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Myrovlytis Trust: Searching for Orphan Disease Cures

FINDING BETTER TREATMENTS AND CURES FOR ORPHAN DISEASES

Change and challenge is in the wind as 2008 comes to an end. The same is true when examining this month's ReachMD XM 160 special series - Focus on Global Medicine. We take a look at both the changes and the challenges impacting global medicine.

There are millions of people around the world with orphan diseases. Who helps these patients with rare diseases, find better treatments and cures. Welcome to this special report on Global Medicine on ReachMD, The Channel for Medical Professionals. I am your host, Dr. Bruce Bloom and joining us to discuss finding better treatments and cures for orphan diseases is Mr. John Solly, charity manager of Great Britain's Myrovlytis Trust, Not-For-Profit organization, dedicated finding treatments and cures for rare disease.

DR. BRUCE BLOOM:

Mr. Solly, welcome to ReachMD.

MR. JOHN SOLLY:

Thanks for inviting me.

DR. BRUCE BLOOM:

So, when was the Myrovlytis Trust founded? What does that mean and what are its aims?

MR. JOHN SOLLY:

See the trust was founded in mid 2007, so it has been around for about a year in London, in Great Britain. It has 2 aims, to promote research in rare genetic disorders that is including, but not limited to Birt-Hogg-Dubé Syndrome, which I guess we will talk a little bit about later. Then, the second aim is to advance education of the public in medical and molecular genetics and in terms of the name, it means Myrrh-Scented and it is related to our source of funding.

DR. BRUCE BLOOM:

So, why do you think the Myrovlytis Trust or the philanthropist put it together? Why would they pick orphan diseases, wouldn't significant funding make a broader impact if you found a better treatment or cures for larger diseases?

MR. JOHN SOLLY:

So, I think there are several reasons here, one though important one is the idea of the quality of opportunity to us or the trust. It is unclear that health should be subject to some kind of rigid utilitarianism. You know the greatest good for the greatest number and so may be health is the special case where, however, rare your disease, you know, you have a chance of being able to live a healthy life. So, here is the trust is motivated by quality of opportunity. We don't really understand why just because your particular rare genetic disorder that you happen to have inherited happens to be very rare, just because that happens you receive an inferior quality of treatment compared with somebody who has a more common disease. We believe that the same level of treatment, the same range of drugs and the same quality of drugs should be available to everybody really regardless of how rare or how common any genetic mutation you have might be.

DR. BRUCE BLOOM:

And have you looked at the disproportionate funding either in Great Britain or around the world as it affects orphan diseases and what does that tell us about, how governments feel about this.

MR. JOHN SOLLY:

So, orphan diseases are the case where the market fails essentially. There are thousands and thousands of orphan diseases and many of them have essentially no money, being put into them at all and so the government recognizes this certainly in the UK and I believe its similar in the States as well and has introduced excellent legislation in the orphan drug acts and equivalent pieces of legislation in different countries and so they are aware of the problem and they are trying to do something to tackle it, but even that isn't quite enough to encourage drug companies to invest in developing drugs for what a very often very, very few patients .

DR. BRUCE BLOOM:

And is it part of the purpose of the Myrovlytis Trust to engage industry and government in doing more around these kinds of orphan diseases, is that part of why you exist?

MR. JOHN SOLLY:

Absolutely, yes. So, based in terms of our specific disease that we are looking at the moment, Birt-Hogg-Dubé Syndrome and more generally we are working mainly within the UK at the moment, we are just in the process of setting up some policy work with Nowgen which is an organization, a genetic center in Manchester in the North of England. Americans hopefully be able to start to develop policy and engage with government to work with this, you know, it is very important program.

DR. BRUCE BLOOM:

So, you mentioned the trust has a focus on genetics. How did they focus on those kinds of orphan diseases versus orphan disease in general?

