Challenges of returning results in a community health setting: insights from the Healthy Nevada Project

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No conflicts to declare

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What is the Healthy Nevada Project?

- Large scale population genetics and health determinants study
- Exome+ sequencing (CLIA/CAP)
- Recruiting as many Nevadans as possible
  - Current IRB approval is 250,000 participants
  - Current cohort = >50,000 sequenced individuals
- Two components:
  - Clinical
    - Reporting on Incidental Findings - currently, CDC Tier 1
    - Risk awareness of **autosomal dominant** inherited conditions
  - Research
    - Investigator focused
    - Leveraging a data-lake of health determinants

Healthy Nevada Project structure V1

CONSENTS: STUDY, RECONTACT, RESULTS, NRS 629.181

SURVEY PLATFORM: BEHAVIOR/ SOCIAL

RECALL: BLOOD/IMAGING

RETURN OF + RESULTS

LGC

FAMILY IMPLICATIONS

Nevada Revised Statutes: Requirements for obtaining, retaining or disclosing genetic information

Outcomes of results being returned directly to individuals

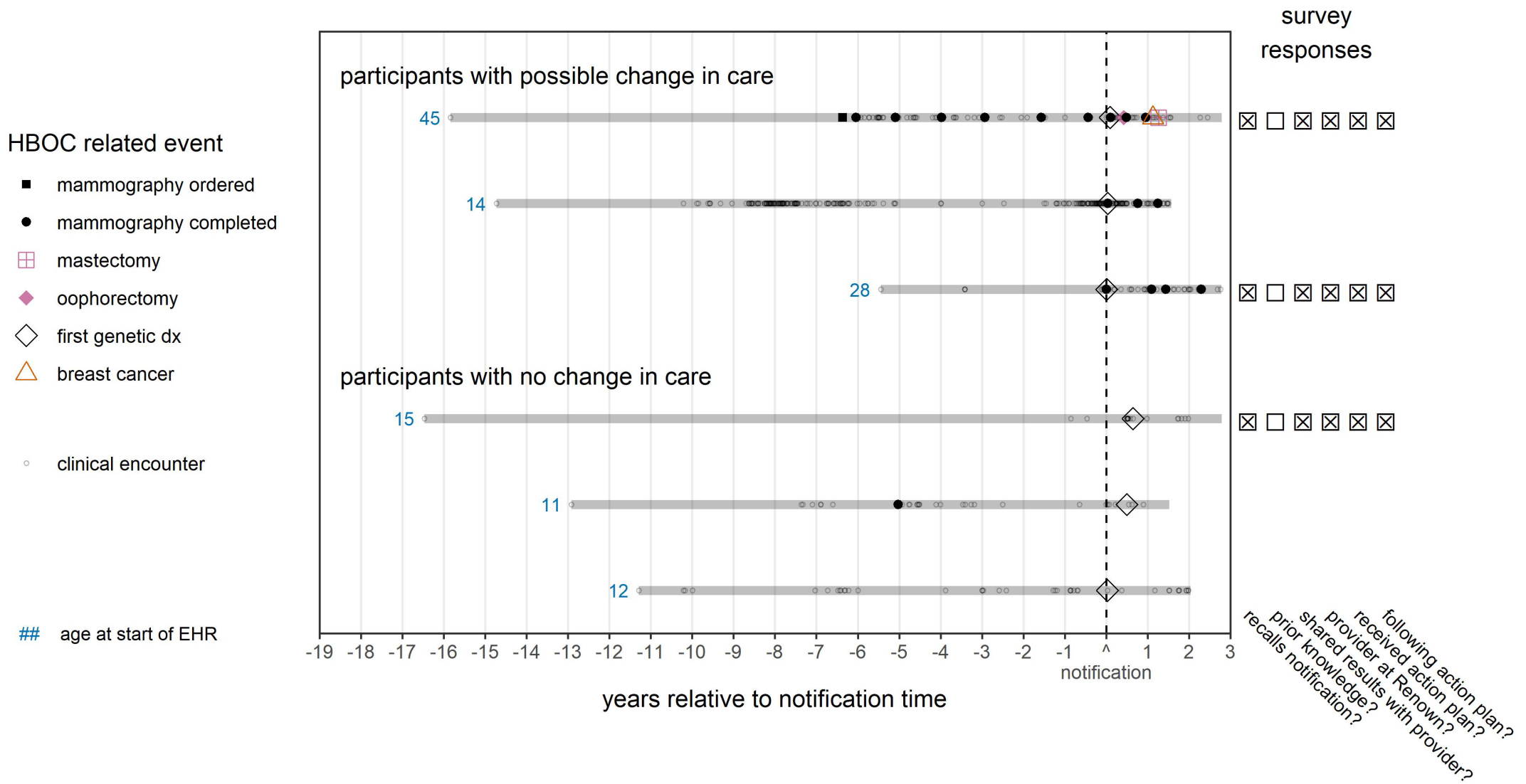
18% of participants were lost to follow up or declined result

