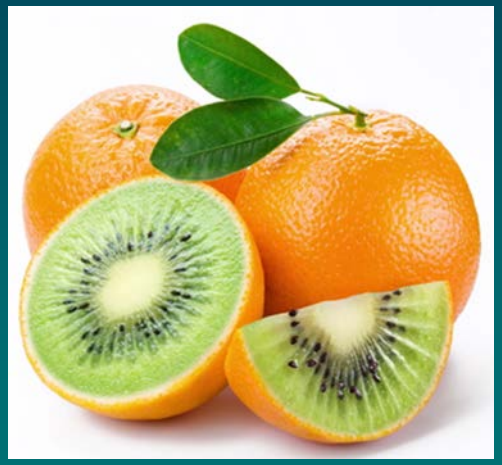
Genomic investigations: health care professional (HCP) and family experiences of managing incidental information in clinical practice

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



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

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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

Not new…

- Medicine: radiology, biochemistry, history taking
- Genetics: karyotyping, mis-attributed paternity, Biobanks

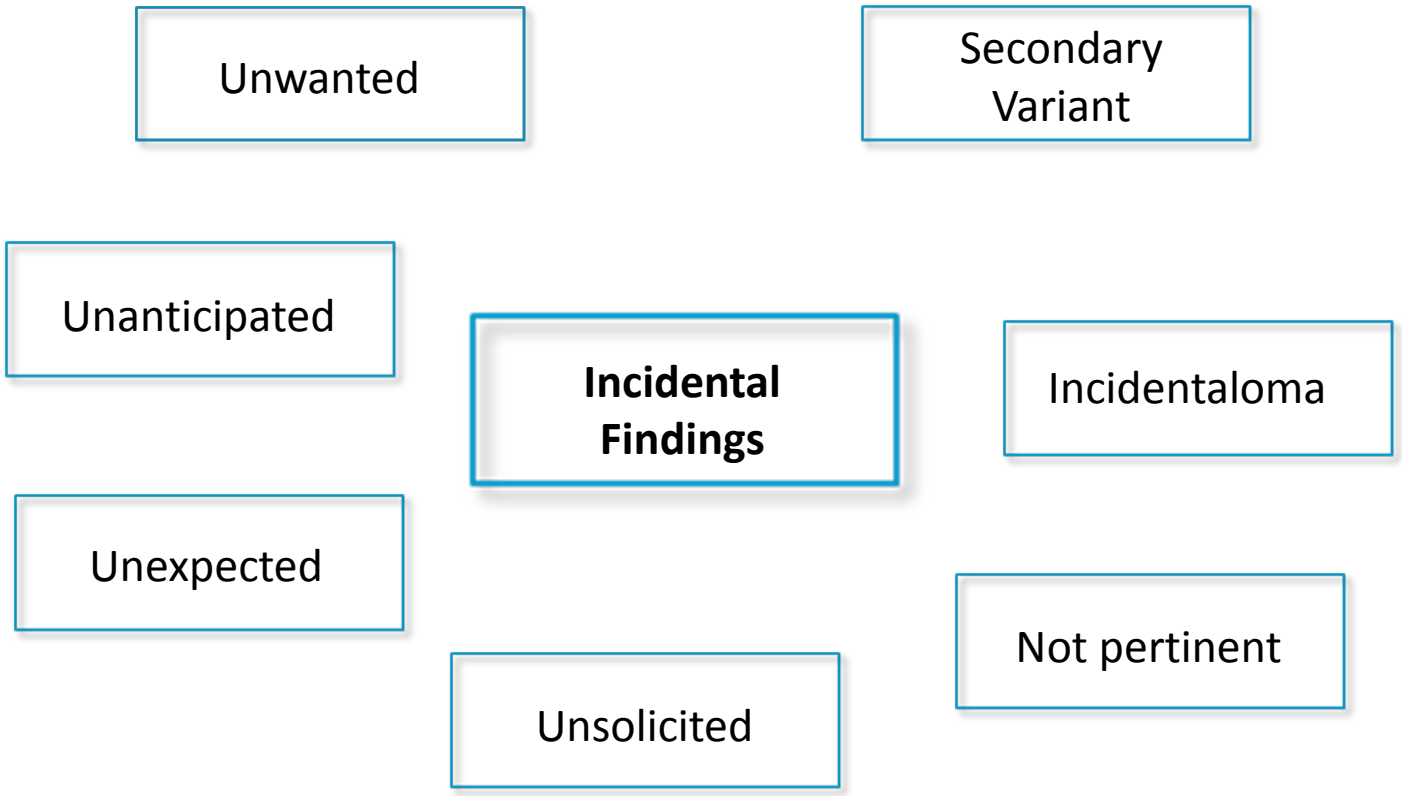
BUT

- Genetic technologies will increase frequency
- Limited research on management of IFs in clinical practice (consent and disclosure)
- Information about risks to others and clinical relevance in the future
- Pre-natal and new born screening

