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THE UNIVERSITY *of* EDINBURGH

Paper

# Human-Precision Medicine Interaction: Public Perceptions of **Polygenic Risk Score** for Genetic Health Prediction



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# Introduction | Precision Medicine (PM)



“one-drug-fits-all”



with other factors

personalised treatments

# Introduction | Precision Medicine (PM)

- **Precision Medicine (PM)** is an innovative approach to healthcare that customises and personalises medical treatment to the characteristics of each individual
- PM applications heavily rely on the ability to effectively process, interpret, and communicate the insights derived from complex, multi-dimensional healthcare datasets
  - mostly biological data with **genetics** in a central position
- PM involves significant computational challenges – with open questions as to how these advanced PM technologies **can be integrated into real-world healthcare practices**
- Here, HCI could play a crucial role

## U.S. Precision Medicine Initiative in 2015



Source: <https://obamawhitehouse.archives.gov/precision-medicine>

# Introduction | Precision Medicine and HCI

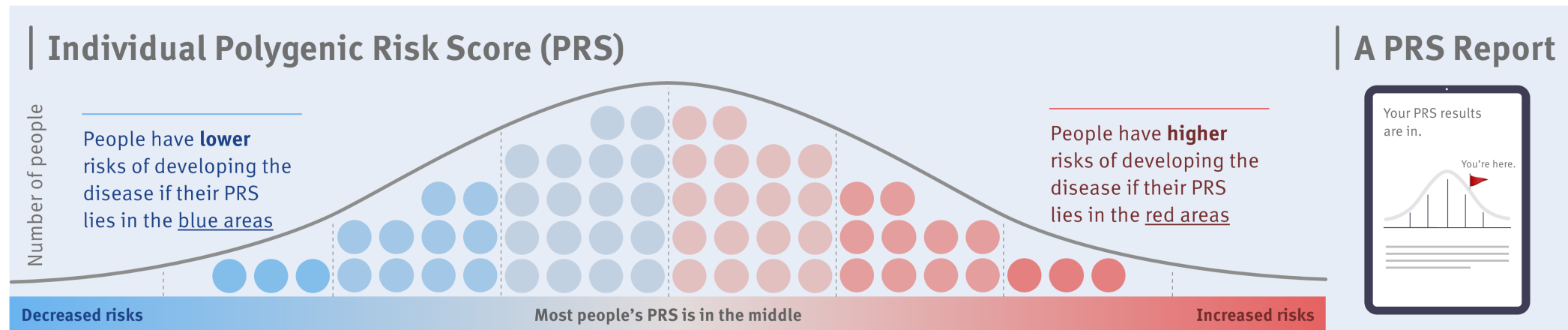
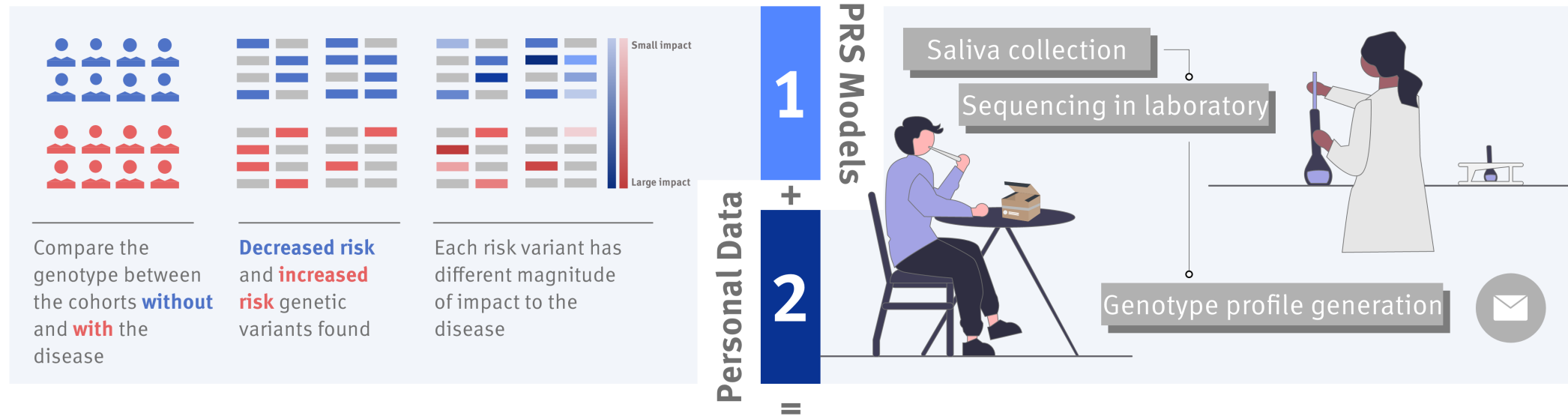
Clinical decision-making support

