Rethinking Clinical Approaches to Genetic Risk

Joey Mackle

Department of Bioethics | Te Tari Matatika Koiora

Faculty of Medicine – Dunedin | Te Kaupeka Whaoira – Ōtepoti

Key Ideas

- Healthcare practitioners take multiple, often implicit, approaches to family involvement in genetic testing decisions
- Each approach brings about different normative commitments to involving or considering ‘the family’
- Any approach to disclosure decisions ought to reflect the relational nature of genetic risk

Acknowledgements

- This presentation draws on research conducted at the Department of Bioethics, University of Otago under the supervision of Associate Professor Josephine Johnston (Bioethics) and Professor Stephen Robertson (Genetics).
- This work was supported by funding from the Maurice and Phyllis Paykel Trust.
- The Health Research Council of New Zealand supported prior research (HRC Ref 23/971) that laid the groundwork for this project.

hrc nz

University of Otago
ŌTĀKOU WHAKAIHU WAKA

THE MAURICE & PHYLLIS PAYKEL TRUST
MPPT
ESTABLISHED 1973

Motivation

- Prior work on HDGC (HRC Ref 23/971)
- Parents wanting to test their children and being turned away
- Parents interests constructed in opposition to minor's interests
  - Whose should be met?
  - Who is the subject of care in this case?
  - What about in non-paediatric clinical genetics?
  - What obligations does a healthcare practitioner have to relatives of a patient?

What is the role of the family in genetic decision-making?

What kinds of decisions?

What kinds of decisions?

HEALTH PSYCHOLOGY REVIEW
2022, VOL. 16, NO. 3, 412–429
https://doi.org/10.1080/17437199.2021.1980083

Routledge
Taylor & Francis Group

OPEN ACCESS Check for updates

Decision-making about genetic health information among family dyads: a systematic literature review

Wai Ki Law[a], Haley E. Yaremych[b,c], Rebecca A. Ferrer[d], Ebony Richardson [a], Yelena P. Wu[e,f] and Erin Turbitt [a,c]

