

Interview with Bob Rinaldi  
Interviewed by Jazzmin, Charity Officer

**Jazzmin:** Hello, my name is Jazzmin, I'm a Charity Officer at the BHD Foundation and Myrovlytis Trust and today I'm delighted to be into interviewing Dr Bob Rinaldi about his experiences with BHD and also getting a bit of that doctor's perspective of what it's like navigating the medical world. Bob it's lovely to meet you today how are you doing?

**Bob:** I'm good this morning my pleasure to be here.

**Jazzmin:** Fantastic so I'd like to start with asking you how and when were you first diagnosed with BHD?

**Bob:** Surprisingly almost a year ago to this day.

**Jazzmin:** Perfect timing.

**Bob:** Yeah a very good time .

**Jazzmin:** What was the first sign and symptoms that you got that made you think oh there might be something going on here?

**Bob:** Well, it's an interesting story Jazzmin. So about 30 years ago when I was in my medical training I actually had a spontaneous pneumothorax and I've been an athlete my whole life. It just came out of nowhere. We had no idea why and of course 30 years ago I don't think anybody knew what BHD was. They just diagnosed me as having a congenital bleb it resolved within about a day and I just kind of went my merry way.

About a year ago I had a physical exam just for my annual check-up with my primary care doc and as part of that they did a heart CT scan looking at calcium score and the blood vessels in the heart (that's basically an indicator of how much plaque build-up there is in in the vessels of the heart). It's a pretty routine study done here in the states anyways they noticed I had a dilated aorta and so they decided they wanted to do an cardiac angiogram to look at the aorta. And you know fortunately when they did the angiogram the technician took the scan down towards the level of the kidneys and it was caught on that scan there was a large about six centimetre by two centimetre mass on my left kidney. I remember my primary care doc called me that evening and said hey I wanted to follow up on the CTA. Your aorta's dilated I'm not worried about that what I am worried about is this large mass on your left kidney. And that hit me like a ton of bricks. My mom passed away from kidney cancer about about seven years ago. So that that hit me like a ton of bricks and I was like oh my god what are you talking about, and he said yeah we need to get you in to see a urologist and figure out what's going on here.

I saw the urologist and they immediately said your risk factors for this is the genetics as your mom had it. I didn't know there was a genetic component to kidney cancer and he said yeah there is. So I met with a geneticist and that's when they diagnosed the BHD. The geneticist was actually funny. I was sitting in the meeting with the geneticist and she began looking at my forehead.

**Jazzmin:** You're like what's going on here?

**Bob:** I'm like what is she doing. I didn't know what BHD was and she was looking at my forehead and she looked at me and she said how long have you had those spots on your forehead and I said well you know for as long as I can remember they've been there for 10, 15, 20 years. She said that's consistent with a diagnosis called Birt Hogg-Dube syndrome have you ever heard of that? I said no no clue what that is. And she said pneumothorax, kidney cancer, and fibrofolliculomas and I'm like just ticking off the boxes and that that's when they diagnosed me and again that was almost exactly a year ago.

That's how I arrived at my diagnosis you know by the grace of god a technician decided to take a scan down far enough and caught the mass in my kidney and it just was one of those very fortuitous things they. It would have been diagnosed eventually but the earlier the better obviously.

**Jazzmin:** Completely and are they or have they done anything for that mass in the kidney.

**Bob:** Yeah I actually went to the operating room for a partial nephrectomy about a month and a half later after they made the diagnosis.

**Jazzmin:** Brilliant and I think what's really interesting about this case is the thing that kind of tipped your urologist off was the fact you have that family history. Making sure doctors are asking that question is so so important and I'm so glad that they caught that and put those dots together.

What kind of monitoring are you having currently for your BHD?

**Bob:** Right now, I'm getting annual MRI scans. I've actually requested every six months at least for the first year to two years. I had my first follow-up MRI scan two months ago and it was clean no recurrence no metastases. I get my second one in September. My main concern with my tumor is it was a stage one but a grade four. Grade four being the most dysplastic (abnormal) type of tumor that there being strictly confined to the kidney. We caught a very aggressive cancer very early and that's been my main concern in wanting to get more frequent MRI scans. It is just to make sure that this thing didn't metastasize and we just didn't catch it on the MRI scan. My urologist had been very agreeable to that at least initially.