MR. JOHN SOLLY:

So, also the funding has an interest in Birt-Hogg-Dubé Syndrome, which is a genetic disease and so many of these orphan diseases have a genetic basis, those were the 2 primary reasons and it feels that at the moment, we rule this new sequencing technology and all this new information that we are finding out every day about genetics and about genes and about what is going on in terms of biology that this is a real opportunity where we are at the start of the 21st century to be able to make a difference to orphan diseases approaching them primarily from a genetic point of view.

DR. BRUCE BLOOM:

Do you have sister organizations in other countries or even within Great Britain that are also looking at the genetic basis of orphan diseases or you sort of one standing out alone right now?

MR. JOHN SOLLY:

So, this feels like a situation where there are organizations, particularly in the States who are ahead of us here in the UK in Great Britain. So, there are several organizations that we have been learning from in the States. In the UK, it is less so, I think, there are some organizations, but the idea of Philanthropy targeted Philanthropy in this manner is I think less developed than it is over with you in the States.

DR. BRUCE BLOOM:

And who have you partnered with so far to move your agenda forward and how did you select those partners?

MR. JOHN SOLLY:

So, the first group is Research Scientist that is the biggest thing for us so far at the moment. We have given about more than 1.5 million pounds in grants to some basic research that is probably well it depends on the exchange rate, but may be 2.5 to 3 million dollars, the first big grant that we gave was to Dr. Eamonn Maher, who works at Birmingham in UK. He is one of the world's leading experts on kidney disease and inherited kidney disease and then we chose other scientists, for their scientific expertise. We are going to work with the best guys. We are lucky that we are able to do that and we chose people according to where the guts were and knowledge of the biology. So, that is the first group that we set scientists and then we are working with patient's organization in the States to BHD family alliance, they work with patients, together we help to organize the first scientific conference about the BHD syndrome and we have been working with other scientists to develop some kind of consortium in Europe. So, these are the partners that we have been working with at the moment.

DR. BRUCE BLOOM:

And how many people attended the BHD symposium and what did you manage to accomplish there?

MR. JOHN SOLLY:

So, we had about 47, I think, people there that included all the main guys, a lot of the initial work into BHD has been done at the NIH. So, we were very lucky that several people from NIH were able to be there including Dr. Marston Linehan and Laura Schmidt as well as all the other main guys from around the world from Europe and from Japan and from everywhere basically and it was the first opportunity for some of these, the research that was the first opportunity for them to meet each other. It's amazing having been working on this disease for many, many years and reading each other's paper and so some of them, you know, had never been in the same room with each other. So, it was very good as a starting point for us to begin to move forward. I think it was a focus point, this is where we are, these are the guys working on it, this is what we know, it's like a baseline. We set the baseline and now it's almost a < ____ >.

DR. BRUCE BLOOM:

In addition to the researchers, were there also patients and applicants at this meeting?

MR. JOHN SOLLY:

There were some, yes. The Cathy Sherman from the BHD Family Alliance, she was involved in organizing it with us, so she was there and a very few other patients as well. It was a scientific meeting, it was not aimed particularly at patients. It was aimed the scientists and clinicians. We felt that there may be scope to have perhaps the patient meeting next year or to keep it to separate, we didn't want it to be kind of okay for everybody. We wanted to focus on 1 aspect and we decided to focus on the science because that is where the cure was going to come.

DR. BRUCE BLOOM:

So, tell me what kind of response you got from Cathy and others around the world when they found out that an organization like yours was going to focus this much effort and funding on this small disease.

MR. JOHN SOLLY:

Everybody is being very positive, without the exception everybody has been very positive. It is one of the most gratifying things and, you know, you can be cynical and say that some of the research scientists, you know, it is an extra source of funding for them and I am sure that is part of it, but even so the patients, they are obviously very, very pleased, it is a wonderful thing for them.

DR. BRUCE BLOOM:

And one of the trust accomplishments was organizing the Myrovlytis Trust scientific thought leader workshop. Tell us what that workshop focused on and what do you hope it would accomplish?

