

# EE217: Population Genomic Screening for Tier 1 Genomic Applications: A Cost-Effectiveness Analysis



Clara Marquina<sup>1,2</sup>, Paul Lacaze<sup>1</sup>, Jane Tiller<sup>1</sup>, Adam Brothie<sup>1</sup>, Zanfina Ademi<sup>2</sup>

<sup>1</sup>School of Public Health and Preventive Medicine, Monash University; <sup>2</sup>Faculty of Pharmacy and Pharmaceutical Sciences, Monash University

## BACKGROUND

- Tier 1 genomic applications are poorly ascertained by the healthcare system, but early detection and intervention could dramatically reduce morbidity and mortality<sup>1</sup>.
- Three genetic conditions are included in Tier 1: Familial Hypercholesterolemia (*Apo E*, *PCSK9*, *LDLR*)<sup>1</sup>, breast and ovarian cancer (*BRCA1* and *BCRA2*)<sup>2</sup> and Lynch Syndrome (colorectal cancer ; *MLH1*, *MLH6*)<sup>2</sup>.

### Aim

To assess the impact and cost-effectiveness of offering population genomic screening to all young adults in Australia to detect heterozygous familial hypercholesterolemia (FH), hereditary breast and ovarian cancer and hereditary colorectal cancer.

## METHODS

Figure 1. Decision tree.