Data privacy and security

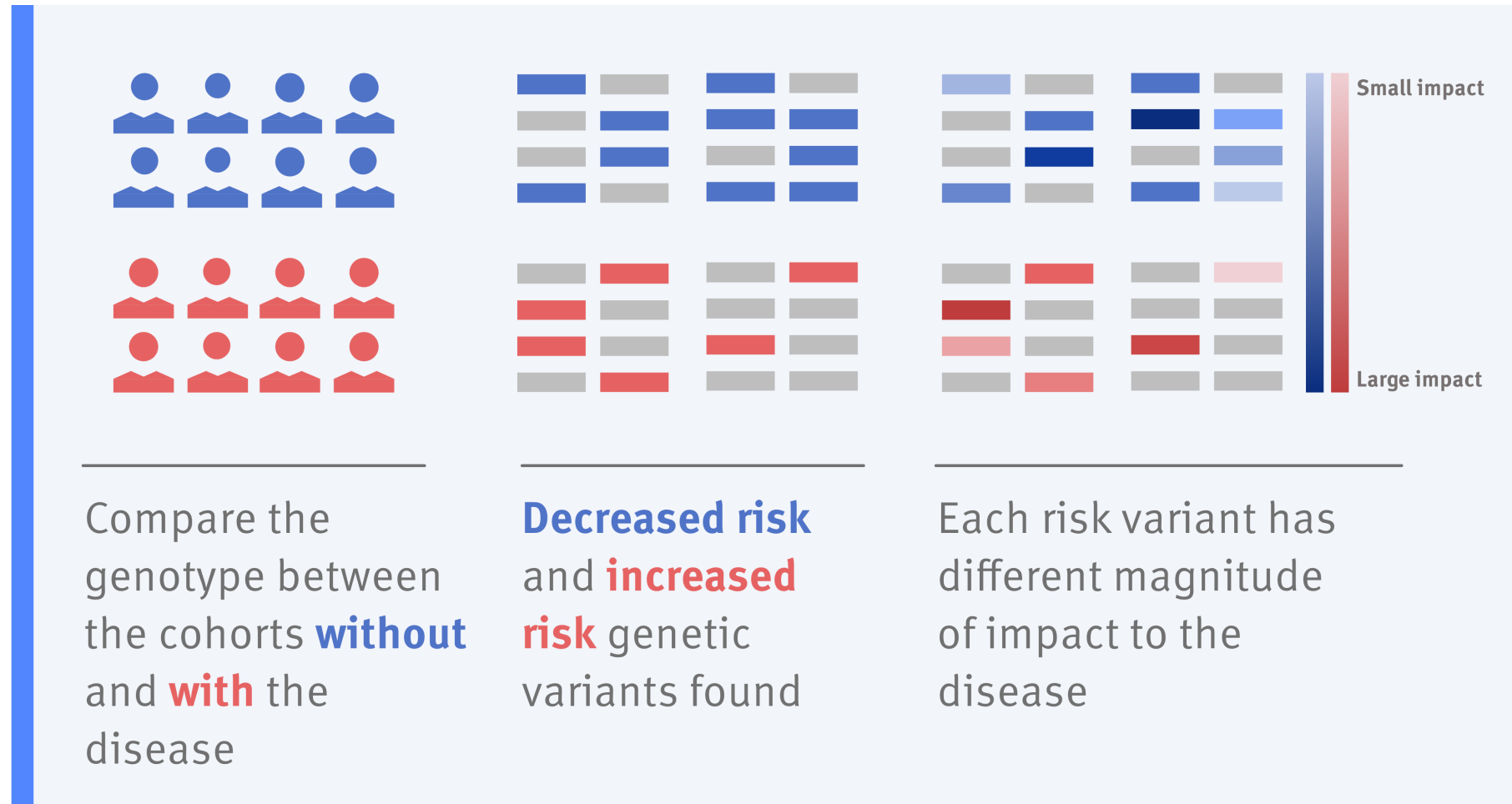
Patient engagement and empowerment

Although there are only a few papers that explicitly use the term “precision medicine” or its