Final Report: Emerging Point-of-Care Testing (POCT) in Genomics

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Conflicts of interest

No conflicts of interest to declare.

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Contents

Abstract 5

Definitions 6

Background 7

Aim 8

Methods 8

- **Search Strategy** 8
- **Selection Criteria** 9
- **Screening** 10
- **Quality Assurance** 10
- **Data Extraction** 11
  - AI-Assisted Semi-Automation of Data Extraction 12
- **Data Analysis** 14

Results 14

- **Identified Studies** 14
- **Overview of Included Studies** 16
- **Number of Products** 17
- **Innovation Pipeline** 19
- **Characteristics of Technologies** 22
  - Sub-Analysis of TRL 5-9 Technologies 22
  - Timeline 27
  - Purpose 28
  - Setting 30
  - Operator 30
  - Sample Type 31
  - Genomic Target 32
  - Condition 35
  - Other Data 37

Discussion & Conclusions 39

Appendix 41

- **Appendix A. Searches** 41
- **Appendix B. Screening Guide** 47
- **Appendix C. Data Extraction Guide** 48

Appendix D. Project Protocol.....51
References.....64

Abstract

There is a pressing need to identify point-of-care tests (POCTs) that can rapidly and accurately analyse genetic variation in humans without requiring specialist knowledge. This horizon scan aimed to provide an overview of the pipeline of POCTs to identify variation(s) in the genome and epigenome that enable the use of genetic information to inform diagnosis, prognosis, and treatment decisions in any clinical area. The pipeline was focused on POCTs with the potential to reach the market in the next five years. Searches were conducted on Embase and MEDLINE from 2019 to the date of the search (19th December 2024). ClinicalTrials.gov was also searched for clinical trial records with a primary completion date of 1st January 2018 onwards. The scan identified 350 unique technologies. The most common purpose was diagnosis (n=266), with 22 for treatment decisions, and three for prognosis. Additionally, there were 55 technologies for which there were multiple purposes, which included different combinations of diagnosis, prognosis, and treatment decisions, and four for which the purpose was not reported. The technologies were being developed for a range of conditions, with 18 different condition classifications as per topics used by the National Institute for Health and Care Excellence (NICE) represented. Cancer was the most common, accounting for 68.9% (n=241) of the technologies. Within cancer, 17 specific types were represented when analysing at the second level of NICE classifications (i.e., sub-conditions within the first level of classifications). To differentiate between 'true point-of-care' technologies, and those that were not (classed as 'near-patient'), a definition was used which included being highly automated, capable of using complex samples, and able to be operable by a non-specialist. Only 36 of the technologies were classed as being true point-of-care, with the others not yet able to be classified as such but presumed to be in development as POCTs. Some of these technologies may evolve into true POCTs within the five-year timeline to market, as defined in this horizon scan. Technology readiness levels (TRLs)[a] were used to determine the market readiness, along with tiers used to group the technologies based on principles including CE mark, clinical trials, high automation, use of complex samples, and clinical validation. Most of the technologies were at earlier stages of development (TRL 3 or TRL 4; n=266, 76.0%), with few at clinical trial stage (TRL 5 or TRL 6; n=5, 1.4%) or CE-marked (TRL 9, n=19, 5.4%). There was also a portion for which limited available information meant that a TRL could not be determined (n=60, 17.1%). Overall, a wide range of genomic POCTs in development were identified, though relatively few were at later stages of development and/or yet able to be classified as being 'true POCTs'.

[a] TRLs defined using the UK Research and Innovation (UKRI) guidance. TRL 3: early proof of concept demonstrated in the lab. TRL 4: late proof of concept demonstrated in real life conditions. TRL 5: technology refined and ready for initial human trials. TRL 6: technology has completed initial trials and demonstrates preliminary safety data. TRL 9: regulatory approval/notification takes place, product is made available to the market.

Definitions

POC: This is our definition of a true 'point-of-care' technology. These technologies are capable of processing complex biological samples, followed by signal amplification and integrated analysis, all in an automated manner. These technologies can conceivably be used by an operator who is not specially trained in laboratory techniques. Any technologies not displaying at least moderate levels of automation and integration, suitable for an untrained operator, will automatically be allocated as 'Near-patient'.

Near-patient: These tests can conceivably be carried out in the same hospital as the patient resides at the time, without needing to send any samples to a specialised genetics laboratory. Due to the varying development levels of technologies, many do not fit into the POC category by not completely fitting the established criteria above. However, these technologies still predominantly refer to themselves in the text as POCTs, and presumably are being continuously developed to reach the POC criteria.