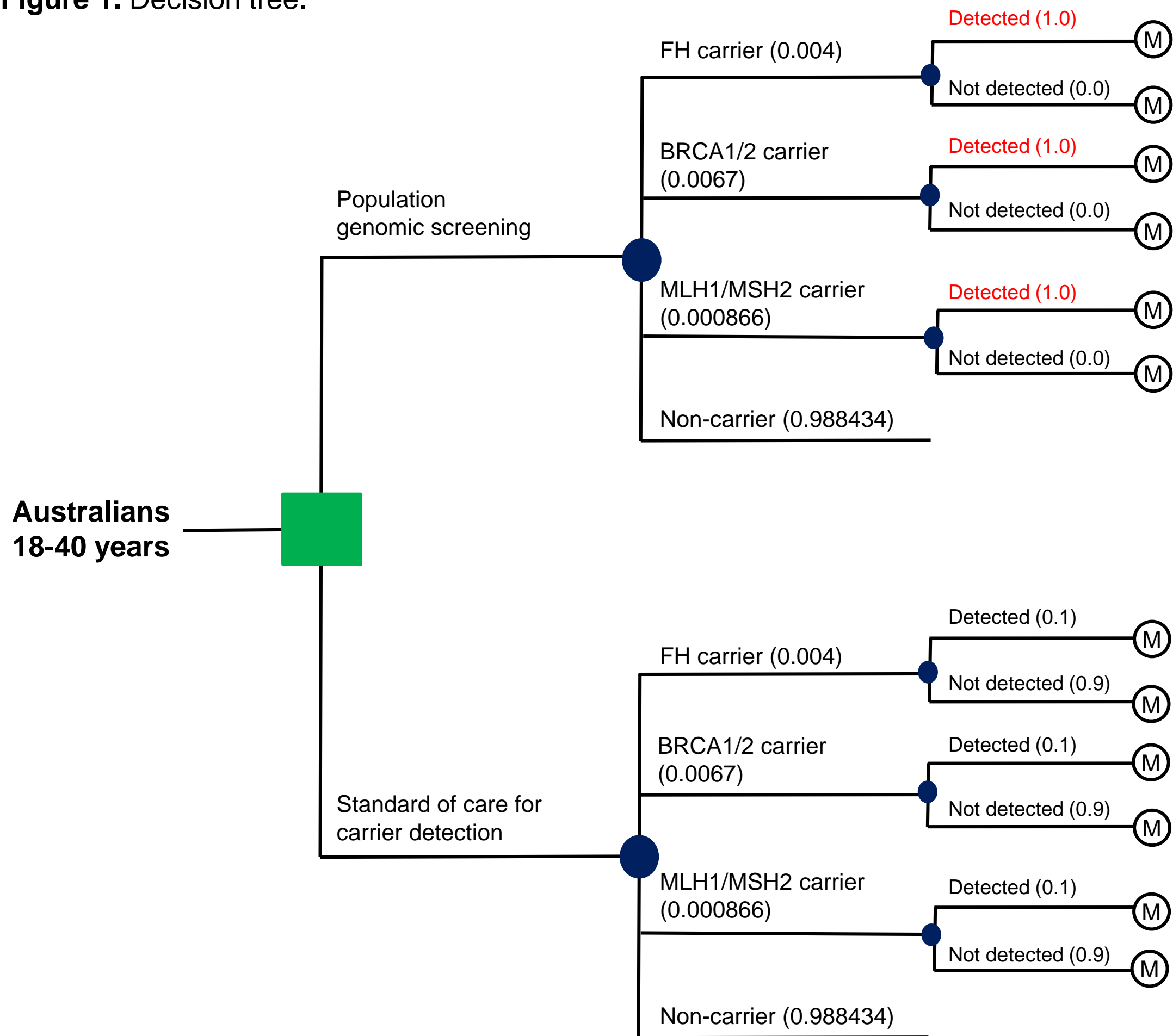


Figure 2A. Markov model for *ApoE*, *PCSK9*, *LDLR* (Familial Hypercholesterolemia)