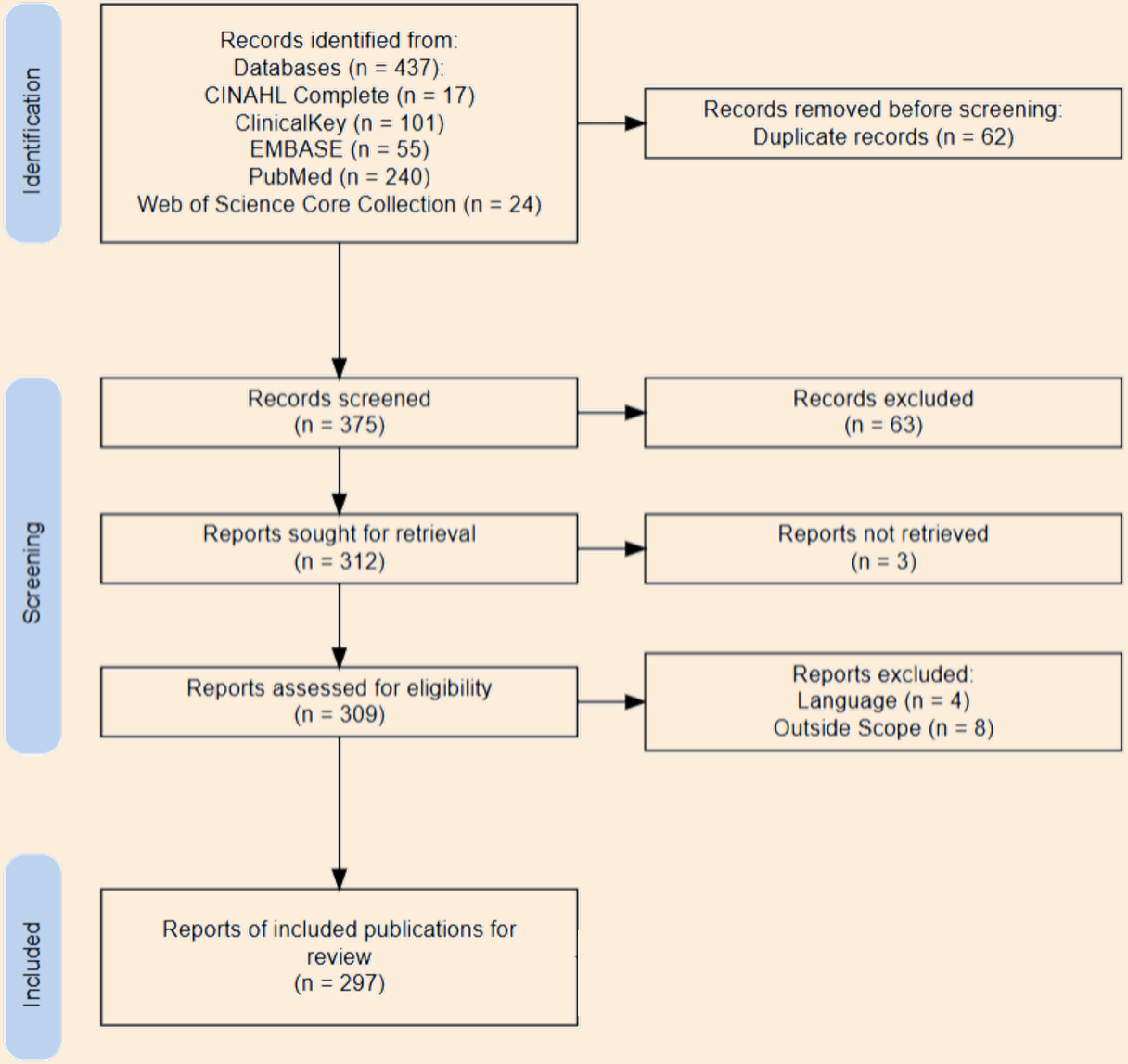
[a]The Discipline of Genetic Counselling, The University of Technology Sydney, Ultimo, Australia; [b]Department of Psychology & Human Development, Vanderbilt University, Nashville, TN, USA; [c]Social and Behavioral Research Branch, National Human Genome Research Institute, Bethesda, MD, USA; [d]Basic Biobehavioral and Psychological Sciences Branch, National Cancer Institute, Bethesda, MD, USA; [e]Department of Dermatology, University of Utah, Salt Lake City, UT, USA; [f]Huntsman Cancer Institute, Salt Lake City, UT, USA

What kinds of decisions?

