Making the Decision to Participate or Decline Predictive Genetic Testing

Dr. April Pike and Dr. Fern Brunger

Memorial University of Newfoundland
School of Nursing
Canada
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MEMORIAL UNIVERSITY
School of Nursing

Overview of Presentation

Introduction

Background of ARVC

Review of the Literature

Overview of Study

Discussion of Findings

Recommendations

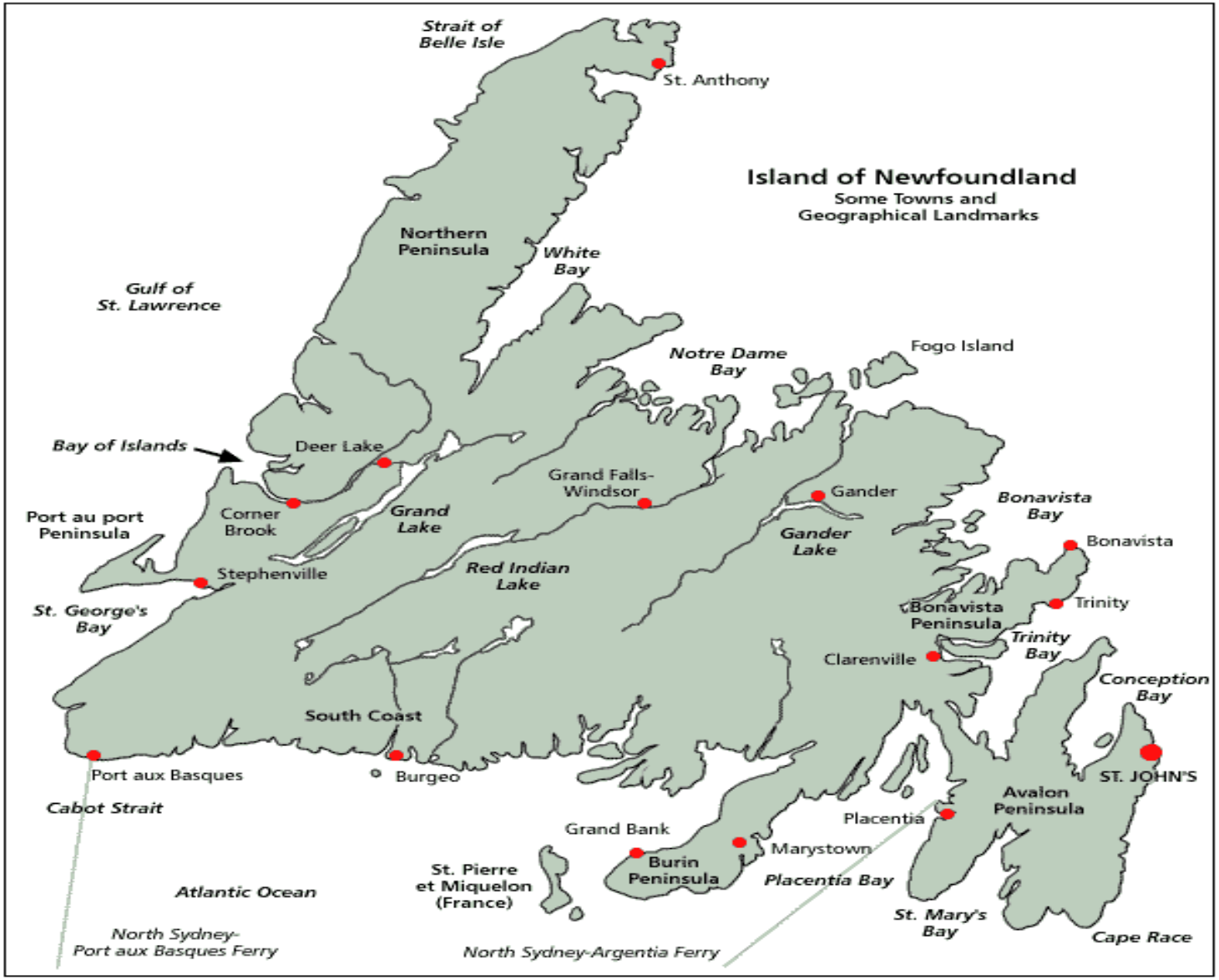
Questions

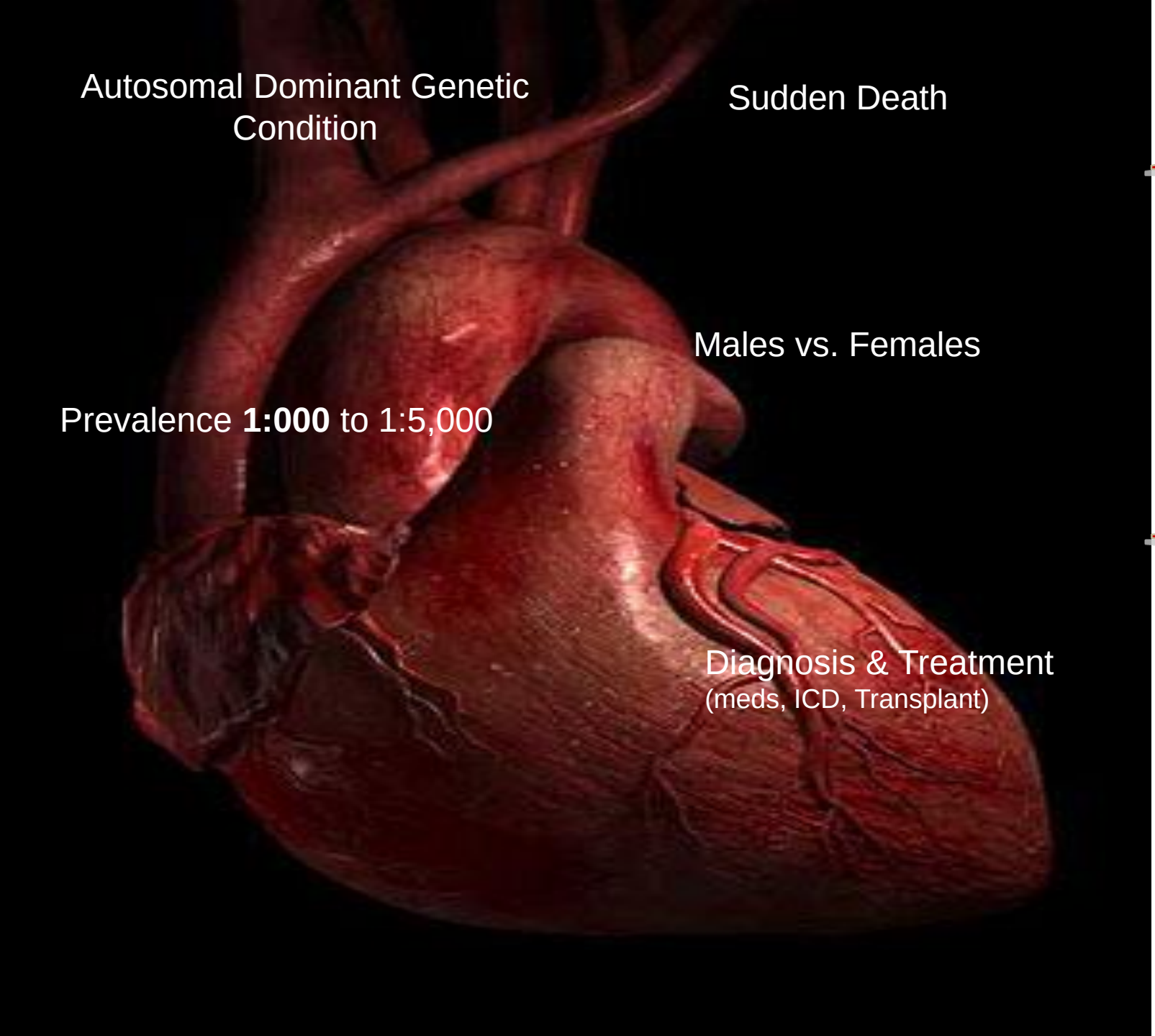
What is Predictive Genetic Testing

PGT foretells the health care outcomes or lifetime risk of acquiring a disease of an otherwise healthy, asymptomatic person

