



Ethics and Genomics

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Integration of social science into research is crucial

Social scientists must be allowed a full, collaborative role if researchers are to understand and engage with issues that concern the public, says [Ana Viseu](#).

16 September 2015



Incidental findings



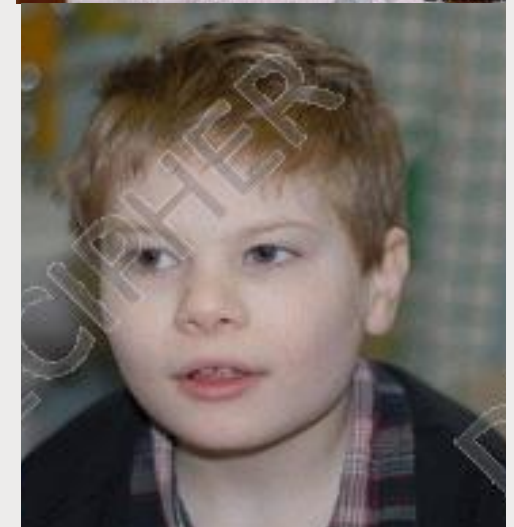
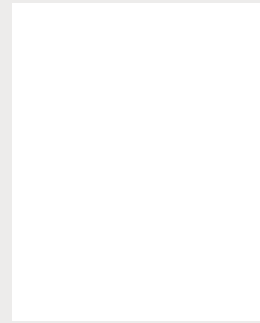
DNA and big data

What to do with
incidental findings
from sequencing
research?

DDD Molecular Project

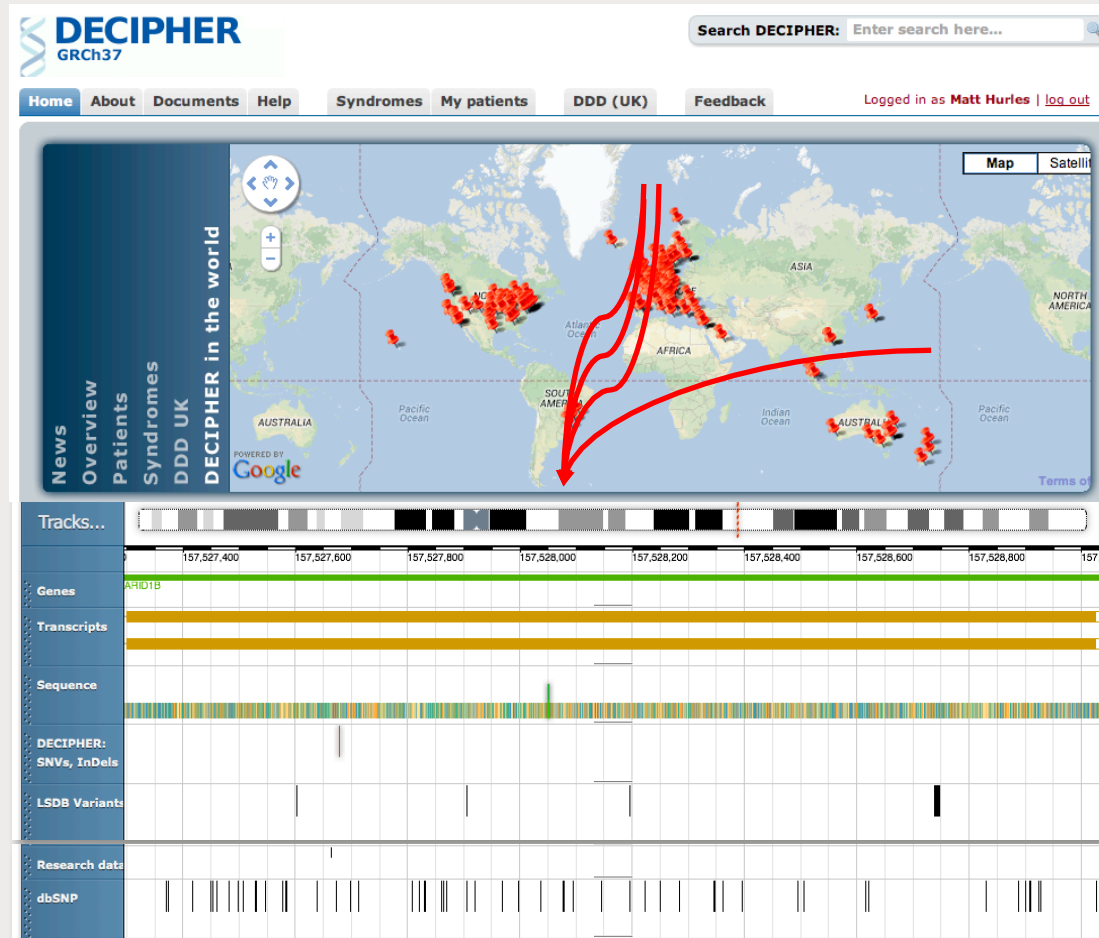
Strategy:

- Recruit 14,000 children plus parents, i.e. 40,000+ samples
- Deep phenotyping
- NHS testing revealed no diagnosis
- Exome Sequence
- Feedback likely diagnoses (yield 36% and increasing)



DECIPHER: Genomic Matchmaking

- Sharing of minimal genotype and phenotype
- Data deposition and visualisation
- Global: 206 centres, >28k patients
- Will include all DDD patients



Over 500 publications citing DECIPHER in past 5 years

PCGF2

G→A Chr17:36,895,854

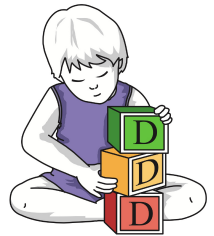


'INCIDENTAL FINDING', OPPORTUNISTIC SCREEN

e.g. BRCA1

PERTINENT
FINDING

Developmental Disorder
gene



In DDD

- We are not exploring or sharing IFs
- Want to focus on the clinical question
- Difficulties with interpretation
- No firm position taken in clinical practice, thus in 2010 establishing a position in research was premature

THE SOCIAL- SURVEY



- ✓ Questions about you
- ➔ Sharing of Pertinent Findings
 - Sharing of Incidental Findings
 - Categorizing Incidental Findings
 - Relations with Risk
 - Raw data
 - Duty of Genomic Researchers
 - Filter of Genomic Information
 - Consent for genomic research
 - Last few questions about you

Sharing of Pertinent Findings

- **Should Pertinent Findings from genome studies be made available to research participants?**
 - Research participants should be able to receive pertinent findings if they want them
 - I don't think pertinent findings from research projects should be available
 - I don't know

« Previous

Next »

Public = 4961



Genomic
researchers = 607



Genetic health
professionals = 533



Other health
professionals = 843



Q: What influences attitudes the most?

A: Our professional background rather than the country we are from



Genetic Health Professionals



Other Health Professionals



Genomic Researchers



Public

Three key messages

- On the whole, all stakeholders would be interested in receiving IFs
- Actionability is important to people
- Genetic health professionals are more conservative
 - *Most realistic about how this would work in clinic*

Middleton A et al (2016) Eur J Hum Genet

Middleton A et al (2015) J Med Genet

Middleton A et al (2015) Lancet

Middleton A et al (2014) Soc Sci Research

Middleton A et al (2014) J Community Genet

Current Genomics England List

- **Bowel cancer predisposition:**
 - *MLH1* (adult only), *MSH2* (adult only), *MSH6* (adult only), *MUTYH* (adult only)
 - *APC* (adult and child)
- **Breast and ovarian cancer predisposition:**
 - *BRCA1* (adult only), *BRCA2* (adult only)
- **Other cancer predisposition:**
 - *VHL* (adult and child), *MEN1* (adult and child), *RET* (adult and child)
- **Familial hypercholesterolaemia:**
 - *LDLR* (adult and child), *APOB* (adult and child), *PCSK9* (adult and child)
- **Cystic Fibrosis :**
 - *CFTR* (*parents only*)

**Millions of
genomes being
researched...**

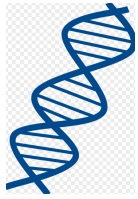
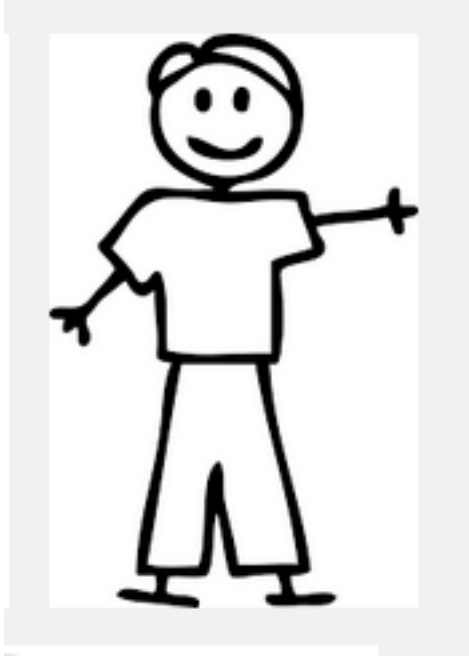


