

KNOWLEDGE GAINED

Recall was cost-effective for women with BRCA1 and BRCA2 aged 20-59 years, and with PALB2 aged 20-69 years.

KEY MESSAGE

Returning genomic data to sample donors resulted in noteworthy health gains and was feasible and cost-effective.

From Biobank Sample to Breast Cancer Prevention

Cost-Effectiveness Analysis of Returning Genomic Data to Sample Donors

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BACKGROUND

- Germline mutations in the Breast Cancer gene 1 or 2 (BRCA1 or BRCA2) and Partner And Localizer of BRCA2 (PALB2) are associated with a high lifetime risk of aggressive breast cancer (BC).
- The FinnGen study combines genotype results and health data from 500,000 sample donors in Finland. The genotypes are available for additional projects. FinnGen is a large public-private biobank study aiming to collect and analyse genome and health data from 500,000 study participants in Finland.
- Returning genomic information to donors could enable prophylactic interventions or intensified screening to reduce BC burden. (Figure 1)

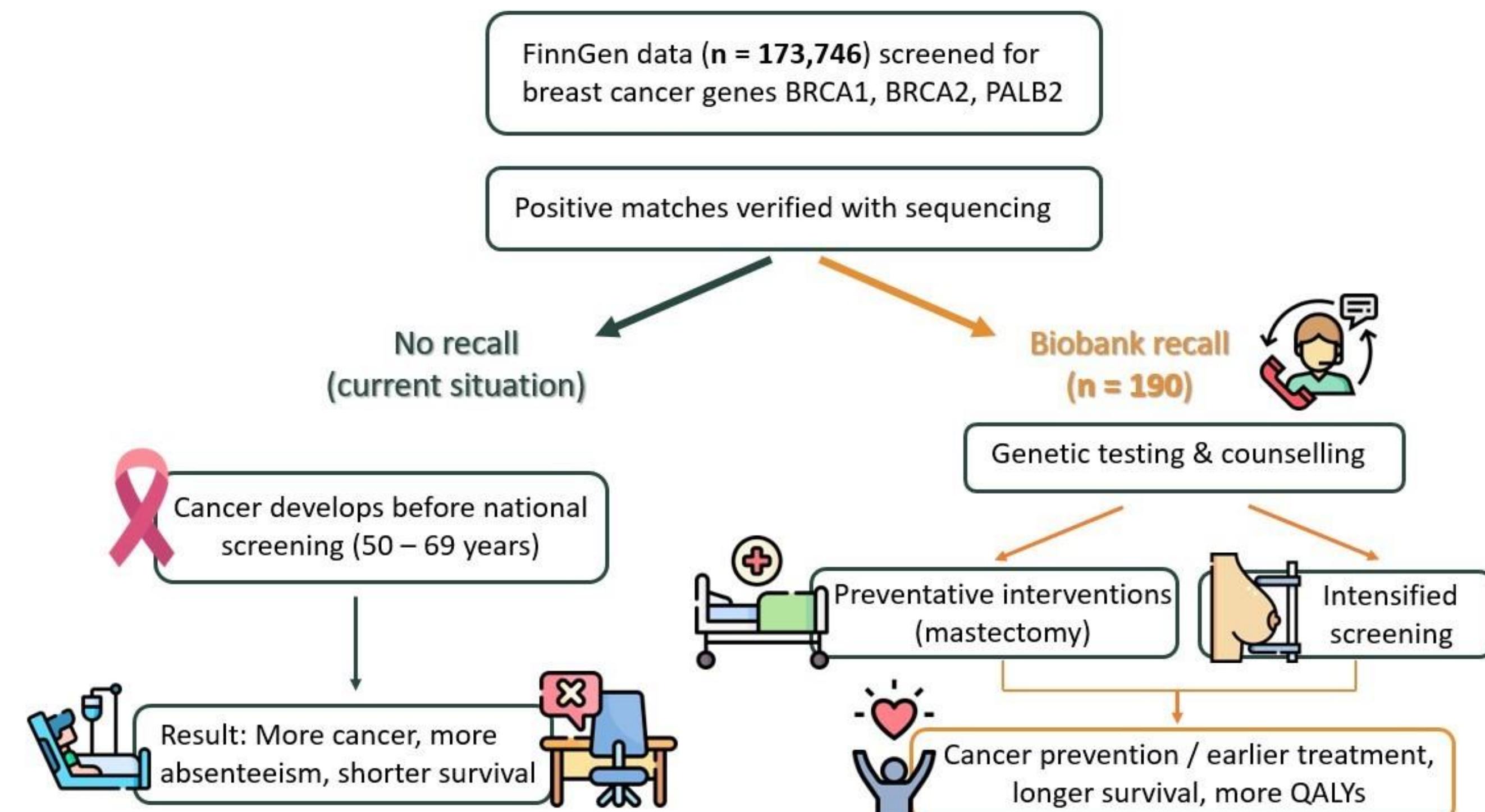


Figure 1.

