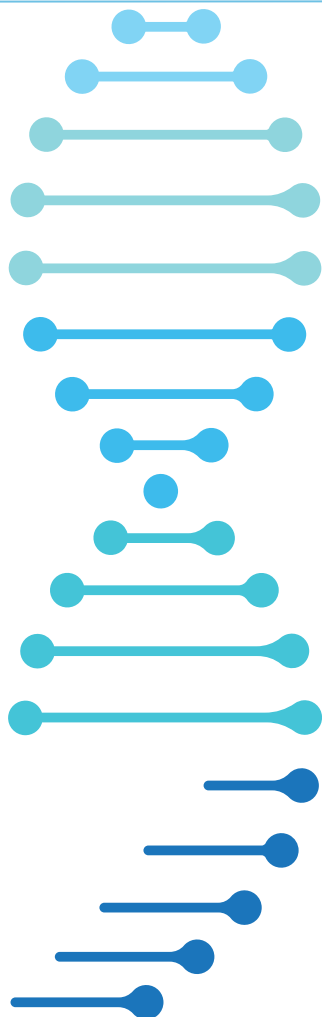


PROPHET TOOLBOX FACTSHEET#3

JUNE 2025



Current landscape of personalized preventive approaches for non- communicable disease: A scoping review

 **ROPHET**

a PeRsOnalized Prevention roadmap
for the future HEAlThcare

In the context of PROPHET, we mapped existing primary studies and clinical guidelines on **personalized preventive approaches** by searching scientific databases and grey literature documents published from 2017 to 2023.

We included **121 publications, of which 60 were primary studies and 61 clinical guidelines.**

Among them overall, **249 personalized preventive approaches** were identified as reported in the figure 1, of which 113 from primary studies (45%), and 136 from clinical guidelines (55%).

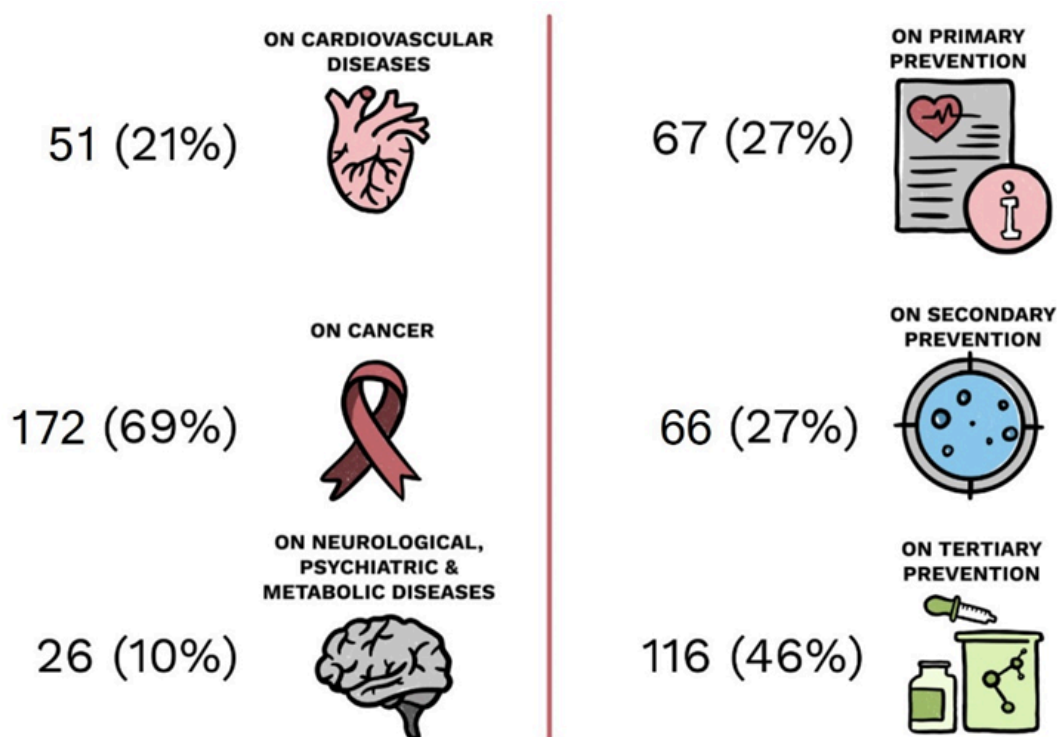
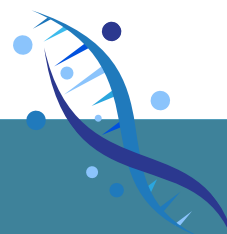


Fig. 1. Personalized preventive approaches classified by level of prevention (right) and type of disease (left).