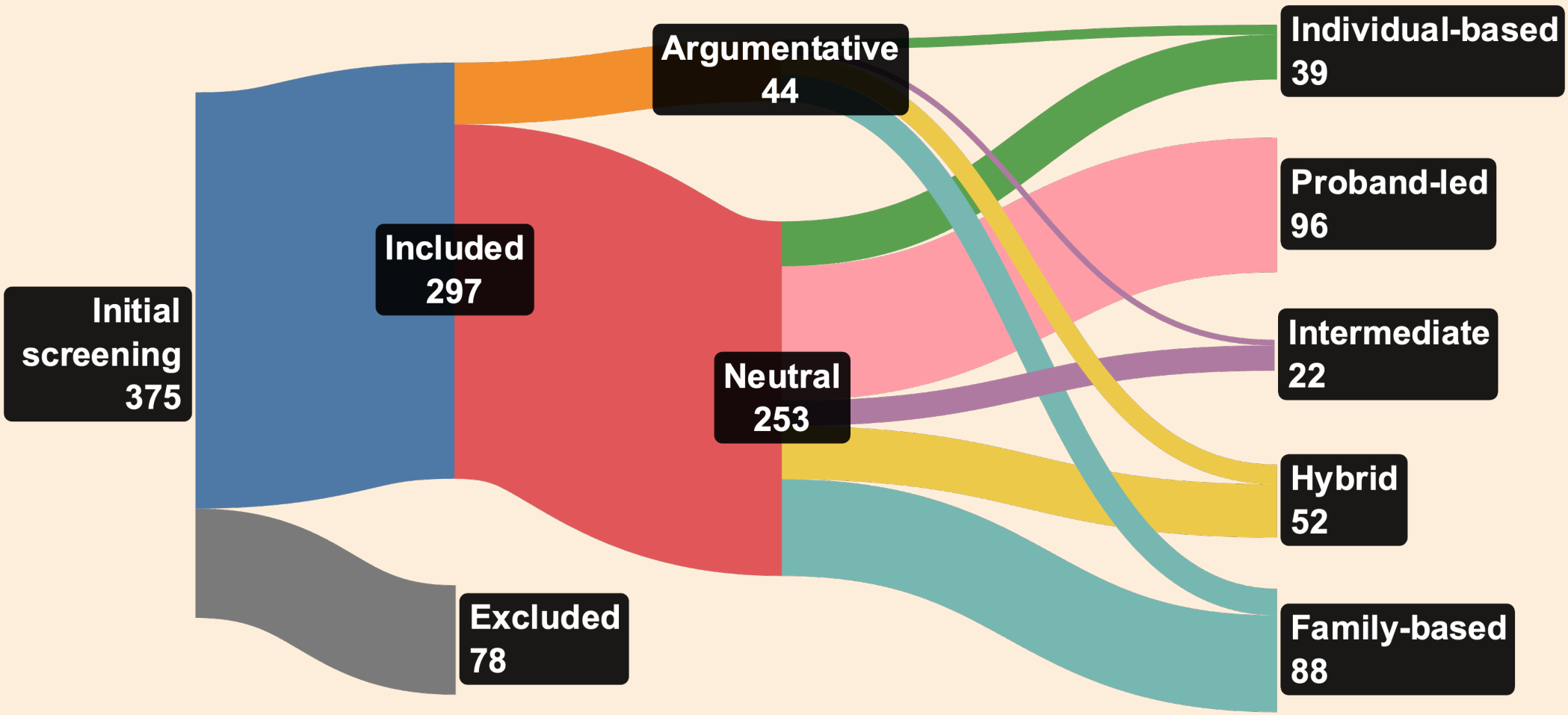
- Decisions about whether to test, and for what
- Decisions about whether and how to act on genetic test results

Literature Review

- Search terms: “genetic”, “family”, “decision-making”, “ethics”
- Inclusion/exclusion criteria
- 437 records across five databases, 375 records screened, 297 reports underwent full-text analysis
    - Method: critical, interpretive, iterative


Identification
Records identified from:
Databases (n = 437):
CINAHL Complete (n = 17)
ClinicalKey (n = 101)
EMBASE (n = 55)
PubMed (n = 240)
Web of Science Core Collection (n = 24)
Records removed before screening:
Duplicate records (n = 62)
Screening
Records screened
(n = 375)
Records excluded
(n = 63)
Reports sought for retrieval
(n = 312)
Reports not retrieved
(n = 3)
Reports assessed for eligibility
(n = 309)
Reports excluded:
Language (n = 4)
Outside Scope (n = 8)
Included
Reports of included publications for review
(n = 297)

Results



Made at SankeyMATIC.com

Key Ideas

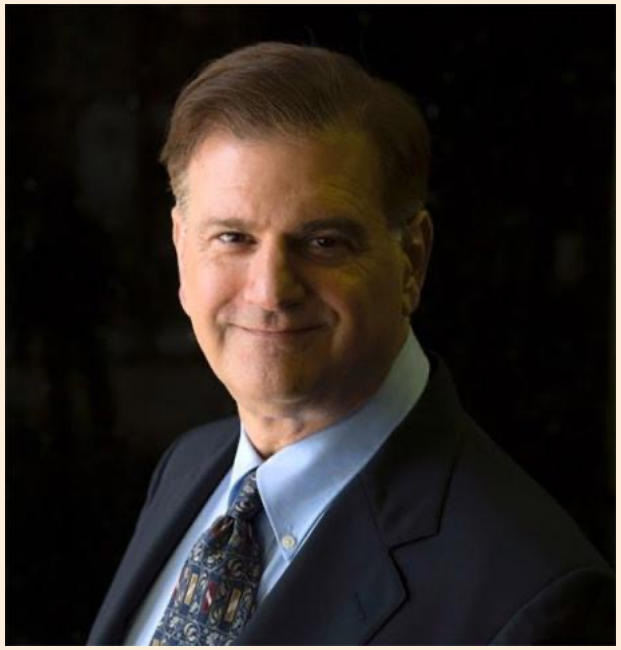
- Healthcare practitioners take multiple, often implicit, approaches to family involvement in genetic testing decisions
- Each approach brings about different normative commitments to involving or considering ‘the family’
- Any approach to disclosure decisions ought to reflect the relational nature of genetic risk

