1 Introduction

I've got a young patient with a family history of Huntington's Disease who wants to have a test to see whether she is going to be affected by the disease as she gets older. She is worried because she knows that her paternal grandmother has it. During counselling my patient disclosed that she is an identical twin. She says that her twin sister is not aware of the family history and says that she does not want her sister to know because she doesn't think that she could cope with this knowledge, particularly because the disease is untreatable. When I told her that I was reluctant to do the test on her without discussing it with her sister – because the fact that they are identical twins means that the test would also be a test on her twin – she said that she didn't want her sister to be involved. To reassure me, she promised that whatever the test result she would not disclose this. The other problem I have got is that in addition to being a test on her twin the test, if positive, would also be test on her father who, she says, also does not know that she has come in for testing. I've tried to encourage her to talk to her sister and father about the test but she says that she's not able to do this. I feel that I have got a duty of care to my patient, but I'm also worried about her sister and father even though I have never met them.

A woman who is trying to get pregnant was recently referred to me because she is a member of a family with a history of a serious X-linked disorder. Her cousin is affected. My patient is interested in using preimplantation genetic diagnosis (PGD) to ensure that she doesn't have an affected baby. We didn't have any information about the mutation in the family so we had to carry out a linkage study to assess her risk. This meant looking at samples from a number of other family members. We were talking about a generation of people in their fifties and over. They were all very happy to provide blood samples. When we tested the samples, however, it became clear that one of them had no genetic markers in common with anyone else in the family, suggesting possible adoption, and another showed non-paternity. These results mean that my patient is not at risk of the condition. What should we do? These events were a long time in the past. Is it acceptable to simply tell my patient that there is no risk of the disease in this part of the family or does she need to know that she is adopted?

> The person concerned is now deceased so it is not possible to go back to them to ask for their consent to share this information. So, we are just left with this and not knowing how to deal with it.

As these two stories show, genetic testing can sometimes tell us more about ourselves and about our relationships with the people around us than we expect, and perhaps, in some cases, more than we really want to know. The stories also show that genetic testing can sometimes end up telling us rather *less* than we might have wanted to know because of the ways that decisions about access to genetic testing and the distribution of the results of such testing are mediated by the complexities of the relationships we have with those around us, such as our family members, and by the policies and practices of the health care institutions and genetics professionals who are – even in the era of the Internet and direct-to-consumer genetic testing – most often the gatekeepers of such testing and advice. What we come to know about ourselves and about our relationships through genetic testing depends to an important degree upon the decisions and values of our relatives and the ways in which they conceive of their relationships with us, and upon the views of the genetics professionals who offer or refuse to offer such tests, and the guidelines by which their practice is informed.

What these stories also reveal is that there is a *co-productive* relationship between genetic testing and decisions about whether or not to make it available, and the nature of our relationships with relatives and reproductive partners. Our knowledge about and understanding of our relationships with others, and consequently the very nature of those relationships themselves, has the potential to be shaped in significant ways by practice in genetics. This is increasingly likely to be the case in the future as genetics inevitably becomes more a part of mainstream medicine and as genetic testing in one form or another becomes a more pervasive feature of life outside the clinic.[1] In both of the stories above, for example, genetic testing and the sharing of the information resulting from such tests (or the refusal to do so) has the potential to radically alter the relationships between those who have been or wish to be tested and others in their families. Just as these stories show that genetic testing can create new ways – or maintain old ways – of being related, they also show that such influences can work in the opposite direction – that is, they help us to see how the particularities and complexities of the ways in which we are related to those around us have the potential to make a difference to the development, use, availability and

[1] PFG Foundation, *Genetics and Mainstream Medicine: Service Development and Integration* (Cambridge: PFG Foundation, 2011).

understanding of genetic technologies and of the genetic information produced by them. A good example of this is the way in which the twin's understanding of the nature of her relationships with her sister and her father in the first story above informs her decision not to include them in her deliberations about testing. Another is the way in which the genetics professional in the second case worries – and the very fact that she does worry – about the implications of information about adoption and 'non-paternity' for her patient's wellbeing. Seen from this angle, what these stories serve to highlight is that just as genetics has the potential to change the nature of our relationships with the people around us, the ways in which we are related also have the potential to change the practice of genetics. The day-to-day practice of genetics, the translation of new genetic technologies, and the transformation of human relationships are interwoven in complex and dynamic ways.