ACMG American College of Medical Genetics and Genomics

Genomics england

Terminology



'One size does not fit all'

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

Other finding

Picked up something else

Unexpected

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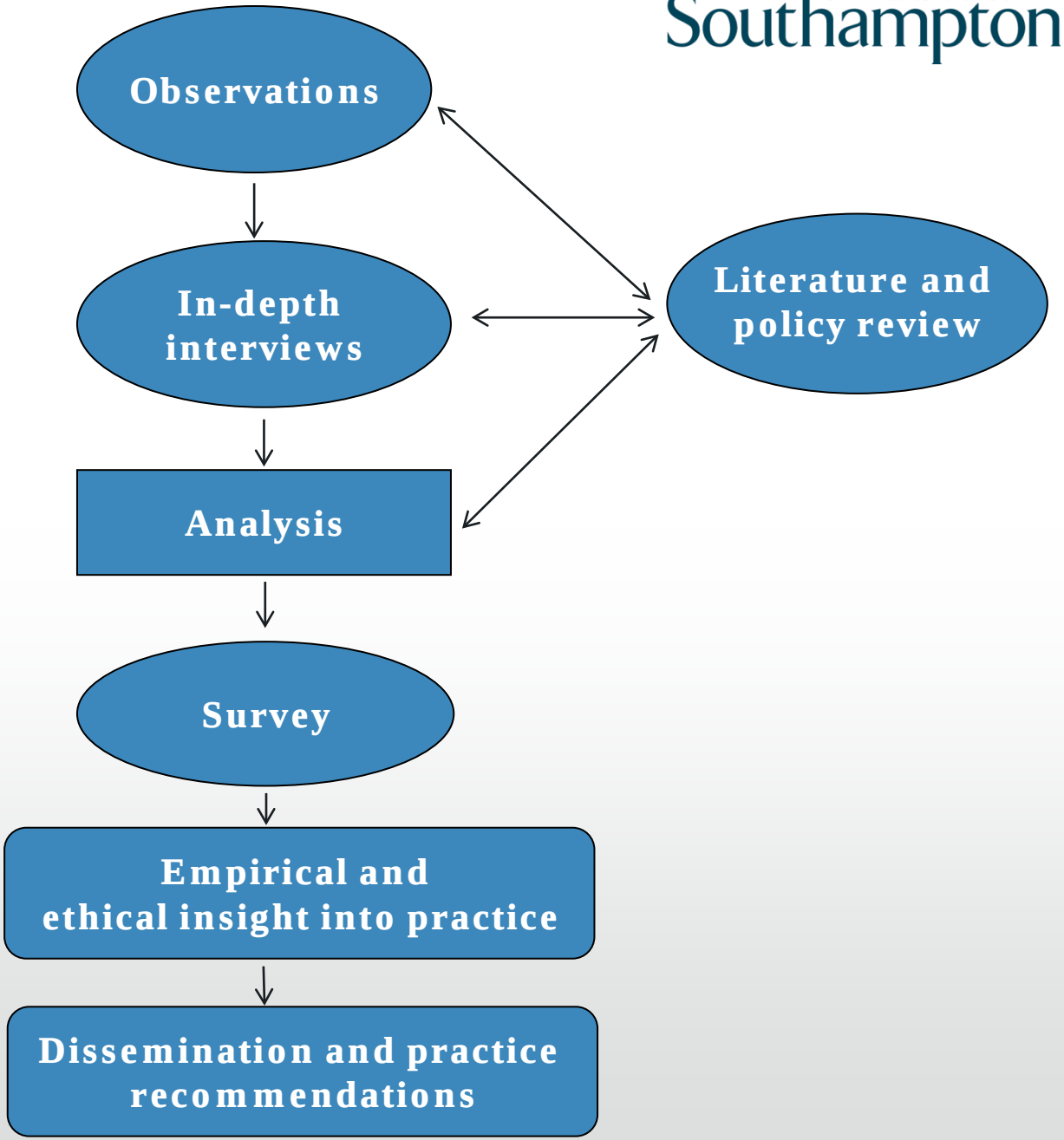
Phase 1