Send-off: In the articles that have been sourced, the use of the term 'POC' has been applied to a number of different technologies. Upon further investigation into development and commercial availability, some of these products are marketed through the lens of convenience for the patient, meaning that the individual deposits a sample at home, and then mails it to the company, where they will carry out the test. Whilst not rigidly fitting in the established criteria for a POCT, for full comprehensiveness, they have been included in the final results.

Specialist: This operator refers to someone who is specially trained in laboratory techniques pertinent to the critical steps of genomics analysis e.g., DNA extraction, followed by amplification and analysis. Importantly, only one specialist step of the whole process is required for it to necessitate a specialist operator. For example, technologies with a high level of automation only after a purified DNA sample is added to the device will still require a specialist to conduct DNA extraction/purification potentially near the site of patient care (but not at the bedside) prior to the use of the technology. Additionally, for technologies with insufficient methodological sections that do not convey the full process in an articulate manner, an assumption that a specialist operator will be required has been made.

Non-specialist: This operator refers to someone who is not specially trained in laboratory technique (e.g., DNA extraction, precision pipetting), and whilst they may not necessarily understand the theory underlying the processes taking place throughout the usage of the technology, they remain capable healthcare practitioners that are well suited to carry out POC testing at the bedside e.g., nurses.

Background

Point-of-Care Testing (POCT) technology is defined by NHS England as 'any medical device and/or system that enables diagnosis, monitoring or screening of patients at the time and place of care by appropriately trained users'.[1] Additionally, according to Public Health England, POCT refers to tests which may be 'performed at or near the site of patient care, undertaken by healthcare professionals who may not be trained laboratory staff. It is a test to support clinical decision making, and for which the results can be available in real-time'.[2]

There are extensive examples of POCT in everyday use, such as blood glucose meters, pregnancy tests, and haemoglobin subtype tests.[3] However, for the majority of genetic-associated conditions, POCT often focus on diagnosing through the physical manifestation (the phenotype) rather than the genetic root cause (the genotype). For example, sickle cell disease is caused by inheriting two copies of an altered Haemoglobin Subunit Beta (*HBB)* gene, which produces a phenotype of sickle-shaped haemoglobin. There are many available POCT that will diagnose sickle cell disease based upon this phenotype; however, scientists have identified hundreds of varying *HBB* genotypes that cause abnormal haemoglobin to form, which may produce alternative reactions to any potential future treatments.[4] Therefore, as our understanding of the underlying links between genetic variations and the functional mechanism of the phenotype they produce evolves, it becomes increasingly important to be able to diagnose an individual on the molecular scale, to properly allocate the correct treatment.

Variations in an individual's genome and epigenome directly affect disease onset, burden, response to treatments, and a host of other factors.[5-7] For diseases with complex genetic factors affecting diagnosis, prognosis, and treatment-response, it is of great importance that there is access to rapid, accurate, and simple tests to inform timely clinical decision making.[8-10] Next-generation sequencing (NGS), the current benchmark for genetic analysis, employs a strategy of genome fragmentation and individual sequencing of these segments.[11] Despite its increasing integration into clinical standards-of-care, NGS is often reserved for later-line treatment following the failure of alternative therapies. This approach however contains limitations: specifically, it requires complicated setup, trained personnel, and a lengthy turnaround time. A study of 1,767 patients (January 2020-December 2021) reported an average of 17.9 days between NGS order and result delivery, during which 33 patient deaths occurred; the subsequent NGS analysis of these fatalities indicated actionable treatment modifications in all cases.[12] Even relatively simple genetic testing using polymerase chain reaction (PCR) workflows, first established in 1985, require specialised reagents made ahead of time depending upon the genetic variation being investigated; again, time-consuming and requiring a specialist.[13] This supports the case for the development of genomic POCT technologies for the rapid identification of disease-associated genetic variations including single nucleotide polymorphisms, copy number variations, gene fusions, aberrant expression, and excessive methylation.