Individual-based Approaches

- Rare in practice; more common in research (relatively)
- Genetic information belongs to the individual
- Consideration of implications for relatives is absent
- Practice example: predictive genetic testing of children for adult-onset conditions
- Subject of care is the individual

Family-based Approaches

- Currently confined to paediatric and reproductive contexts
- Genetic information treated as a shared resource
- Few attempts to extend family-based thinking to other areas of genetics or wider medicine (e.g., family covenant)
- Subject of care is the family unit



David J. Doukas

Jessica W. Berg

Target Article

The Family Covenant and Genetic Testing

David J. Doukas, University of Pennsylvania Jessica W. Berg, Case Western Reserve University

The physician-patient relationship has changed over the last several decades, requiring a systematic reevaluation of the competing demands of patients, physicians, and families. In the era of genetic testing, using a model of patient care known as the *family covenant* may prove effective in accounting for these demands. The family covenant articulates the roles of the physician, patient, and the family prior to genetic testing, as the participants consensually define them. The initial agreement defines the boundaries of autonomy and benefit for all participating family members. The physician may then serve as a facilitator in the relationship, working with all parties in resolving potential conflicts regarding genetic information. The family covenant promotes a fuller discussion of the competing ethical claims that may come to bear after genetic test results are received.

Michael Parker

Anneke Lucassen

ORIGINAL ARTICLE

Concern for families and individuals in clinical genetics

M Parker, A Lucassen

J Med Ethics 2003;**29**:70–73

See end of article for authors' affiliations

Correspondence to: Dr M Parker, The Ethox Centre, Institute of Health Sciences, University of Oxford, Oxford OX3 7LF UK; michael.parker@ethox.ox.ac.uk

Revised version received 22 May 2002
Accepted for publication 9 July 2002

Clinical geneticists are increasingly confronted with ethical tensions between their responsibilities to individual patients and to other family members. This paper considers the ethical implications of a "familial" conception of the clinical genetics role. It argues that dogmatic adherence to either the familial or to the individualistic conception of clinical genetics has the potential to lead to significant harms and to fail to take important obligations seriously.
Geneticists are likely to continue to be required to make moral judgments in the resolution of such tensions and may find it useful to have access to ethics training and support.

