CENTRE for HUMAN GENETICS

NUFFIELD DEPARTMENT of MEDICINE

UNIVERSITY OF OXFORD

Ethical Issues of Genomics in Healthcare

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Rachel Thompson

CELS/Lucassen Group, Centre for Human Genetics, University of Oxford

Rachel.thompson@well.ox.ac.uk

Clinical Ethics Law & Society (CELS)

Led by Professor Anneke Lucassen and based at the Centre for Human Genetics, Oxford.

We are clinicians (including genomic medicine), social scientists (sociology; anthropology; STS), and ethics specialists.

We work on ethical and societal issues around genomic medicine, research, and healthcare

We take an interdisciplinary approach to complex problems- future-facing and in response to historic and current challenges.

Closely linked with the **Centre for Personalised Medicine (CPM),** University of Oxford

Today we will…

Introduce some key ethical issues in genomic medicine and research

Genomic medicine (clinical genomics)-decision-making; uncertainty; familial information sharing

Use an example clinical case to examine how these issues are encountered in practice

Research- specific ethical challenges; diversity, benefit sharing

Centre for Personalised Medicine- Schools Art Competition and public events

Consent and confidentiality in genomic medicine

What we might think of as ‘standard’ ideas of consent are challenged by genomic medicine

I consent but genetic information is not just about me

Familial information sharing

Considerations for children- Ceri’s story

Best interests decisions- such as in PALOH (Jessica’s talk- newborn genetic testing- hearing loss prevention)

Uncertainty: prediction; diagnosis; clinical significance and ‘results’

Clinical Case example: Ceri's story

Questions to consider

Who holds an interest in Ceri's case?

Whose interests are most important?

What do you think family members should be told? By whom?

How might we ensure that Ceri can access information about her risk as an adult?

