

Recontacting and cascade testing Sharing data and information with patients

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Introduction and background

- Vigdís Fjóra Stefánsdóttir
- Genetic counsellor PhD
- Landspítali National University Hospital, Iceland
- Member of the ESHG Public and Professional Policy Committee
- I have nothing to disclose

Topics

Iceland

Genetic
counselling

Recontacting

Consent

Biobanking
from patients'
perspective

Cascade
testing

Information to
patients

The future

Iceland

Nordic country

Nordic values

Nordic trust

Nordic standards

Some discrepancies

Want to belong

Author: Simone de Haan
Beverwijk, Netherlands



Why is this important?

Biobanks have a vast amount of information

Our DNA can be traced

Health issues can be traced

Relationship to others can be traced



Combine this with comprehensive health records, population databanks, bank accounts, school records and genealogy data many generations back...


Biobanks

- Laymen's knowledge of biobanks
 - Collection
 - Consent
 - Storage
 - Usage
- Biobank-based research

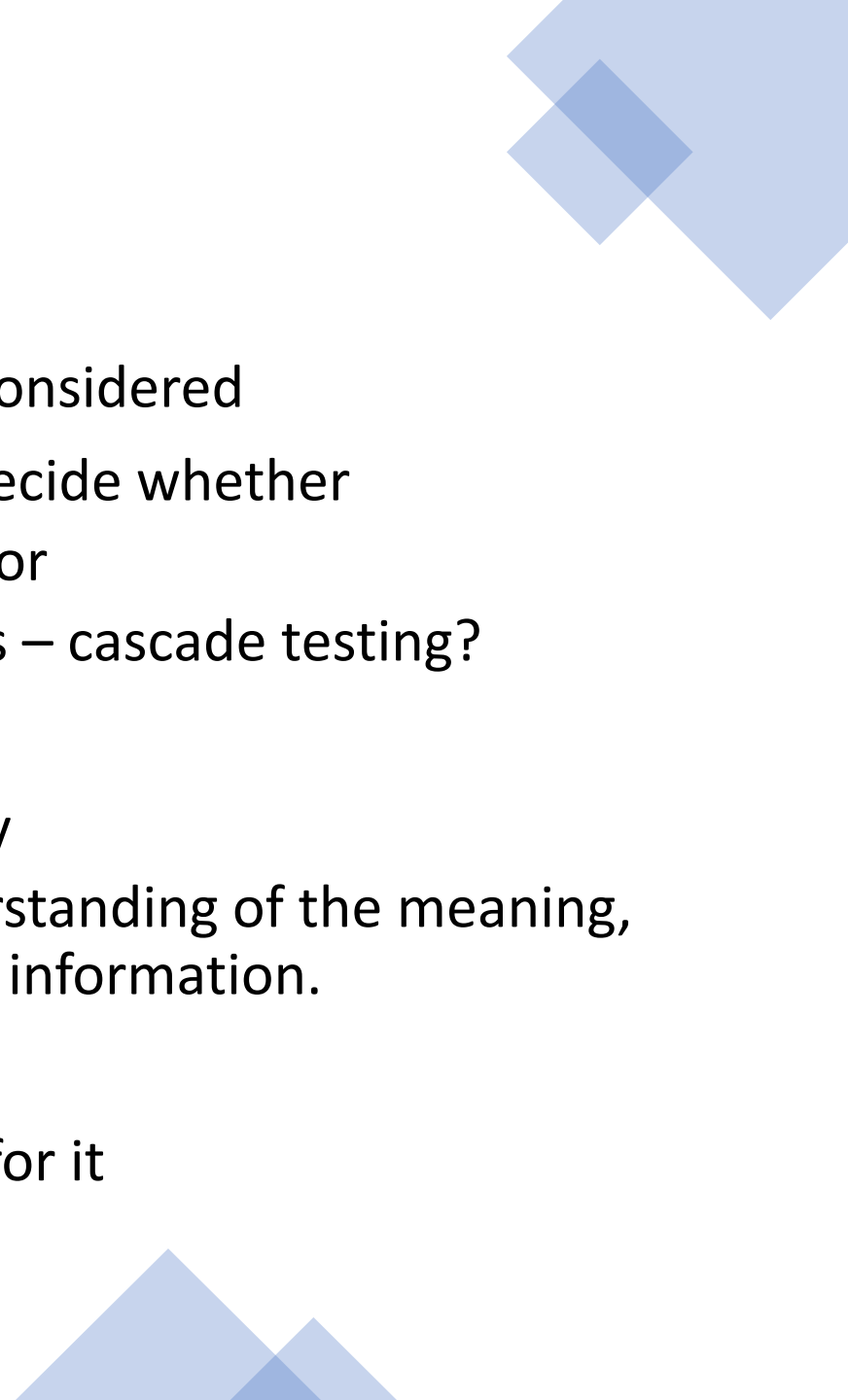
The big debate

- Researchers, policymakers and ethicists debate the return of findings from genomic research to participants
 - Is it appropriate to return results?
 - Which results should be returned?
 - Only significant results? Other?
 - The “best” way
 - How to consent
 - Clinical confirmatory testing
- The data obtained through research is vast and can be very important





Return of individual results - research

- Should be routinely considered
 - Participants should decide whether
 - they receive and/or
 - share their results – cascade testing?
 - Consent
 - Communication is key
 - to promote understanding of the meaning, and limitations of information.
 - Expensive – account for it
- 



Consent - biobanks