Because of their significance for people's lives and for the kinds of relationships they have with the people around them, many of the questions posed by the use of genetics inevitably have a moral or ethical aspect. Sometimes these are questions which people struggle with as patients or as family members. In the first story above, for example, the twin wrestles with the tensions between her desire for the Huntington's test, her recognition that the results of the test will be of relevance to her sister, and her concern about the potential harms that might happen to her sister if the results were to turn out to be positive. Does she – she wonders – have an obligation to talk to her sister about the test? If so, might this nevertheless be outweighed by concerns about her sister's wellbeing or by her own right to gain access to health care without the requirement to seek permission from or take on responsibility for, her relatives? In addition to the moral difficulties experienced by patients and their families, the potential uses of genetics and of the information generated by genetic tests can also present difficult ethical questions for genetics professionals – for the counsellors, doctors and laboratory staff who work in clinical genetics. In many cases, such as in the first story above, the ethical challenges are to some extent shared by the patient and the genetics professional. Here, in addition to the worries confronting the patient herself, the genetics professional is also faced with a tension between her duty to do the best for her patient and her sense that she has responsibilities for the wider family. There are also some situations in which genetics professionals face difficult ethical challenges of which patients may be completely unaware. In the second story, for example, to ask the patient about whether she would want to know that she was adopted would probably be to reveal to her that she is. The question of whether or not to disclose this information

is something the genetics professional must wrestle with alone, or with the help of her colleagues.

In the stories above, genetics professionals are called upon to make difficult moral decisions with the potential to impact upon the social as well as the medical lives of their patients and with significant implications for the distribution and use of genetic technologies and information. This reflects the key role genetics professionals play at the interface between technology, science, health care practice and contemporary human relations. The moral complexities of this role are striking both in their particularities and in their scope. For not only does the genetics professional deal with individually difficult cases with morally significant implications for the lives of specific patients and their families, the fact that she and her colleagues encounter such problems on a day-to-day basis, as a profession, means that in her practice the genetics professional plays an important role both in the development and implementation of genetic technologies and in the forming, sustaining and transformation of human relationships more broadly.

There has been extensive discussion of the implications of genetics and of genomics in the media and in the academic bioethics and social science literature. Much of this has focused on the implications of developments in genetics for patients and their families. These debates are familiar, if not over-familiar. Despite this, there is very little on the relationships between developments in genetics and the contexts into which they are being or may be translated. This is the domain of the genetics professional: a dynamic and complex space of moral commitments and material practices in which genetic tests are carried out, and their implications unravelled, in real and material ways with patients and their families over time. This book aims to provide a rich account of the moral world of the contemporary genetics professional at a key moment in its development.

The work informing this book has its origins in three conversations I had with clinical geneticists in 1999 and 2000. The first of these was with Anneke Lucassen at Oxford in the summer of 1999 shortly after I had started a new post at the Ethox Centre.[2] Anneke was a consultant geneticist at the Regional Genetics Unit in Oxford at that time[3] and our conversation started because she was attending a 'masterclass' in medical ethics that was being run by the Ethox Centre and on which I was a tutor. The masterclass format combined seminars on various topics in

[2] The Ethox Centre is a multidisciplinary bioethics research centre in the University of Oxford (www.ethox.ox.ac.uk).
[3] Anneke is now Professor of Clinical Genetics at Southampton University.

medical ethics with an opportunity for participants to work on a piece of ethics writing with one-to-one tutorial support. Anneke, who had come to the masterclass because of her interest in medical ethics, had brought with her a couple of cases from her own practice which she had found ethically challenging and which she wanted to write up. In these cases, genetic tests had identified, as an 'incidental finding', misattributed paternity. As we worked on developing the paper over the course of the week, Anneke and I had a number of fascinating discussions about the ethical aspects of day-to-day practice in clinical genetics.[4] These were of interest to me both in themselves and for two further reasons. First, I had a long-standing fascination with the importance of relationships in ethics and, in particular, with the relationships between individuals and families[5] and the cases that Anneke and I were discussing seemed to offer new and productive ways of thinking about this issue. Second, and more prosaically, my new position at Oxford required me to spend the equivalent of one day a week providing clinical ethics support to health professionals in the local hospital. As our conversations developed, the regional clinical genetics service – where Anneke worked – began to emerge as an ideal setting in which to begin this work.[6] Anneke discussed this possibility with the clinical team, who proposed that I give a presentation at one of their weekly case conferences. This was to be the second conversation I had that would lead to the writing of this book.