**Jazzmin:** I think the thing is with a lot of BHD associated kidney tumours they are quite slow growing but there are cases where they are faster and as in your case higher grades and this is something we're very interested in finding more about at the BHD Foundation. We've actually developed a registry recently and what we're really hoping to get from it is to get a better idea of different kidney cancers people are getting and also the monitoring that's needed because your doctor said every year MRI's and you kind of went every six months which is perfect for you, but then other doctors are saying every three years if there haven't been any kidney cancers. Trying to find what is best is so important and it's what we're hoping the registry will eventually do. We're very excited that we've got that up and running and we are trying to get as many people as possible involved in it because the big thing with registry is as I'm sure you know is it is all about numbers and BHD being rare it can be difficult to get enough people involved to answer those research questions.

**Bob:** Absolutely. You know you speak to the rarity of it that's really interesting. My wife is very curious about it obviously and I'm on the Facebook page and it's it's fascinating to

see it seems like every day there's a new person you know being introduced to the group and that just speaks to the fact that this is rare but I also at my core I feel it's not quite as rare as we think it is. Maybe it's out there and we just aren't recognizing it and awareness is so important.

**Jazzmin:** Completely it's amazing that in the literature (the literature is quite outdated now) it says there's 600 families worldwide and as you said you look at that Facebook group and it's so much more. I think that Facebook group has been such a haven for so many people because when your doctor's not heard of the condition when you've heard of no one else with the condition it must feel something's quite isolating so having that connection is so important.

So has being a physician has that helped you manage and understand BHD and how?

**Bob:** It's been helpful. it's also been stressful you know as a physician we always laugh that sometimes we know too much right. So that side of it for me has been a struggle. I do know too much in some ways and that doesn't let me quite relax into things as easily. It's been helpful in the sense that I've known immediately where to go for information factual information. I have my resources on the internet that I that I know are appropriate resources as everybody knows the internet's full of you know misinformation. I do know the resources that I've used as a clinician. I went straight to those and those are all publicly accessible. I was able to go to those, I was able to get a better understanding of what BHD was and then immediately dug down into the tumor types and the staging and the grading aspect of it. My background has been very helpful in me gaining a fairly in-depth understanding of the disorder fairly quickly.

By the same token I just mentioned it's also kind of been a struggle between my very logical brain my physician brain and then my emotional person's brain. I've been trying trying to rectify those two things and I've been fortunately or fortunately enough I've been able to lean into that sort of logical physician brain to allay many of my fears.

**Jazzmin:** That's good but as you said sometimes you do need to be that patient as well as that doctor and finding that balance must sometimes be challenging.

**Bob:** It can be and the one thing I didn't want to do when I first was diagnosed with this (for better or worse this is how I managed it personally) was I didn't want this to define me you know as a person. So it's yes I've got this diagnosis I understand what the ramifications are for this. I understand what my future is. I understand this isn't going away but I'm also not gonna let that define me. I'm gonna continue to do everything I love to do in life and continue to do everything I want to do. And yes this is there but you know outside of very very close friends of mine and my family I have not really let a lot of people know that I've got this. I've found that to help me, I'm not putting it in the background not compartmentalizing not ignoring it, you can't do that with BHD you can't but I've just found that if I move on with life and don't let this define me that's been very helpful as a physician again knowing a lot about the disease.

**Jazzmin:** I think that's very wise word. Another person has said to me also that they're just going to kind of continue on their life occasionally have BHD dates with their mum where they go to the doctors have all their tests but it's not going to stop them doing anything. You've now got that information to look after yourselves and do the right things

and keep monitoring which is information that you wouldn't have if you didn't have the diagnosis and it'd still be there but now you can be informed and look after yourself.

So, what advice would you give to other people who've been diagnosed with BHD when talking to their doctors about BHD and just in general navigating the healthcare system? We will be getting the kind of American perspective on this I realized. It'd be great to hear what your thoughts are on that.

