Interview Transcript — Dr Ilene Sussman

***We're at the Vontz Center at the University of Cincinnati for the Fourth BHD Symposium. My name is Jill Woodward and I'm here with Ilene Sussman, the Executive Director of the VHL Family Alliance. Ilene, can you talk about the goals and activities of the VHL Family Alliance?***

The VHL Family Alliance was founded in 1993 by three family members or patients themselves, because they felt isolated and with a rare disease or a rare condition that they didn't know anyone with the same...to be able to speak the language. And of course they were having problems being diagnosed and being considered seriously by clinicians. And so they founded the organisation to, first of all, to create a community of support so that there was not a feeling of isolation. And that's something that really has grown. We have contact with about 90% of all diagnosed cases of VHL throughout the world. So there is no longer a feeling of isolation. And of course there was patient education. People didn't know anything about the disease at that time, or the condition, and so now we have created forms to educate the patients and support them, particularly in times of crisis, and to connect them with other patients, as well as to help them and to empower them to speak to their clinicians. And so the next phase of course is to be able to work closer towards a cure with the researchers as well as the healthcare providers and that's the direction that we're going in now, as well as continuing our extremely important efforts in terms of education and support and also feelings of hope for patients.

***Can you tell me a little bit more about the history of the VHL Family Alliance?***

It was really a grassroots effort to create a community, and of course in 1993 the world was very different. So now that we have technologies through things like Facebook and what we have as in Inspire, another online support community, their vision of creating a community, a family, as it were, of VHL patients has really come to fruition.

***How can we encourage doctors who are often very busy to consider rare disease as a diagnosis?***

It‘s a challenge. It’s definitely a challenge because doctors are trained to see horses rather than zebras as we would say, or as Dr Groopman says, Jerry Groopman says. There are resources for clinicians out there such as, particularly with the internet, such as UpToDate, WebMD. And when they are baffled, when they reach a point where they don’t know the answers, they do refer to these resources. And so we are working with places like UpToDate, not as much with WebMD, but particularly with UpToDate on getting—making sure the information about, in this case, VHL is accurate and up to date. And that they have resources within that website to get the information. We also provide our patients with handbooks, VHL 101, everything you need to know about VHL: the guidelines for diagnosis, the best treatments that are out there at this current time; in fact the fourth edition is just coming out as we speak. And we provide this for the patients so that they can ask the right questions and also provide information to their clinician. Patients need to be empowered and need to be educated so that they can ask questions.

***How did you become involved with the VHL Family Alliance?***

I just started about six months ago as the VHL Family Alliance Executive Director. I saw the advertisement and it talked about some, as I hate to use the word but—an unusual disease. And there was something in the back of my mind because at my previous position I had a very close colleague who talked about this quote-unquote weird disease that his wife and family had had, where tumours were constantly growing and as you removed them more would grow. And it didn’t take long for me to do the research, to put two and two together and realise that this was actually the disease that my friend had been talking about. So I felt sort of a connection there and felt it was interesting and important to do this type of work.

***Does the VHL Family Alliance collaborate with other organisations and how might that help your work?***

Absolutely. There is so much overlap in so many of these rare diseases. So for example BHD, where you have the kidney issues, the risk of kidney cancer, and the importance of screening, has the commonality. So clearly that’s an area where we would love to work with the BHD patients. We actually are doing a similar thing with HLRCC which is a new organisation, a new patient advocacy organisation, and we’re helping them with their website, we’re helping them create their online community. They actually have just completed their first handbook

and are getting ready to post it on their website and after that edition we'll help them get to a second one which can be printed. So we can use our models to help other organisations as well as our infrastructure; for example, we are already a 501(c)(3), so donations can come through us and [are] earmarked for different things. As well as wanting to work together with the researchers and the clinicians to help educate them and to help find a cure for all these diseases.

***What are the largest needs right now for understanding von Hippel-Lindau syndrome?***

That's a very good question. There's a lot known about the molecular mechanism but there's still a lot more unknown—what the downstream effects are. Of course what are the best tools for prevention or for treatment—that's essential. The other thing is we know a lot about the genetics and the background of VHL, but what we don't really have firm data is the history: individual and family history. We know it's genetic but we don't know why it is somebody in one family presents differently than someone else in that same family. We don't know the effect of environment. We don't know the effect of other external drugs. For example, just now that we're starting to understand pregnancy in VHL—but actually a study has been done in France. We don't have that information here. So creating a registry so that we can track history in a scientific and a searchable way is essential. And also to link that to a tissue bank so that we can give tissue to researchers or provide it to them so that they also have the history of that patient, which is going to effect and could also help in their research. And then thirdly that registry can also be used to engage patients in clinical research.

***There are currently clinical trials for VHL and VHL-related symptoms; can you talk about the process of getting a drug from the bench to the bedside?***

You have to start in basic research. When I was in research, my Master's advisor used to tell me that ten out of every experiment you did, ten times—ten of them were negative results. It's a very long and slow process. Once it hits, you have an idea and you let's say find a molecule that could have an impact on a given treatment or a given disease or a symptom, it then has to be taken into animal models and tested in there, larger and larger. Once that's done, it then has to go from the lab into the human where it has to be scaled up; if it's a protein, made sure that you can purify it and clean it and produce whatever it is in an appropriate fashion. And of course what happens in a mouse doesn't necessarily happen in a

human being. In the case of VHL it's also more complicated, because it hits so many different organs or sites within the body that it could be beneficial in one area and possibly cause harm in another, so that obviously complicates it. And it's expensive and the resources need to be there in order to improve our healthcare.

***And that brings me to: what is your feeling about the state of rare disease research and funding as a whole?***

Well, research funding is very difficult these days, as a whole. NIH has done a lot of cutbacks. Those that are being funded are being delayed; they're not getting all the money they had hoped for, they had been originally planning to get. So that's just the general environment in which we live. In the case of rare disease, there's also creating that interest. And again by using teams of rare diseases with commonalities, that interest can be raised to a higher level. It also requires a lot of interest from pharma because clearly the dollar speaks. There's a lot of money in diabetes. We have a real crisis in our society around diabetes. So the pharmaceutical companies see the benefit of it financially. That's not always the case in a rare disease. So there's also the humanitarian effort that needs to take place. In addition to that, it's not as—because of the newest regulations in our society or within pharma, it makes it even harder to get funding for rare disease research through pharma because they cannot speak about and they cannot work with certain types of areas, if that's not the primary indication of the drug. It just makes life much harder.

***What have been some of the challenges and rewards of working in the medical non-profit sector?***

Well, I'd like to talk more about the rewards. Because I did work in the for-profit medical research community and since going into non-profit I have found it exceptionally rewarding. That a patient can call and speak to me for two seconds and I maybe have just said hello and all they're saying is "thank you, thank you for all you do". How much more rewarding can life be? The challenges—there are many. There are challenges in any job that you take: in any political environment, anything. But, you may not get the financial benefit from being in a non-profit, but you certainly get the human contact and the feeling that you're really helping somebody out.

***Ilene Sussman, thank you very much for taking some time to talk with us today.***

Thank you.