Over the course of the next four weeks, in order to prepare for the talk, I built on my earlier work on genetics[7] by reading as much as I could of the bioethics literature and by talking to people, including Anneke, who I knew had worked in this area. My presentation to the clinical genetics team would be the first time I had met health professionals since my arrival in Oxford and I wanted to make sure that I did a good job. I wanted the presentation to be interesting, to emphasise the importance of the ethical dimensions of genetics and, hopefully, to convince the

[4] This paper was published in *The Lancet* as: A. Lucassen and M. Parker 'Revealing false paternity: some ethical considerations' (2001) 357 *The Lancet*, 1033–1035.

[5] This interest was in large part formed by my work, over the course of the earlier decade, with young homeless teenagers and families in conflict. See for example: M. Parker, 'Children who run: ethics and homelessness', in B. Almond, *Introducing Applied Ethics* (Oxford: Blackwell, 1995), pp. 58–70.

[6] I later provided clinical ethics support in a number of other settings in the hospital, including adult and neonatal intensive care and emergency medicine.

[7] M. Parker and D. Dickenson, *The Cambridge Medical Ethics Workbook* (Cambridge University Press, 2001); M. Parker, R. Williamson and J. Savulescu, *Ethical Issues in Genetics Research: An Introduction for Members of Australian Human Research Ethics Committees* (Melbourne: Cooperative Research Centre for the Discovery of Genes for Common Human Diseases, 2003).

genetics team that they might benefit from a regular meeting with an 'ethicist'. On the morning of the presentation, one of the senior consultants from the genetics unit arrived at my office to accompany me on the short walk over to the clinic. As we set off across the car park on a lovely sunny day, she told me how much the team were looking forward to the session. She said that ethical issues were, increasingly, a major part of their practice and that the geneticists worried about this a lot. This made me feel a bit more confident about my talk. She then mentioned, as an aside, that the team had once before invited someone to 'give a talk on ethics' and 'that', she added portentously, 'was exactly what he had done'. Without realising or intending its impact on me, she finished by saying that they had never invited him back.

It was clear to me that I had seriously misjudged the nature of the event but I had no idea what the alternatives might be. As we continued our walk, I tried to decide what to do. Should I give my prepared talk as planned, making it as interesting as possible but accepting from the outset that this was not likely to lead to an invitation to return? Or should I try to come up with a different kind of improvised approach in the five minutes available to me? In the end, I decided that the only sensible thing to do was to introduce myself, say something about the work I had been doing with Anneke on the difficulties of dealing with unexpected information, and then to ask them what they themselves considered to be the most important ethical issues in their everyday practice.

We arrived at the meeting a few minutes early, as a clinical case discussion involving a twin who had requested a test for Huntington's Disease was coming to a close. The meeting room was no larger than the average living room. Its walls were covered by shelves of cardboard boxes which contained, I assumed, medical records. At its centre was a large table surrounded by approximately ten people, and behind these, seated against the walls, were about another twenty. As I listened to the discussion of the case, it became clear that the people in the room came from a variety of different backgrounds: genetic counsellors, nurses, laboratory staff, consultants and registrars. There was even a medical anthropologist, sitting in the corner, quietly taking notes. When it came to my turn to speak I introduced myself, mentioned my work with Anneke, and went on to say something – I can't remember what – about the ethical aspects of the case they had just been discussing in their case conference. I concluded by enquiring whether these kinds of issues arose very often in their practice. In this way, my second conversation began.