DNA and big data

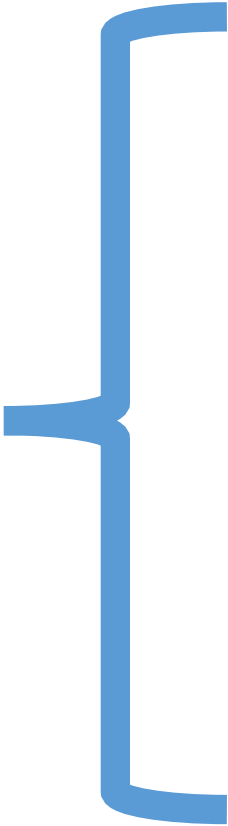
Overview

- Why is data sharing necessary?
- What are the risks from donating your data?
- Attitude research

**Why is data
sharing
necessary?**



compare



What are genomic databases being used for?

- For use by medical doctors
 - *is my patient's genome result the same as someone else's?*
- For use in non-profit research
 - *What's the prevalence of a particular genetic change across the UK?*
- For use in for-profit research
 - *Is this medicine working in people with a particular genetic change?*

How we described
this to the public

**What are the risks
of donating data?**

Risks

- Anonymous data is linked back to person
- Person is identified
- Personal information available online
- Could be accidental
- Could involve malicious intent
- There are serious penalties

**Attitudes from
public?**

Your DNA, Your SAY

[reset answers and start again](#)



- Welcome
- Online footprint
- Data access by others
- Access by medical doctors
- Access by non-profit researchers
- Access by for-profit researchers
- Perceived harms
- Expectations of information
- Trust
- Socio-demographic questions
- Submit your response

Welcome



First 1117 responses

- Spread of ages, socio-demographics, 50/50 men/women
- 57% know nothing about genomics ('public') rest are health pros/researchers, or patients
- Preliminary data

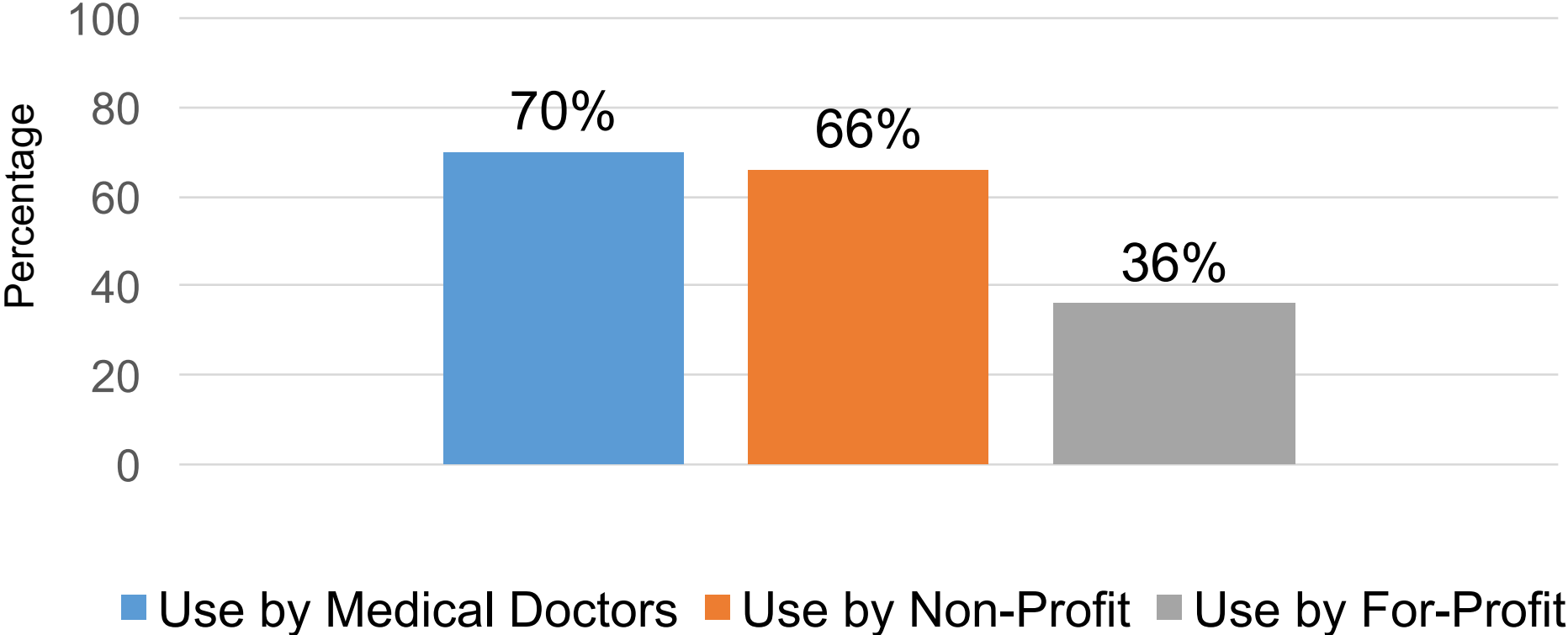




Q: Would
you donate
your data?