METHODS

The methods of the economic evaluation are summarised in the table below using the PICOSTEPS framework.⁶

COMPONENT	CONTENT
Patients	Women with mutations (BRCA1/2, PALB2) conferring high risk of breast cancer (BC), aged 20-79 years
Intervention	Recall by Helsinki biobank, resulting in prophylactic interventions, intensified screening, or no change
Comparator	No recall
Outcomes	Number of BC cases, deaths due to BC, costs (euros in 2022), life-years (LY) and quality-adjusted life-years (QALY), net monetary benefit (NMB) at 37,364 €/QALY gained
Setting	Cost-effectiveness modelling using a cohort simulation model
Time	Lifetime, 3% per annum discounting
Effects	Prophylactic bilateral mastectomy reduces risk of BC to 9%, and intensified screening can prevent BC through detection of pre-cancerous lesions ⁴
Perspective	Finnish societal perspective
Sensitivity analyses	Type of mutation, age group

CONCLUSIONS

- Returning genomic data to sample donors and genetic counselling resulted in noteworthy health gains and was feasible and cost-effective.
- Further studies regarding the impact of ovarian cancer and of prophylaxis in relatives are warranted and should further improve the simulation outcomes.

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RESULTS

- FinnGen data freeze 7 consisted of 173,746 female donor samples, of which 190 individuals were estimated to proceed to recall.
- The recall resulted in an average additional lifetime cost of € 3,509/recalled woman (€ 1,444 recall process, € 7,084 screening and prophylactic interventions, and – € 5,888 offset for avoided BC treatments), Figure 2.

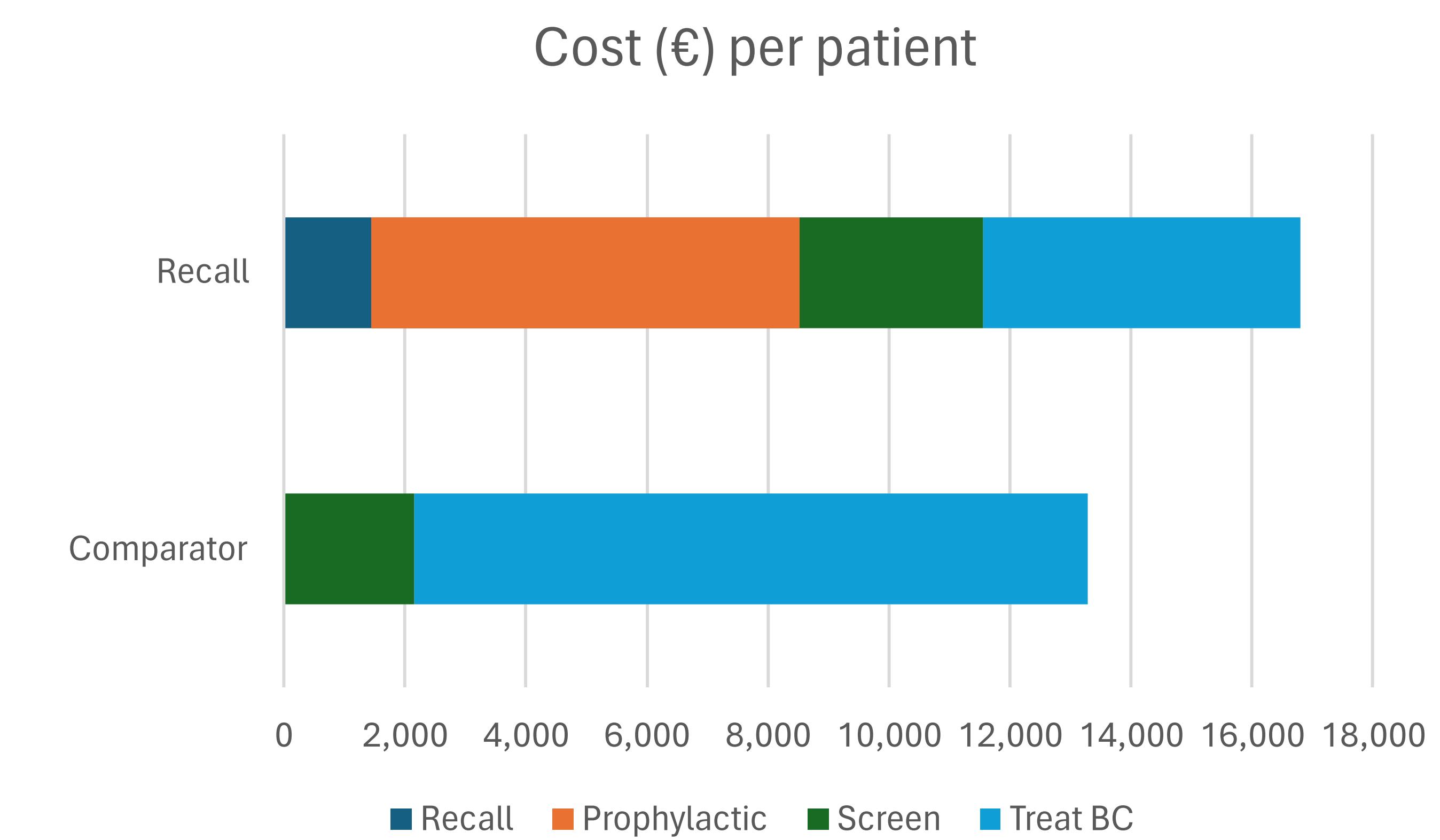


Figure 2.