Some key concepts

Confidentiality

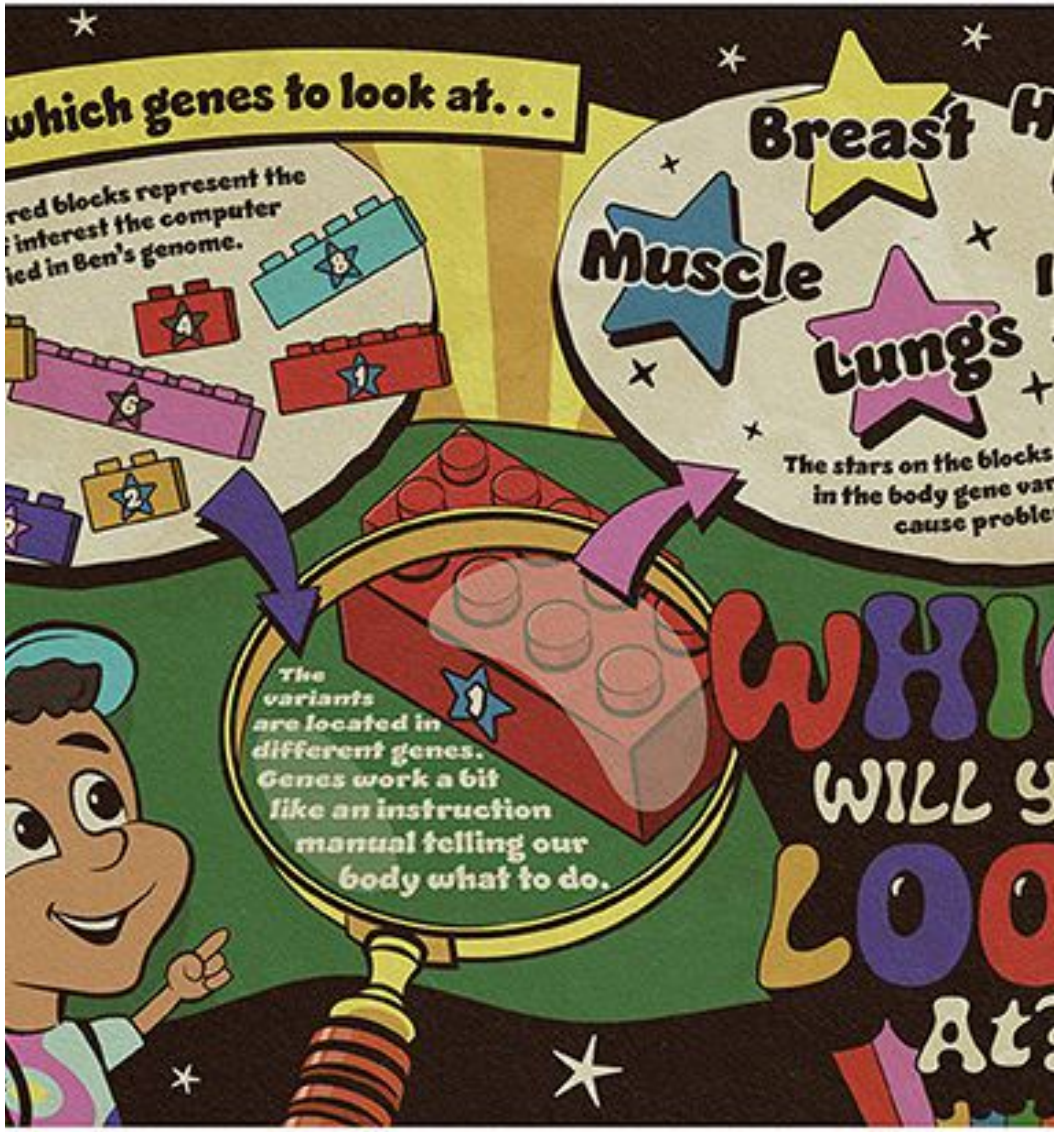
Familial information sharing

Best interests- basis for decisions about or on behalf of others

Predictive vs diagnostic testing

Uncertainty-intrinsic to genomic information

Incidental findings- something extra found in a test



Clinical Case Example: Ceri's story

Ceri is a 4-year-old girl with heart problems and a cleft palate. Her paediatrician requests genomic testing to investigate her health problems.

Testing is carried out and finds thatCeri only has one working copy of the BRCA2 gene (a deletion). **This has no relevance for Ceri's health in childhood (and does not explain the problems that led to the test)**. However, this BRCA2 finding might mean her adult relatives have an increased risk of breast cancer and some might be eligible for screening and/or interventions to reduce their risk.

Ceri's story continued…

Ceri may have inherited her BRCA2 deletion from one of her parents. This means that they and/or their relatives may have an increased risk of cancer.

They might benefit from screening or risk-reducing interventions such as surgery, but they might not know to access this if the BRCA2 finding is not communicated.

For Ceri, there are no recommended actions until adulthood.

What should we consider when deciding whether to test a child or adult?

What 'incidental findings' do you think we should share with a child and their parents? With an adult?

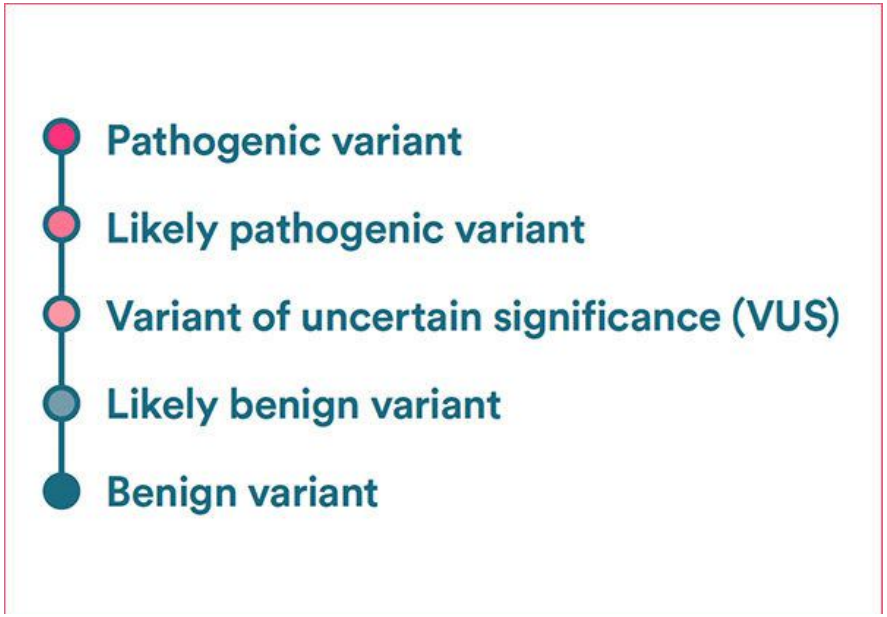
What rights do relatives have to information that may affect their health decisions?