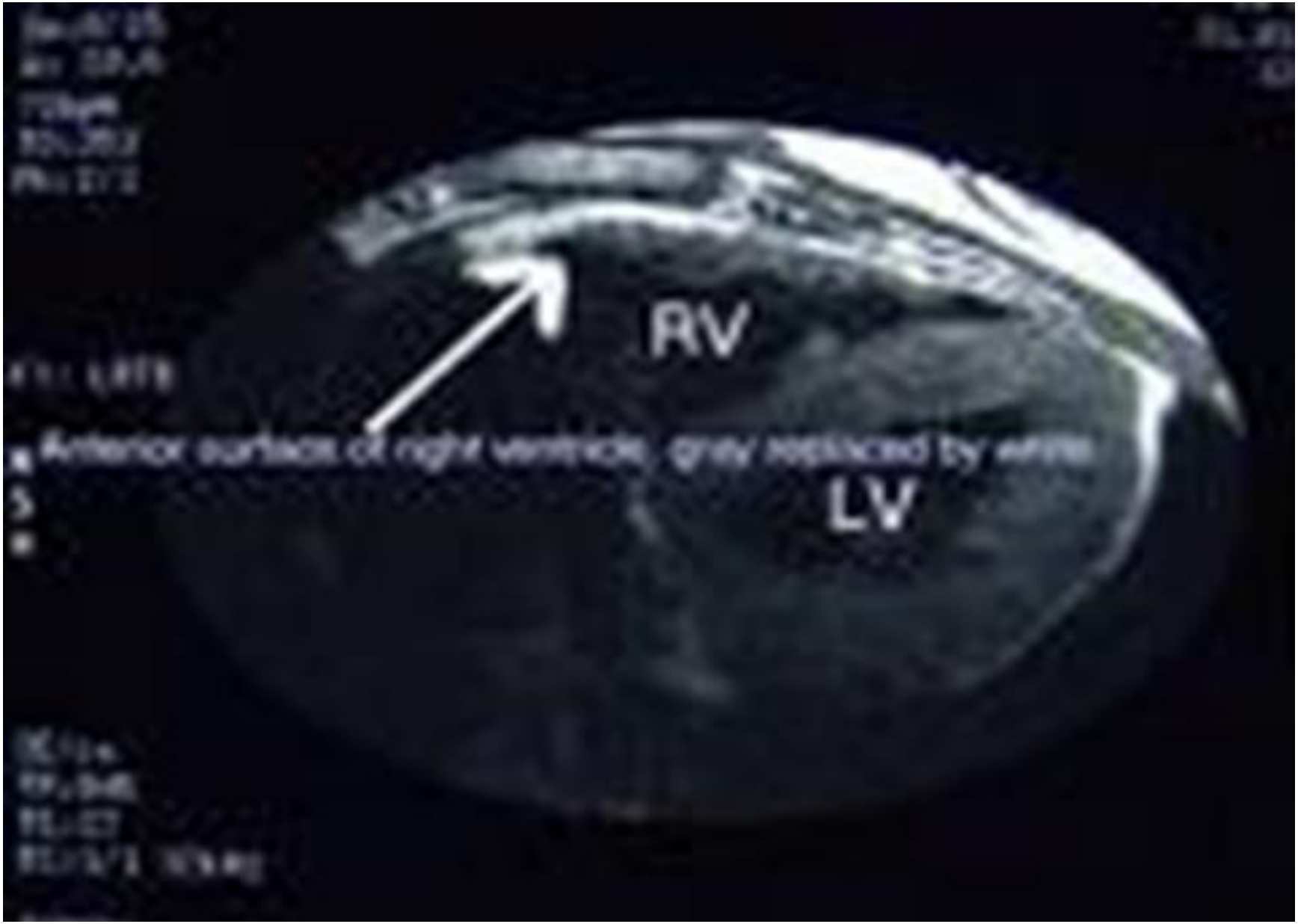

NORTH
Northwest
Northeast
WEST
EAST
Southwest
Southeast
SOUTH
Arctic Ocean
YUKON
Whitehorse
NORTHWEST TERRITORIES
Yellowknife
NUNAVUT
Iqaluit
Hudson Bay
NEWFOUNDLAND & LABRADOR
St. John's
BRITISH COLUMBIA
ALBERTA
Edmonton
SASKA-TCHEWAN
Regina
MANITOBA
Winnipeg
QUÉBEC
Québec City
ONTARIO
Ottawa
PRINCE EDWARD ISLAND
Charlottetown
Fredericton
NEW BRUNSWICK
Halifax
NOVA SCOTIA
Atlantic Ocean
Pacific Ocean
Victoria
Lake Superior
Lake Huron
Lake Michigan
Lake Ontario
Lake Erie