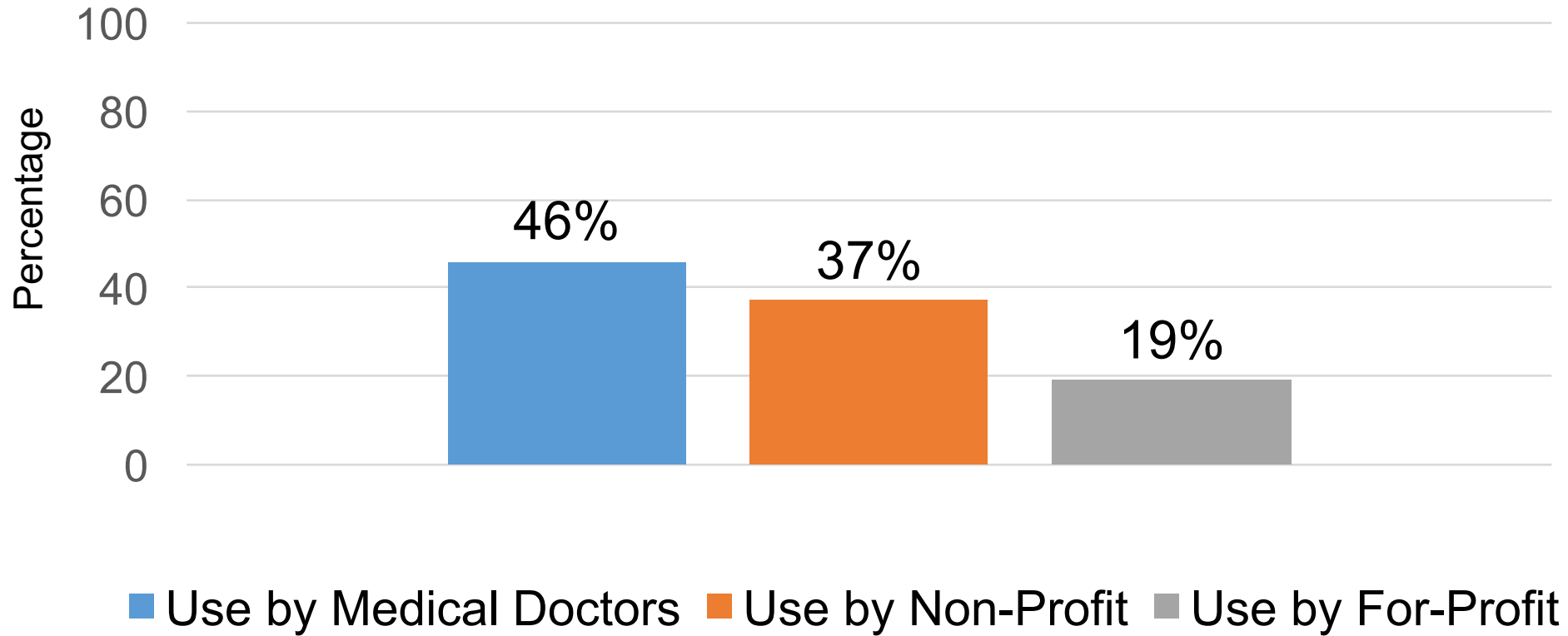
Connected to genomics world (through work or being a patient or research participant)

“Yes, I’d donate my DNA and medical data for...”



Public

“Yes, I’d donate my DNA and medical data for...”



Concern about
pharma making
profits from free
donation

**Those who know
about genomics
are more likely to
donate**

The plan

- Translated into multiple languages (Russian, French, Polish started, Japanese, Arabic, Swedish planned)
- Data collection through 2017-2018



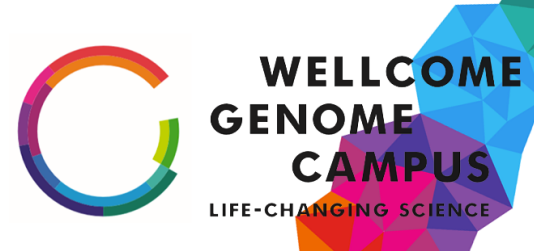
There is an urgent need to raise consciousness

What can genomics do for me?

How can I talk to relatives about it?

What sort of society do I want to live in?

It couldn't have happened
without.....



www.GeneTube.org

DDD Team

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Nigel Carter

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