The National Institute of Health and Care Excellence (NICE) provides guidance on all new innovative healthcare technologies for UK healthcare providers. As of March 2025, there are very few NICE-recommended genomic POCTs. One example is the Genomadix CYP2C19

System, which is used to detect specific mutations in the cytochrome P450 enzyme gene, *CYP2C19*.[14] This directly affects treatment-decisions regarding the antiplatelet drug clopidogrel, which is metabolised by the CYP2C19 enzyme. Certain *CYP2C19* mutations impair the individual's ability to metabolise clopidogrel, thereby reducing the therapy's effectiveness for those individuals. The Genomadix system only requires the input of a buccal swab, followed by automatic and integrated DNA extraction, PCR amplification, and fluorescence-based detection of *CYP2C19* alleles within 55 minutes.[14] This test meets the ASSURED criteria (Affordable, Sensitive, Specific, User friendly, Rapid, Equipment free, Delivered) for POCT technologies, compiled by the World Health Organization (WHO), who stated that the test in question should be: affordable; sensitive; specific; user-friendly; rapid and robust; equipment-free; and deliverable to end-user.[15]

New discoveries are made daily to the underlying genetic aetiology of complex diseases. As genetic testing becomes mainstreamed into standard patient care they may be increased interest in using POCT technologies for testing in certain settings alongside traditional laboratory testing.

Aim

The aim of this project is to provide an overview of the pipeline of POCT to identify variation(s) in the genome and epigenome that enable the use of genetic information to inform diagnosis, prognosis, and treatment decisions in any clinical area. To achieve this, the objectives are to identify and describe POCT technology (standalone tests) in genomic medicine with the potential to reach market in 5 years or under.

In this report, we provide a comprehensive overview that includes the background, methodologies, and findings from our searches. We outline the criteria utilised for data collection, the analytical techniques applied, and the key insights derived from our horizon scan. This structured approach ensures a thorough understanding of the subject matter and facilitates informed decision-making.

Methods

Search Strategy

Searches were conducted on Embase and MEDLINE on the Ovid platform on 19th December 2024. Considering the risks of unmanageable volumes of non-relevant results (due to the lack of standardisation in terminology and the nuances of inclusion criteria within a vast and rapidly emerging field), a variety of approaches in terms of combinations of search terms were employed to achieve a practical balance between specificity and sensitivity. These approaches included, for example, using not just direct synonyms for 'POC', but also results that may refer to 'simplified' or more 'rapid' testing processes. Results were not limited by article type but were restricted to material published in 2019 or later.

MEDLINE and Embase were chosen due to being the most comprehensive bibliographic databases indexing medical and biomedical journals likely to be relevant to this project – plus the Ovid platform has functionality to implement complex, nuanced searches. Searching databases with broader or more technology-focused coverage was considered, but with less sophisticated search interfaces and more material less related to potential clinical use, the risk of getting an unmanageable volume of irrelevant results was deemed too great.

A preliminary search was also conducted on the ClinicalTrials.gov trial registry. This search was screened separately to the Embase and MEDLINE search. The nature of trial registry records means it is difficult to search for relevant records without already knowing the names of technologies being used. Therefore, ClinicalTrials.gov was searched again, later, for the specific product names (and related terms) identified during extraction. Reviews which were identified from the database searches were also used to find additional relevant studies.

The details of the search strategies are provided in Appendix A. Searches.

Selection Criteria

The inclusion criteria for the review were determined a priori in terms of Population, Intervention, Comparator and Setting (PICOS):[16]

Population: Any (children and adults)

Intervention: POCT technology to identify variation(s) in genetic expression in humans only, excluding those for viruses and bacteria. Variation can be to DNA, RNA, or epigenetic.

Comparator: Standard-of-care lab testing, if available.

Outcomes: For the technology to be developed to commercialisation.

Setting: Clinical setting outside of a specialist genomics lab e.g., clinic, bedside OR at a non-specialised hospital laboratory lab. The final developed test should be able to feasibly be carried out by a non-specialist operator (e.g., clinician) for results in real time.

The inclusion criteria specified that POCT will be performed at or near to the site of patient care. Due to the timeframe selected for analysis (2019-2024) and overarching objectives of this horizon scan, i.e., identifying genomic POCT technologies with the potential to reach market in 5 years or under, therein lies a maximum time of 11 years for potential development between a technology reported in 2019 and the cut-off date (2030) for commercialisation.

It is conceivable that, although a technology identified during this scan may currently appear distant from the ASSURED criteria—such as having limited automation or requiring sample preparation by specialist personnel—these limitations may be addressed by 2030.

For this reason, there have been several technologies identified and included that require specialist operators and complex steps, at the request of the stakeholder. Following final extraction of data, these were divided into separate tiers to benefit the stakeholder's goals.

There were nine tiers, with each corresponding to various principles of POCTs, including CE-marked, high automation, use of complex samples, and clinical validation.