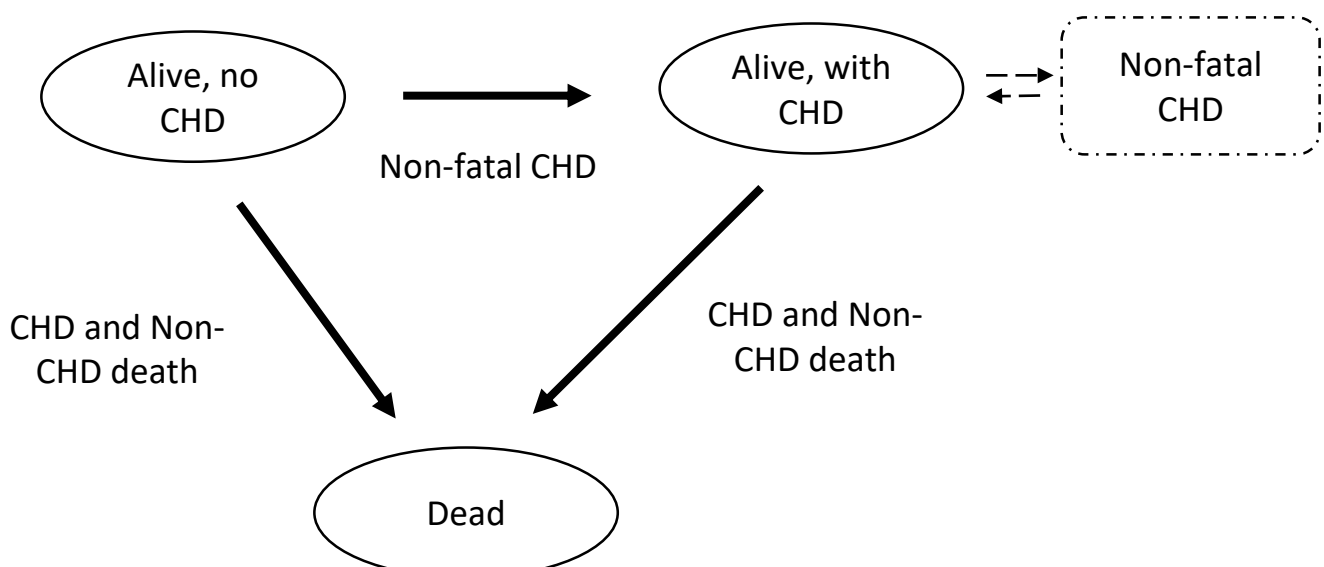


Figure 2B. Markov model for *BRCA1/2* screening (Breast cancer & Ovarian cancer)

