when I was 20, and it's getting more and more and looking at my mom she has a definitely more severe than I have. And she's come to peace with it as well but like I said it's just, it's just nice knowing what it is you know and that it's not something that you could have prevented.

Katie: Yeah, and having to live with something for the rest of your life. And with the idea that it actually might get worse to some extent, is you know a difficult thing to balance and I'm really glad that your mum has come to peace of mind with it. It doesn't affect her as much as

it might do other people. So, you mentioned that you try to have some laser treatments and stuff, but how did you go about finding a good dermatologist.

Julia: Finding a really good dermatologist actually took me until I was diagnosed, because in the same month, I was diagnosed with Birt-Hogg-Dubé syndrome, I was diagnosed with skin cancer. So, on my forehead. And that is something that to my knowledge, hasn't been directly associated with BHD but could potentially, of course be was a basal cell carcinoma, on my forehead that I saw growing for three weeks or so and you know, if you have a lesion on your skin that looks suspicious and doesn't go away within that timeframe, then you should definitely see a dermatologist and I saw her and she biopsied it and came back positive for cancer. So, I have had a little surgery and that that was an I mentioned that I also worked on Birt-Hogg-Dubé syndrome, I was just recently diagnosed, like I said that was by that time a week ago, so it was really within the same month. And that was kind of when I found out an expert in Birt-Hogg-Dubé and all in general skin in dermatology. So, before that. Like I said, I really don't blame any physician, but you get all kinds of treatments offered, and you take them because you're kind of desperate, you know, you try, you try this and you try that, and you spend rather good amount of money on laser treatments and, and then the lesions come back after three, four months, you know, and it's, yeah, it's not only painful but like I said, also from a monetary perspective quite burdensome. So, yeah, I've done laser treatments I've gotten tons of facials skin, dermabrasion, things like that. And, yeah, but when I was diagnosed I kind of found my, my routine that works for me, works for my skin, and that I'm very happy with together with my dermatologist my oncologist and the entire team that helps me

Katie: I am glad you have got a really good support network. Yeah, finding a dermatologist that knows about BHD does sound like a difficult thing. And I think many people would struggle with, so it's really useful to know kind of your story and I'm kind of almost stumbled into it. What would you say that people could do the public patients us a charity, could do to help raise awareness of BHD within dermatology?

Julia: Interviews like this. Yeah, I think just the, I mean, the good thing about the Birt-Hogg-Dubé community is that we are so well connected, you know, and that we, we are, we've always relied on being on spreading the word and being out there and connecting with other clinicians, patient advocates, patients. And I think this is one of our big strengths that we that we that we're able to do that, so I think having more meetings, also participating in bigger meetings, you know, as a, as a smaller group, but representing an orphan disease and as we're moving more and more towards the, I don't necessarily like the term of personalised medicine. I mean the term has been abused over the years, but I think the point being is that we're all individuals, you know, and we all need different cares, and I think as we're moving towards that individual patient care more and more, it becomes it becomes less of a hurdle to have a very rare disease. So, yeah, I think what we're already doing is great, spreading the words, participating in bigger conferences, being out there raising awareness and there's a lot of, now that I've started being in the network, there's a lot of people who actually care about and do a lot of brilliant work. So, I'm very, very confident that this, this will continue, and this will raise more awareness.

Katie: Great the future can be bright for BHD. Just to talk a little bit more about some of the treatments, you've received. It sounds like you've probably almost had everything under the sun, that can be offered to you. How did you find out about kind of what the different treatments were? And what did you take into consideration when thinking about potential treatments.

Julia: I'm always like I said I've always tried a lot of things but that's also my nature. I'm always curious to see what's out there and you know it might or might not work so that's really kind of a single person study I would say I always describe it like that. On the other hand, there is no there's no right now there's no targeted skin treatment. So, if there was one, or if there was one in development, especially for folliculin induced skin lesions, I would definitely be one of the first volunteers to participate in that. On the other hand, Yeah, like I said, the more general, more common treatments I would say, I just, I just tried them and figure out if they work for me or if they're too harsh for me I also at the same time have very sensitive skin, I don't know if this is associated to Birt-Hogg-Dubé or not but everyone in my family from the Birt-Hogg-Dubé side has very extremely sensitive skin so it was a lot of troubleshooting I would say, and now I found. I found a dermatologist, that offers really gentle facials and yeah like I said I found my routine. So, I've learned, I've definitely worked with with the skin lesions.

Katie: That's great to hear. So, you talked a little bit about how you been able to come to terms with your skin, quite well, and you found your routine that works for you. Have you had to kind of think about any of the other aspects of BHD so the kidney/lung symptoms and kind of it being an inherited condition and having to come to terms with that as well?