After asking this question, an hour of heated discussion ensued in which one person after another described difficult situations they had been involved in, only to be followed by another who agreed on

the importance of the issue, but who took a different view about what should be done and who could support their position with another case from their own experience. Although I had said no more than three or four sentences during this discussion, it was agreed at the end of the session that the team's agenda would include a regular slot dedicated to the discussion of ethics, that I would facilitate this, and that someone in the genetics unit would work with the team and with me to identify cases for each monthly meeting. The first of these meetings took place in 1999 and they have taken place more or less every month since. The approach that I had unintentionally adopted, in which I facilitated the discussion rather than led it, had worked very well and the event served as a model for much of the work I have done in bioethics ever since.

The third and final conversation, and perhaps the most significant with regard to the writing of this book, also took place in Oxford. In Spring 2001, Angus Clarke, Tara Clancy, Anneke Lucassen and I obtained a grant to bring together a small number of genetics professionals from across the United Kingdom to talk about the ethical issues arising in clinical genetics.[8] With the consent of those present, this meeting was taped and transcribed. When I looked back at the transcript as I prepared to write this introduction, I was struck by the similarities between the discussion at that workshop and the one in the clinical genetics unit which I have described above. The plan had been that we would introduce the aims of the meeting at the start of the day and then follow an agenda organised around a number of key issues which Angus, Anneke, Tara and I had identified in advance as important. What happened was very different. In my role as the facilitator of the meeting, I began by suggesting that we introduce ourselves and say something about the kinds of ethical issues that we thought were important in genetics practice. Although there were no more than twenty people in the room, it took the entire length of the meeting (about four hours) to complete these 'introductions'. As each person set out what they thought were the interesting issues, and gave cases as examples, others joined in and gave their own, sometimes contrasting, examples, going on to provide richer and richer accounts of the nature of the problems they were facing and the different ways in which these problems might arise.

We never got on to the agenda. We did, however, allocate time at the end of the day to thinking about how some of these issues might be addressed. One of the most important ideas to come out of that discussion was a suggestion that a regular national ethics forum should be

[8] This symposium was funded by a grant from the Wellcome Trust (SYM/3/99).

established at which anyone working in clinical genetics could present and discuss cases with the aim of sharing experiences and potentially working towards models of good practice. The idea for the format had its origins in a national forum which had already been successful in relation to the clinical aspects of practice – the Dysmorphology Club.[9] Angus Clarke, Tara Clancy, Fiona Douglas, Anneke Lucassen and I agreed to take on the task of organising the forum, and the first meeting of what came to be known as the 'Genethics Club' took place later in the same year.

Our aim in establishing the Genethics Club was to provide a regular forum at which anyone working in clinical genetics in the United Kingdom could present and discuss ethical issues arising in their practice. We knew from our experience and from the discussions at the meeting described above that there was significant diversity of experience across the different clinical genetics units in the United Kingdom and our idea was that the Genethics Club should be both a space in which people could feel comfortable discussing the difficulties they faced and an opportunity to work towards shared models of good practice through its encouragement of communication between regional units.

At the time of writing, the Genethics Club has met thirty times, being attended on average by about thirty people (ranging from a low of twenty at one meeting to sixty-five at another). Because the meetings are always attended by a significant number of new people, they begin with a welcome and an introduction from either myself or Anneke about the origins and purpose of the Genethics Club. As part of this introduction, one of us – usually me – outlines the Genethics Club's approach to confidentiality. The policy is straightforward. Those who present cases are asked to ensure that they are anonymised. This is important because although, if the meeting is to function effectively, the issues and cases discussed need to be real, the meetings are usually attended by people such as myself who are either not directly involved in the care of the patients or are not health professionals at all. Given the nature of clinical genetics in a relatively small country, there is also sometimes the possibility that the families discussed may be recognisable to clinicians from other regional centres. For this reason, the general rule we have adopted is that when genetics professionals go back to their clinical teams and discuss the issues that were raised at the Genethics Club, this should be done in a way which ensures that as far as possible

[9] For more information, see www.clingensoc.org/Dysmo/index.htm (accessed 1 August 2011).

the families are not recognisable to their colleagues. Following these introductory remarks, the time available for the day is divided up to ensure there is space available for discussion of all the cases that have been brought along. On average about ten formal case presentations are made at each meeting.

