

Paper Summary

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Title: How Clinicians Conceptualize “Actionability” in Genomic Screening

Authors: Kellie Owens, Pamela Sankar, Dina M. Asfaha

DOI: <https://doi.org/10.3390/jpm13020290>

Year: 2023

Publication Type: Journal Article

Discipline/Domain: Genomic Medicine / Medical Ethics

Subdomain/Topic: Actionability in population genomic screening

Eligibility: Eligible

Overall Relevance Score: 90

Operationalization Score: 75

Contains Definition of Actionability: Yes

Contains Systematic Features/Dimensions: Yes

Contains Explainability: Partial

Contains Interpretability: Partial

Contains Framework/Model: No

Operationalization Present: Yes

Primary Methodology: Qualitative

Study Context: Population genomic screening in primary care

Geographic/Institutional Context: United States; multiple health systems

Target Users/Stakeholders: Primary care providers, clinical geneticists, genetic counselors, genomic program

Primary Contribution Type: Empirical qualitative analysis

CL: Yes

CR: Yes

FE: Yes

TI: Partial

EX: Partial

GA: Yes

Reason if Not Eligible: n/a

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How Clinicians Conceptualize “Actionability” in Genomic Screening

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Actionability in population genomic screening

****Contextual Background:****

The paper explores how clinicians define, assess, and operationalize “actionability” in the context of genomic screening.

****Geographic/Institutional Context:****

United States; multiple health systems with varying resources and genomic screening programs.

****Target Users/Stakeholders:****

Primary care providers, clinical geneticists, genetic counselors, public health genomics program managers.

****Primary Methodology:****

Qualitative interviews (n=35) with purposive and snowball sampling.

****Primary Contribution Type:****

Empirical qualitative analysis.

General Summary of the Paper

This qualitative study investigates how clinicians—primary care providers, clinical geneticists, and genetic counselors—conceptualize and operationalize “actionability” in the context of genomic screening.

Eligibility

Eligible for inclusion: ****Yes****

How Actionability is Understood

Definitions range from narrow (“pathogenic change with known treatment or surveillance saving lives”) to broad (“any genomic finding that could inform clinical care”).

> “Actionable would have to be a truly pathogenic change, for which there is a known treatment or surveillance option.”

> “I think of [actionability] really broadly: anything that has the potential, either now or in the future, to modify the course of a disease.”

What Makes Something Actionable

- Strong evidence of pathogenicity and penetrance
- Efficacy, burden, and availability of interventions
- Severity of potential disease
- Potential for life modification, clinical trial enrollment, or psychosocial benefit
- Alignment with patient’s values and personal utility
- Institutional capacity to act on the result

How Actionability is Achieved / Operationalized

- **Framework/Approach Name(s):** No formal named framework; draws on ACMG, CDC Tier One, and other guidelines
- **Methods/Levers:** Variant classification (ClinVar, ClinGen), professional guidelines, institutional policies
- **Operational Steps / Workflow:** Evidence assessment (now vs. later), determination of available interventions
- **Data & Measures:** Variant pathogenicity, penetrance, disease severity, trial data, population-level science
- **Implementation Context:** Varies by health system—resource-rich systems expand scope; resource-limited systems focus on core

> “Because we’re a safety net hospital... we would return results on the CDC Tier 1 conditions... probably within a few weeks.”

> “We don’t want to leave patients hanging, because... reality happens before the ideal does.” — Genetic counselor

Dimensions and Attributes of Actionability (Authors’ Perspective)

- **CL (Clarity):** Yes — clarity of variant classification and intervention pathways emphasized.
- **CR (Contextual Relevance):** Yes — tailored to institutional resources and patient population.
- **FE (Feasibility):** Yes — dependent on institutional capacity and follow-up infrastructure.
- **TI (Timeliness):** Partial — tension between acting now vs. waiting for more evidence.
- **EX (Explainability):** Partial — some emphasis on transparency of evidence and interpretation.
- **GA (Goal Alignment):** Yes — decisions reflect health system priorities and patient benefit.
- **Other Dimensions:** Personal utility, psychosocial impact, marketability (for some institutions).

Theoretical or Conceptual Foundations

- ACMG actionability guidelines and secondary findings lists
- CDC Tier One genomic conditions

- Concepts of clinical validity, clinical utility, and personal utility
- Precision medicine vs. evidence-based medicine paradigms

Indicators or Metrics for Actionability

- Evidence level for pathogenicity/penetrance
- Strength of intervention evidence (RCTs for PCPs; mechanistic plausibility for geneticists)
- Severity of disease outcome
- Institutional capacity metrics (follow-up care rates)

Barriers and Enablers to Actionability

- **Barriers:** Lack of consensus on evidence standards; insufficient infrastructure; provider genetics training
- **Enablers:** Established guidelines (ACMG, CDC Tier One); institutional investment; belief in patient benefit

Relation to Existing Literature

Builds on prior patient-centered studies of actionability by focusing on clinician perspectives; confirms that

Summary

Owens et al. (2023) provide a qualitative investigation into how clinicians conceptualize “actionability” in g

Scores

- **Overall Relevance Score:** 90 — Strong explicit and implicit definitions, systematic features, and clinical
- **Operationalization Score:** 75 — Clear description of decision-making processes and contextual factors

Supporting Quotes from the Paper

- “[My definition of] actionable would have to be a truly pathogenic change... that impacts a lifetime of me
- “Anything that has the potential, either now or in the future, to modify either life choices or medical treatm
- “We don’t want to leave patients hanging... reality happens before the ideal does.” (p. 6)
- “Because we’re a safety net hospital... we would return results on the CDC Tier 1 conditions.” (p. 10)

Actionability References to Other Papers

- Berg et al. (2016) — semiquantitative metric for evaluating clinical actionability
- ACMG SF v3.1 (Miller et al., 2022)

- CDC Tier One Genomics Applications
- Lázaro-Muñoz et al. (2017) — subjective judgments in selecting medically actionable genes
- Kohler et al. (2017) — personal utility in genomic testing