Screening

The screening was carried out by four reviewers and performed in two stages: title and abstract, followed by full text. At title and abstract screening, a 10% sample was initially screened independently and blindly by two reviewers. Conflicts were resolved through discussion with all four reviewers to ensure consistency with decisions going forward. The remaining records were then single screened. At full text screening, a sample of 20 records were screened independently by the four reviewers and conflicts again resolved through group discussion, followed by single screening. These 20 records were selected by taking five randomly from each of the four reviewers' allocations. A single screening approach was undertaken due to the large number of records to screen. During the screening process, a guide (Appendix B. Screening Guide) explaining the eligibility criteria was followed by the reviewers to ensure consistency in decisions, and any queries were addressed by another reviewer(s). If a decision could still not be made, then the record was included at this stage.

Quality Assurance

To ensure the accuracy and reliability of our decisions, we implemented a rigorous quality assurance process. Specifically, we sent a representative random sample of 20 of the included studies to clinical scientists and a clinician at NHS England for verification and validation. Random sampling was employed to ensure that every study included had an equal chance of selection. This minimised selection bias and enhanced sample representativeness. This approach also strengthened the validity and generalisability of the findings. The clinical scientists and clinician reviewed our selections, allowing us to confirm the validity of our choices and make any necessary adjustments to screening decisions.

Throughout the project, a document of the questions asked, and answers received from the stakeholders was kept to track decisions made. This collaborative approach enhanced the credibility of our research by incorporating the insights of experienced clinical practitioners, thereby ensuring that our findings are robust and trustworthy. Internal quality assurance was also embedded throughout the horizon scan. The Preferred Reporting Items for Systematic reviews and Meta-Analyses (PRISMA) flowchart[17] was used to report the flow of studies through the screening process. Throughout the screening and data extraction processes, a second reviewer checked any records where the first reviewer had queries. A second reviewer was involved to check queries to ensure the reliability and accuracy of the screening and data extraction processes, thereby minimising the risk of bias and enhancing the robustness of the findings. Following data extraction, two reviewers cleaned the data for consistency, and further quality checks of the data were also undertaken at this stage. Following completion of the data analysis, and feedback from the stakeholders, a further check was conducted for clarity. This consisted of checking a random 10% (n=40) sample of the included records for the publication type (i.e., 'article' which referred to a study containing one potentially relevant technology, or

'review' which referred to a study containing multiple potentially relevant technologies). This extra check revealed no records in the random sample which had been misclassified.

Data Extraction

The following data items were extracted from each of the included studies:

- Product information
  - Developer
  - Test description/name
  - Cost
- Product application
  - Genomic target
  - Condition/indication
  - Time to results
  - Setting (POC; near-patient)
  - Operator (non-specialist; specialist)
  - Purpose (diagnosis; prognosis; treatment decisions)
  - Sample type
  - Preparation
  - Clinical validation
  - Amplification type
  - Analysis type
- Diagnostic accuracy
  - Sensitivity
  - Specificity
- Stage of development
  - Technology readiness level (TRL)

Following data extraction, named products were searched via Google and BEAUDAMED[b] (a search tool based on European database on medical devices (EUDAMED)) for further development information, including CE and UKCA marks. Due to limited information found on UKCA marks, as well as the UK transitional arrangements extending the acceptance of CE marked medical devices on the market,[18] CE marks were used as the primary data for differentiating products with approval. The TRLs were determined based on the UK Research and Innovation (UKRI) guidance for activities associated with different technology readiness levels (available at: https://www.ukri.org/publications/activities-associated-with-different-technology-readiness-levels/). Where a CE mark or clinical trial(s) were identified in association with a product, this was used to inform the TRL. However, due to the majority of the products not having this information, the clinical validation data extracted was primarily used to determine the TRLs.

Data extraction was first carried out in duplicate on a sample of 10 records by all seven reviewers performing data extraction in order to pilot the data extraction form and to ensure

[b] https://beudamed.com/ (accessed 13/03/2025)

consistency across reviewers. Discrepancies with the data extracted between reviewers was identified by comparing the individual data extraction forms completed. Discrepancies were resolved through discussion and any amendments made to the extraction guide (Appendix C. Data Extraction Guide) as necessary to ensure that all reviewers agreed and had clarity on the data extraction items before moving forward. The remaining records were split between the reviewers and data extraction performed by a single reviewer. A second reviewer resolved any queries raised by the first reviewer. The single-reviewer approach was adopted to manage the extensive number of records requiring data extraction within the given timeframe. Following completion of data extraction, the data was cleaned for consistency by two reviewers. This involved removing duplicates, correcting errors, and standardising formats across the dataset. To standardise the condition/indication, the NICE conditions and diseases classification was used. However, the specific conditions as extracted were also kept in for detail. The reviewers also checked for missing values and outliers to ensure the data was accurate and reliable. This thorough process ensured the dataset was ready for further analysis.