Classification according to prevention levels

In **primary prevention**, 50% of the 67 mapped approaches were from primary studies, mainly targeting cardiovascular diseases, and 50% from guidelines primarily focused on cancer as depicted in Fig. 2.

Among these, one strategy from primary studies in cardiovascular diseases involves polygenic risk scores (PRS) profiling and disclosure among high-risk subjects in order to encourage the adoption of healthier lifestyles or the uptake of other preventive therapies.

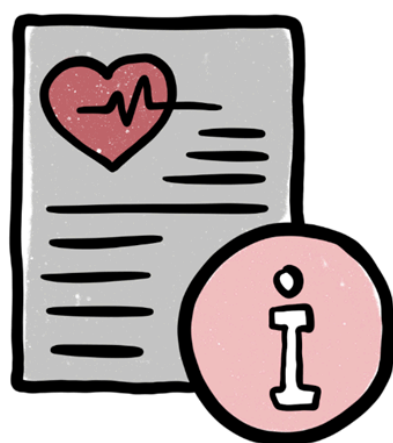


For example, in a randomized pilot study conducted in Estonia, individuals in the highest 20% of the PRS for Coronary Artery Disease received tailored counselling and preventive therapy, leading to a greater uptake of statins and improved LDL cholesterol levels. However, the use of PRS-based interventions is currently absent from clinical guidelines, reflecting the limited and still inconclusive nature of the supporting evidence.



On the other hand, an established approach in cancer prevention, as reported in various guidelines, involves genetic screening for *BRCA1/2* mutations which are primarily associated with breast, ovarian, and prostate cancer. This approach targets healthy individuals with cancer family history or other risk factors and those identified as high risk through testing may receive risk-reduction interventions, such as prophylactic surgery, lifestyle modifications, and chemoprophylaxis.

ON PRIMARY PREVENTION







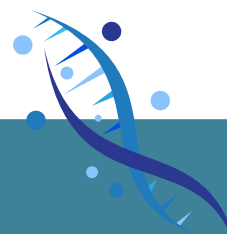
		Clinical Guidelines	Primary Studies
	Cancer	23	7
	Cardiovascular	10	19
	Metabolic Diseases	0	6
	Others	0	2

Fig. 2: Number of clinical guidelines and primary studies per type of disease (primary prevention)



Secondary prevention included 66 approaches, 73% from guidelines mainly concerning cancer screening and only 27% from primary studies (Fig. 3).

Cascade testing refers to the performance of genetic counselling and testing in blood relatives of individuals who have been identified with specific genetic mutations.

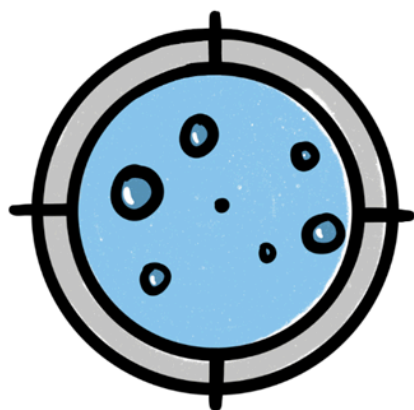


Cascade genetic screening and personalized follow up are recommended for individuals with a family history of colorectal cancer (CRC) or early-onset CRC. In patients with hamartomatous polyposis syndromes, such as Peutz-Jeghers or juvenile polyposis syndrome, the US Multi-Society Task Force recommends the use of multigene panel testing when clinical criteria are met. If a pathogenic variant is detected, endoscopic surveillance is advised starting in childhood or adolescence, with regular follow-up. This personalized approach enables early detection and removal of precancerous polyps, aiming to prevent progression to CRC.