Where is Newfoundland




Autosomal Dominant Genetic Condition
Sudden Death
Males vs. Females
Prevalence **1:000** to 1:5,000
Diagnosis & Treatment
(meds, ICD, Transplant)

ARVC


RV
Anterior surface of right ventricle, grey replaced by white
LV

ARVC

1970s
Family History of Sudden Cardiac Death • first documented

1983
- One NL person who had dissection of right ventricular free wall
(N=8 participants)*

1988
First ARVC family identified •

1990
- Human Genome Project

1993
ARVC Blood samples destroyed •

1994
- US research team enters NL
(N= 4 participants)*

1997
NL ARVC research team starts •

1998
- US research team discovery of ARVC to chromosome 3p25 in NL population
- International Cardiomyopathy research group
- ARVC Haploytpe analysis for NL ongoing in Germany and London

2004
Cardiomyopathy clinic, • NL Eastern Health

2005
- ARVC Haploytpe analysis started in NL under Dr. Young
(N= 13 participants)*
- ICD as a proven treatment for ARVC

2006
AMGGI Funding ($ 9.3 million) • Dr Young's laboratory for ARVC

2007
- Discovery ARVC gene (TMEM43) in NL
(N=4 participants)*

General Literature on Genetics

Quantitative: Genetic testing does not cause long term psychosocial distress *(Collins et al., 2007; Heshka et al., 2008).*

Qualitative: Individuals living in families at risk for a genetic condition do experience psychosocial distress *(Andersen et al., 2008; Duncan et al.,,2008; McAllister, 2002,2003; Davies et al., 2007, Howell et al., 2006; Sobel & Cowen, 2003).*

One's perception of being at risk for a genetic condition and response **(Including the decision to have PGT)** is related to the meanings assigned to the specific factors in one's life *(Cox, 2003, Etchegary 2005,2009; d'Agincourt-Canning, 2005; McAllister, 2003).*

Gap in Literature

"Transient Nature of Risk Assessment"

How **meanings assigned to risk** and one's **health care decisions** are shaped and reshaped alongside the process of **NEW** gene discovery

Little literature examining how people make decisions related to PGT

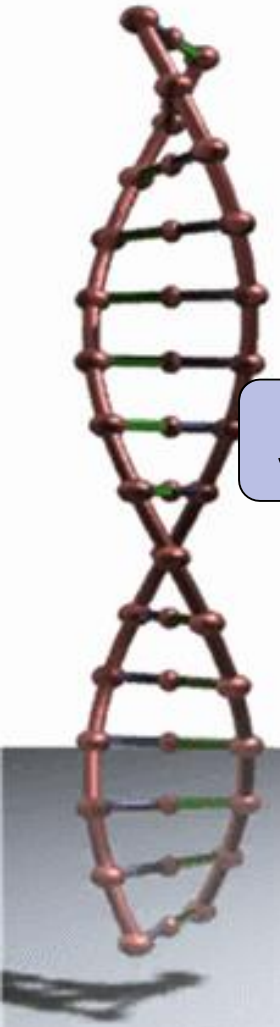
No Qualitative Research on ARVC

Substantive Theory and Model

Purpose of Study

To understand the experiences of individuals as they make the decision to participate in (or decline) genetic testing for ARVC at different phases of scientific discovery.

Grounded Theory (Glaser & Strauss 1967)



Substantive Theory

- **CONSTRUCTS (3)**
- **CATEGORIES**
- **PROPERTIES**
- **“INDICATORS”**

Individual Interviews & Focus Groups N=30
Ethical Approval

Findings

Two approaches to making the decision to engage in genetic testing

(a) develops gradually over time

(b) happens so quickly that it is felt as a fait accompli.

