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Discrete choice experiment to measure patient preferences for massively parallel sequencing genetic testing of colorectal cancer risk

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Background

- 20% to 30% of colorectal cancers (CRCs) are hereditary
- ~5% caused by highly penetrant inherited Mendelian pathogenic variants
- **Lynch syndrome** is the most common cause of Mendelian CRC risk

Genetic Testing for CRC risk

- Testing for Lynch syndrome is currently **recommended** for all newly diagnosed CRC patients
- Multistep process, time-consuming, 80% sensitivity
- **Massively parallel sequencing** (MPS) can detect other less prevalent forms of CRC risk

Objectives

Use discrete choice methods to enumerate:

1) Patients' **personal utility** for information on genetic causes of CRC

2) **Preferences** for MPS testing

3) **Willingness-to-pay** (WTP) for a MPS test that better identifies genetic causes of CRC

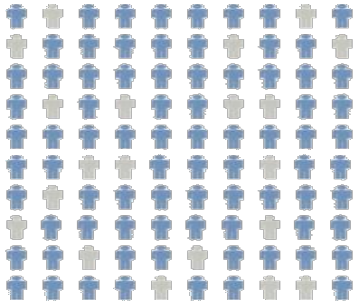
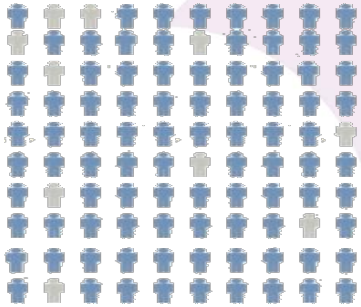
Discrete Choice Experiment (DCE)

- Assume technologies can be described by their characteristics (**attributes**), from which individuals derive utility

- Ask people to **state their preferences** in hypothetical scenarios involving two or more competing alternatives, termed **choice tasks**

Example of Choice Task

Which Genetic Test do you prefer?

Test Characteristic	Genetic Test A	Genetic Test B	No Genetic Testing
Number of individuals tested who receive a definitive genetic diagnosis causing CRC	80 individuals out of every 100 who are tested receive a genetic diagnosis 	90 individuals out of every 100 who are tested receive a genetic diagnosis 	You will not receive a genetic test, nor a conclusive genetic diagnosis
Number of genetic tests you will undergo. *Each genetic test will require a clinic visit and a blood draw*	3 genetic tests	1 genetic test	Not relevant
Total time waiting for results of all genetic tests	3 weeks	3 months	Not relevant
Total cost to you of all testing	$1000	$425	$ 0
Which genetic test do you prefer? *(Check one box only)*	**Genetic Test A ☐**	**Genetic Test B ☑**	**Neither Test ☐**

Approach

- Setting: New Exome Technology in (**NEXT**) Medicine Study, Seattle, WA, from **2012-2016**
- Error-component mixed logit model
- Relative importance scores
- WTP and predicted uptake of policy scenarios