alternatives to define their work, the field has implicitly explored relevant themes through various HCI studies.

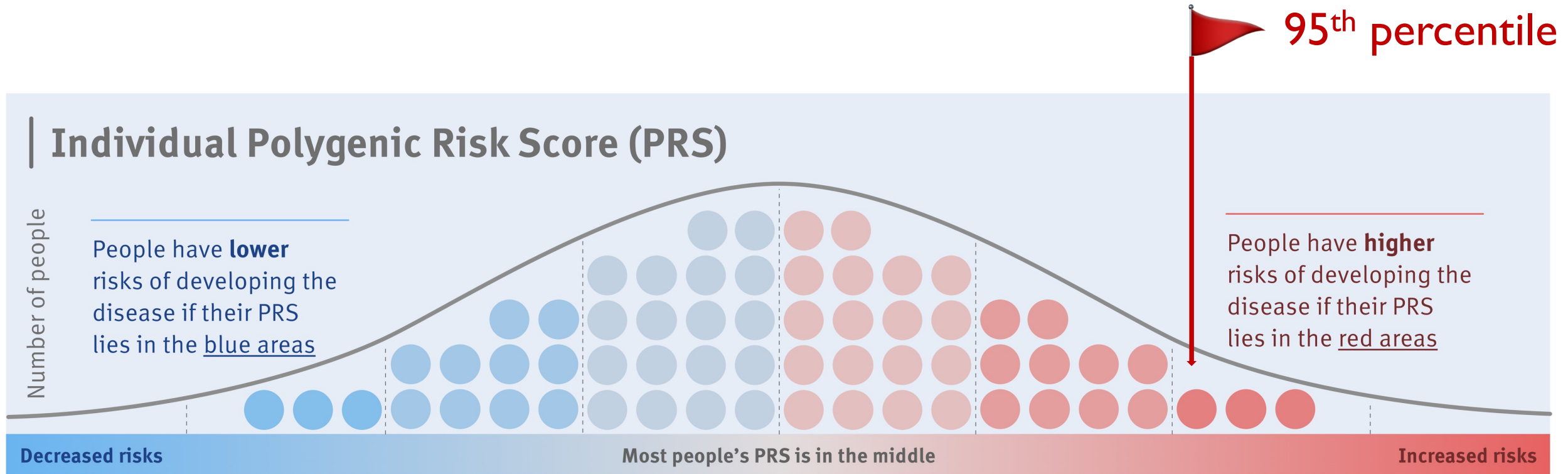
# Introduction | Polygenic Risk Score (PRS)