Genomic medicine, uncertainty and ‘results’

We tend to think of medical testing as ‘finding facts’- uncovering things that ‘naturally’ exist- by using the right equipment and expertise they can be found.

Analyzing patients’ genome tests has been described as a bit like interpreting abstract art, in which different people might see and value different things.

Thinking about clinical or genomic data in this way can help us work with uncertainty and engage with underlying assumptions



Genomic Medicine Research

ELSA of Genomic Medicine: Research

Ethical considerations- some shared with clinical/healthcare, some specific to research. The boundaries between healthcare and research are increasingly blurred.

Research purposes and justifications: Public interest; responding to common disease burdens; rare disease; population health; health economics and political drivers.

Who is research for? - potentiality; benefit sharing; prioritisation; challenges to consent; timelines and intergenerationality; immortal data

Who don't we see and why does it matter? Diversity in genomics; equity; access to healthcare and research participation; public involvement

Diversity in genomics research- some headline numbers

People of European genetic ancestry represent ~80%

of people in genome-wide association studies (GWAS)

Some polygenic risk scores are ~4x

more accurate for people of European genetic ancestry than of African genetic ancestry

Approximately 7% of significant associations

have been discovered in individuals of African ancestry

While only 2%

of genome-wide association studies (GWAS) participants are of African genetic ancestry

Diversity in genomics research- why does it matter?

Fairness and accuracy- for the benefit of all

Problematising conceptions of family, identity, and health in genomic medicine and research- constructed categories; meanings to participants and communities

Remembering that- complex combinations of factors shape our (ill) health- Genomics may be one part of the story.

Social determinants of health and existing inequities- avoiding reductionism.

Diversity should be understood intersectionally, and as a means not an end.

Key takeaways- ethics and genomic medicine

Genomic data are both individual and shared and there may be multiple interests to consider

‘Standard’ understandings of e.g. consent are challenged by genomic medicine and research

Research usually does not benefit participants directly and can take many years to inform practice.

Diversity in genomics is important, but not an end in itself. Diversifying data must be accompanied with efforts to increase equity in healthcare and research

Complex multifactorial conditions, research topics require interdisciplinary approaches- there are many ways to contribute.

Ethics in action

Embedding ethics (ethical preparedness)

Ethical working requires a multifaceted and ongoing approach.