MR. JOHN SOLLY:

So, that was focusing on renal gene therapy. So, this is an area where we were looking at something that is useful for our own disease, but also may be a little bit more widely applicable. So, we invited 15 of the leading renal gene therapists and other gene therapists and other kidney specialists from around the world 2 days meeting in London in August. We had about 15 guys and the idea was to identify the current barriers to progress and how we can overcome them and how we can move forward with renal gene therapy. The first clinical trial was with 14 years ago in 1994 and yet there is no approved drug, gene therapy drug, as it were for renal cell carcinoma for cancer of the kidneys. So, we are receiving some barriers and some challenges that people have been working on it over the last 14 years and we feel that the lot of very exciting research, particularly in the last couple of years, can be applied to the kidneys in terms of targeting particularly and so we feel that the time is right really for us to bring all that together and have a big push with renal gene therapy which will be useful for us, but also useful for the 12,000 Americans who die every year from renal cell carcinoma, 3500 Brits who die every year from renal cell carcinoma, so much more widely applicable, not just a narrow disease.

DR. BRUCE BLOOM:

And is just one of the reasons why working in the orphan disease phase make so much sense that any kind of breakthrough for Birt-Hogg-Dube or any other orphan disease is likely to have wider applicability.

MR. JOHN SOLLY:

I think that is one of the main reasons, yes. I mean obviously some of the, some of things you find out are going to be just to do with your gene and just to do with your disease, but a lot of it isn't and there was some big guts out there and some big prizes just waiting to be filled, waiting to be gun after.

DR. BRUCE BLOOM:

And you talked about kidney cancer, is that a component of Birt-Hogg-Dube and what are some of the other signs and symptoms of this disease?

MR. JOHN SOLLY:

There are 3 different symptoms, but the answer to your question is yes. About may be a quarter or a third of people develop kidney cancer, that is one of the symptoms. One of the others is growth or bumps on the skin, particularly the face and your torso and then the third one is growth on your lung and the risk of collapsed lung. So, the skin bumps, they don't kill you, but they are disfiguring and they can cause a lot of emotional distress I if you have a lot of them, particularly on your face and the collapsed lung typically doesn't kill you, you know, in the western world, but again it can cause problems and then obviously the kidney cancer can be fatal.

DR. BRUCE BLOOM:

And are all 3 of those big areas of the disease, are they related to the same genetic problems or are they separate genetic problems.

MR. JOHN SOLLY:

It is a very good question. They are all related to mutation of the main gene, so they are related, but we don't understand why there is variation in most why some guys only get the lung symptoms, some guys only get the kidney symptoms, and why people get more severe phenotype or less severe phenotype. So, that is something that we are looking out at the moment.

DR. BRUCE BLOOM:

And what resources are you willing to put towards BHD and what is your goal or your endpoint for this kind of research and treatment?

MR. JOHN SOLLY:

So, the resources were putting money that is our big thing we work with the best guys that we can get, you know, we make use of the best expertise that we can find anywhere in the world and we provide money to developing the therapies and treatments for BHD syndrome. At one point, if you have BHD syndrome, it doesn't matter, where there are drugs that can take away your skin lesions, say you don't have the disfigurement and the psychologic, you know, the emotional distress where there are drugs that can remove the risk of pneumothorax and where there are drugs of treatment which mean that the kidney cancer is not a problem. We are not going to cure it, but we can diminish it or we can put it out in the time, you known, in your life span that it becomes irrelevant, that is our endpoint.

DR. BRUCE BLOOM:

I would like to thank our guest, Mr. John Solly, charity manager of the UK's Myrovlytis Trust. For joining us to discuss the search for better treatments and cures of orphan diseases, especially Birt-Hogg-Dube syndrome.

I am your host, Dr. Bruce Bloom and you have been listening to a special report on global medicine from ReachMD, The Channel for Medical Professionals. Please visit our website at www.reachmd.com, which features our entire interview library available through on-demand pod casts or call us toll free with your comments and suggestions at 888-639-6157 and thank you for listening.

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