71% of participants with T1pos findings shared results with providers

"*However, a sufficiently specific genetic diagnosis appeared in the EHRs and problem lists of only 22 and 10%, respectively, of participants without prior knowledge*"

(Elhanan et al. 2022, https://doi.org/10.3389/fgene.2022.866169)

Genetic dx doesn't always lead to care change



Root causes of return failures

1. The result was not directly put into the patients medical record (caveat for patients with no EHR)
2. Results were returned by outside genetic counselors with limited coordination with patient care team
3. Results return more successful when call / contact came from Renown
4. Poor provider education about CDCT1 positive results and follow-up clinical decision support

Healthy Nevada Project structure V2

CONSENTS: STUDY, RECONTACT, NRS 629.181

SURVEY PLATFORM: BEHAVIOR/ SOCIAL

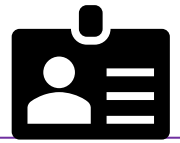
RECALL: BLOOD/IMAGING

RETURN OF RESULTS

All CDCT1 results (and other future results) returned to patient **AND** medical record

Nevada Revised Statutes: Requirements for obtaining, retaining or disclosing genetic information

Healthy NV Workflow – Clinical Integration

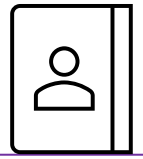


1

Coordinators identify potential participants with an upcoming appointment in Epic

2

Sends informational message about study to patient prior to appointment



3

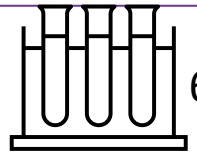
Brochure is handed to patient during the visit and is encouraged to participate by provider

4

Patient is referred to schedule later through MyChart, or encouraged to enroll before leaving with a coordinator

5

Coordinator schedules visit and answers questions and the patient signs consent through Epic



6

Coordinator places DNA Sequencing order and scans kit into order and into manifest. Collects patient sample

7

Ships to sequencer. DNA is sequenced and results returned to Epic

8

Positive results are returned by RN and then referred for Genetic Counseling (if desired). Negative results are returned by referring provider. All results are available through MyChart and through the Helix Customer Portal

Strengths of this approach

1. 100% return of results – all controlled in house with “Renown” label (CDCT1 +; n=116)
2. Much easier to monitor clinical decision support steps as patients flagged with CDCT1 conditions and follow-up workflows
3. Approach benefits our rural, underserved population that does not have as many clinical touchpoints
4. Better data to improve diversity and address historical inequities of care

Drawbacks of this approach

1. Large focus on the healthcare system as most recruitment occurs within system
2. Clinic-to-clinic variability in receptivity of genetic screening likely augments health disparity
3. Documenting in EHR require participant and provide-side follow-up
4. Cascade screening of family members outside health system is challenging but screening is warranted! (case study)
    1. Attention to family needs
    2. Often requires case-by-case interactions
    3. Extends provider education needs

Case Study

- Large multiracial family
- Mother and Father both have *BRCA2* pathogenic variants
- Family located in and out of Nevada
- One of the sibling asked for help in explaining risks to other family members

Resultant Action

- Privacy, HIPAA, ethics, etc., precludes reaching out to family for cascade screening
- HNP provides materials for CDCT1 positive individuals to provide to family members possibly affected
- Sibling arranged for a voluntary information session at the hospital and online with PI and Study physician with expertise in returning results and family med., in attendance
- Attended by >20 family members who was an FDR to someone with a pathogenic finding

Take home points

- Returning results and ensuring best post-return care is difficult and relies on participant education and engagement for success
- Rural and non-EHR integrated settings present challenges for benefiting the underserved population
- Provider education and engagement is key to effective results return, follow-up, and documentation

Thank you HNP Team!