In addition to these recommendations, several other guidelines addressing personalized cancer screening were identified compared to primary studies, indicating a more advanced stage of evidence development and the consolidation of tailored secondary prevention strategies.

ON SECONDARY PREVENTION



Cancer

46

14



Cardiovascular

2

3

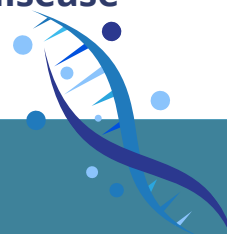


Others

0

1

Fig. 3: Number of clinical guidelines and primary studies per type of disease (secondary prevention)



Tertiary prevention accounted for 116 approaches, with 53% derived from primary studies and 47% from guidelines, focusing mostly on cancer and cardiovascular diseases (Fig. 4).

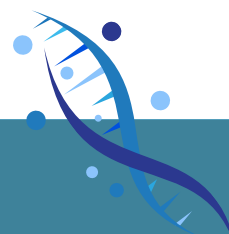


One example of tertiary prevention from primary studies is a randomized controlled trial conducted in the United States that evaluated the use of pharmacogenetic profiling in older polypharmacy patients receiving home health care after hospital discharge.



Since nearly half of the most commonly prescribed medications in the US are metabolized by *CYP450* enzymes, patients underwent testing for *CYP450* and related gene variants. Results were integrated into a clinical decision support tool (CDST) to inform prescribing, with the aim of optimising drug selection and dosing to minimise adverse reactions. The intervention led to a significant reduction in hospital readmissions and emergency department visits among patients with common chronic conditions such as chronic obstructive pulmonary disease (COPD), gastroesophageal reflux, and hypothyroidism. Although this approach is not yet included in formal guidelines, this study illustrates how pharmacogenomics can enhance medication safety and personalize care in complex, high-risk populations.

An established guidelines-based strategy involving pharmacogenomic testing is based on genotyping *DPYD* genes, to ensure proper dosing of anticancer drugs. This approach is commonly applied in the treatment of colorectal cancer, where patients with reduced DPD enzyme activity are at increased risk of severe toxicity from fluoropyrimidine-based chemotherapy (such as 5-fluorouracil or capecitabine). Identifying these patients through *DPYD* testing allows for personalized dose adjustments or alternative therapies, reducing adverse effects and improving treatment safety.



ON TERTIARY PREVENTION








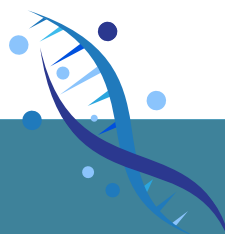
		Clinical Guidelines	Primary Studies
	Cancer	44	38
	Cardiovascular	5	12
	Metabolic Diseases	1	2
	Neurological/psychiatric disorders	5	5
	Others	0	4

Fig. 3: Number of clinical guidelines and primary studies per type of disease (tertiary prevention)

Conclusion

The review highlights a significant focus on personalized prevention in oncology in the past few years, as reflected by the large number of recently issued guidelines. In parallel, there is growing interest in cardiovascular diseases, with substantial original research focused on personalized primary prevention approaches. In contrast, the application of personalized prevention in neurological, psychiatric, and metabolic disorders remains limited, with most of the published works stems from primary studies and is concentrated in tertiary prevention. Pharmacogenomics is the leading approach in these areas, playing a pioneering role in translating research into clinical practice by improving medication safety and treatment outcomes.



However, the distribution of primary studies and directives across the three preventive levels anticipate challenges in generating evidence of clinical utility in primary and secondary prevention, with most approaches falling under tertiary prevention.

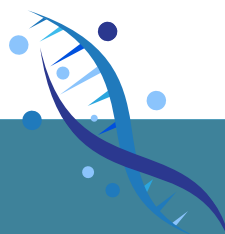
While primary studies reflect current research advancements, guidelines indicate well-established approaches with longstanding evidence. Many emerging technologies are currently being evaluated in research settings but have yet to be integrated into standard care. Bridging this gap will be key to advancing the implementation of personalized prevention across a broader spectrum of non-communicable diseases.

**For those who wants to read the paper, it is available at:
<https://doi.org/10.1371/journal.pone.0317379>**



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