Study Participants

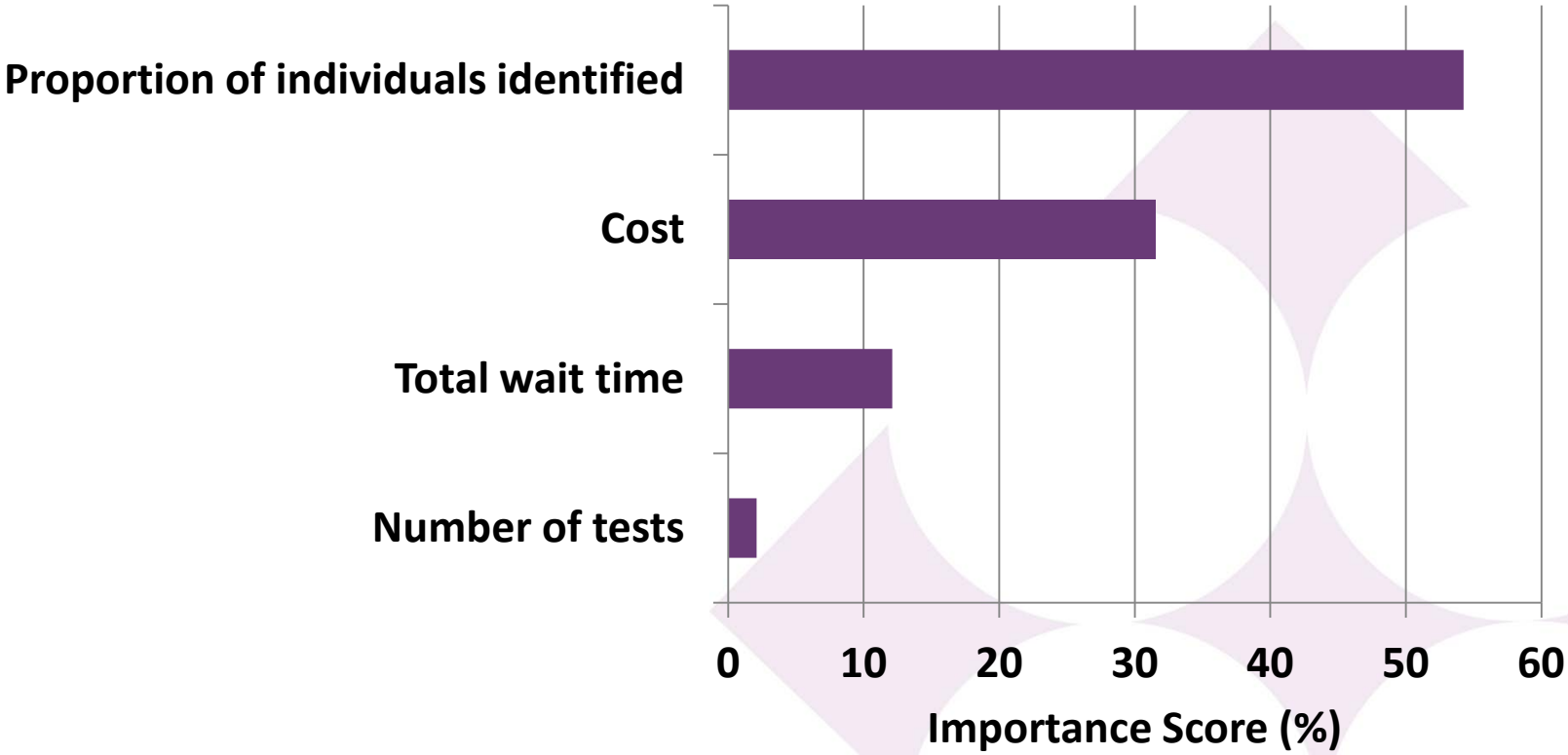
- 184 patients enrolled in NEXT Medicine study from 2012 to 2016
- 122 participants completed the DCE
  → **response rate=66%**
- 44% had personal history of CRC
- 88% had family history of CRC/polyposis

Model Estimates

Attribute and level	Part-worth utility, mean	Part-worth utility, SD	Part-worth utility<0
Proportion of individuals identified			
40/100	-1.65*	1.62*	84.5%
60/100	Reference	-	-
80/100	1.62*	1.30*	10.6%
90/100	2.08*	2.25*	17.8%
Number of tests	0.04	0.38*	46.2%
Total wait time (Months)	-0.16*	0.15*	84.7%
Cost ($)	-0.0010*	-	-
Opt out of testing	-5.36*	-	-
Opt in for testing	0 (assumed)	6.29*	-

*p<0.05, SD=standard deviation

Relative Importance of Attributes



Prevailing Policy Scenario

Patients have one testing option: either undergo traditional testing for CRC risk or receive no testing

Traditional Diagnostic Testing
40% of individuals receive a definitive diagnosis
Undergo **3 tests**
Wait **3 months** for results

New Policy Scenario 1

Patients choose between two testing options:

Option 1: Traditional Testing	Option 2: MPS Testing (1)
40% of individuals receive a definitive diagnosis	**80%** of individuals receive a definitive diagnosis
Undergo **3 tests**	Undergo **1 test**
Wait **3 months** for results	Wait **3 weeks** for results

Incremental WTP = US$1,600 (95% CI: $1,255, $1,946)

80% choose MPS testing (95% CI: 74%, 87%)
2% choose traditional testing (95% CI: 0%, 4%)
17% choose no testing (95% CI: 12%, 23%)

New Policy Scenario 2

Patients choose between two testing options:

Option 1: Traditional Testing	Option 2: MPS Testing (2)
40% of individuals receive a definitive diagnosis	**90%** of individuals receive a definitive diagnosis
Undergo **3 tests**	Undergo **1 test**
Wait **3 months** for results	Wait **1.5 months** for results

Incremental WTP = US$1,876 (95% CI: $1,365, $2,387)

85% choose MPS testing (95% CI: 77%, 93%)
2% choose traditional testing (95% CI: 0%, 3%)
13% choose no testing (95% CI: 7%, 19%)

Conclusion

- Patients preferred genetic tests **identifying more individuals** with a diagnosis and involving a **shorter wait time**
- Average WTP for MPS ranged from **US$1,600 to US$1,876**
- Patients value information on genetic causes of CRC and **replacing traditional diagnostic testing with MPS testing will increase patients' utility**

Acknowledgements

- **The authors have no conflicts of interest to declare.**

- **Research support:** The National Human Genome Research Institute and National Cancer Institute under Grants No. U01HG0006507 and U01HG007307; and by the National Institutes of Health Common Fund/National Institute of Aging under Grant No. U01AG047109.

- **Disclaimer:** The views expressed in this presentation are the authors' own and not an official position of the funder.