Approach taken by the participants were influenced by 6 factors

(1)available & relevant PGT
(2) numerous losses
(3) onset of physical signs and symptoms

(4) gender
(5) relational responsibility
(6) family support

Available & Relevant PGT

“ They [researchers] just wanted to do some testing, and it wouldn’t affect our lives.” (1980’s)

“ I started to think about the problems of knowing and not knowing…what if you don’t have life insurance…what happens if there is something wrong?”(1998)

“ I’m going to get testing when I’m 40.” (2005)

“ I wanted to know. I don’t remember any stress making the decision to get it done. It wasn’t a big decision.” (1980’s-1998-2007)

Losses

"My third brother was 43 when he passed away suddenly…In the back of my mind I was thinking this could happen to me."(1980s-1998-2007)

"I never really knew about ARVC until recently. Mother kept studying it, finding out what she could." (2007)

You're kind of wondering about it [reason for deaths], so you don't acknowledge it. It's like being in a house and you don't know if this place is haunted, but there's weird stuff going on so I won't look (1998)

Physical Signs/Symptoms & Gender

" I had some pain in my chest and I went to see what was going on." (2007)

"I came from a generation of all girls. ARVC was never thought of as a problem because most females don't get this disease. There were no boys in the family until I had my son. Next thing you know he's 30 years old and drops down dead. No one ever knew that it could be passed on through the women. It was only then we started to realize that ARVC [could] be passed down through the women in the family to the men." (1980s,1998,2007)

Relational Responsibility

" I have two daughters. I have to know for them."(1998)

" I want my son tested but I don't want him tested. I'd like to bury my head in the sand , but for his sake I can't."(2005)

" I didn't really want to be tested…I'd rather just go through life not knowing and not know at all." (2005).

Family Support

“ I can’t let you test [son] because my husband is not here, and I could not deal with it at the time.” (2007)

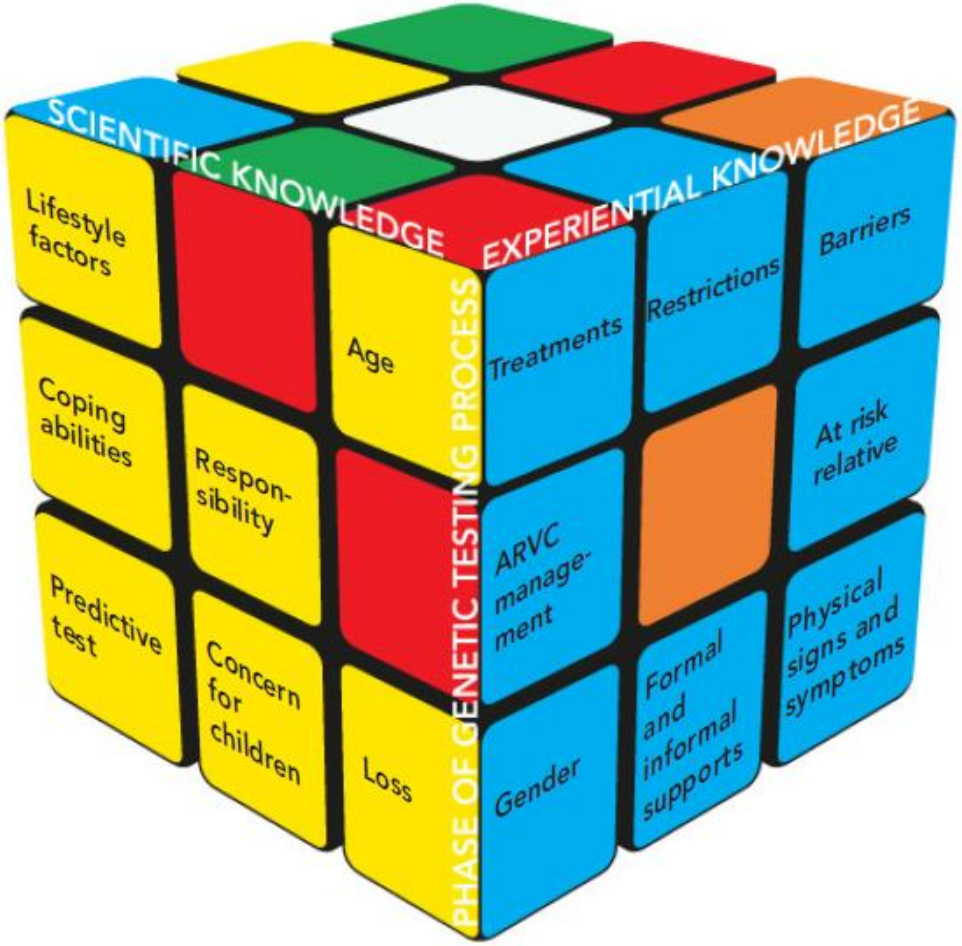
“Eight of us went in and had blood work.” (1998)

Key Message

"The concept of risk is pragmatic, fluid and transient. The meaning assigned to being at risk & the decision to engage in PGT or not evolves from the juxtaposing of scientific knowledge against experiential knowledge and phase of the genetic testing process.