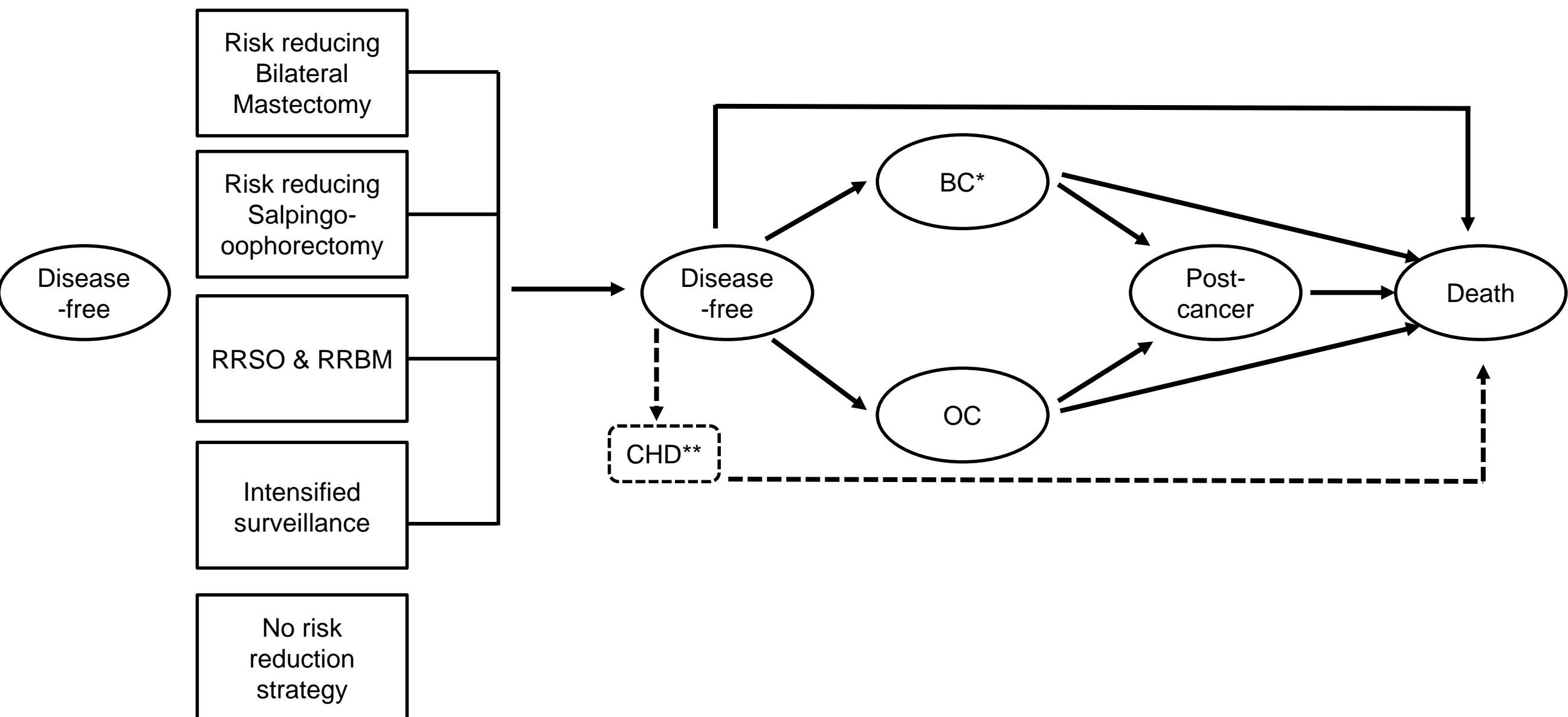
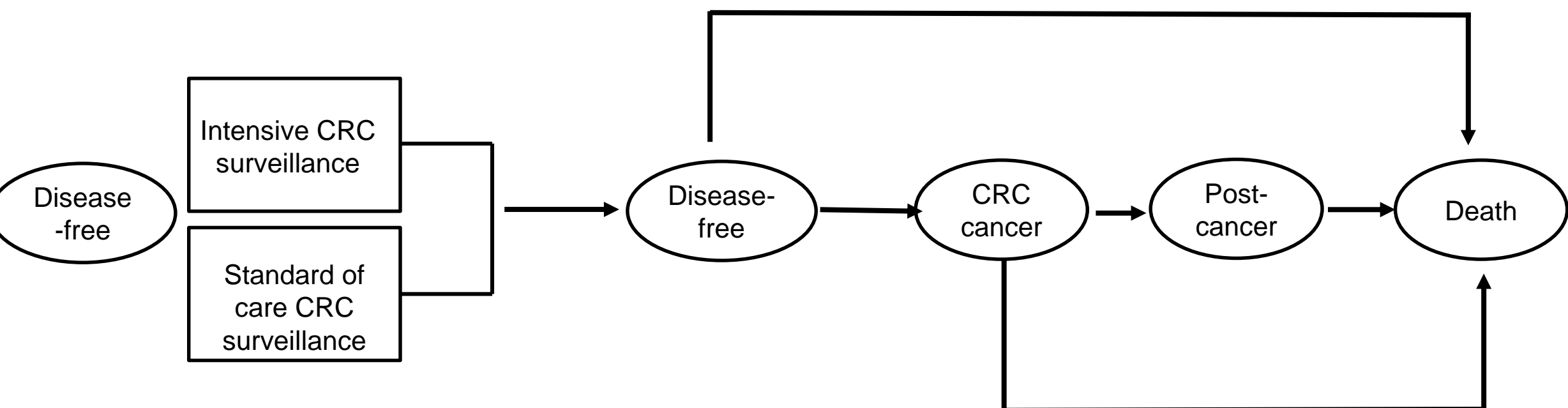