It is important to point out that, as the story above of the origin and development of the Genethics Club illustrates, this is very much a book about ethics and genetics *in the Genethics Club*, and as encountered by the genetics professionals who have participated in the Genethics Club, rather than a book about 'genetics'. Nevertheless, in providing a space – one might call it a laboratory – for ethics, the Genethics Club has arguably *changed* genetics (genetics as practice) in significant respects.[10] The story above also serves to highlight the significance of my own role in, and to some extent outside of, the Genethics Club. When asked, I have sometimes described this role as follows:

> My role in the Genethics Club is essentially concerned with facilitating the discussion in a way which doesn't interfere too much but is attentive to the morally significant features of the cases presented and of the subsequent discussion. I organise the space and lunches, etc., and, inevitably, I've done a certain amount of behind-the-scenes work to encourage people to bring cases along. But primarily my role has been to ensure that anyone who wants to speak has the opportunity to do so and that as many voices as possible are heard.

Essentially, this has been my role. But, particularly as I have gained experience over time, my participation in the discussion has been more active than this. For example, I have sometimes used my experience of involvement in the regional genetics service in Oxford, my reading of the literature and my experience of previous Genethics Club meetings, to challenge positions taken when it has seemed to me that a too-easy consensus has been reached, to introduce issues that I think have been neglected, or to remind those present of the views taken in relation to similar cases in previous meetings. These aspects of my role have been made possible at least in part by the fact that, since the first meeting of the Genethics Club, I have kept reasonably detailed notes of the cases presented and, as much as has been compatible with my role as facilitator, set down the range of positions taken in the subsequent discussion. On four or five occasions, the group has agreed to record the Genethics Club meetings as a way of preserving the debate, identifying possible topics for invited plenary presentations and capturing enduring themes

[10] I develop this point further in Chapter 6.

and issues for further, future discussion. In the chapters that follow, I have included short extracts from these notes and transcripts.[11]

But, who are the 'genetics professionals' who attend the Genethics Club? Clinical genetics services in the United Kingdom are provided through twenty-three Regional Genetics Centres, which can cover very large populations – the centre in Manchester, for example, serves a population of about five million people.[12] Services are provided through the centres themselves, through outreach clinics in District General Hospitals or on hospital wards, and – sometimes – through visits to patients' homes. The majority of genetics professionals who attend the Genethics Club work in these regional centres. Like those in genetics services in many other countries, the staff members in Regional Genetics Centres fall, broadly speaking, into three main categories. *Clinical Geneticists* are medical doctors (physicians) who have undergone specialty training in genetics after general medical training (or, sometimes after specialty training in a related field such as paediatrics). This means that some of the clinical geneticists in the regional genetics centre will be fully trained consultants and others will be junior doctors in training. The role of the clinical geneticist is diagnosis, risk estimation and the management and support of patients and families affected by or at risk of inherited conditions. *Genetic Counsellors*, who are also sometimes referred to as specialist genetic nurses, genetic associates or genetic co-workers, will either have completed a Masters in Genetic Counselling or have undergone several years postgraduate professional training in counselling and some training in genetics and will also be a qualified health professional – most often a nurse. The role of the genetic counsellor is to help individual patients and their families to understand information about the nature of the genetic condition, appreciate the inheritance pattern and risk of recurrence, understand the options available, make decisions appropriate to their personal and family situation, and make the best possible adjustment to the disorder or risk.[13] The clinical geneticists and genetic counsellors at the twenty-three regional clinical genetics centres provide services related to a range of types of inherited disorders including neuromuscular conditions, eye diseases, neuropsychiatry, adult and paediatric endocrinology, cardiac, deafness, dysmorphology, infertility, and a range of other adult and

[11] I return to discussion of my role in the Genethics Club, and to my analysis of the Genethics Club as an 'ethico-ethnographic' research technique in Chapter 7.
[12] PFG Foundation, *Genetics and Mainstream Medicine*.
[13] Association of Genetic Nurses and Counsellors website www.agnc.org.uk/howtobecomeaGC.htm (accessed 24 July 2011).