- Usually (informed) consent is available for but...
- not always and sometimes just forgotten or...
- samples are used for different studies without reconsenting

Consent to studies and clinical testing

The informed consent is very important

But how informed is it?

How does it compare on a reading scale?

Many pages of gibberish?

What about those that are unable to understand and consent

- Children
- People with various disabilities
- People who do not use computers (in case of electronic consent)

Do people read the consent?

Remember the content of the consent

Even remember taking part in a study?

Dynamic
consent – the
solution to all?

Traditional consent

- Narrow
- Broad
- Paper
- Electronic

Dynamic

Health portals



Cascade testing

- Definition of Cascade testing (CT)
 - To offer relevant family members testing after a pathogenic variant has been found in an index patient
- What is the first thing coming to mind following positive results?
 - The children – how to inform them
 - Other relatives
- Information to relatives
 - The index patient (most common)
 - The genetic counsellor (in some cases)
 - The lab (unusual in clinical testing)



Communication is the core of genetic counselling

- Listen to the counsellee
- Evaluate the situation and state of mind
- Explain
- Inform



Monies


Positive test results – then what?

Cost of informing

- Information and consent
- Clinical confirmation
- Surveillance

Cost of surveillance

- No
- But...to send out nationwide letters giving positive information from biosamples!
- A consent is an agreement between the researcher and the subject – if you are going to inform people – tell them and include it in the consent



Is it illegal to give
research results in
Iceland?

Web-based return of results

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<https://doi.org/10.1038/s41431-020-0665-1>