It is out of this intersection of these three contextual dimensions (and the 6 identified factors) that decisions about PGT are made"

The Shifting Faces of Risk



Key Message

This decision develops gradually over time or is fait accompli.

The decision evolves with each new experience and gene discovery.

These pathways can merge momentarily or change completely and are contingent on the meaning assigned to being at risk for ARVC

Recommendation

The creation of a relational space within which to provide psychological counselling and assessment for the six identified factors that shape the decision to engage in predictive genetic testing.

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ORIGINAL RESEARCH

Making the Decision to Participate in Predictive Genetic Testing for Arrhythmogenic Right Ventricular Cardiomyopathy

April Manuel · Fern Brunger

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Abstract This paper describes the experience of predictive genetic testing for Arrhythmogenic Right Ventricular Cardiomyopathy in the context of novel gene discovery. Two approaches to making the decision to engage in genetic testing were apparent: the decision to be tested either (a) develops gradually over time or (b) happens so quickly that it is felt as a "*fait accompli*." Six key factors that influenced the particular approach taken by the participants were identified: (1) scientific process—available and relevant predictive genetic test; (2) numerous losses or deaths within the family; (3) physical signs and symptoms of disease; (4) gender; (5) sense of relational responsibility or moral obligation to other family members; and (6) family support. This study found that at risk individuals juxtapose scientific knowledge against their experiential knowledge and the six identified factors in order to make the decision to participate in genetic testing. Recommendations include the creation of a relational space within which to provide psychological counselling and assessment for the six identified factors that shape the decision to engage in predictive genetic testing.

Keywords Arrhythmogenic right ventricular cardiomyopathy · Risk · Psycho-social factors · Decision-making · Predictive genetic testing

A. Manuel (✉)
Faculty of Nursing, Memorial University, 300 Prince Phillip Drive, St. John's, Newfoundland and Labrador, Canada A1B 3V6
e-mail: admanuel@mun.ca

F. Brunger
Division of Community Health and Humanities, Faculty of Medicine, Memorial University, St. John's, Newfoundland and Labrador, Canada A1B 3V6

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Introduction

The Human Genome Project (HGP) led to a proliferation of predictive genetic tests for a wide range of hereditary conditions. In its wake, social scientists have been examining how individuals at risk for a genetic condition make the decision to participate in genetic testing. This article reports on a segment of a larger study that examined how individuals assign meaning to being at risk for Arrhythmogenic Right Ventricular Cardiomyopathy (ARVC) in the Canadian province of Newfoundland and Labrador (NL) (Manuel 2013). That study focused on how individual understandings of risk in relation to ARVC are constructed out of the intersection of scientific knowledge (that is, biomedical models of inheritance and disease causation and genetic knowledge about ARVC), experiential knowledge (the cumulative knowledge of everyday life, such as a family history of numerous deaths to heart disease, presence of disease symptoms, gender), and phase of the genetic testing process (pre, during or post-testing). The study captured participants' accounts of their experiences of being "at risk" and making the decision to undergo genetic testing, in relation to the evolving science of ARVC genetics, including clinical testing (1980s), availability of haplotype genetic testing (1998), and refinement of testing with the introduction of a definitive predictive genetic test (2007).

This paper focuses on how individuals in the pre-testing phase make the decision to engage in predictive genetic testing. Specifically, it examines how decisions are made out of the intersection and mutual transformation of experiential knowledge and scientific knowledge. This study is unique in that it examines how personal meanings of risk are shaped by, but also reshape, the science of novel gene discovery.

ARVC is an autosomal dominant heart condition that primarily affects young males (Hodgkinson et al. 2009). Although women can have ARVC, they experience symptoms to a lesser degree and later in life (Hodgkinson et al. 2012).

Thank You

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Individuals & Families Living with ARVC

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