AI-Assisted Semi-Automation of Data Extraction

After extracting data from 200 full texts and the clinical trial records (n=8) manually, we switched to a semi-automated, human-in-the-loop workflow for the remainder of the data extraction (161 records). Full data extraction was not performed on the remaining records (n=35) due to either being duplicates (including Genomadix and Genedrive) or excluded. For the duplicates, records were checked to ensure it was the same technology and indication; the most recent or a combination of records were then used as the information source. The PRISMA flowchart shows the total number of studies included and excluded.

Briefly, for the semi-automated data extraction we used OpenAI's 'o1' model, a Large Language Model (LLM) trained with reinforcement learning algorithm. According to OpenAI, it was created for complex reasoning and problem solving tasks with long in-and output contexts[c]. The specific version number of the o1 LLM used during this work is o1-2024-12-17. We implemented an automated extraction pipeline in Python, including PDF-to-text conversion with PyPDF2[d] and automated request and retrieval of LLM output data directly using the official OpenAI Python library[e].

We used data from six already extracted full texts to develop prompts for 16 data extraction targets per included full text (*developer*, *description*, *cost*, *target*, *indication*, *time*, *setting*, *operator*, *purpose*, *sample*, *preparation*, *validation*, *amplification*, *analysis type*, *sensitivity*, *specificity*). For each item, we prompted the LLM to return extracted text as well as a verbatim excerpt from the original text that was used to inform the model's extraction decision.

We then used the six development texts to conduct basic checks for factuality, for example to check if the LLM is returning made-up (hallucinated) data in the absence of valid information within the source text. We specifically prompted the LLM to return "NF" (not found) if it is unable to answer any extraction question. During this quality-checking phase, we identified

[c] https://platform.openai.com/docs/models#o1 (last accessed 28/02/2025)
[d] https://pypi.org/project/PyPDF2/ (last accessed 28/02/2024)
[e] https://pypi.org/project/openai/ (last accessed 28/02/2024)

that some extractions were false-negative (i.e., the information existed in the full text, but automated extraction was incomplete or missing) but we did not identify hallucinations. Two data extraction items, "operator" and "setting" are difficult even for humans to extract which is why we decided to apply extra caution with them.

Going forward, we decided to use a semi-automated, human-in-the-loop approach. Reviewers used the automatically extracted data and relevant excerpts to expedite the process of finding and verifying information in the full text. The following is an example of a case where the LLM extracted data and excerpt for genomic test target from:[19]

- Extracted data: "*BRCA 1 gene*"
- Extracted context: "*Herein, we developed an integrated micro fluidic chip (IMC) that could extract cfDNA from plasma and automatically detect and quantify mutations in the OvCa biomarker BRCA 1.*"

We used the extracted information and the excerpt to quickly locate and manually confirm the relevant information in the full text. The use of AI-assistance resulted in more efficient data extraction speeds, which, combined with an increase in resources, meant that the data extraction could be completed within the timeframe given the large number of included records.

Data Analysis

From the included records, a list of unique technologies (products) was produced. To assess the market readiness of the technologies, a tier system was used in addition to the TRLs. This was to provide additional development information due to limited available information on which to determine the TRLs. The tier system comprised of nine groups which were based on principles relating to the POCTs development: CE mark; clinical trials; high automation; use of complex samples; clinical validation. The tiers corresponded to the following:

- Tier A: CE-marked products
- Tier B: Associated clinical trials
- Tier C: High automation, clinical validation and use of complex samples
- Tier D: High automation and clinical validation
- Tier E: High automation and use of complex samples
- Tier F: Clinical validation and use of complex samples
- Tier G: Clinical validation
- Tier H: Use of complex samples
- Tier I: Other

'Other' encompasses any identified technology for which the principles defining Tiers A-H (i.e., any one or more of CE-mark, clinical trials, high automation, clinical validation, or use of complex samples) did not apply. Further detail on these tiers and the POC aspects used to inform them is described in the subsequent Results section.

