

Myotonic Dystrophy: A 40 Year Perspective

Transcript of a 2011 MDF Interview with Professor Sir Peter S. Harper.

Female Speaker:

Can you please give me your name?

Peter Harper:

Peter Harper.

Female Speaker:

And can you tell me what you do?

Peter Harper:

I'm a medical geneticist. That's to say, I...I trained in medicine, did internal medicine, but always had an interest in genetics, and then trained in that, mainly at Johns Hopkins with Dr. Victor McKusick, but I carried on with medical work as well, both in America and after I got back to Britain.

Female Speaker:

And tell me, I read your book, and I...I know that it's brought comfort to, you know, possibly thousands of families at this point. What drove you to write it?

Peter Harper:

The timing of the small book, which is Myotonic Dystrophy: The Facts, it was good timing because I had just completed my large textbook, and so I had really all the facts in my head, and that meant that I could sit down and just write the book, which I did and in fact, I wrote it over the course of a week. But you could say there was about 30 years of information and experience went into it. It was just a question of having some spare time to get it down on paper, and I had a weeks' vacation, and a very nice isolated place, which allowed me to do just that. So I wrote the first and main draft of the book, really straight out of my head, without any reference material around, and then polished it up from there.

Female Speaker:

What why did you think it was necessary?

Peter Harper:

From the very beginning, the thing that struck me about families with myotonic dystrophy has been that they've always said they need something simple and easy to understand that tells them about the condition. So I wrote it from the point of view of a patient, but also the family as a whole and because I'd had many years of experience looking after and talking with myotonic dystrophy families, first in America, then in Britain. Well, it was fairly natural writing it. I put myself in the position of one of them and...and wrote around that.

Female Speaker:

What do you think, you know, just looking at it, possibly you know sort of objectively even, what kind of impact do you think it had on them, or what have you been told maybe, or...or what do you personally think as...as someone who is in contact with these families? What kind of impact did it have on them to see this information written down so from their perspective? You know, really what kind what kind of impact did it have?

Peter Harper:

I think it's had a big impact, much more than I expected, because there really wasn't anything like it before my textbook on myotonic dystrophy, wasn't aimed at families. It was aimed at professionals, and so it really needed something that was written very specifically for the families, and I've been amazed, actually, how big an impact it's had, and I think it's partly due to the fact that the support groups have done a very good job promoting it. Um back in Britain, the leader of the myotonic dystrophy support group, Margaret Bowler, she...she won't let anybody through the door of a meeting until they've bought a copy of the book, and of course it helps. The royalties go back to the support group, and much the same has happened over here with people promoting it a lot. Uh I just like to think and hope that it deserves the promotion it's had; certainly it's had an impact.

Female Speaker:

It's incredible when you when you read it... it...it struck me immediately because I had been reading a lot of different research, dense research, about a really complicated condition, and I read it and I went, it reminded me of something that I had been given a few years back when my father-in-law was dying of cancer, and it was the hospice sort of this is what's going to happen, and what...what struck me was that cancer is from here to infinity. You know there's like a million different ways that this thing is going to present itself to families. Why is there such a commonality in the condition when they have myotonic dystrophy? That...that...do you know what I mean? Like there was universality

to what you were writing about, that it was like...this is what...this is what it's going to feel like to be going through this journey.

Peter Harper:

Yes it is. There are a lot of features in common and people can learn from the experience of another family. At the same time though, it's very different, and varying in different families, very variable, and some people may have serious problems with one aspect and none of another, and for a second patient, it might be the reverse. But there is a lot in common, and the main themes, which one can bring out, that I tried to bring out in that book. There's a huge amount in common, and I...I think a lot of people can identify with it and say, "Oh yes, that's just what happened in our family."

Female Speaker:

When you began your research there was no genetic test for myotonic dystrophy and anticipation wasn't accepted as a true biological phenomena. Tell us about the families you met and... and sort of what, possibly just...just briefly, what...what they went through before treatments. I think you called it the dark ages at one point.