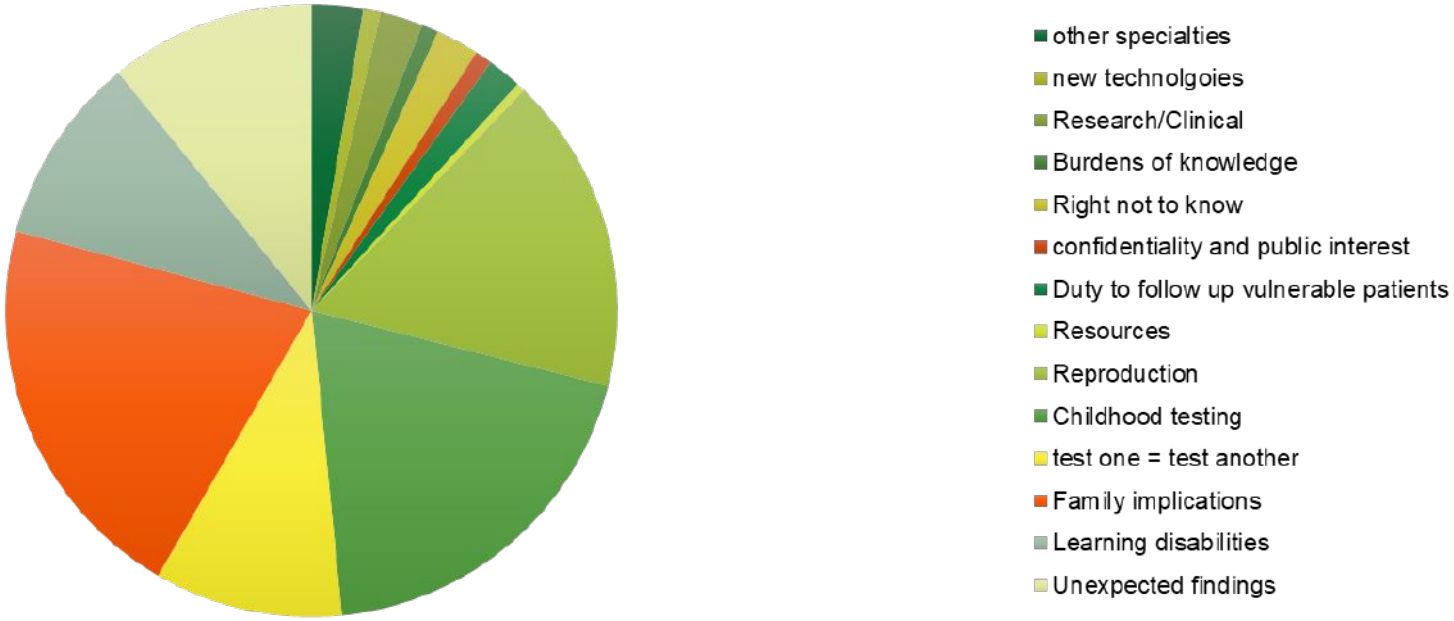
For example…

- Ensuring that ethical issues can be openly discussed as they arise in healthcare and research
- Meaningfully including the patient experience– developing ethical preparedness of healthcare professionals *and* patients and families.
- Engaging with ethical and societal considerations at *all stages* of healthcare and research

including ethics in teaching and training- beyond compliance

The Genethics Forum

What topics are discussed?



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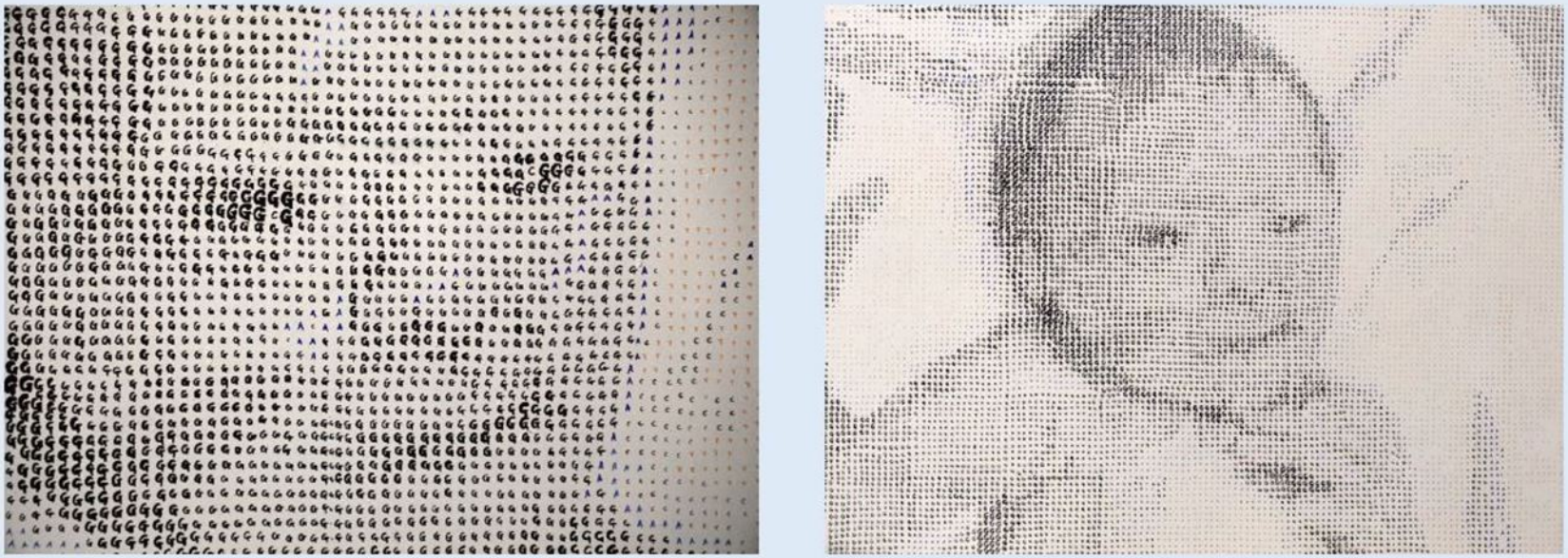
Exploring personalised medicine from a range of perspectives

