Technoleg Iechyd Cymru
Health Technology Wales

Topic Exploration **Report** [1]

Registries for the identification, tracking and surveillance of people at risk of inherited cancers

What is a Topic Exploration Report?

Topic Exploration Reports are not health technology assessments. These reports provide a high-level briefing on new topics submitted to Health Technology Wales and are not based on exhaustive or systematic literature searches. Instead, they rely on a focussed scan of key resources.

What evidence is used in a Topic Exploration Report?

Priority is given to summarising the most relevant or useful evidence, rather than covering all possible evidence. Information reported is typically based on abstracts and study authors' own conclusions, rather than detailed scrutiny of full texts.

What are the aims of a Topic Exploration Report?

Topic Exploration Reports offer an overview of the available evidence on a topic and aim to highlight any uncertainties or gaps in the evidence. These reports outline the quantity and type of evidence found, but no critical appraisal or formal evidence synthesis is conducted.

How should a Topic Exploration Report be used?

Topic Exploration Reports can be used to indicate what evidence may be available for a topic, and do not provide definitive guidance on how a technology should be used. The evidence presented within the reports should be interpreted with caution.

[1] Cyfieithu dogfennau HTW wedi'u cyhoeddi o'r Saesneg i'r Gymraeg
Translation of published technical HTW documents from English into Welsh

Topic exploration report number	TER591
Topic	Registries for the identification, tracking and surveillance of people at risk of inherited cancers
Summary of findings	Cancer is often diagnosed at a late stage when treatment options are limited, and prognosis (likely course of the condition and its outcome) is worse. The term hereditary cancer syndrome is used to describe conditions with such an inherited gene change, and includes familial adenomatous polyposis (FAP) and Lynch syndrome. People with a hereditary cancer syndrome have an increased risk of developing certain types of cancer. Registries have been developed for the identification, tracking and surveillance of people at risk of inherited cancers. The use of registries and associated systems may allow earlier detection of cancer, potentially improving treatment options, quality of life and overall prognosis. Furthermore, surveillance data may form the basis of cancer research, helping to plan and evaluate cancer prevention and control interventions. Genomic testing, recommended for people who are either known to be at risk, or those whose clinical diagnostic data detect they are potentially at risk, may trigger inclusion in a registry. One systematic review of the impact of registration and screening on colorectal cancer incidence and mortality in FAP and Lynch syndrome was identified. No randomised controlled trials were included. A significant reduction of colorectal cancer incidence and mortality was reported in participants with registration and screening. Five of the forty three studies provided evidence for complete prevention of colorectal cancer-related deaths during surveillance. There is a five-month Small Business Research Initiative (SBRI) project in Northern Ireland to create a population-wide registry to support surveillance and tracking of people at risk of inherited cancer, and also to support research and clinical trials. It is unclear when results will be available. HTW researchers did not identify any economic evaluations. There is a lack of clinical and cost-effectiveness evidence on registries for the identification, tracking and surveillance of people at risk of inherited cancers. It is unclear whether the use of registries will translate into improvements in important clinical outcomes.

Introduction and aims

Cancer is often diagnosed at a late stage when treatment options are limited, and prognosis (likely course of the condition and its outcome) is worse. Up to 12% of cancers diagnosed are linked to an inherited gene change. The term hereditary cancer syndrome is used to describe conditions with such an inherited gene change, and includes familial adenomatous polyposis (FAP) and Lynch syndrome. People with a hereditary cancer syndrome have an increased risk of developing certain types of cancer, including breast, ovarian, prostate, colorectal and brain cancer.

Registries (information systems that collect, store and manage data) have been developed for the identification, tracking and surveillance of people at risk of inherited cancers. They may be used with clinical decision support systems and may involve the use of artificial intelligence. The use of registries and associated systems may allow earlier detection of cancer, potentially improving treatment options, quality of life and overall prognosis. Furthermore, surveillance data may form the basis of cancer research, helping to plan and evaluate cancer prevention and control interventions. Genomic testing, recommended for people who either are known to be at risk, or those whose clinical diagnostic data detect they are potentially at risk, may trigger inclusion in a registry.

