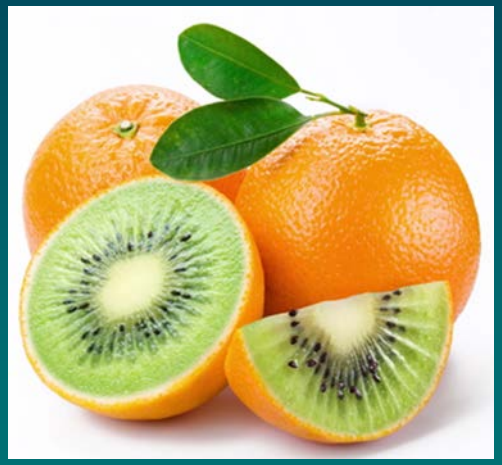
Genomic investigations: Health Care Professional (HCP) and family experiences of managing incidental information in clinical practice

Gillian Crawford
Clinical Doctoral Fellow/
Principal Genetic Counsellor
Clinical Ethics and Law at
Southampton (CELS)



gc@soton.ac.uk
www.soton.ac.uk/cels

23rd September 2014

Phenotype ➡ Genotype

Targeted counselling and testing

Low chance of uncertain and incidental findings

Genotype ➡ Phenotype

Broad counselling and testing

Increased numbers of uncertain and incidental findings

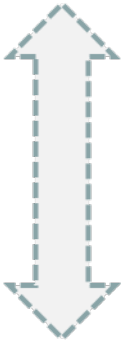
Abnormalities of potential clinical significance that are unexpectedly discovered during routine testing, unrelated to the original purpose of the investigation

Incidental to what?
Unexpected to whom?