ORIGINAL ARTICLE

Concern for families and individuals in clinical genetics

M Parker, A Lucassen

J Med Ethics 2003;**29**:70–73

See end of article for authors' affiliations

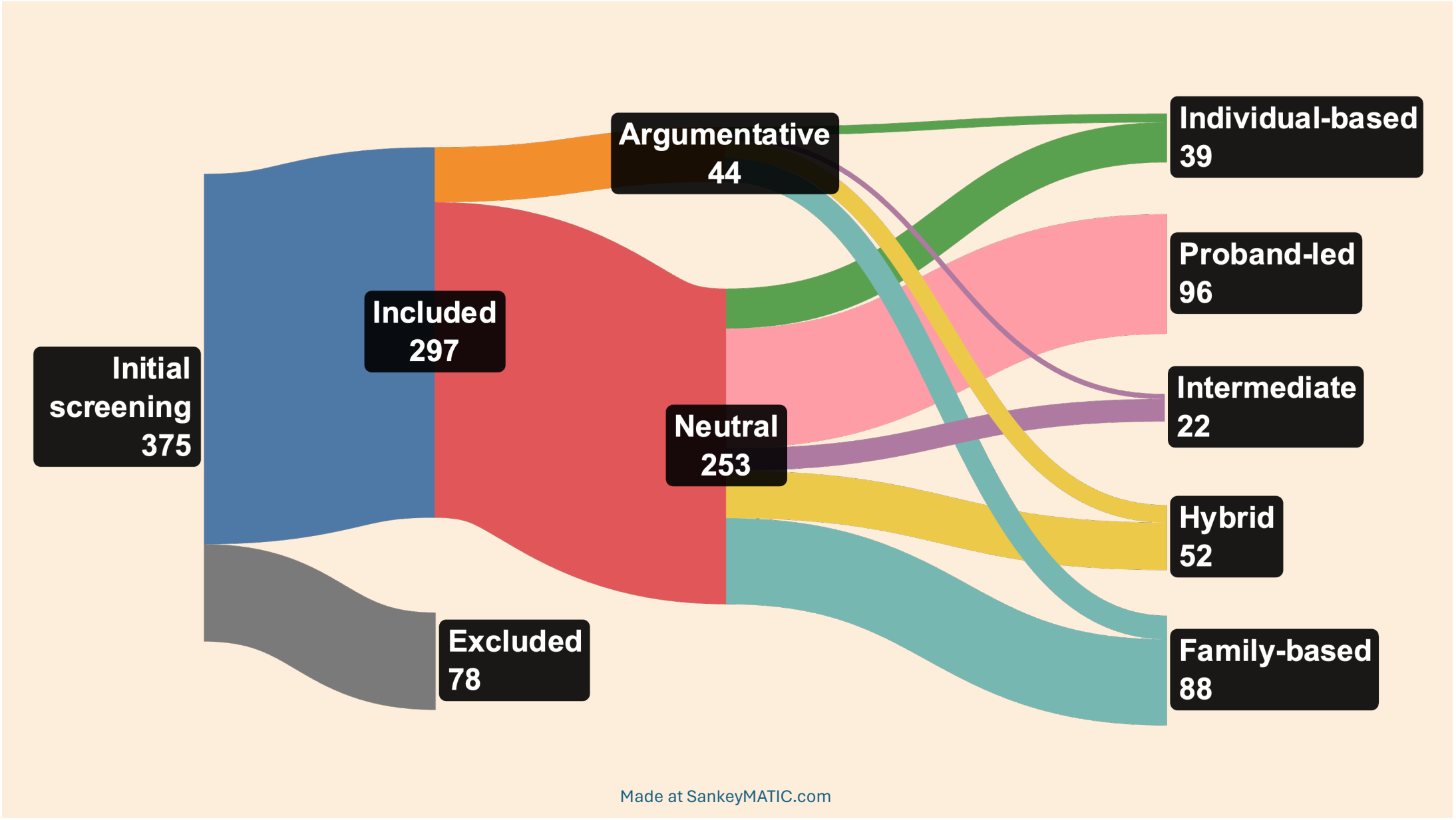
Correspondence to: Dr M Parker, The Ethox Centre, Institute of Health Sciences, University of Oxford, Oxford OX3 7LF UK; michael.parker@ethox.ox.ac.uk

Revised version received 22 May 2002
Accepted for publication 9 July 2002

Clinical geneticists are increasingly confronted with ethical tensions between their responsibilities to individual patients and to other family members. This paper considers the ethical implications of a "familial" conception of the clinical genetics role. It argues that dogmatic adherence to either the familial or to the individualistic conception of clinical genetics has the potential to lead to significant harms and to fail to take important obligations seriously. Geneticists are likely to continue to be required to make moral judgments in the resolution of such tensions and may find it useful to have access to ethics training and support.

Middle-ground Approaches

- Proband-led (dominant)
- Intermediate
- Hybrid


Initial screening 375
Included 297
Excluded 78
Argumentative 44
Neutral 253
Individual-based 39
Proband-led 96
Intermediate 22
Hybrid 52
Family-based 88
Made at SankeyMATIC.com

Key Ideas

- Healthcare practitioners take multiple, often implicit, approaches to family involvement in genetic testing decisions
- Each approach brings about different normative commitments to involving or considering 'the family'
- Any approach to disclosure decisions ought to reflect the relational nature of genetic risk

What is the role of the family in genetic decision-making?