# Introduction | PRS calculation



# Introduction | PRS calculation



# Research Questions

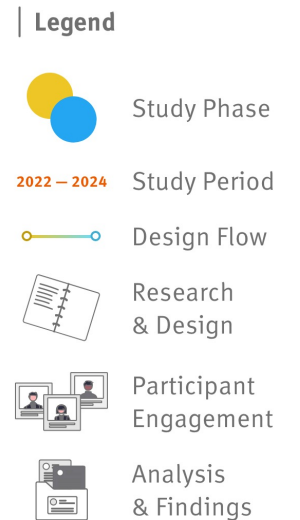
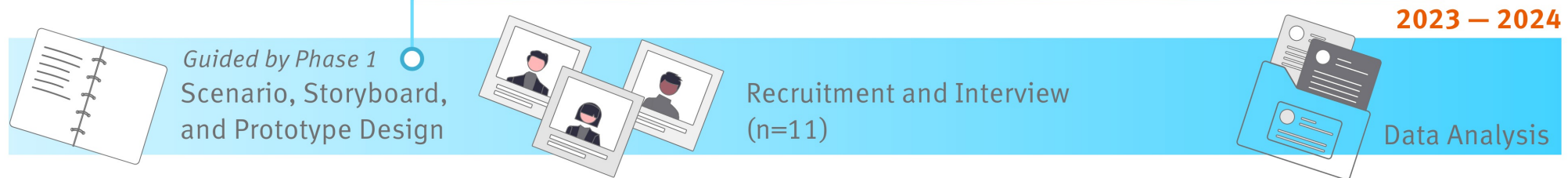
- 1 What are the **challenges and barriers** perceived by participants of using PRS services?
- 2 What are the **benefits** perceived by participants of using PRS services?
- 3 How do **people's experiences and stories** influence their informed decisions about potentially using PRS services in the future?