Clinical question that arises from signs/ symptoms/ FH

Duty of care in clinical practice

Clinical benefit



Research into the sequence of genomes

Altruistic participation; duty of contract

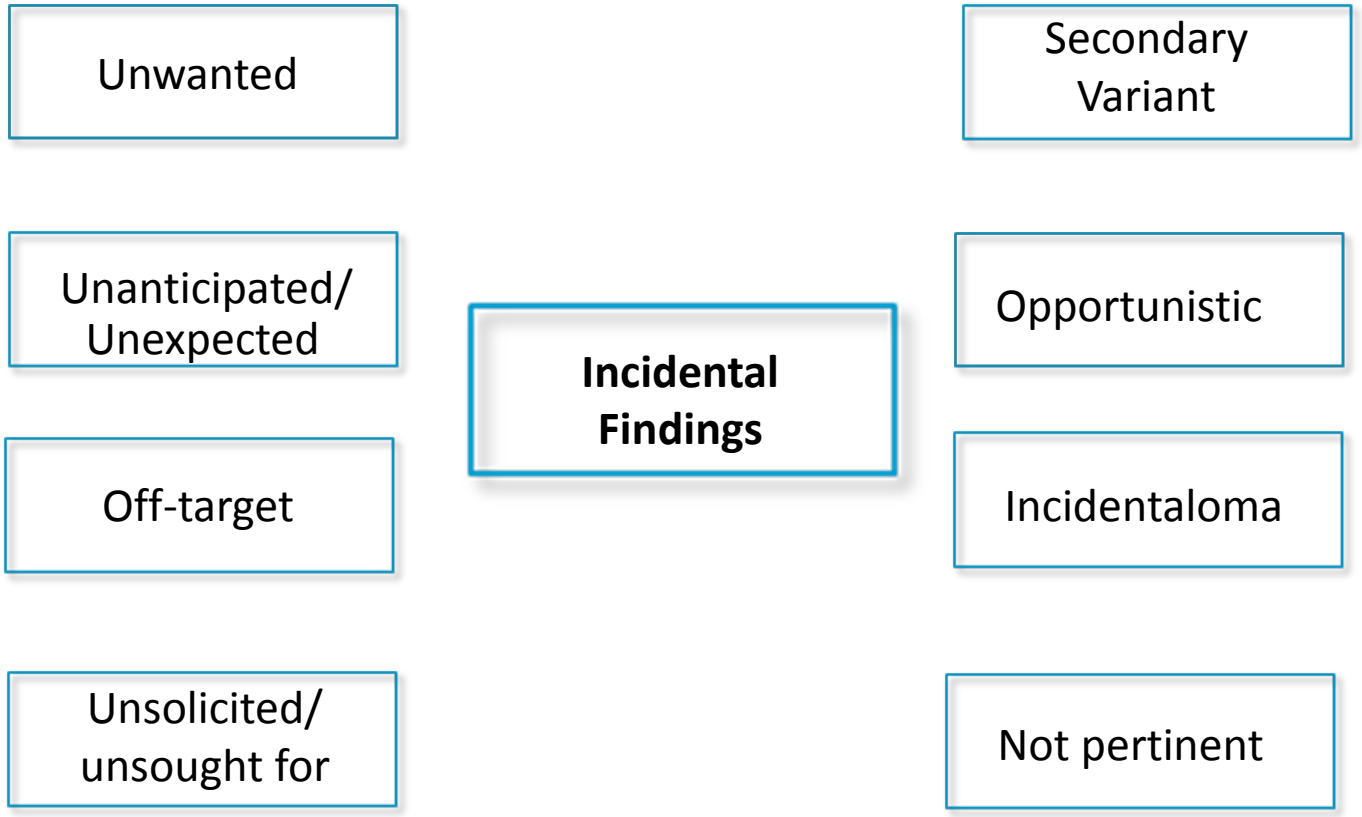
Consent

Disclosure practices may differ

Not new…but…

- Genetic technologies will increase frequency
- Limited research on management of IFs in clinical practice (consent and disclosure)
- Pre-natal and new born screening

Terminology



'One size does not fit all'

