Quantifying The Value Of Knowing: Measuring Patient Community Preferences For Genomic Testing In Rare Diseases

RWD182

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Background

Genomic testing can have life-changing impacts for the rare disease community. However, HTA assessment can be challenging due to the lack of evidence around how people value different types of genomic testing benefits, including **health benefits** (clinical benefits & diagnostic benefits) and **non-health benefits** (personal benefits - also referred to as **"the value of knowing"**, and research benefits). For example, Australia's Medical Services Advisory Committee have acknowledged that it would be desirable for the societal value placed on the value of knowing to be captured more formally (Norris et al., 2022). The present study was conducted to fill this evidence gap in partnerships with decision-makers, academics and patient representatives.

Objectives

The Key research questions to answer were:

1. How do patients and carers who are impacted by rare diseases value the health and non-health benefits of genomic testing, including the value of knowing?
2. What preferences do patients and carers have about data sharing and associated risk-benefit trade-offs?
3. How do disease background and demographic characteristics influence preferences?

Methods

Discrete Choice Experiment (DCE)

An online survey, including **two Discrete Choice Experiments (DCE),** was disseminated to **adult patients and carers impacted by genetic rare diseases in Australia.**

In the DCEs, participants were shown a series of choice scenarios with different **hypothetical testing alternatives** (see example scenarios). in each scenario, participants evaluated the tests and **chose their most preferred option or 'opted out'**. Attribute levels varied between each scenario according to a statistical design.

A staged co-design approach was taken to develop the DCE:

- *Rapid literature review*
- *Collaborative instrument development with multiple rounds of discussion and feedback*
- *Community consultation (N=18 rare disease patient group leaders)*
- *Cognitive interviews (N=3 rare disease patient group leaders)*

This study recieved ethics approval from Bellberry Human Ethics Committee Australia

DCE1 focused on different benefits/outcomes of genomic tests

Attribute	Genomic Test A	Genomic Test B
Chance of diagnostic benefits	5 out of 10 will receive diagnostic benefits (50%)	7 out of 10 will receive diagnostic benefits (70%)
Chance of clinical benefits	7 out of 10 will receive clinical benefits (70%)	5 out of 10 will receive clinical benefits (50%)
Chance of personal benefits	3 out of 10 will receive personal benefits (30%)	3 out of 10 will receive personal benefits (30%)
Links to research	No	Yes
Wait time	No wait	1 month
Out-of-pocket-costs	$300	$900
I would choose	Genomic Test A	Genomic Test B

Example scenario from DCE1

DCE2 focused on data sharing and storage considerations

Attribute	Genomic Test A	Genomic Test B
Whether secondary findings are shared with you	An expert panel decides what is shared with you	An expert panel decides what is shared with you
Whether findings are shared with researchers	Requirement for data to be shared with researchers	Requirement for data to be shared with researchers
Whether findings are shared with life insurers	No requirement for data to be shared with life insurers	Requirement for data to be shared with life insurers
Who stores the data	Government organisation	Not-for-profit organisation
Where the data is stored	Overseas	Australia
Wait time	9 months	9 months
Out-of-pocket-costs	$50	$600
I would choose	Genomic Test A	Genomic Test B

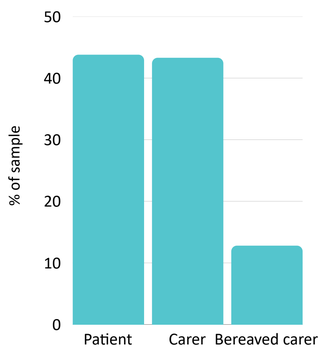
Example scenario from DCE2

In addition to the two DCEs, the survey included questions on disease history, treatment experience, previous genetic/genomic testing, and quality of life (patients only) via the EQ-5D-5L.

Results

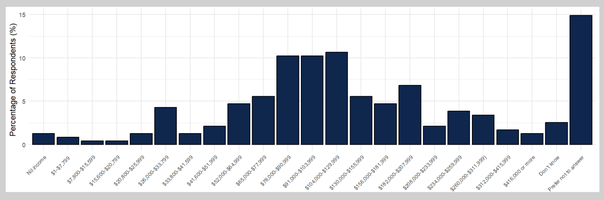
Sample characteristics

- **Total sample:** 235 participants
- **Recruitment:** primarily led by Rare Voices Australia (RVA) who shared the study information with their network of community partners.
- **Secondary recruitment pathway:** specialist healthcare panel company, PureProfile
- **Median survey time:** 28.7 minutes.



