

Anna Middleton - Genetics and ethics

Anna Middleton, Wellcome Trust Sanger Institute

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Kat - You're listening to the Naked genetics podcast with me, Dr Kat Arney. Still to come, our gene of the month is forever young. But now it's time to return to our main topic of mass genome sequencing, focusing on the 100,000 Genomes Project. Although many people - particularly in the scientific and rare diseases communities - are keen to see this kind of research happen, there are significant concerns around ethics, data, privacy and more. Genetic counsellor and social scientist Anna Middleton, based at the Wellcome Trust Sanger Institute outside Cambridge, is investigating how to bring forward more informed discussions about the ethics of genetics. I started by asking her to explain what some of the key issues are.

Anna - The key ethical issues really relate to expectations, managing expectations properly and an understanding of what can be delivered and what can't be delivered. With the 100,000 Genomes Project is the opportunity for three groups of patients - so cancer, rare diseases, and infectious diseases - to access sequencing technology. By doing that, they may gain answers to a diagnosis, to a range of treatments that are possible for them, or they may gain absolutely nothing. The promise of the delivery of answers is really bandied around but actually, may not offer anything useful at all. I think patients seem to go in and engage with it with their eyes open. As in any genetic test, if you get results related to anything, the results are often relevant to not only yourself but also to your immediately family as well - so your siblings, your parents, your children. So, patients may go for a test for one thing and come out with answers relevant to their whole family. And so, that raises ethical dilemmas for different people and so it sort of really brings to the heart of this that consent is the most important thing, and that people really understand what it is that they're signing up for, they understand what the options are in terms of results, and they go into it with their eyes open.

Kat - As I see it, one of the challenges with this kind of technology is we're moving towards being able to do whole genome analysis, we're moving towards an era of the thousand dollar genome where you can have everything tested for regardless of whether it's relevant or not. Is it that the technology has raced ahead of our ability as a society to understand this and to cope with it and to think about it?

Anna - Well, the technology has raced ahead and just because it's easy and relatively cheap to look at 20,000 genes, that does raise the question of, well, should we? But in terms of healthcare, policy's really been shaped around answering those specific clinical questions. So, in a healthcare setting, really,

you're not going to get an analysis of 20,000 genes in one go and have your whole genome delivered to you on a plate. Really, what they're going to be doing is just fine-tuning it to answer those specific clinical questions. So, I think it's a little bit misleading to be thinking that there's going to be an absolute deluge of data that people can't cope with. That's probably not what we're dealing with say, in the NHS.

Kat - Obviously, there are scientific research going on into sequencing, but there's also research going on into the ethics and the public understanding of this. Tell me a bit about the work that you're doing to try and understand what people think, what people understand, and to shape the policy in how things do go forward?

Anna - Yes. There's lots of assumptions about how people might want to use a technology and what they'd want from it, but very little empirical data that actually asked people what they want from it. And so, I designed the GenomEthics study which is a very large scale survey to try and get people to engage with this. The way we did that was to create ten short films that sit in the survey and they describe the ethical issues raised by genomics. The films really ended up being a great hook to get people interested in the topic. And they helped the survey to go viral and we had 7,000 responses from 75 different countries. And that's given us a really large data set to try and understand what people want from this and what they think about it. The overwhelming response is that people are really excited by this. So, they like the idea of knowing what's in their genes. They feel connected to it, they're inspired, they're interested. So, that was a really nice finding. Then we asked people, "What would you actually want to know? Would you like to know about genes linked to serious life-threatening conditions that can be prevented? Would you like to know about serious life-threatening conditions that can't be prevented? Would you like to know about information relevant to your children? Would you like to know about information relevant for when you're older later in life? The things that aren't relevant now, but maybe in the future." As we went through different categories of information, as the categories became less serious, or less treatable, people were less interested.

So, mostly across the board, irrespective as well, they're most in the world were interested in data relating to serious life-threatening conditions where some action could be taken to protect the person against the condition. They were least interested in uncertain data or receiving raw sequence data. But actually, what was so fascinating was that even for uncertain data and even for very low risk data relating to health conditions that have a very low chance to actually happening. People were still saying, "Yes. If you know it, I'd like to know it too if this is interesting and useful for me."

Kat - How would you like to see things moving forward in this area – the 100,000 Genomes Project, Genomics England is launching, they're starting to gather people. There's large scale sequencing projects going on all over

the world. How would you like to see things moving forward in our public understanding and also, in terms of some of these ethical issues?

Anna - To me, I feel it's incredibly important to have a very robust and sensitively delivered public engagement exercise. It's a real opportunity missed if people don't understand what this technology can offer and are possibly fearful of it or just confused by it. We really need to explain what it can offer, explain what it can do and help people to have a populist conversation about it. One of the next projects that I'm working on is looking at really how to turn genomics from something that's currently quite anti-social into something that's quite social. I mean, how do you start a conversation about it with people who have no clue what it means? And so, I'm looking at working with people from the advertising industry to try and get really simple messages about what this can actually offer out into the public and to get people talking about it. And then once people are talking about it then they can choose to engage with this or not. But at the moment, we're at this situation where the science is moving so fast and it's going to be implemented in clinics so quickly, but the public aren't really there with it. We need to start having national and international conversations about what it could actually do.

Kat - I'm a science journalist, I'm a science broadcaster. I've spent a long time trying to get across messages about how genes work, what's in our genes, and what they can do for us. Obviously, people who are listening to the Naked Genetics have some kind of interest in genetics. Do you have any tips maybe for me or for my listeners about what we can do, how we can get engaged in those conversations, or where we can go for more information?

Anna - How to actually start a conversation about this is incredibly difficult. We don't yet know really what the hooks are to start that conversation. You know, is it something about, we're all related to each other? Is it something about our identity? Is it even something as simple as – well Angelina Jolie has had a test for something genetic and starting the conversation that way? I mean, how to actually bring this into conversation will be really, really difficult and that's something I'm currently trying to research. We need to understand from a social sciences perspective what actually touches people, how it's meaningful for them, and how it connects them. I mean, my mission is to do more work with film and to try and discover the metaphors that people like to use to explain genetics and genomics. If I can create a series of films that have no spinning double helixes in there, then I'll feel I've made a contribution.

Kat - Anna Middleton from the Sanger Institute, and if you'd like to have a go at her Genomethics survey yourself, it's online at <http://genomethicsblog.org/>