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VIDEO INTERVIEW TRANSCRIPT

Harper, Peter: transcript of a video interview (06-Jun-2015)

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Note: Video interviews are conducted following standard oral history methodology, and have received ethical approval (reference QMREC 0642). Video interview transcripts are edited only for clarity and factual accuracy. Related material has been deposited in the Wellcome Library.

Harper, Peter: transcript of a video interview (06-Jun-2015)*

Biography: Professor Peter Harper (b. 1939) is Emeritus Professor of Human Genetics at Cardiff University. He has been closely involved with the identification of the genes underlying Huntington's disease and muscular dystrophies, and with their application to predictive genetic testing. He has also been responsible for the development of a general medical genetics service for Wales. He is a Consultant to the 'Makers of Modern Biomedicine Project' for the History of Modern Biomedicine Research Group, Queen Mary University of London.

[1]. BECOMING A GENETICIST, THE INFLUENCE OF CYRIL CLARKE & VICTOR MCKUSICK

I think I'd been interested in genetics for a very long time, scientifically, and I'd always wanted then to try and combine it with medicine. At the time I was in Medical School, there didn't really look much chance of doing that, but then I found that things were becoming possible, and I was lucky enough to be able to work first with Cyril Clarke in Liverpool, who was a practising physician who very much was a pioneer in early medical genetics. And then from there I went on to Baltimore to work with Victor McKusick, and that really gave me the skills with which I was able to come back and practise as a clinical geneticist. But at the same time I'd trained in adult hospital medicine, internal medicine, and so I kept that up as well, because at the time I began clinical genetics, it was far from clear whether there would be any long-term jobs in the field. And also before my medical degree was finished, I'd actually been at Oxford where I worked with basic geneticists. When I was at Oxford, I was able to spend a lot of time in the Zoology Department where there were very good basic geneticists in population genetics working. And the scientific aspects really enthused me very greatly, and so I wanted to carry on something scientific as well as something medical, and genetics gave the opportunity to combine the two.

[2]. MYOTONIC DYSTROPHY & HUNTINGTON'S DISEASE

Well, in terms of which contribution I may have made, would I feel was/has been the most valuable, or at least valuable in my own view, there have been two diseases which I've worked with for well over 30 years, more like 40 in fact, which have kind of interacted. One of them is myotonic dystrophy, one of the muscular dystrophies. The other has been Huntington's disease, a serious brain degeneration. And I started off by doing my Thesis on myotonic dystrophy. That work began in 1969, and it's never really stopped. And in terms of Huntington's disease, my work began almost immediately after I came back to Britain in 1971, and again it's gone on pretty continuously since then.

What I had no idea about when I started was that these two disorders, which at a clinical level don't seem very similar, one a brain disease, the other a muscle disease, proved to be due to the same genetic mechanism. And I feel extraordinarily fortunate to have been able to follow the story and the development of these two diseases right through from the clinical description and the study in families through to the mapping of the genes involved, and then to the discovery, which I and my colleagues were very much involved with in both, of the basic molecular defect. And even more remarkably, this turned out to be the same type of molecular defect in both, a trinucleotide repeat of unstable DNA. And that the two had the same mechanism so the two have interacted. And then from there I was able to apply the new discoveries in terms of the gene to helping families in terms of prediction of those who did and didn't have the gene, and now that work has

* Interview conducted by Professor Tilli Tansey, for the History of Modern Biomedicine Research Group, 06 June 2015, in Glasgow. Transcribed by Mrs Debra Gee, and edited by Professor Tilli Tansey and Mr Alan Yabsley.

gone on with others to the stage where trials for therapy are in progress. So I feel very fortunate to have seen this progress from beginning to end.

[3]. *PRACTICAL GENETIC COUNSELLING*

Apart from any of the research and actual practical and clinical work which I've been involved with, I would perhaps rank my book *Practical Genetic Counselling* as being a valuable contribution, mainly because it's spread around the world, and for more than 30 years seems to have been the book most used by people giving genetic counselling. And that it's really had an influence in a number of countries where really genetics was very undeveloped, like Russia and China and many other countries. And I have always been amazed that it's continued to be useful for over 30 years, and so I feel that has been a contribution, and I've enjoyed meeting the very many people who have told me that they found it useful in their work.

[4]. DID ANYTHING IN PARTICULAR GO WRONG?

You asked me what the greatest problem or mistake I've encountered, or made, has been. Nothing actually stands out individually, and I don't regret having worked in the places I have worked, first in America and then very largely in Cardiff, more recently over a long time. Yes, a few missed opportunities, but nothing sticks in my mind as being particularly problematic, more a question perhaps that if I had done something, more might have come out of it.

[5]. CHANGES IN MEDICAL GENETICS

You asked how had I seen things change over my career? A huge amount has changed. If I stop to think about it, when I began in medical genetics, well, first of all medical genetics hardly existed as a specific field. It's just beginning to. And what one could do was very limited. One could spend time with families, give good genetic counselling, listen to people, and help quite a lot in those general ways. But when you got down to the question of what specific things could you actually do to help with this genetic disorder, there was virtually nothing. And then I've seen it change so that now one cannot only offer genetic counselling and give good, accurate ideas about risk, one can very often now tell whether somebody is or isn't carrying a harmful gene, equally predict whether or not are they likely to develop a disorder. And then, now, there are beginnings increasingly of effective treatment as well as good management. So there's a lot basically one can offer which just was not possible before.

[6]. WHAT DO YOU PREDICT FOR THE FUTURE OF MEDICAL GENETICS?

I'm not going to try and predict what will happen in the next 20 years. I hope that there will be sensible advances, advances which are driven by people's needs and wishes, rather than purely by technology or industry or any of the other things which seem to be rather powerful drivers at present. Whether that will be the case remains to be seen, but I would hope that the sound principles that are present now in medical genetics practice will progressively spread through all clinical specialties, and that people will become more educated. And that those involved in promoting the field do so in a responsible and not an exaggerated way,

[END OF TRANSCRIPT]

Further related resources:

1. Christie D A, Tansey E M (eds) (2003) *Genetic Testing*. Wellcome Witnesses to Twentieth Century Medicine, vol. 17. London: Wellcome Trust Centre for the History of Medicine at UCL.
2. Jones E M, Tansey E M (eds) (2013) *Clinical Cancer Genetics: Polyposis and Familial Colorectal Cancer c.1975-c.2010*. Wellcome Witnesses to Contemporary Medicine, vol. 46. London: Queen Mary, University of London.
3. Jones E M, Tansey E M (eds) (2014) *Clinical Molecular Genetics in the UK c.1975-c.2000*. Wellcome Witnesses to Contemporary Medicine, vol. 48. London: Queen Mary, University of London.

4. Jones E M, Tansey E M (eds) (2015) *Human Gene Mapping Workshops c.1973-c.1991.* Wellcome Witnesses to Contemporary Medicine, vol. 54. London: Queen Mary University of London.
5. Reynolds L A, Tansey E M (eds) (2010) *Clinical Genetics in Britain: Origins and Development.* Wellcome Witnesses to Twentieth Century Medicine, vol. 39. London: Wellcome Trust Centre for the History of Medicine at UCL.
6. Zallen D T, Christie D A, Tansey E M (eds) (2004) *The Rhesus Factor and Disease Prevention.* Wellcome Witnesses to Twentieth Century Medicine, vol. 22. London: Wellcome Trust Centre for the History of Medicine at UCL.