Julia: Definitely. Like I said, the lung is, at least for me pretty manageable. I'm probably not doing as many sports as I as I would do if I if I wasn't diagnosed. But the kidney, like I said it's fine right now. The one thing I keep remembering when I was when I was newly diagnosed, I saw I was in the department for reproductive genetics. And I was told that I would have to have IVF if I ever wanted children. I don't have children right now. And so, it's an autosomal dominant disease, the likelihood is 50 percent that I that I give that to my children, and I was told that I, I should definitely consider IVF to kind of eradicate the disease. And I have a little bit of a, I have a little bit of mixed feelings about this, because I think it's very important that we emphasise on that we, like I said that we spread the word that we raise awareness for disease, and that we're doing more and more research on it, and really getting to the to the basics and developing, developing treatments. On the other hand, I personally, and this is, this is a very personal decision. And I don't say that this is right or wrong, but for me I would never consider IVF, for Birt-Hogg-Dubé. I definitely can see how this can be a point to be discussed, but I for example would never would never consider that I mean, like I said, None of the people in my family had kidney cancer and I can definitely see how this could definitely be an argument towards that, but we know so little about Birt-Hogg-Dubé and how the single, the single mutations affect the family and we've also seen that it can have tremendous differences even within the same family. But for again for my family, my mother is 75 and she is relatively healthy I don't think she takes any medication, actually. She recently broke her arm but that's pretty much all I remember. My grandmother was almost 90 when she passed. So, I would want to choose, or I wouldn't want to go through something, as invasive as IVF, just in order to eradicate the follicular mutation in embryos so but like I said this is a very, very personal decision and I'm not saying that this is right or wrong, it's just a thought that I had when I was when I was diagnosed and that I thought about quite a lot. So yeah, just wanted to share that.

Katie: Thank you for sharing that with us. It's a very personal thing and we do really appreciate that. So, you can live a perfectly kind of happy, healthy life with BHD. It's such a big decision to go down that route.

Julia: I think, I mean I always, I try it. I always say, and again, it's a family with BHD is affected in a in a in a dramatic way in terms of metastasize kidney cancer or something like that this is a totally different story but if you look at other mutations, for example like BRCA mutations that will give you a very very high likelihood of developing a GYN cancer over time, in about 80 to 90%, I definitely think that this is a different story you know and I can definitely see how this is being treated completely different than, than for example Birt-Hogg-Dubé and like I said, it's, it's possible to live with it, and I think it's at this point it's, it's, we should, we should really focus on developing individual care packages and treatments, and, and, and getting to know more, more about Birt-Hogg-Dubé in order to really find mechanisms that we can tackle over time, rather than the aspect we just discussed. So, yeah, I really think that getting to know more about it through an excellent network such as you're providing that, that would be that that is right now, the most important, the most important topic.

Katie: Definitely and having the knowledge of BHD you can introduce screening measures, and lots of other things to try and, you know, monitor your kidneys so the chance of developing kidney cancer is much reduced, and therefore the chances of it metastasizing and being even more serious is also greatly reduced as well. So, just wanted to know what your advice would be for someone who is struggling with both physical and mental health aspects for their skin bumps and BHD symptoms more generally.

Julia: My advice would be to get in touch with other BHD patients. I know there's great patient networks, very well-connected kind of all over the world. My mother is in the Munich area, she has a great oncologist that connected her also with with different patients and yeah, I definitely think that talking to other patients and their experiences is helpful. Doctors can definitely help in that term. But I really think that reaching out to other patients that are out there is definitely very helpful in learning to accept this disease and live with it and just be aware of things that you can and cannot do and screenings that you should and should, yeah, that you should have over time so yeah, I definitely think that the networking that the networking helps.

Katie: I think that's really great advice. We can say that there is a great Facebook page on BHD. So, if anyone is watching and struggling to find a network of people. We at the BHD foundation can help you with this, and we will be more than happy to put you in touch with people. And I think I have one more question, which is, what would you love to see in the future, in terms of improving the quality of life for BHD patients.

Julia: Treatment for skin bumps. A nontoxic one, ideally. As we discussed before, more broader knowledge about how this fits in to others similar conditions that have been described. Really trying, I'm talking from a molecular pathologist level now, like really trying to understand where it fits in how its regulated, what the exact function is having, Yeah, having more basic knowledge about Birt-Hogg-Dubé, and then I think in terms of quality of life, like I said, targeted targeted skin treatment would be a dream of mine and I'm very sure for many, many affected BHD patients. The monitoring, I want to say I mean, if you're, if you're in a country that offers a great health care system. The monitoring is great when once you're once you're diagnosed and once, you're aware of it. The monitoring works very well I think the tricky part is not being diagnosed and I, as I as I read more and more about BHD I really started to think and talk to, talk to many, many well established, researchers in the field. I started to think about what the what the actual number of BHD patients is and how many are out there that have never been diagnosed, I mean like I said my own stories. I had the first symptoms when I was 15, I was diagnosed when I was 20, so 15 years ago and I was diagnosed three years ago when I was 34. So, I think what I would like to see in terms of

quality of life is being that patients can be diagnosed earlier which brings us back to our original point, how can we spread the word and how can we make sure that people are more aware of Birt-Hogg-Dubé. So, I think this is, this is a very very important message.

Katie: Definitely and raising awareness to something we are very keen to do to and we would love to get involved with you, and other patients, trying to establish more ways of how we can raise awareness of BHD, among all different groups of people from the general public, all the way researchers and clinicians as that will be probably the most important thing and early diagnosis can make a huge difference to someone's life.

Julia: Yeah, and I'm very, very much looking forward to more in person meetings and really hoping that the that the annual Birt-Hogg-Dubé meeting is coming back in person soon so we can, we can definitely I guess network easier. Like I said also participate and spread into, into bigger conferences, kidney, lung, I mean it's such a, such a broad field you know where definitely patient advocacy groups, and or scientists should should participate in. So much very looking forward to in person meetings as well.

Katie: fingers crossed for our BHD symposium happening in person next year. I think that is all the questions I have for now. So, I just like to thank you so much Julia for answering these questions, taking the time to talk with me and just really appreciating your honesty about your journey with BHD and how it's affected you and I really think that a lot of people who will watch this will be really inspired.

Julia: Yeah, Thank you so much Katie. This was really great.