HIPAA, ethical, and logistic considerations in cascade testing

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Implementing Precision and Equitable Public Health in Cascade Testing for Genetic Disorders - A CDC Mini-Symposium and Virtual Panel Discussion
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It is a privilege to greet you today from the traditional land of the Duwamish People, what is now Seattle, Washington. I begin my remarks acknowledging and elevating a resilient people – a people who are still here.
Actionable result → Patient → At-risk relatives notified → Relative makes informed decision → Cascade testing
Current standard: patient-led disclosure

Disclosure to relatives incomplete
- BRCA 73%-75% (Taber 2015, Graves 2014, Fehniger 2013)
- Lynch 67%-98% (Stoffel 2018, Graves 2014)

Uptake of cascade testing is low
- 21%-44% HBOC,
- 41%-94% Lynch (Menko 2019)
Care team support for patient-led disclosure

PMC7926393 2021; PMID: 34994636 2021

- Family letters
- Family genetic counseling
- Communication support / training
Why not notify an at-risk relative?

- Burden
- Unaware of benefit to family
- Strained relationships
- Concern about accuracy, genetics knowledge
- Concern about stressing relatives
- Patient, relative privacy
Why is patient-mediated disclosure the current standard?

- Care and coverage systems designed for 1:1 care
- Clinician duty, scope of practice
- Liability
- Patient, relative privacy
Why can’t someone other than the patient notify at-risk relatives?

- Direct contact programs combined with patient-led contact reach more relatives than patient-led contact alone.
- People conceptualize shared genetic information, family implications.

Oh, yeah. I think [health system-led risk notification] would be wonderful ... it would be so much easier instead of me trying to spit out what I think they need to have done or whatever. You folks could do it in a more -- they know what they’re talking about and in a more business-like, you know, informational way than I could.
Alternate paths for disclosure

- **Clinician**
- **Patient**
- **Relative**
- **Cascade testing**

Actionable result

- **Provider-mediated disclosure**
- **Patient-mediated disclosure**
- **Public health-mediated disclosure**
Research questions:

Is direct contact of relatives to recommend cascade testing consistent with HIPAA privacy rule?

Is direct contact acceptable to U.S. patients and families?
Is direct contact of relatives to recommend cascade testing consistent with HIPAA?

- Multidisciplinary working group
- Family vignette, literature review

HIPAA: Health Insurance Portability and Accountability Act of 1996
Patient-mediated approaches

Considering only HIPAA and not any relevant state laws, may the covered entity (provider):

give their patient information or a ‘consent to contact’ form for family members? Yes.
Provider-mediated approaches

Considering only HIPAA and not any relevant state laws, may the covered entity (provider):

- directly contact their patient’s adult relatives to recommend testing? 
  - with patient permission ✓
  - without patient permission * ✗
  - over the patient’s objection * ✗

- directly contact the relative’s provider to recommend testing

*absent public health approach
Public health approach

Considering only HIPAA and not any relevant state laws, may the covered entity (provider):

directly contact public health authorities to report a patient’s genetic risk?

- No, unless state public health law makes genetic risk a ‘reportable condition’
Are direct contact programs acceptable to patients and families?

Roberts, M.C.; et al. *Health Aff (Millwood)* 2018, 37, 801-808

Menko, F.H et al. *Fam Cancer* 2019, 18, 127-135

Current work
NHGRI-funded R01HG010144 (Henrikson)
Focus: HBOC, Lynch

Aim 1: Co-design direct contact program with patients and families (complete)

Aim 2: Feasibility trial of KPWA-led direct contact (in process)
Scene: It's now the care team's job to contact Irene

Molly asks Dr. Lee to tell Irene

Scene: Dr. Lee consults

Dr. Lee consults w/others to determine what to include in a call or letter.

Scene: Dr. Lee calls

He calls; no answer.
He sends letter.

Scene: Dr. Lee is unable to contact.

Scene: Other means are used, and Irene finally responds.

Irene returns call and is invited to meet Dr. Lee or others

Scene: Irene returns call and is invited

2nd letters & calls
Design round 2 - video
The potential health benefit of notifying relatives of actionable genetic risk is the main rationale for direct contact programs.

Regardless of the feelings I may have, ultimately after I calm down, I would want to know if I had a greater risk of dying like my sister did at 41. [relative]
Aspects of direct contact programs were new and raised concerns about whose duty it is to notify relatives and about how privacy would be maintained.

SPEAKER 1: Where the doctor knows that someone is at high risk, I mean, shouldn't they contact somebody? ...It's a tradeoff, the privacy versus the health -- the lifesaving information. That's what it amounts do, doesn't it?

SPEAKER 2: Maybe. But I think we're forgetting that, like, Molly can also reach out. It's not, like, if the doctor doesn't do it, they're not going to get this information.
I kind of wonder if, like, if you need consent to do that, like, in your initial intake with Kaiser or, you know, how every so often, is your contact information up to date or all that. And you, like, say can we contact you based on family member information, like, would it be okay if we reached out to you if we find something that might be pertinent to you based on a family member.

Participants thought that direct contact of relatives should be a program, not an individual provider’s responsibility. Pre-consenting programs were frequently suggested.
Requirements for direct contact of relatives
Henrikson 2021 PMID: 34200550 J Pers Med

U.S.-based health system-led direct contact of relatives
- Has clear potential benefit
- Should take a programmatic approach
- Should include early, pre-disclosure consent of relatives
- Should complement patient-mediated disclosure
- Should allow relatives to control information flow
Current work

- Direct contact intervention with patient preference for relative contact
  - Letter with phone follow up
  - Relative consultation with genetic counselor
- Pilot study (n=60 probands), KPWA
- Outcomes: acceptability, feasibility, limited efficacy
Thank you
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