Patient perspective

Other finding

Picked up something else

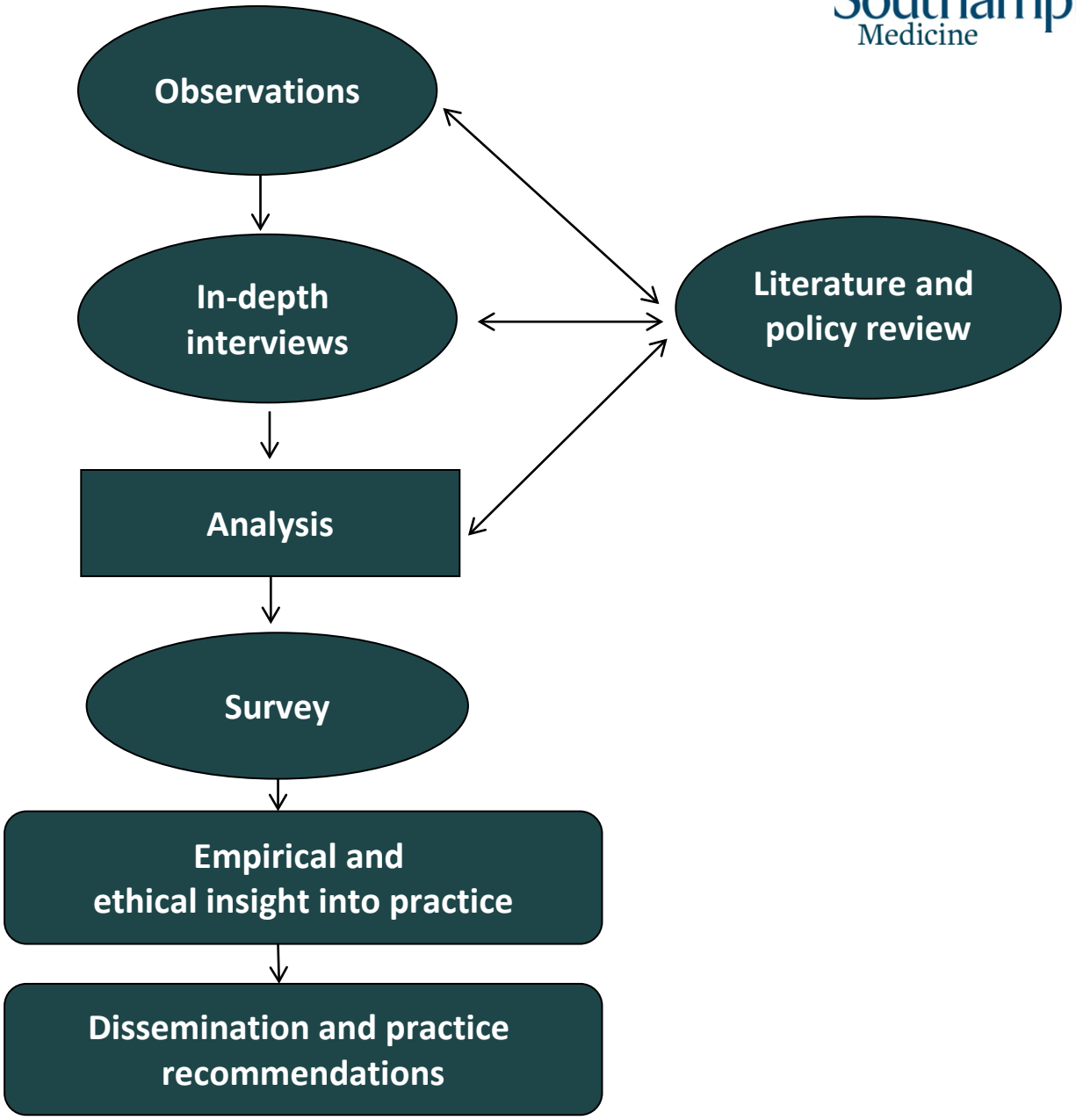
Unexpected

Crawford G et al (2014) A more fitting term in the incidental findings debate: one term does not fit all situations. *European Journal of Human Genetics 22 957*

Phase 1

Phase 1

Phase 2



Research aims

- Describe current practice in a range of clinical settings
- Explore lay and professional views and experiences of genetic IFs arising in the clinical setting
- Generate information to advise policy on the consent and disclosure practices of IFs in clinical practice

Interview sample

HCP Interviews (n=32)

- Genetic specialists: 22
- Paediatric specialists: 7
- Adult physician:1
- Clinical scientist: 2

Family interviews (n=16)

- 16 participants from13 families
- 3 sets of parents
- Number of families with an IF: 5

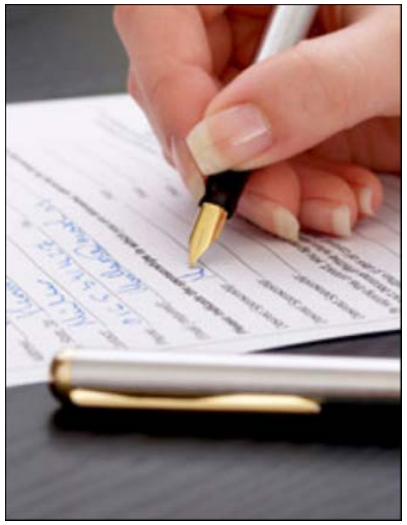
Reason for test	IF discovered	Intervention
DD aCGH	22qdel including SMARCB1 Schwannomatosis	Regular review
Muscle problems aCGH	FBN2 mutation Marfan's phenotype	Cardiac screening
Behavioural and social problems aCGH	Y chromosome deletion (infertility)	N/A
DD aCGH	Deletion including MSH6	N/A Referral for bowel screening as an adult
RP Gene panel	Mutation in LRP5 gene Osteoporosis	Bone density screening

Findings: Consent and Disclosure

- The possibility of an IF is not consistently discussed
- No common practice when IFs are discussed