Figure 2C. Markov model for *MLH1/MLH2* screening (Colorectal cancer)



### References:

1. Buchanan AH et al., *Genet Med* (2018); CDC, Office of Public Health Genomics

## RESULTS

Model Outcomes	Standard of care	Population genomic screening	Difference
Non-fatal CHD cases	40,185	37,878	-2,307
Total cancer cases	26,678	15,980	-10,698
Total deaths	71,704	65,908	-5,796
YLL	1,581,626	1,654,255	72,629
QALYs	1,320,541	1,426,673	106,132
Genomic Screening costs		\$1,664,848,400	\$1,664,848,400
Healthcare costs	\$3,763,949,123	\$3,126,779,262	-\$637,169,861
Total costs	\$3,763,949,123	\$4,791,627,662	\$1,027,678,539
ICER (AU\$/YLL)			\$14,150
ICER (AU\$/QALY)			\$9,683

Table 1. Combined model outcomes. CHD, Coronary heart disease; YLL, Years of life lived; QALYs, Quality-adjusted life years; ICER, Incremental cost-effectiveness ratio

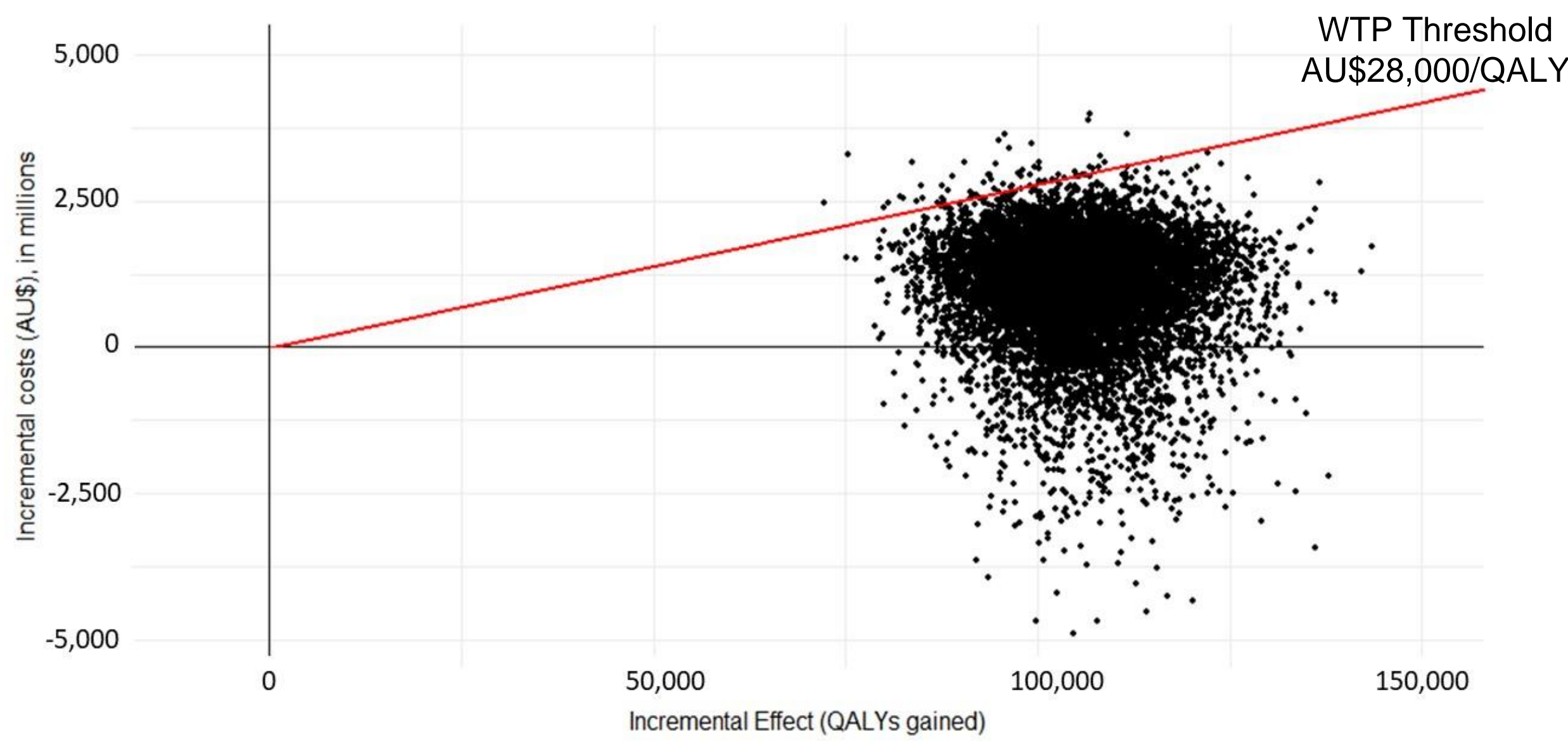
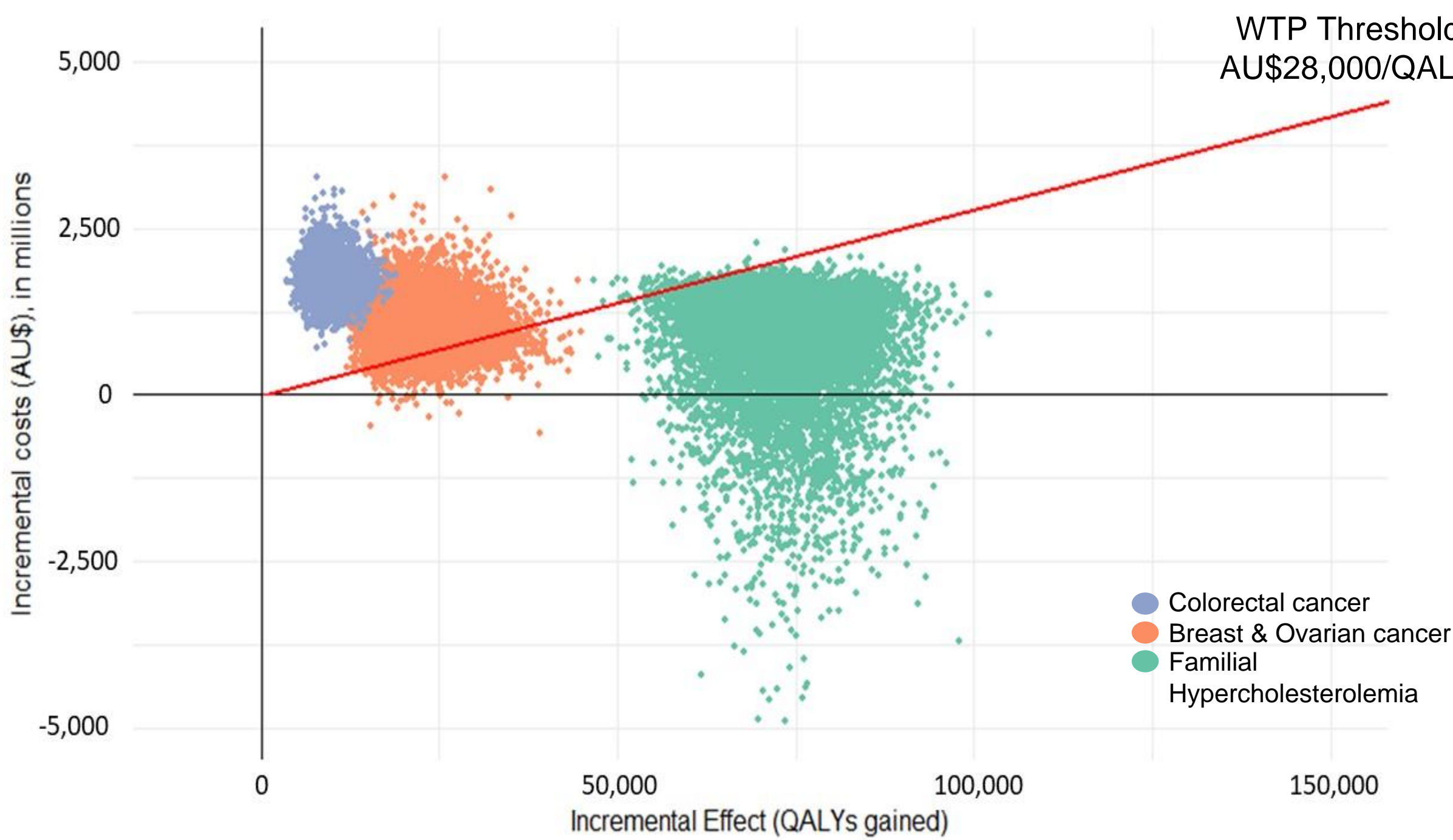


Figure 3. PSA Combined model results. QALYs, Quality-Adjusted Life Years

Figure 4. PSA Individual model results. QALYs, Quality-Adjusted Life Years



## CONCLUSIONS

Based on our model, offering population genomic screening to all young adults could be cost-effective from a public healthcare system perspective in Australia, at testing costs that are feasible (AU\$250 per test).



### Contact details

Clara Marquina, PhD  
Monash University, Melbourne, Australia  
[clara.marquina@monash.edu](mailto:clara.marquina@monash.edu)  
Twitter @claremarquina