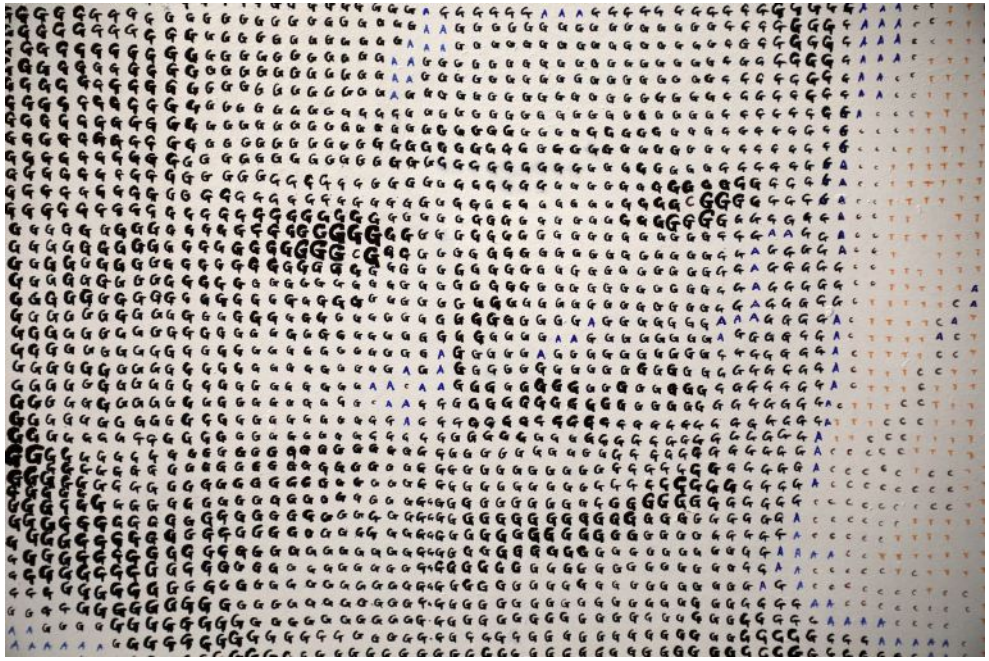
About the Centre

CPM Schools Art Competition 2024 winner



First place: Laranya, aged 13 from Worksop College, Nottinghamshire

Close-up image and artist's description



'When viewing the picture up close you only see the letters, but when you look from a distance you can see the face of the baby. This shows that when examining our DNA, you must look with a microscope, and these tiny proteins make up a whole person.'

Laranya, aged 13

References and resources

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