Phase 2

Phase 3



Study aim

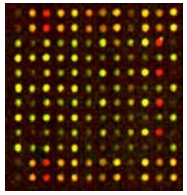
Investigate lay and professional experiences and views about the ethical and practical issues raised by the discovery of IFs

Research question

With the advent of new technologies how are, and how should, clinically relevant IFs be incorporated into practice, taking into account both practical and ethical considerations ?

Cohort

Array CGH testing mainly



Sample

HCP Interviews (32)

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

Family interviews (16)

- Total participants:16 (13 families)
- 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

Families' (5) experiences of IFs

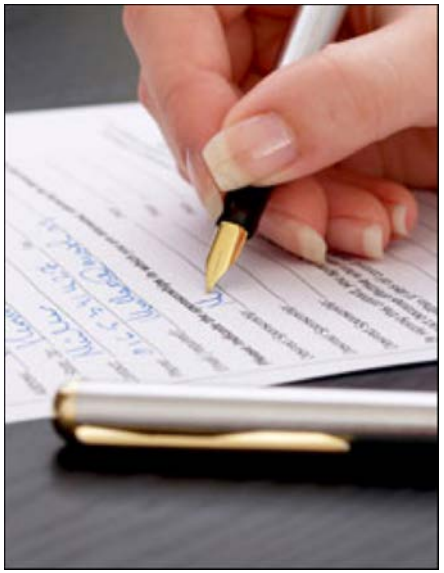
- Surprised by IF result
- No regret at receiving IF (interventions/future treatment)
- Would want further IFs
- Desire for information

"…because there was another part of it that was quite relieved, 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…" *FAMINT011b*

Findings

Consent (HCPs and families)

- The possibility of an IF is not consistently discussed



- No common practice when IFs are discussed

(use of examples, consent forms)

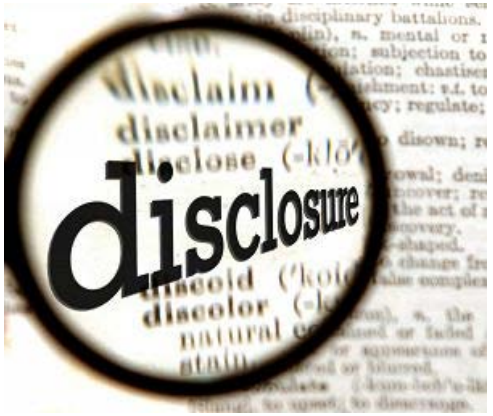
- Consensus that possibility of IFs should be discussed

Disclosure (HCPs and families)

- Currently no exploration of what information families want
- Both groups think it is reasonable to have choices

BUT

- Hypothetical decision-making
- Sustained wishes
- Implementation and ongoing challenges
- What happens to genomic 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. I think it's difficult."

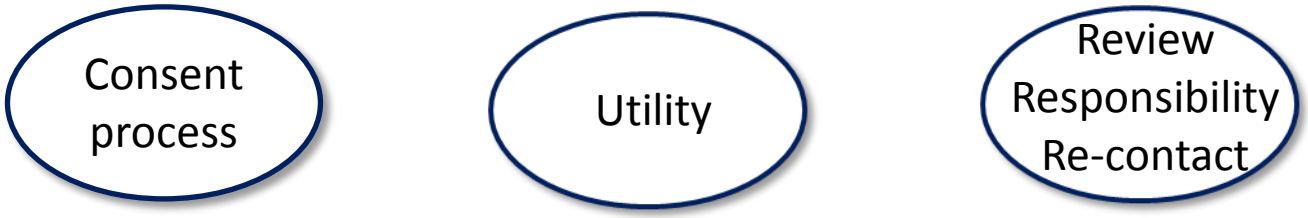
HCPINT028

"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."

FAMINT008b

Ethical considerations

Patient choice ⟺ Clinical judgement



Family led disclosure model ⟺ Professional led disclosure model

To consider…

- The place for non-directive counselling?

- What will the consent process look like?

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

- Challenges at the clinical/research/commercial interface

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