# Research Questions

## | Phase 1 — Survey (RQ1/2)

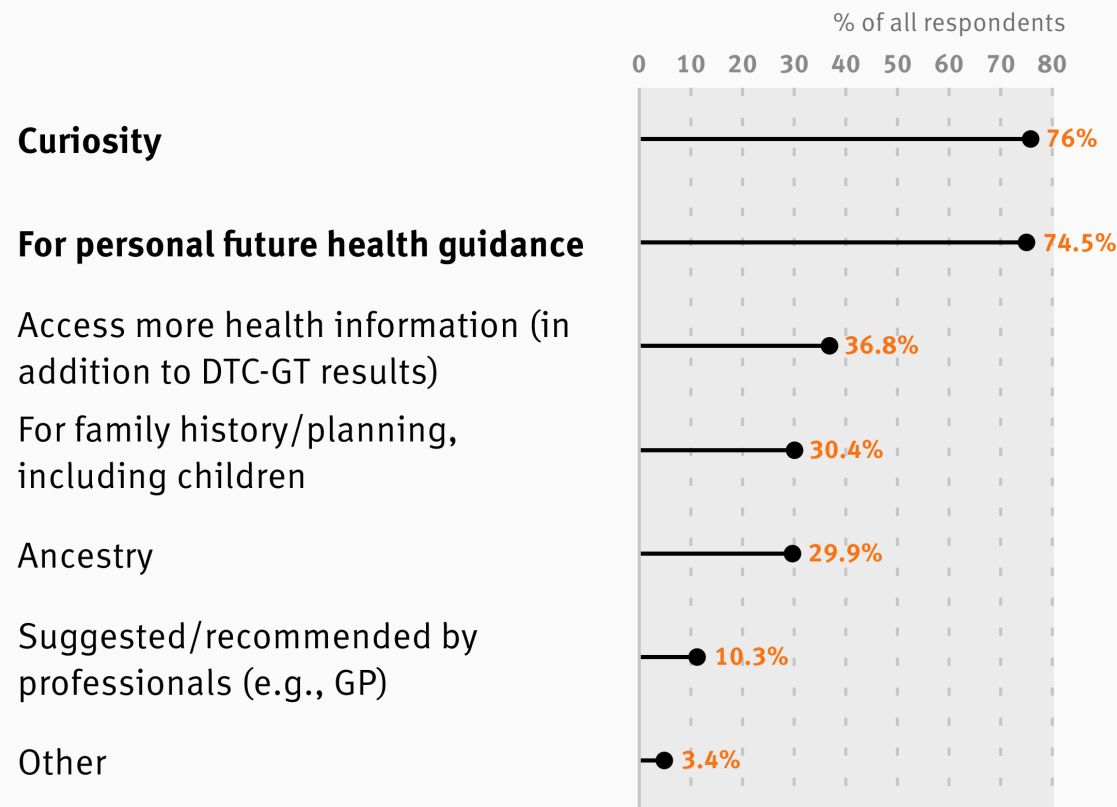


## | Phase 2 — Interview (RQ3, and further RQ1/2)

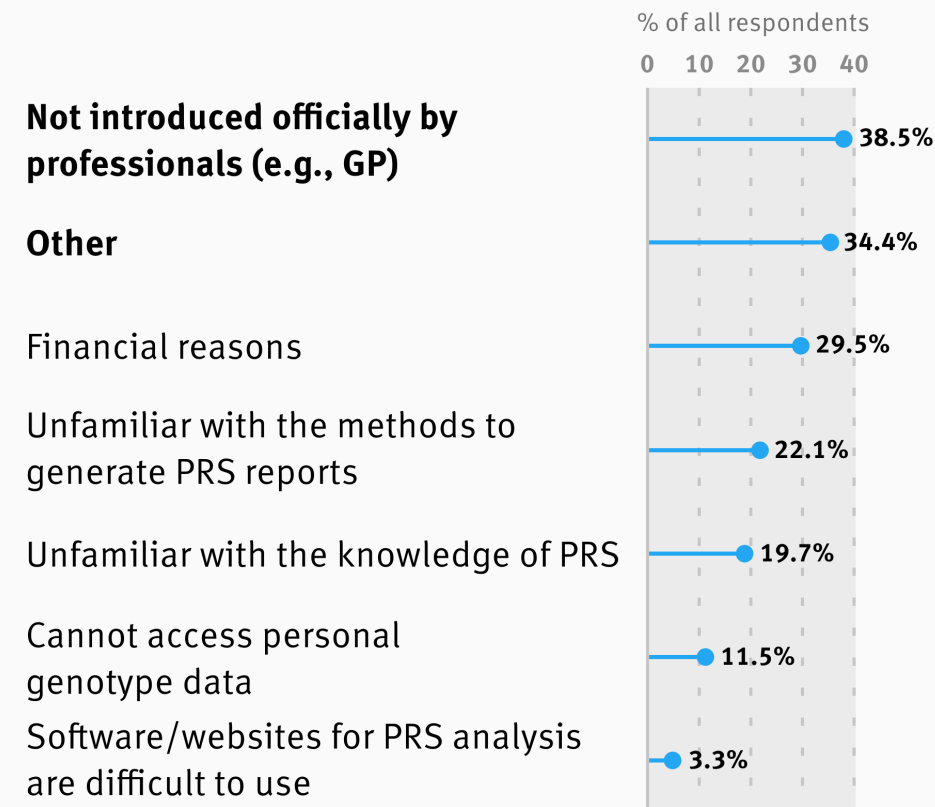


# Survey Findings | Motivations

## Reasons why people are **interested** in using PRS services



## Reasons why people are **uninterested** in using PRS services



# Survey Findings | Ten barriers to PRS usage

1

Privacy

2

Mental Health

3

Interpretation

4

Insurance

5

Ageing

6

Ethics

7

School

8

“Next Steps”