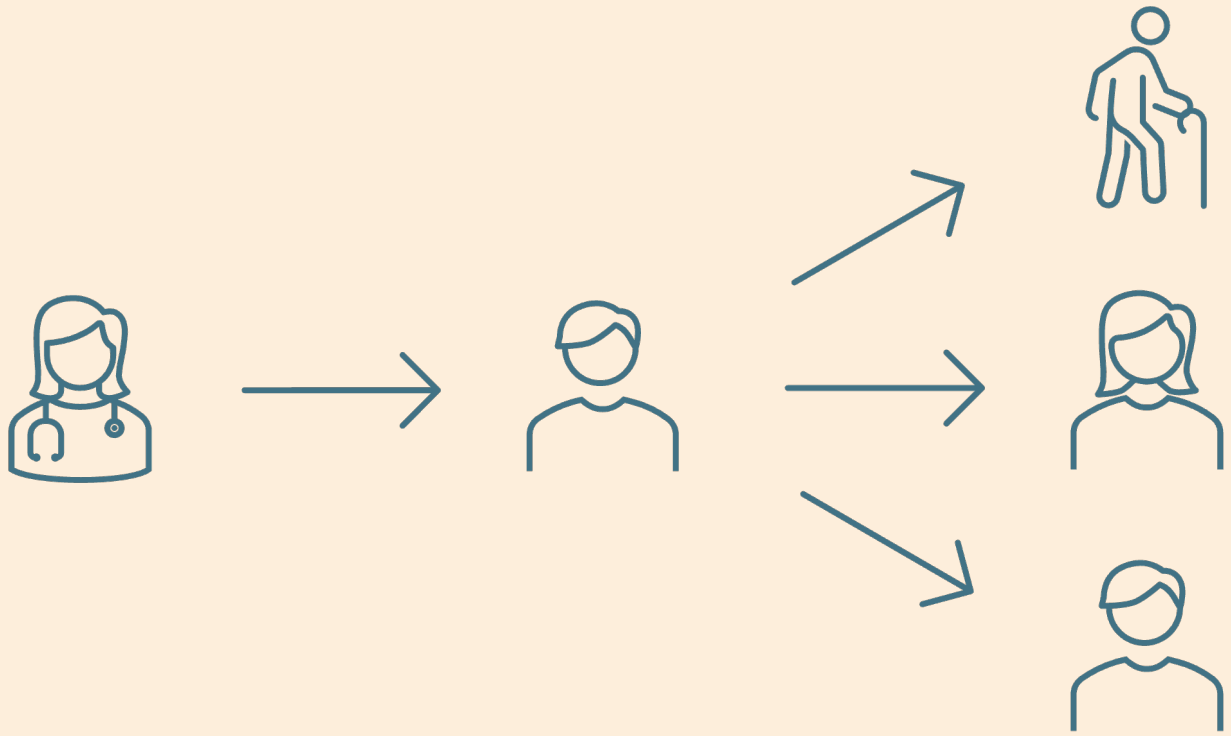
What kinds of decisions?

- Decisions about whether to test, and for what
- Decisions about whether and how to act on genetic test results

What kinds of decisions?

- Decisions about whether to test, and for what
- **Decisions about whether and how to act on genetic test results**

Proband-led Approach to Disclosure



THE AMERICAN JOURNAL OF BIOETHICS
2025, VOL. 25, NO. 10, 9–21
https://doi.org/10.1080/15265161.2024.2388719

Taylor & Francis
Taylor & Francis Group

TARGET ARTICLE

Breaching Confidentiality in Genetic and Non-Genetic Cases: Two Problematic Distinctions

Madison K. Kilbride

University of Utah

ABSTRACT

Ethical questions about confidentiality arise when patients refuse to inform relatives who are at risk of a genetic condition. Specifically, healthcare providers may struggle with the permissibility of breaching confidentiality to warn patients' at-risk relatives. In exploring this issue, several authors have converged around the idea that genetic cases differ from non-genetic cases (e.g., involving a threat of violence or the spread of an infectious disease) along two related dimensions: (1) In genetic cases, the risk of harm is already present in an at-risk third party, whereas in non-genetic cases, it is not; and (2) In genetic cases, the patient does not create a risk of harm to a third party, whereas in non-genetic cases, the patient does. I argue that these distinctions do not exclusively differentiate genetic from non-genetic cases and should not bear on the permissibility of breaching confidentiality. Instead, such determinations should be based on other considerations.