Results

Identified Studies

Following de-duplication, 4,616 records were identified from the database search of Embase and MEDLINE for screening. De-duplication was performed using EndNote. Following title and abstract screening, 890 records were brought forward to full text screening. This included 138 reviews, of which 107 were found to not be relevant. Non-relevance meant that the reviews did not contain any genomic POCTs that were within scope. From the remaining 31 reviews, an additional 26 non-duplicate technologies from 24 records were identified. Of the other full texts, 411 were included following full text screening. The clinical trial registry search of ClinicalTrials.gov retrieved 139 records, of which 8 were included. The other 131 clinical trials were excluded due to not containing identifiable POCTs which were within scope, or due to the trial being terminated. In total, 444 records were initially included for data extraction. This large number of records was due to the expansion of the inclusion criteria and resulted in an increase in resources required to carry out the data extraction. At data extraction, a further 40 studies were excluded due to being out of remit. Therefore, data extraction was performed on 404 records in total (396 studies and 8 trials) (Figure 1).

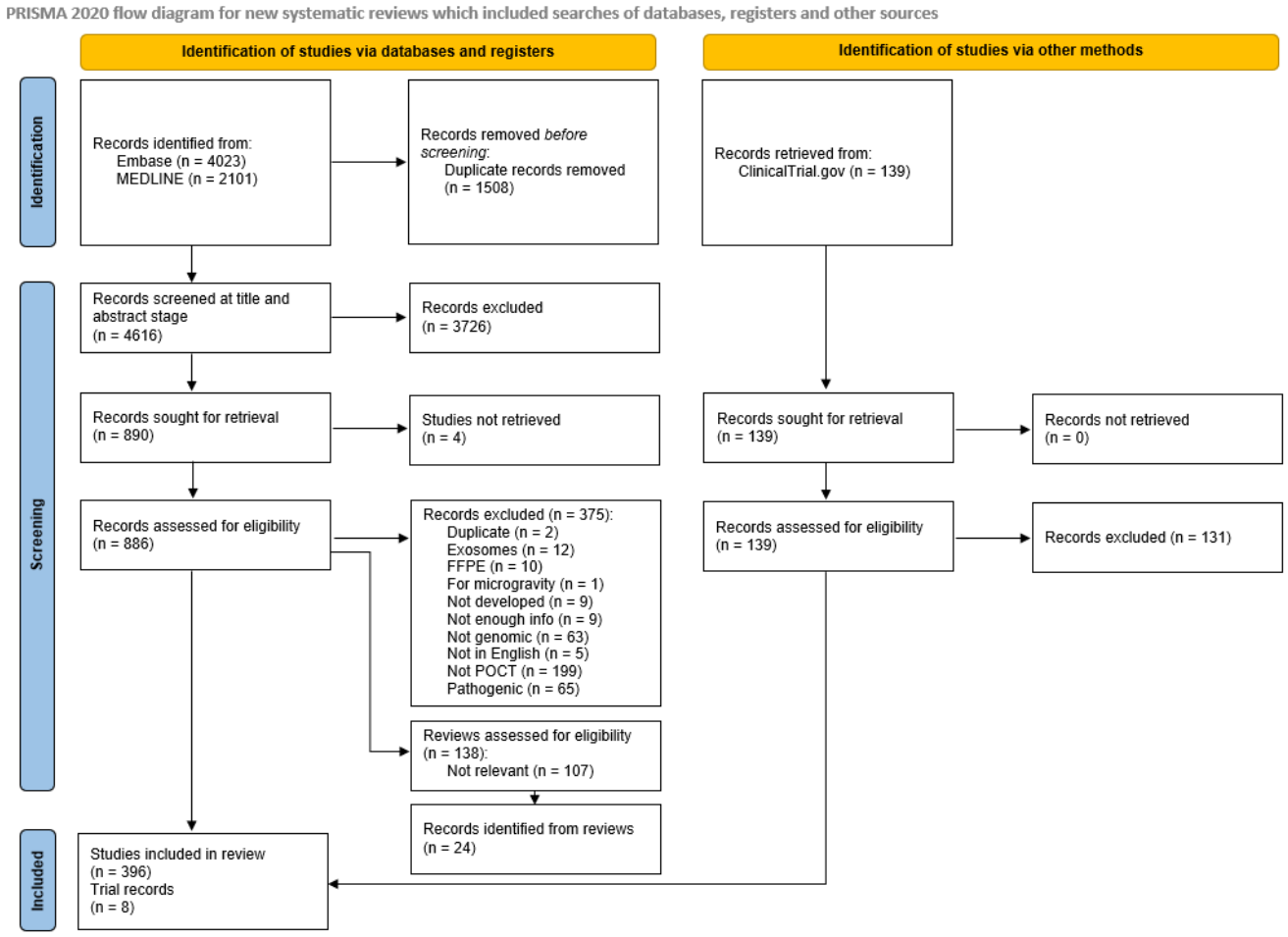

Figure 1. PRISMA flowchart. FFPE: formalin-fixed paraffin-embedded. POCT: point-of-care test.