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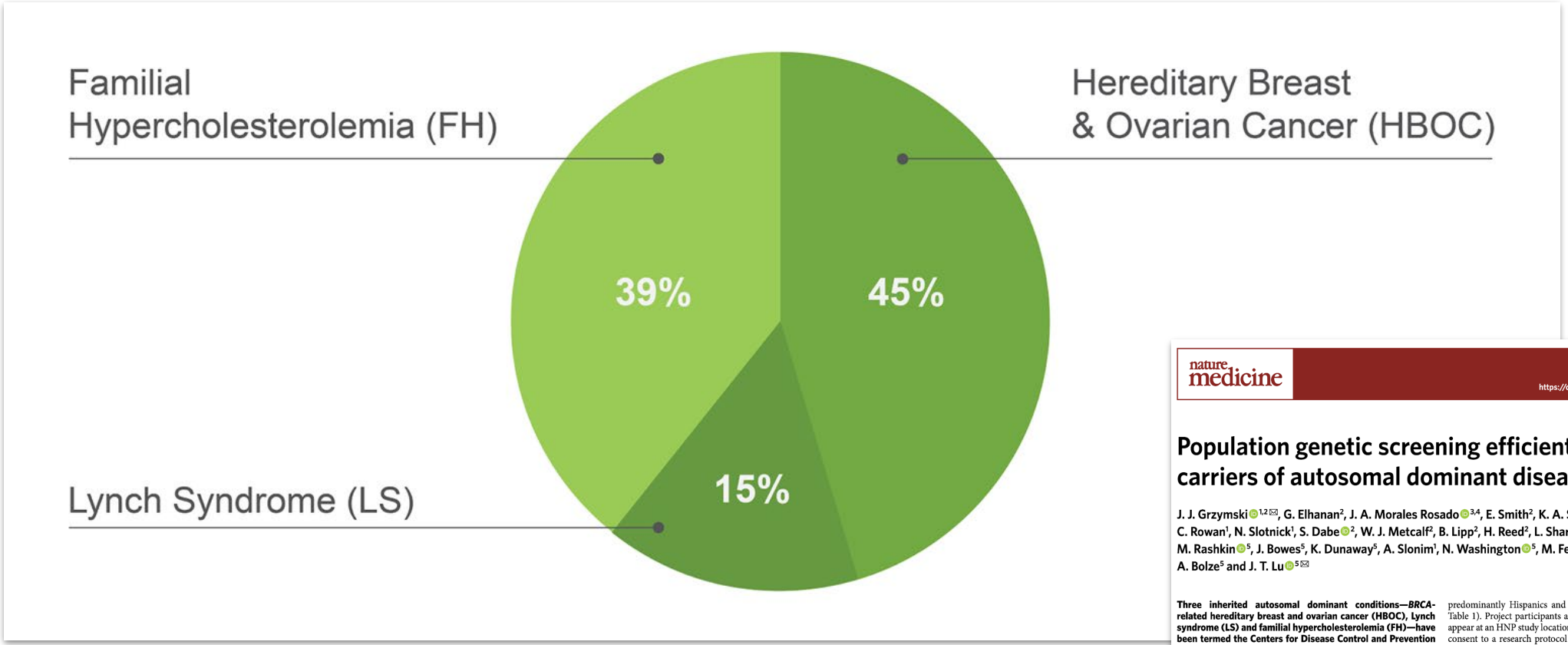
Dr. Leslie Greenberg
Shaun Dabe
Tracy Metcalf