9

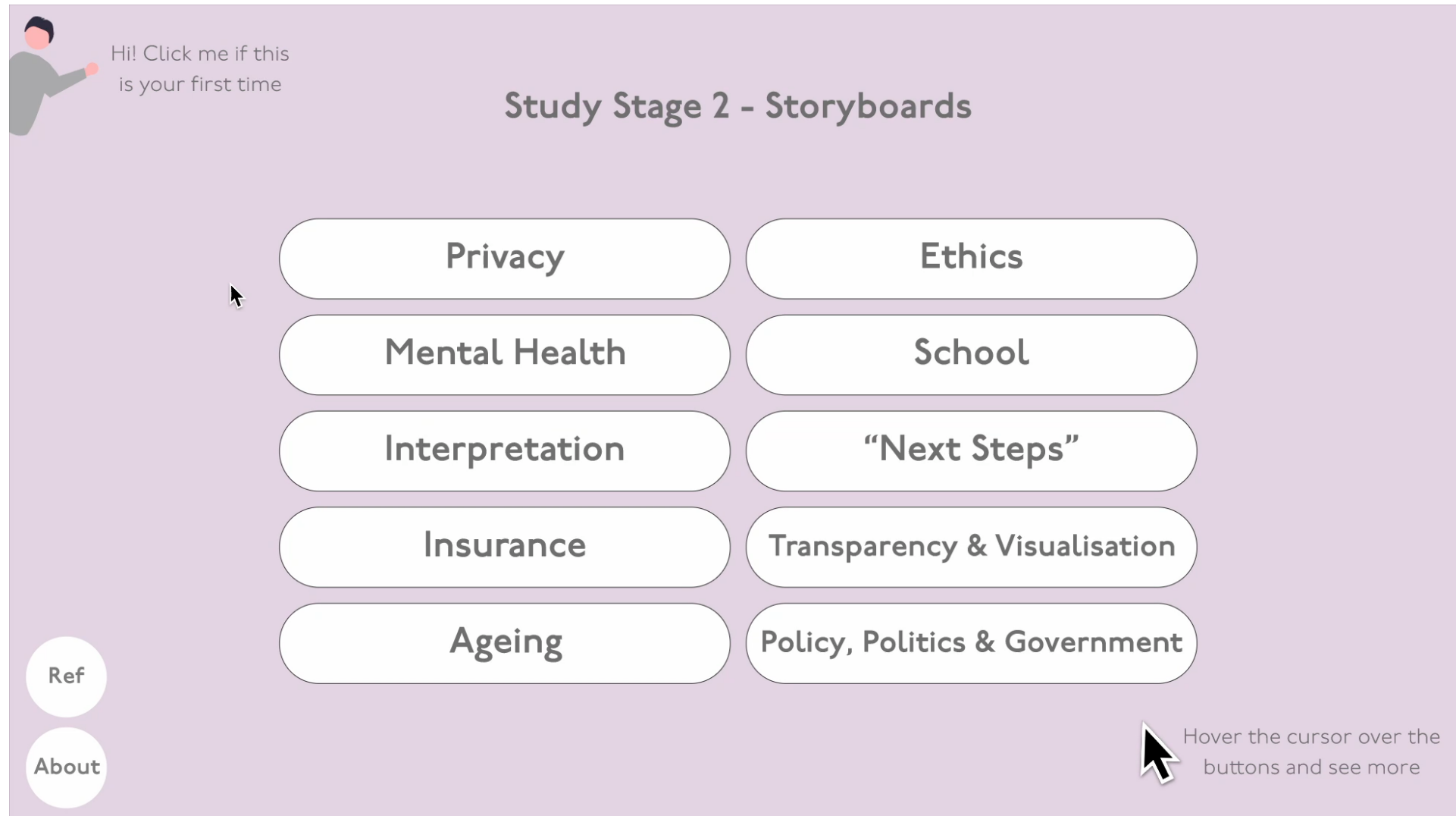
Transparency &  
Visualisation

10

Policy &  
Governance

# Interview | Interactive Storyboard Prototype

Access via  
[yuhaosun.com/perceprs/storyboard](http://yuhaosun.com/perceprs/storyboard)



# Interview Findings | Five themes

## ① Personal health background and proactivity

- Further health information
- Filling gaps in health history
- Proactive health and disease management

## ② Complexity, interpretation, and accessibility of information

- Complexity of jargon
- Interpretation from healthcare professionals
- Seeking trustworthy information online to understand PRS

## ③ Precision and inclusivity

- Demographic bias in genetic studies and database
- A holistic approach to health risk assessment

## ④ Psychological impact and preparedness

- 'Next steps' and psychological impact
- Psychological preparedness
- Different and individualised approach to health management

## ⑤ Trust, privacy, and society

- Privacy concerns and consent
- Trust and transparency
- How will PRS shape society: redefining fairness and embracing innovation?