Table 1 provides an overview of the reasons for exclusion which were used throughout the full text screening.

Table 1. Overview of reasons for exclusion.

Exclusion Reason	Explanation	Example
Not POCT	The most common exclusion reason. These full texts did not sufficiently meet the criteria for a POCT for various reasons including but not limited to: 1. It didn't encapsulate sample to answer workflows, just one part of the process 2. Only identified biomarkers for POCT 3. Optimisation of non-POCT workflows	1: https://doi.org/10.1039/d1ra01732b 2: https://doi.org/10.1093/bjd/ljae105.047 3: https://doi.org/10.3389/fgene.2022.931996
Non-genomic	Only analyses protein (or other analyte) levels.	https://doi.org/10.1039/C9TB01455A
Not medical	Not designed for use in healthcare e.g., forensic identification.	https://doi.org/10.1016/j.fsigen.2022.102788
Pathogenic	POCT for infectious diseases.	https://doi.org/10.1016/j.aca.2024.343041
FFPE	Uses formalin fixed paraffin embedded tissue samples which have been excluded by the stakeholder's request.	https://doi.org/10.1016/j.ab.2020.114087
Exosomes	Exosome quantification POCT, not the genetic material inside. Excluded at the stakeholder's request.	https://doi.org/10.1038/s41378-023-00617-w
Non-human	POCT for biodiversity or forensic applications.	https://doi.org/10.3390/genes10060468

Overview of Included Studies

Most of the included studies were published articles. There were also conference abstracts/posters, preprints, and protocols. The year of publication of the included studies ranged from 2019 to 2025 (Figure 2).

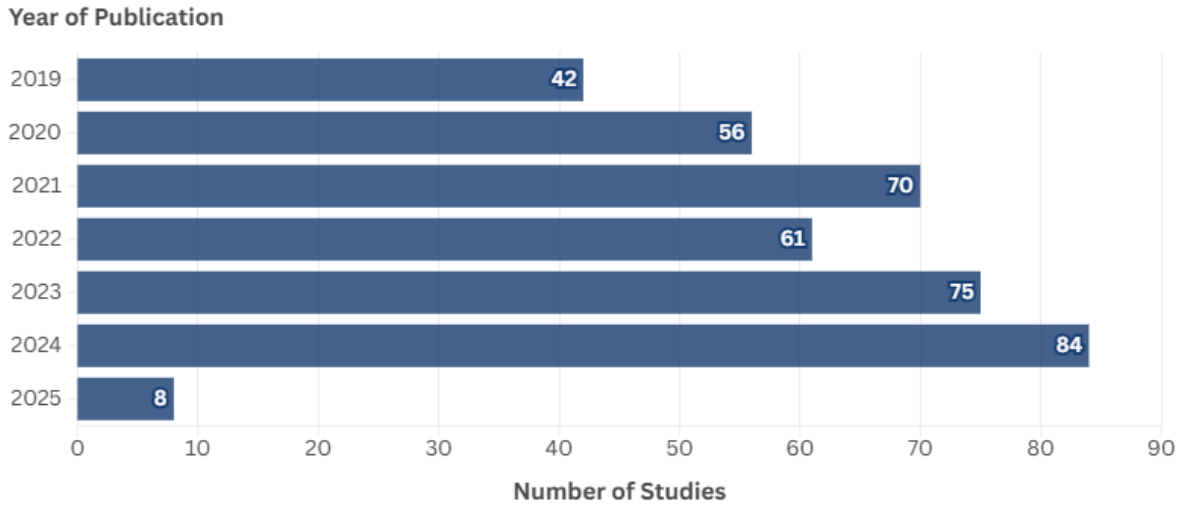

Figure 2. Year of publication of the studies included at data extraction.