All the HNP Genomic Ambassadors

1 in 75 had actionable medical findings across three conditions

Breakdown of patients with actionable medical findings by condition



nature medicine

LETTERS

https://doi.org/10.1038/s41591-020-0982-5

Check for updates

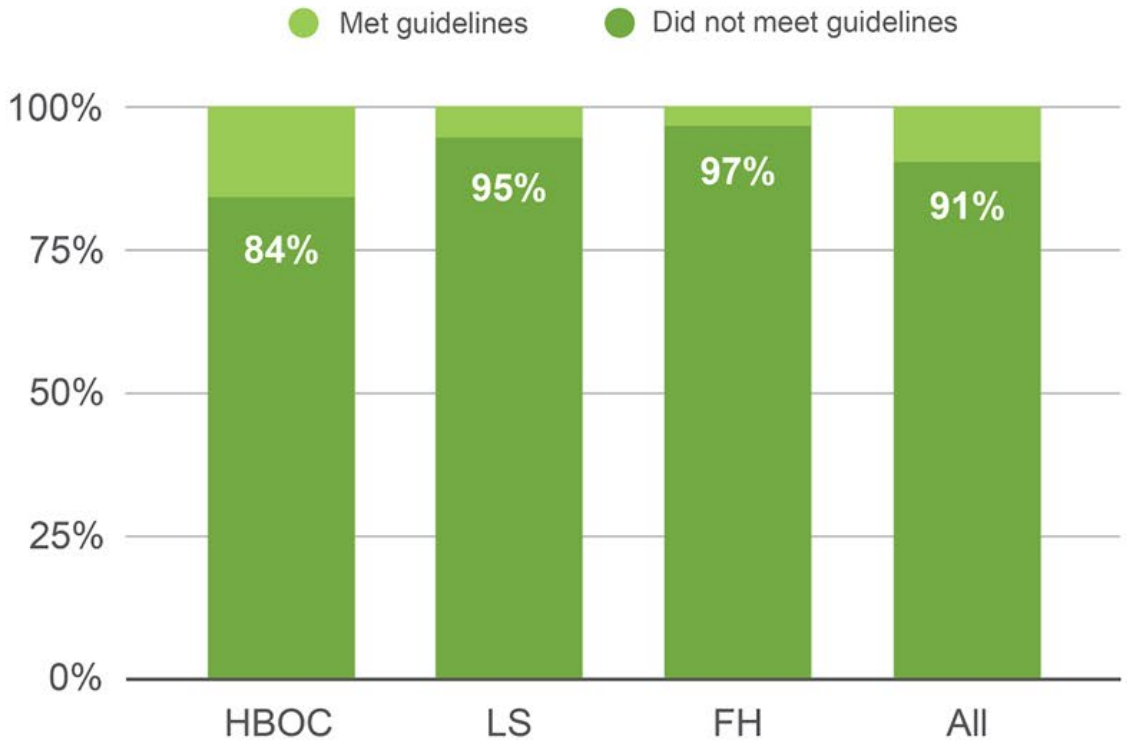
Population genetic screening efficiently identifies carriers of autosomal dominant diseases

J. J. Grzymski[1,2], G. Elhanan[2], J. A. Morales Rosado[3,4], E. Smith[2], K. A. Schlauch[2], R. Read[2], C. Rowan[1], N. Slotnick[1], S. Dabe[2], W. J. Metcalf[2], B. Lipp[2], H. Reed[2], L. Sharma[5], E. Levin[5], J. Kao[5], M. Rashkin[5], J. Bowes[5], K. Dunaway[5], A. Slonim[1], N. Washington[5], M. Ferber[3,4], A. Bolze[5] and J. T. Lu[5]

Three inherited autosomal dominant conditions—*BRCA*-related hereditary breast and ovarian cancer (HBOC), Lynch syndrome (LS) and familial hypercholesterolemia (FH)—have been termed the Centers for Disease Control and Prevention Tier 1 (CDCT1) genetic conditions, for which early identifica-

predominantly Hispanics and Native Americans (Supplementary Table 1). Project participants are at least 18 years of age and must appear at an HNP study location for in-person consent. Participants consent to a research protocol that includes (1) clinical 'Exome+' sequencing and (2) linking of sequencing data with electronic

Over 90% did not meet guidelines for genetic testing



Of These Individuals

<20% had any documented suspicion of inherited genetic disease in the medical record

<40% had family history of relevant disease on subsequent ascertainment