Peter Harper:

Well certainly when I started, which was in America, I went up and down the whole of the Eastern part of the United States and that took about two years, and I must have seen several hundred families during that time, collecting information. Partly to try and localize the gene, but also it soon became clear one had to look at the condition as a whole, and look at it clinically, and also because of the distances I was seeing people in their homes and I think that was very valuable. But the common feature was that these people had had very little information. They may have had some medical information, but very often they didn't understand that. They didn't have any ongoing care and support, with very few exceptions, and they certainly didn't have anything simply explained, and I think one of the reasons why I got a big welcome from the families was that I was able to tell them things which were really helpful, and because they really had had almost nothing before and that made me realize that having simple, yet accurate information is pretty vital.

Female Speaker:

The...there are two things that you talked about; one was _____ (0:07:59.5) to maybe tell me this in your own words, you talked about it being such a variable disease. Like maybe you could just tell me that again, what did you mean, or possibly just...just tell me that again really. I'd...I'd love to have that.

Peter Harper:

Well, however you look at it, the condition is variable. It's variable in its age at onset. Some children have severe problems from before birth; others have nothing at all until old age. So it's variable in that. It's variable in severity of muscle involvement and weakness. Some people really have almost no muscle involvement. They may have other quite serious problems, and a...a number have serious muscle involvement, and then it's variable because just about every system of the body can be affected. Not just the muscles but the heart, the eye, hormonal problems, the brain, wide range of systems, and so you really do have to have a very broad and thorough approach.

Female Speaker:

Is it the most variable disease you've ever seen?

Peter Harper:

Yes, no doubt about that...and I...I think ...I think it's probably just about the most variable condition there is and so you really do have to be aware of all the different aspects, and that creates practical problems because very often the patients may first be seen by somebody who has no experience of muscle disease, and they don't _____ (0:09:44.5) that it's a generalized condition, and so part of the battle is making sure that all these different specialists in different areas are aware of the whole picture, not just of a bit of it.

Jackie:

Could you just say that again? In the...the first part of that by saying myotonic dystrophy is the most variable, variable, variable disease you've ever seen.

Peter Harper:

Sure.

Female Speaker:

That would be fantastic. So I guess my question was, is myotonic dystrophy the most variable disease?

Peter Harper:

Myotonic dystrophy probably is just about the most variable disease there is. Uh it's variable in terms of its age at onset. Some affected children have severe problems from before birth, and yet other patients may only develop problems in old age. It it's affected in terms of the severity of the muscle disease. Some people may have it badly, and others

may have hardly any muscle symptoms, even though they have other problems, and then in terms of the systems affected, almost any system in the body can be involved, whether it's heart, brain, gastrointestinal problems, hormonal problems, quite apart from the muscles. So it's very variable and part of the battle is to ensure that all of the different specialists don't just see their part of the picture, but are aware of the condition as a whole.

Female Speaker:

You also said at one point that about the previous 90 years of knowing about myotonic dystrophy and what we've learned in the last, I think you said the last decade, I was wondering if you could reiterate that for me, for us, today?

Peter Harper:

Up until the gene was isolated, really nobody had any clue as to what the basis of the condition was, and without knowing something about the basis of...of any disorder, you're really working in the dark as far as treatment goes. You don't really know what lines to pursue. As soon as the gene was isolated, that gave immediate advances on the genetic front. Genetic tests were very helpful in some families. It meant there were many people at risk who you could tell there is no risk, we've ruled it out, and other options for those who were carrying the gene.

So that was the starting point, and then very rapidly people began to make some progress in terms of how does the defect in the gene cause the condition, and that proved a lot more complicated than people thought initially, and this is what we're seeing progress in now, and there's real understanding of how the genetic changes cause the muscle and the other pathology, and now this is really leading to rational approaches to treatment. So one doesn't just have to base approaches to treatment on speculation anymore, you can look at very well documented changes which you know underlie the causing of the disease, and then try and attack those using a range of approaches, and that's just getting to the stage where it's going to impact on treatment.

Female Speaker:

So you would say in the last 10 years we're...we're moving, is it sort of, much rapidly, much more much quicker?

Peter Harper:

What I'd say is that the last 10 years have dug the foundations and produced the knowledge in terms of what causes the actual disease changes, and now as a result of that 10 years' of knowledge, it's becoming possible to start to devise likely therapies, first using models, and now it's getting to the point where I think proper trials of therapy in patients will be feasible in the very near future.