**Bob:** Yeah there's this nasty tangled healthcare system it's ridiculous. In my specialty area I've leaned into my own personal experience as a physician. I work with children with physical impairments and disabilities and of course congenital and genetic disorders and rare disorders have that. So I see a lot of patients who have things that aren't described or we don't know a lot about. So I've always leaned on my patients to educate me because they come in knowing more about it than I do. Of course as physicians we can't learn everything in medicine we don't know all the diseases you know and I think it's a very important skill for a physician to say I don't know because we don't. I didn't know what BHD was. So I lean on my patients to educate and I love it when a patient comes in with information for me and says this is what we have, these are the websites where you can go to learn more about it if you don't know about it and that kind of builds in that relationship that I have with my patients. You know second third visit they'll bring in the information because I'll ask for it so it really is a mutual process or a process of mutual education between me and my patients. In my understanding what they've got and you know where those resources are and then me helping them understand more in depth what this disease means and what the process is behind it. Um there's that part of it.

As a patient I think it's very important to help educate doctors. I think as a patient it's also important to be an advocate for yourself. Nobody's going to be an advocate for you but you I've found that out the hard way in in many aspects of healthcare so be an advocate for yourself. If your physician doesn't understand the diagnosis or doesn't doesn't want to take it seriously provide them with the information be the squeaky wheel. Believe me you have to be, you have to be because you're the advocate yourself. I think that's important to understand, if you're not getting the answers you want to hear that you need to hear then look for other places to get those answers. Whether that's reliable online resources, whether that's second opinions, don't be afraid to get second opinions. I always support my own patients if they're seeking second opinions and I think any physician who's worth their salt would support a second opinion. So be an advocate for yourself, seek information, mutually educate your physicians and hopefully they reciprocate that.

**Jazzmin:** What I'm going to do for every anyone watching this interview put a list of resources underneath it. Obviously there's the BHD Foundation but Bob it'd be great to get the resources you think are reliable and compare to our list and we can kind of collate lists for underneath this video for people to have a look at.

The final question I've got is what can the BHD Foundation do to educate more doctors. I know you've kind of touched upon it by giving patients those resources to be able to take to doctors but are there any other tips and tricks you think we can do as a charity?

**Bob:** Yeah I think the biggest thing is visibility. As physicians we don't get a lot of training in a lot of these rare disorders and we only find out about them through patients who come in with it right or if we happen to learn something about it in the literature that we're reading. As a physical medicine rehab doc I never would have stumbled across

BHD ever it's just not in our literature it's not what we do. My knowledge base never would have existed had I not gotten it. So I think visibility is one of the strongest things. At least from what I've found one of the best exposures I've had as a physician is at annual meetings when foundations are at our annual meetings, and they are providing information directly to us at annual meetings. it's sort of a captured audience. I think reaching out through annual meetings, annual conferences and being visible with those and providing information for those is important. Again I think it's very grassroots too. I think patients have to educate physicians about these things. The media can help, social media certainly helps with regard to getting word out and increasing awareness. So it's tough you know as you mentioned seven thousand rare disorders and it's hard.

**Jazzmin:** I'm really glad you said about annual meetings and conferences because we have created a new BHD leaflet with the idea that we're not trying to educate doctors completely about BHD but we're trying to make them think kidneys, lung problems could this be BHD or I've read something that is linked to that. We've created this leaflet and we're trying to get it at a few different annual meetings because at the moment traveling is not the easiest thing. So that is kind of one of the routes we're going down and really good to hear you say that you think that could be a viable way to increase visibility of BHD. Very powerful way to do it I think.

**Bob:** Jazzmin absolutely if you've got a captured audience of a thousand, two thousand physicians, 500 physicians who are going to see that information and think huh what's that and read about it and that'll stick in the back of their mind.

**Jazzmin:** Even if one or two doctors remember it and see that patient and catch that patient early enough to get them starting on the monitoring it's completely worth it because it's benefited that person.

**Bob:** Absolutely yup.

**Jazzmin:** Well thank you so much for talking to me today it's been an absolute pleasure and good luck with all the fantastic work you're doing as well.

**Bob:** Thank you, thank you I appreciate it