- With the recall, 50 BC cases and 13 BC deaths could be avoided. This would generate 88.0 additional life-years and 45.7 additional QALYs, clearly favouring recall.
- NMB was positive for women with BRCA1 and BRCA2 aged 20-59 years, and with PALB2 aged 20-69 years. (Figure 3)

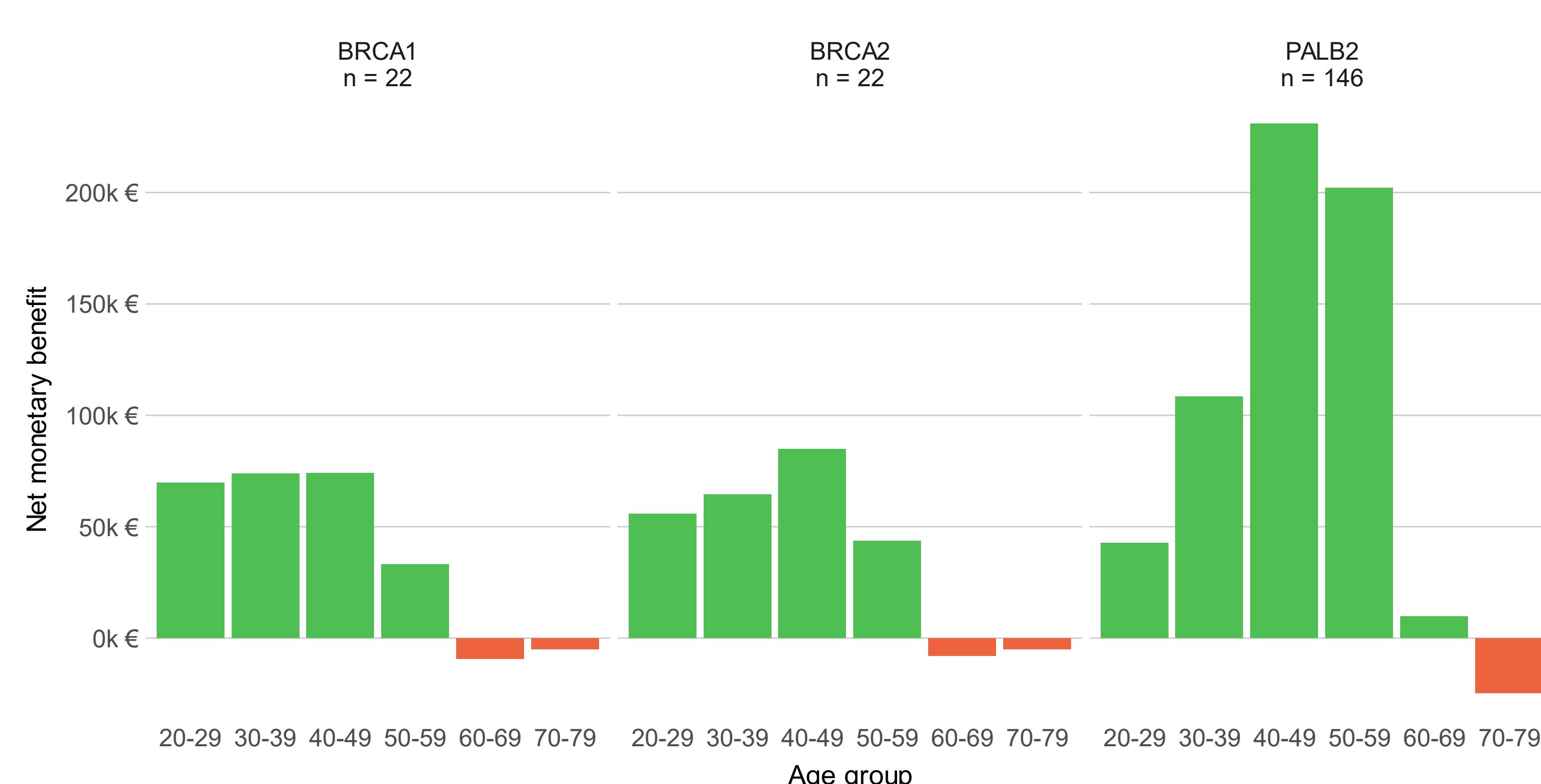


Figure 3.

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