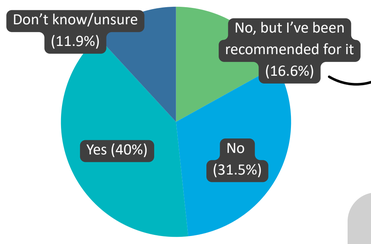
Demographics	N (%)
Living or caring for someone with rare cancer (vs non cancer)	51.5%
Female	57.4%
Metro/city area	70.6%
Working full-time	46.0%
50 years or older	47.2%
Median EQ-5D-5L score (patients only; n=103)	0.795

Annual household income



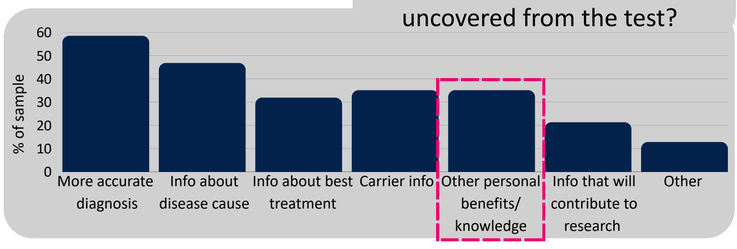
Genomic testing experience

Have you/the person you care for undergone genomic testing?

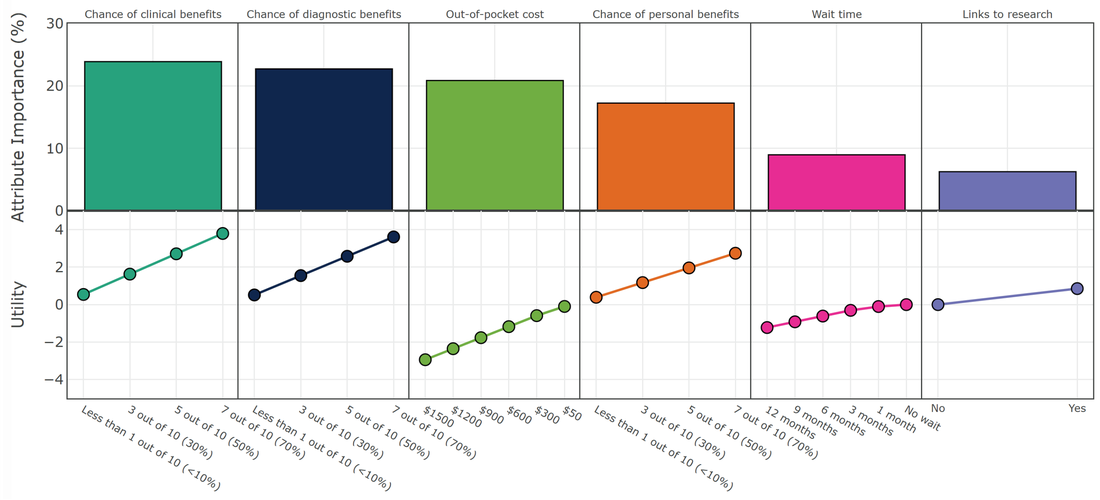


The most common reason people have not received genomic testing despite being recommended for it is **COST** (reported by 53.8%) $

What information was uncovered from the test?



DCE1- Mixed Logit Model (MLM)



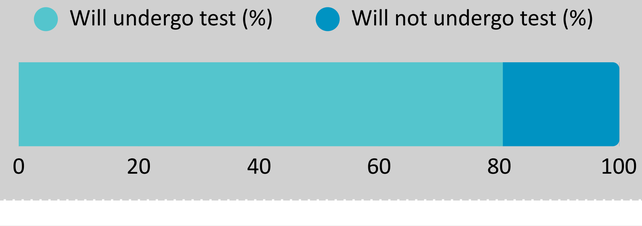
In DCE1, the most important attribute was the **chance of clinical benefits,** followed closely by the **chance of diagnostic benefits, out-of-pocket costs,** and **personal benefits.**