This topic was identified following an application for a Small Business Research Initiative (SBRI) project by Future Perfect (Healthcare) Ltd.

Health Technology Wales (HTW) researchers searched for evidence on the clinical and cost-effectiveness of registries for the identification, tracking and surveillance of people at risk of inherited cancers.

Evidence overview

Registries for the identification, tracking and surveillance of people at risk of inherited cancers are digital health technologies and have been determined to be a Tier C technology according to the Evidence Standards Framework for Digital Health Technologies. Technologies within this classification include those that inform or drive clinical management.

Health technology assessments and guidance

HTW researchers did not identify any Health Technology Assessments or guidance specifically relevant to the topic under consideration.

Systematic reviews

A systematic review of the impact of registration and screening on colorectal cancer incidence and mortality in familial adenomatous polyposis and Lynch syndrome was carried out by Barrow et al. (2013). A total of 43 studies were included, of which none were randomised controlled trials (RCTs). A significant reduction of colorectal cancer incidence and mortality was reported with registration and screening (FAP: 33/33 studies; Lynch syndrome: 9/10 studies). Five studies (FAP: 2; Lynch syndrome: 3) provided evidence for complete prevention of colorectal cancer-related deaths during surveillance. Pooling of data for meta-analysis was not possible due to clinical and statistical heterogeneity.

Individual studies

No further clinical trials were included within the scope of this TER.

Economic evaluations

HTW researchers did not identify any economic evaluations.

Ongoing studies

This topic was identified following an application for a Small Business Research Initiative (SBRI) project by Future Protect (Healthcare) Ltd. This is a five-month project in Northern Ireland to create a population-wide registry to support surveillance and tracking of people at risk of inherited cancer, and also to support research and clinical trials to improve their prevention and treatment options. It is unclear when results will be available.

Evidence overview

No ongoing studies that are due to complete in the next 6-12 months were identified as part of the HTW literature search.

Areas of uncertainty

- The clinical evidence focuses on a small population of inherited cancers, and the systematic review by Barrow et al. (2013) does not include any RCTs. It is unclear whether the use of registries for the identification, tracking and surveillance of people at risk of inherited cancers will translate into improvements in important clinical outcomes.
- This technology would be reliant on accuracy/reliability of genomic testing, which would impact any outcomes.
- There is potential for the use of artificial intelligence with this technology. However, no evidence evaluating its use was identified.
- No economic evaluations were identified.
- There is a lack of clinical and cost-effectiveness evidence on registries for the identification, tracking and surveillance of people at risk of inherited cancers.

Literature search results

Health technology assessments and guidance
No health technology assessments or guidance were found within the scope of this topic exploration report (TER) that referred to the use of inherited cancer registries.
Evidence reviews and economic evaluations
Barrow P, Khan M, Lalloo F, et al. (2013). Systematic review of the impact of registration and screening on colorectal cancer incidence and mortality in familial adenomatous polyposis and Lynch syndrome. British Journal of Surgery. 100(13): 1,719-31. doi: https://doi.org/10.1002/bjs.9316.
Individual studies
No individual studies were found within the scope of this TER.
Ongoing research
No ongoing studies that are due to complete in the next 6-12 months were identified as part of the HTW literature search. However, there is a five-month Small Business Research Initiative (SBRI) project to create a population-wide registry to support surveillance and tracking of people at risk of inherited cancer, and also to support research and clinical trials. It is unclear when results will be available.

Date of search	February 2025
Concepts used	At risk; Future Perfect; genetic; hereditary; inherited cancer registries; oncology.

Proposed research question and evidence selection criteria (if selected)

Proposed Research question	**Are registries for the identification, tracking and surveillance of people at risk of inherited cancers clinically and cost-effective?**

	Inclusion criteria	Exclusion criteria
Population	People at risk of inherited cancer	People at risk of non-inherited cancer People already diagnosed with cancer (unless predisposed to inherited cancer)
Intervention	Inherited cancer registries	
Comparison/ Comparators	Standard approach without registry	
Outcome measures	Time to diagnosis Cancer incidence Mortality Health related QoL Resource use Economic outcomes	

Proposed speciality	**Cancer**