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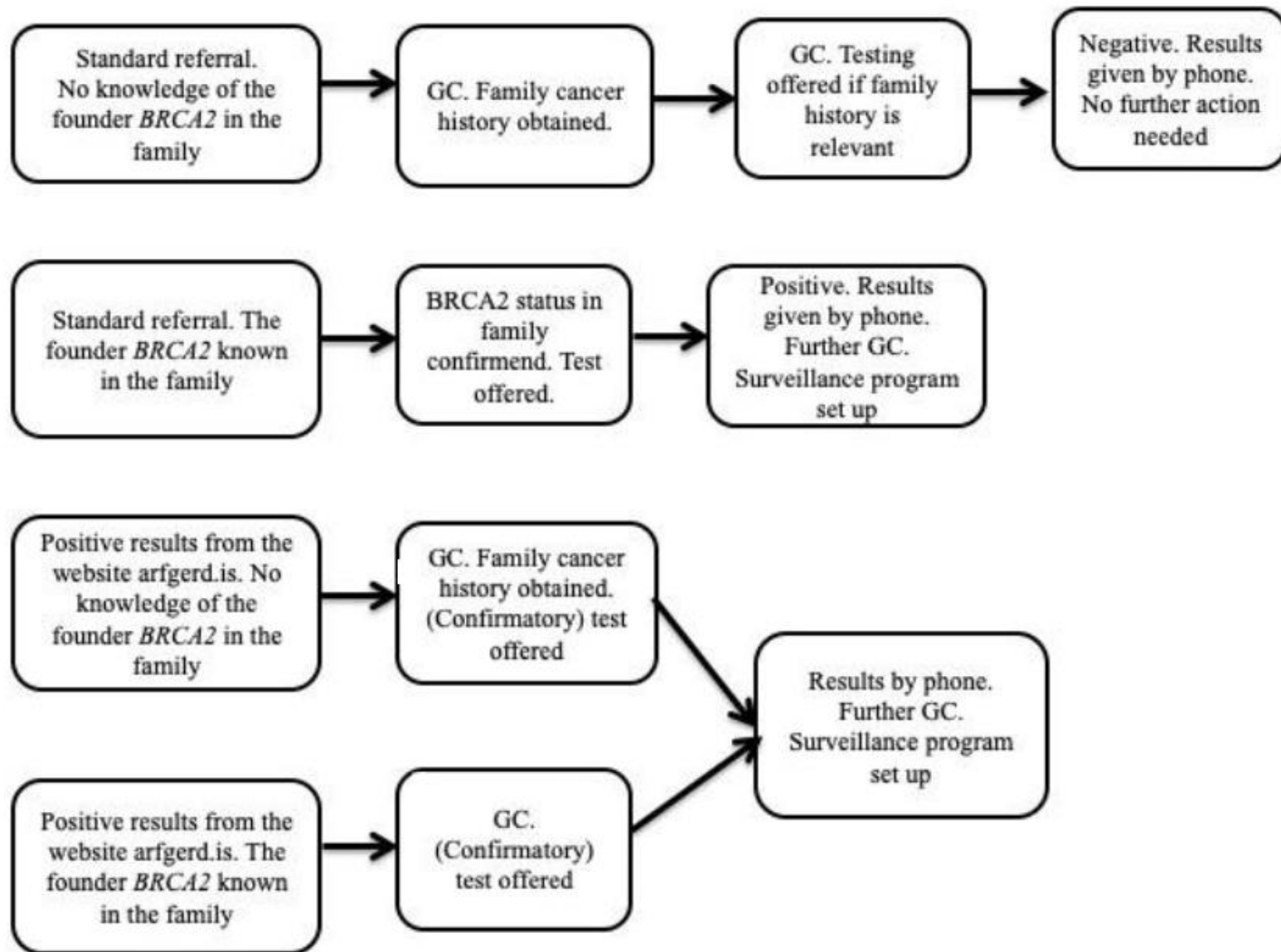


Web-based return of *BRCA2* research results: one-year genetic counselling experience in Iceland

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The outcome

- Carrier rate 0.7-0.8% - appr. 2400 individuals
- 46.000 had signed on to the website (verbal information from deCODE)
- Appr. 37.000 had results
- Over 17.000 (36%) had no sample available
- 5000 donated a new sample
- 352 positive
- Only 195 of the 352 contacted the GC unit
- Additionally 129 relatives

Information
on the
website

Long – but clear

FAQ

Where to go if positive
(Landspítali)

Emotional impact

Emotion	No. of positive through website (%)	
Shock	26	(13%)
Surprise	46	(24%)
Discomfort	5	(3%)
Fear for relatives	6	(3%)
Other negative reaction	24	(12%)
Not surprised / expected the result	61	(31%)
Positive reaction	26	(13%)


How to do
this better?

Information, clear information, easily understood information

Have people stop and answer questions in several places in order to keep on

Have someone standing by to answer questions by phone or electronically

Have the option to send the results to a genetic counsellor who will explain



*„Learning from the past, taking
the best, and moving forward“*

Professor Peter Harper.



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