# ‘Genetic Medicine: What are the ethical issues?’ Study Day



Saturday 16 January 2016

10.00-16:00

Lecture Theatre C, Building 65

Avenue Campus

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| **10:00** | Arrival and coffee  |
| **10:15** | Welcome and introduction to the day*Gillian Crawford, Principal Genetic Counsellor/Clinical Research Fellow* |
| **10:30** | New technologies in genomics: “the potential and the hype”*Anneke Lucassen, Professor/Consultant in Clinical Genetics* |
| **11:30** | Coffee Break  |
| **11:50** | Whose eyes on your genome? Confidentiality and information-sharing in genomic medicine*Sandi Dheensa, Post-doctoral Research Fellow* |
| **12:30** | How sci-art helps us to engage with the issues*Halina Mikolajek, Post-doctoral Research Fellow, Centre for Biological Sciences* |
| **12:40** | Lunch (Cafe area) |
| **13:40** | World café *Angela Fenwick, Associate Professor in Medical Ethics and Education* |
| **14:40** | Coffee Break |
| **15:00** | Genomic testing: managing incidental findings and uncertainty*Gillian Crawford* |
|  | Pre-conception genomic testing: is implementation in healthcare possible and desirable?*Mirjam Plantinga, Research Fellow* |
| **15:45** | Final questions*Led by Anneke Lucassen* |

**Abstracts**

**New technologies in genomics: “the potential and the hype”**

***Anneke Lucassen***

Massive leaps in technology over the last decade have enabled us to analyse a person’s entire genetic code far more quickly and cheaply than in the past. However, it turns out that interpreting our genetic code is often much harder than many expected and raises a range of ethical issues. These will be explored in this introductory session.

**Whose eyes on your genome? Confidentiality and information-sharing in genomic medicine**

***Sandi Dheensa***

Imagine you have a rare condition, the cause of which is unknown. After several inconclusive tests, your doctors offer to sequence your whole genome. A genetic explanation for your condition is found, but so is lots of other possibly significant but unrelated information. Should your genome sequence, and those of other patients, be shared with medical researchers, so they can explore the information in more detail? What about with researchers working for pharmaceutical companies? What if it were sometimes possible to identify the sample came from you?

Now imagine that your sequence shows you are at risk of a particular cancer, unrelated to the original condition for which you had testing. Should doctors share this information with your family members, who might also have inherited the same genetic finding? What about if you are a family member, and the tested person is refusing to share their information: should doctors share it anyway? These are just some of the difficult considerations related to privacy, confidentiality, and information-sharing in genomic medicine. In this interactive presentation, you will hear about current practice in the NHS and in research, as well as what the future might hold.

**World café**

***Angela Fenwick***

This is an informal way of engaging people in conversations about issues that matter. In a café setting we will have the opportunity to discuss some of the ethical issues raised during the course of the day, facilitated by a member of the Clinical Ethics and Law team.

**Genomic testing: managing incidental findings and uncertainty**

***Gillian Crawford***

Rapidly declining costs and increasing availability of genetic testing means that we can now examine an individual’s entire genome. Possible results from these tests include those relating to the original reason for testing but also other unrelated ones, so-called Incidental Findings- IFs. Suppose your child went for testing because they had developmental delay and while a genetic explanation for this was found the test also discovered that they were at significant risk of developing bowel cancer as an adult. Incidental findings, especially when identified in children, may not bring health consequences for many years and whether and how this information is fed back to families, requires consideration. Other findings of uncertain clinical significance can also be discovered. Where this is the case further testing may be needed to better understand their meaning. The potential complexity of such information means that careful attention to how patients are prepared prior to any tests and how results are fed back to them is required.

**Pre-conception genomic testing: is implementation in healthcare possible and desirable?**

***Mirjam Plantinga***

Every one of us is, usually without knowing, a carrier of one or more gene abnormalities. Since we all have 2 copies of each of our genes, having one altered or abnormal copy will not cause any health problems because the normal copy of the gene compensates this alteration. However, if we have children with another carrier of the same gene abnormality then there will be a 1 in 4 chance (25%) per pregnancy that children inherit a genetic condition from us because they inherit both abnormal copies. With this sort of genetic inheritance, there is often no family history of the condition, so that the first diagnosis in a family is unexpected.

With a pre-conception carrier (PCS) test, it is possible to test, before pregnancy, whether future parents are carriers of the same genetic condition and therefore have an increased chance of having an affected child. Offering PCS testing to a general population would allow couples who are both carriers to be identified pre-conception so that the information can be incorporated into their family planning. This talk will explore whether responsible implementation of population based expanded carrier screening in healthcare is possible and desirable.