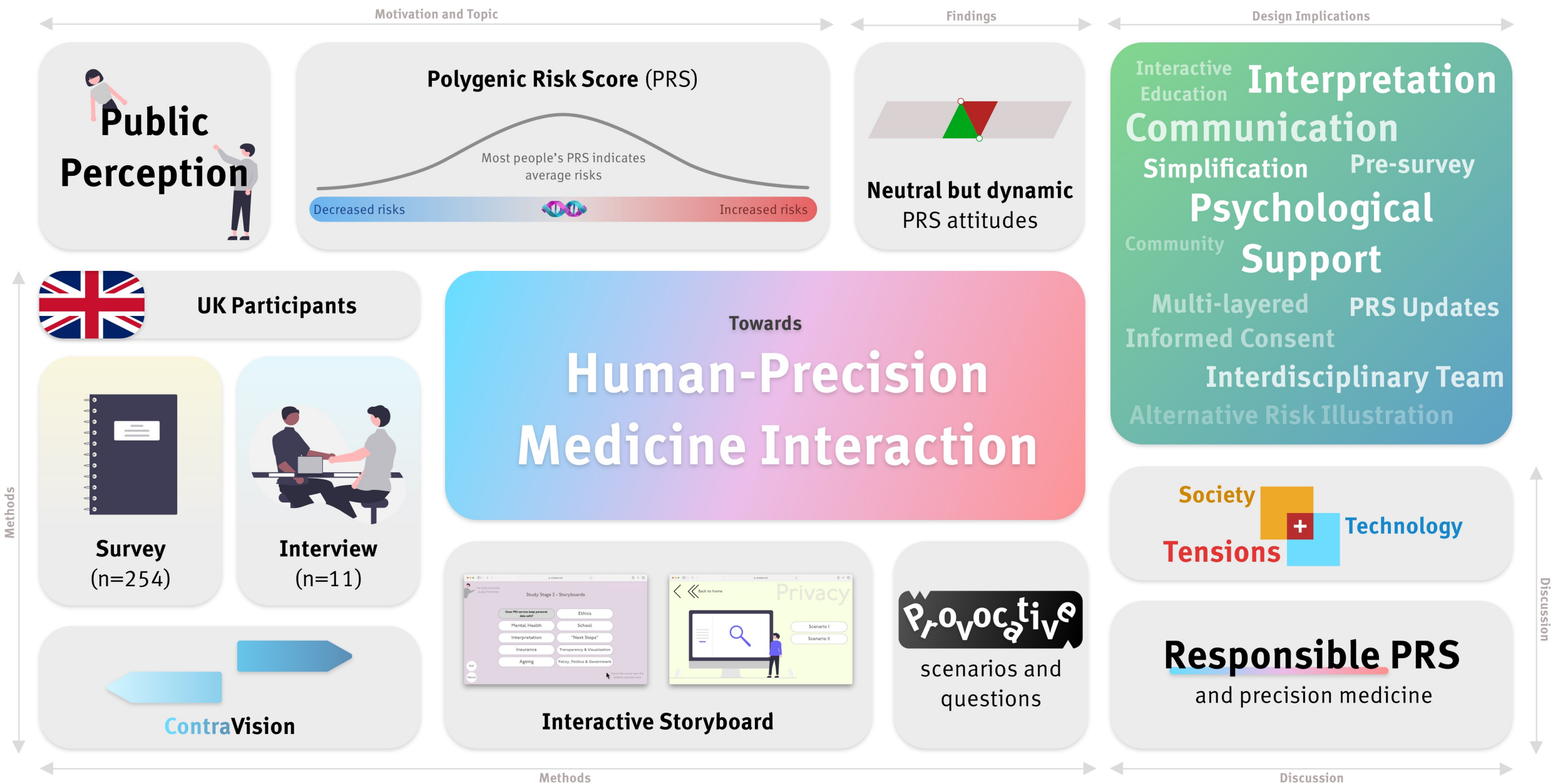
# Understanding PRS as a complex socio-technical system

Non-linearity and probabilistic nature

Delayed and indirect feedback loops

Risks of data governance and  
commercialisation

Challenges in regulating genetic data  
misuse



# Human-Precision Medicine Interaction (HPMI)

Terminology	“Human-Computer Interaction” + “Precision Medicine”
Scope	Undefined – we send an invitation to explore
Future work	Complex health data communication and interpretability / systemic collaboration and redesign

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Colin Farquharson  
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Edinburgh Futures Institute  
Anonymous reviewers

Visit [yuhaosun.com/PercePRS](https://yuhaosun.com/PercePRS) for slides, supplementary materials, and more details on this research about Perceptions of PRS

Paper

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I'll be on the job market this autumn!



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