KEYWORDS

Confidentiality and privacy; genetics (clinical); philosophy; professional ethics; professional-patient relationship

Necessary Conditions for Breaching Confidentiality

A. At-risk party is identifiable

B. Information has significant utility

C. Risk of harm is serious

D. Disclosure does not endanger the patient

E. Potential harm is imminent

F. Unlikely the at-risk party will learn of their risk from another source

G. (Risk involves a public health threat)

Necessary Conditions for Breaching Confidentiality

A. At-risk party is identifiable

B. Information has significant utility

C. Risk of harm is serious

D. Disclosure does not endanger the patient

E. Potential harm is imminent

F. Unlikely the at-risk party will learn of their risk from another source

G. (Risk involves a public health threat)

THE AMERICAN JOURNAL OF BIOETHICS
2025, VOL. 25, NO. 10, 37–40
https://doi.org/10.1080/15265161.2025.2543741

Taylor & Francis
Taylor & Francis Group

OPEN PEER COMMENTARIES

Structural Features of Genetic Risk and the Need for a Relational Approach to Disclosure

Joey Mackle, Stephen P. Robertson and Josephine Johnston

University of Otago

In clinical medicine, the prevailing ethical and legal frameworks give primacy to individual autonomy and patient confidentiality. Genetic information is typically treated as belonging to the individual patient, even when it carries significant implications for biologically-related family members. The responsibility—and burden—for initiating disclosure of risk information to relatives ordinarily lies with the patient, with limited scope for professional intervention. Kilbride's (2025) paper fits squarely within this individual-centered approach. While consistent with law and conventional medical ethics, it sits uneasily with the relational structure of most genetic risk, which is inherently shared and significant for others beyond the individ- and risks to genetic relatives is real and present from the outset.

In response to the dominant individual-centered approach, many scholars have proposed relational alternatives (Doukas and Berg 2001; Parker and Lucassen 2004; Weller, Lyle, and Lucassen 2022). These relational approaches vary in how they conceptualize autonomy, responsibility, confidentiality, and the unit of care in genetics, but they recognize that individuals are embedded within networks of relationships and in particular families, and reflect how these relationships shape how risk is experienced, shared, and responded to (Lillie et al. 2022). We understand that a biological connection to relatives does not

THE AMERICAN JOURNAL OF BIOETHICS
2025, VOL. 25, NO. 10, 37–40
https://doi.org/10.1080/15265161.2025.2543741

Taylor & Francis
Taylor & Francis Group

OPEN PEER COMMENTARIES

Structural Features of Genetic Risk and the Need for a Relational Approach to Disclosure

Joey Mackle, Stephen P. Robertson and Josephine Johnston

University of Otago

In clinical medicine, the prevailing ethical and legal frameworks give primacy to individual autonomy and patient confidentiality. Genetic information is typically treated as belonging to the individual patient, even when it carries significant implications for biologically-related family members. The responsibility—and burden—for initiating disclosure of risk information to relatives ordinarily lies with the patient, with limited scope for professional intervention. Kilbride's (2025) paper fits squarely within this individual-centered approach. While consistent with law and conventional medical ethics, it sits uneasily with the relational structure of most genetic risk, which is inherently shared and significant for others beyond the individ- and risks to genetic relatives is real and present from the outset.

In response to the dominant individual-centered approach, many scholars have proposed relational alternatives (Doukas and Berg 2001; Parker and Lucassen 2004; Weller, Lyle, and Lucassen 2022). These relational approaches vary in how they conceptualize autonomy, responsibility, confidentiality, and the unit of care in genetics, but they recognize that individuals are embedded within networks of relationships and in particular families, and reflect how these relationships shape how risk is experienced, shared, and responded to (Lillie et al. 2022). We understand that a biological connection to relatives does not

Genetic Risk Is Inherently Relational

- Familial
- Probabilistic
- Transgenerational

Necessary Conditions for Breaching Confidentiality

A. At-risk party is identifiable

B. Information has significant utility

C. Risk of harm is serious

D. Disclosure does not endanger the patient

E. Potential harm is imminent

F. Unlikely the at-risk party will learn of their risk from another source

G. (Risk involves a public health threat)

Key Ideas

- Healthcare practitioners take multiple, often implicit, approaches to family involvement in genetic testing decisions
- Each approach brings about different normative commitments to involving or considering ‘the family’
- Any approach to disclosure decisions ought to reflect the relational nature of genetic risk

Thank You

joey.mackle@postgrad.otago.ac.nz