The model picked up **preference heterogeneity**

- e.g., chance of personal benefits was relativley more important to carers than to patients (19% vs 15% attribute importance); out of pocket costs was relativley more important to those under 50 (24.6%) than those 50 or over (17.8%), as well as those with children (24.2%) compared to those without (18.5%) (**see QR code for utility paramaters)**

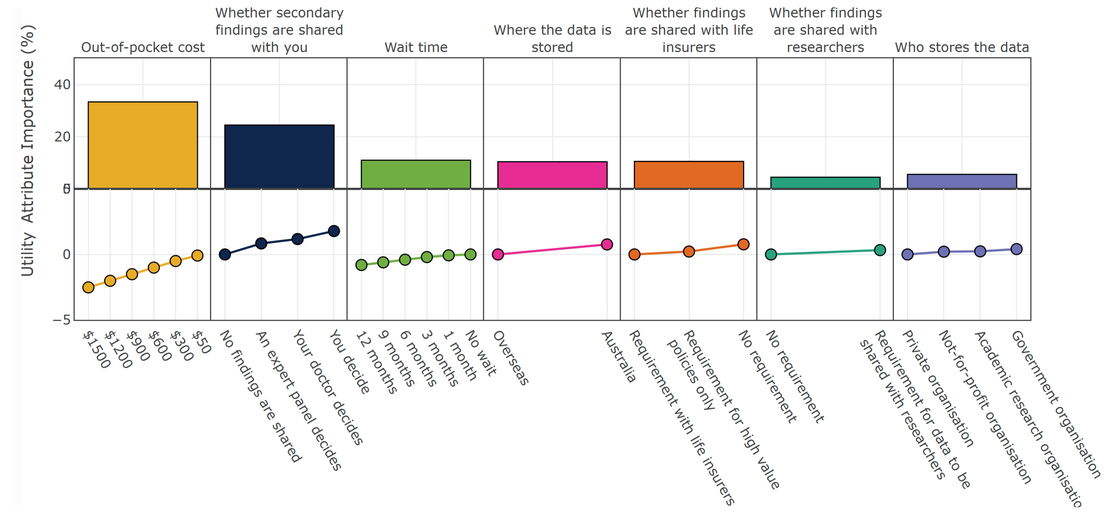
Chance of diagnostic benefits	10%
Chance of clinical benefits	10%
Chance of personal benefits	70%
Links to research	No
Wait time	12 months
Out-of-pocket-costs	$800

Community Uptake: Hypothetical Scenario



If there is a **low chance of receiving direct medical outcomes**, but a **high chance of receiving personal benefits**, uptake is estimated at **80.6%**, with participants willing to wait 12 months and pay $800 OOP

DCE2 - Mixed Logit Model (MLM)



In DCE2, the most important attribute was **out-of-pocket costs,** followed by **whether secondary findings are shared with you.**

Preferences for data storage and sharing are:

- for **you** to decide what secondary findings are shared wth you
- for data to be stored in **Australia**
- for there to be **no** requirement to share findings with **life insurers**
- for there to be requirements to share data with **researchers**
- for data be stored by a **government organisation**

The model for DCE2 also picked up preference hetergeneity which can be explored via the online dashboard (see QR code).

Conclusion

While clinical and diagnostic outcomes were most important to participants, most would still opt for testing with minimal chance of these health benefits if chance of personal benefits were high—**highlighting the importance of "the value of knowing" to the rare disease community, and the value of information in general**. This is further reflected in participants' strong preference to access secondary findings and choose for themselves which secondary findings are shared. Despite a high desire for genomic testing, findings also suggest **cost remains a significant barrier** when public reimbursement is unavailable. These preferences can **enhance HTA decision-making** around the value of new and existing genomic testing technologies, from the perspective of people impacted by rare diseases.

Scan this QR code to view a Decision Support System (DSS) or 'dashboard' that has been developed to support the visualisation of study results. For optimal viewing, please use a laptop or large tablet device.

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Norris S, Belcher A, Howard K, Ward RL. Evaluating genetic and genomic tests for heritable conditions in Australia: lessons learnt from health technology assessments. J Community Genet. 2022;13(5):503-522. doi:10.1007/s12687-021-00551-2