Female Speaker:

Out of all of the, you know, genetic disorders and diseases and conditions that you've looked at, and looking at it in comparison to what else is going on out there in the medical field, how would you compare myotonic dystrophy and the...the sort of leaps and bounds they're making in their in their treatments and therapies, is this one of the kind of bright stars out there as far as trying to that may give us hope, that we are we are getting smarter, we're figuring these things out. Do you know what I mean? Like is this something in the medical community is like wow, we...we're...we're figuring things out here with myotonic dystrophy?

Peter Harper:

Well, given the complexity of the condition, I would say yes. I...it...it is one where one can be very optimistic. Of course there are some genetic disorders where it's very much a matter of finding out what's missing and just putting it back, and that is not always simple, in fact very often not simple, but the conceptual level is something you can go for straight away. With myotonic dystrophy, it's a lot more complex because it's involving different proteins and very different types of medical problem. Even knowing whether you're doing any good or not is quite a complex problem because you don't just have to ask is a person getting stronger, or stopping getting weaker; you've got to look at the heart and all the other systems too. So that's complicated, but there really is progress. Compared with other conditions, well I would say there's progress in a number of major genetic disorders, and I'm thinking of those affecting muscle and brain, especially. But yes, I would say there's real progress and real reason to be optimistic, and very importantly real reasons for the patient community to be prepared and closely linked to the medical research community so that proper trials can be organized, because it will take a...a lot of patience and a lot of effort to make sure that these trials are done as they should be.

Female Speaker:

Do you think MDF is in a good position to help these patients organize and get be available for those kind of trials? Is MDF going to play a role in that? Are they are they the ones that could play the role in that as a facilitator?

Peter Harper:

Yes, I think I think very much so. I think Myotonic Dystrophy Foundation and its counterpart in the U.K. are in a good position, because they are in very close touch with the day to day problems of patients and families. There's also a role I think for broader associations. Now in America it's MDA, and there's a corresponding association in Britain, and in France the AFM. These are bodies that are much better set up on the fundraising for research on a big scale, but the more specific and focused societies, like Myotonic Dystrophy Foundation, are certainly well placed to link up the families with the research workers and even if they're only able to fund the seed _____ (0:18:35.4) for research projects then they can perhaps get things to the stage where bigger funding agencies can then take things on from there.

Male Speaker:

Well just that what would you... what would you say to encourage philanthropy towards one of those kind of organizations? What...what would be your encouragement to them to...to make them see it as reasonable?

Peter Harper:

I think if they came to a meeting, a family meeting like this or indeed a little bit of a scientific meeting, then that would show them the kind of things which are going on and show them the work and devotion and ...and the progress and the close links between families and research workers and I think that seeing that would make them realize that this is a very good place for them to donate, and also thinking in terms of the fact that it might not need a massive donation to see some results. Although a massive donation, I'm sure, would be extremely welcome and helpful, but I think to see for themselves what's going on is probably the best bet for any potential donor.

Female Speaker:

Can I just ask you what...and this is...this is pretty much it, what...what...what inspires you about the people that you've met over this the course of your career?

Peter Harper:

Well, in working with the families of the many years, I'm inspired by the resilience and persistence, and toughness of the families having to cope with very difficult problems, and also by the fact that they are really so receptive to having anything that could help, whether it's information or progress in research. I think they've often been dealt a...a very difficult hand of cards in life and have a lot of problems, and they realize that not all these problems are going to go away. But the fact that there is this work going on and such outstanding scientists contributing, I think gives them hope and gives everybody hope, and certainly one consequence of this has been for people working in the field that many of the people who've started working on myotonic dystrophy, and I'm thinking perhaps of the lab scientists especially, they stay very loyal to the disorder and rather than jump off and start working on something else and give up working on myotonic dystrophy, they've stuck with the condition over many years and so there is a real community of research workers alongside the family community.

Female Speaker:

That's lovely. Thank you. What do you do when you're not going to myotonic dystrophy conferences and clinical geneticists, what do you do outside of...what are your passions outside of that?

Peter Harper:

Well, family keep me pretty busy actually, grandchildren and still children, and so there's a lot...a lot to do with them; hill walking, we've got beautiful mountains in Wales, so I do a lot of hill walking; music, my wife and I enjoy singing, early music especially; bird watching, I usually bring my binoculars to any meeting I'm going to, including this one. There isn't a huge amount of spare time, even though I'm now meant to be retired, but that's the way it is.

Female Speaker:

Thank you so much. I appreciate it.

Jackie

Yeah, that was beautiful Peter. Thank you.