  (use of examples, consent forms)
- No discussion about the information

  families want



"…But you know it was a bit of a shock, because no we didn't know that they could have found anything else out. Nobody said that….No, nobody ever mentioned…I didn't really realise and I thought they were looking for one thing; didn't know that you could sort of find all these other things out…" FAMINT013 (IF discovered)

Lay and professional experiences of IFs

Family members

- Surprised by IF result
- No regret at receiving IF (interventions/future treatment)
- Would want further IFs
- Desire for information

HCPs

- Nearly all IFs were disclosed
- HCPs found disclosure challenging
- Often used cases in subsequent consultations
- Reported that patients adjusted well to this information

"…because it's actually a twist of fate if you like, a bit of luck, that had [son] not had these underlying problems he could have developed… this condition later in life and been unaware of it. So actually the fact that he knows about it, it gives him a kind of … a lucky break… because he'd got the opportunity to get screened for it regularly…" *FAMINT011 Father*

"…it proved very hard work, explaining that [juvenile polyposis IF] and the mother getting very upset and quite angry… I mean the irony actually was that probably the risk with this single allele being deleted…was uncertain, but almost that was kind of worse because then there was the accusation that you bring something up and yet you still don't really know if it's important or not." HCPINT013 Consultant Geneticist

Findings: Meeting expectations

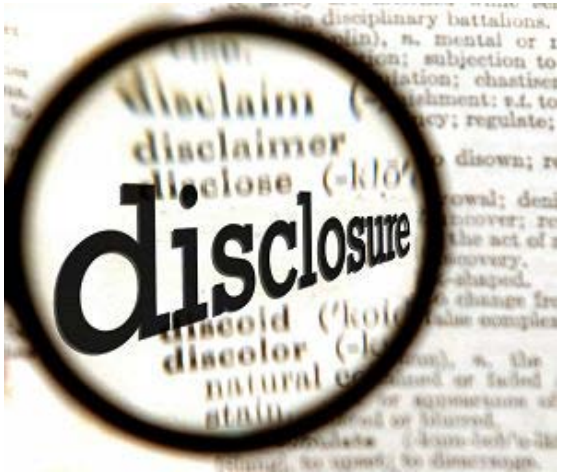
- Consensus that possibility of IFs should be discussed
- Reasonable for patients to have choice on IFs disclosed
- Challenges of offering choice:

- Hypothetical decision-making

- Sustained wishes

- Recording and complying with decisions

- Follow up and storage of information



Hypothetical decision-making

"I think it's really difficult and I think for a lot of people they wouldn't be able to give that [IF choices]. And I think even myself, if somebody asked me that, I think, well it would depend what it is … and clearly you don't know what your incidental finding is until you find it, so you can't counsel somebody and ask them about every single eventuality." *HCPINT028*

"You can get a feeling, a gist of someone's preference, whether… someone would want to know everything, know nothing or a few gradations in between. That's probably about as good as you can get."

FAMINT008 Father

Themes about consent and disclosure practice

What is consent?

- Information
- Information and involvement
- Permission

Who decides?

- Patients
- HCPs
- Shared

Themes about consent and disclosure practice

- What is 'the result'?
- Clinical practice and research
- Genome as a future resource
- Patient expectations
- Mainstreaming
- Follow up and re-contact
- Familial implications

To consider…

- The place for non-directive counselling?

- The consent process?

(who, documentation, dynamic, broad, opt-in, opt-out, mainstreaming)

- Challenges at the clinical/research/commercial interface

Acknowledgements

All participants

Professor Anneke Lucassen

Dr Angela Fenwick

Dr Tara Clancy

Dr Claire Foster

NHS
National Institute for Health Research
Trainees Coordinating Centre

FONDATION BROCHER

University Hospital Southampton NHS
NHS Foundation Trust

Defining and managing incidental findings in genetic and genomic practice (2014) Shiri Shkedi-Rafid, Sandi Dheena, Gillian Crawford, Angela Fenwick and Anneke Lucassen. *J Med Genet*. E-pub.