From the eight relevant clinical trials from the initial ClinicalTrials.gov search, four unique products (Genomadix CYP2C19 System, Xpert BCR-ABL Ultra, Xpert Bladder Cancer Monitor, and an unnamed qPCR-based assay) were identified; the named three were also identified from the database search. The products identified from all sources were combined in the spreadsheet containing the list of products.

Number of Products

The list of included studies was translated into a list of products. Following de-duplication (as some technologies had multiple studies), 350 unique technologies were identified. Each technology was categorised into one of nine tiers according to descriptions in Table 2. There were 19 in Tier A, four in Tier B, 30 in Tier C, four in Tier D, nine in Tier E, 31 in Tier F, 73 in Tier G, 52 in Tier H, and 128 in Tier I. The tiers were mutually exclusive, with a technology being categorised into only one based on the principles mentioned. It is important to note that while there is some correlation between the tiers and TRLs (e.g., CE-marked products would all be classified as both Tier A and TRL 9), the two systems do not directly translate, hence the tiered system also being used to provide additional development information.

Table 2. Tiers used to group the technologies.

Tier	Description
Tier A: CE-marked products	These are already available, commercial products. Some of these are used in ongoing clinical trials, however these trials have not been included due to these products already being CE-marked.
Tier B: Associated clinical trials	These are named technologies used or studied in clinical trials identified through either the initial clinical trials database search or our second search of identified named products.
Tier C: High automation, clinical validation & use of complex samples	These are the most advanced tests that encapsulate all three main qualities in a POCT technology. They are in order of number of human subjects used in clinical validation in the article associated with them. Importantly, we have classified both serum and plasma as 'complex' sample types.
Tier D: High automation & clinical validation	These require purification of DNA prior to use in the automated technology. They are in order of number of human subjects used in clinical validation in the article associated with them.
Tier E: High automation & use of complex samples	These technologies can use complex samples and are highly automated. They are grouped together by sample type.
Tier F: Clinical validation & use of complex samples	These technologies are not automated but can use complex samples and have human clinical validation. They are grouped together by sample type.
Tier G: Clinical validation	These technologies have clinical validation but require DNA/RNA extraction and are not automated. They are in order of number of human subjects used in clinical validation in the article associated with them.
Tier H: Use of complex samples	These technologies require DNA/RNA extraction and are not automated. They are in order of number of human subjects used in clinical validation in the article associated with them.
Tier I: Other	These technologies do not sufficiently meet any of the three main qualities of a genomic POCT and are not grouped in any particular order.

The tiers were determined by the main three qualities associated with POCT technologies near-market as discussed with the stakeholder:

- **High level of automation** – The end goal of these technologies is to be used by an unskilled operator at the bedside and in a timely manner; this necessitates a "sample-in-answer-out" workflow that requires minimal hands-on time and reduces human error.
- **Use of complex samples** – Similarly, in efforts to reduce hands-on time and keep testing at the bedside, the use of complex biological matrices (blood, urine, saliva) dramatically reduces the time to results compared to if nucleic acid extraction is required; it also demands a less specialised skillset of the operator.
- **Clinical validation** – The greater the level of testing conducted on real-world cases and subsequent results to utilise for the further development of the technology, the more reliable and closer to market the technology will be.

Innovation Pipeline

The TRLs were determined based on the UK Research and Innovation (UKRI) guidance 'Activities associated with different technology readiness levels' (available at: https://www.ukri.org/publications/activities-associated-with-different-technology-readiness-levels/). Nine TRLs are defined (Table 3).

Table 3. Criteria used to determine the TRLs.

TRL	Associated Activities
1	Preliminary idea with well characterised theoretical case
2	Principles are demonstrated through experimentation
3	Early proof of concept demonstrated in the lab
4	Late proof of concept demonstrated in real life conditions
5	Technology refined and ready for initial human trials
6	Technology has completed initial trials and demonstrates preliminary safety data
7	Technology completes secondary trials and demonstrates further evidence for safety and efficacy
8	Technology completes late-stage trials and has all evidence required to prove safety and efficacy
9	Regulatory approval/notification takes place, product is made available to the market

Most of the technologies were determined as being at TRL 3 or TRL 4, with relatively few at higher levels (Figure 3). Specifically, the earlier stages of development (TRL 3 or TRL 4) accounted for 266 (76.0%) of the technologies, while five (1.4%) were at clinical trial stages (TRL 5 or TRL 6), and 19 (5.4%) were CE-marked (TRL 9). There was also a portion for which limited available information meant that a TRL could not be determined (n=60, 17.1%).

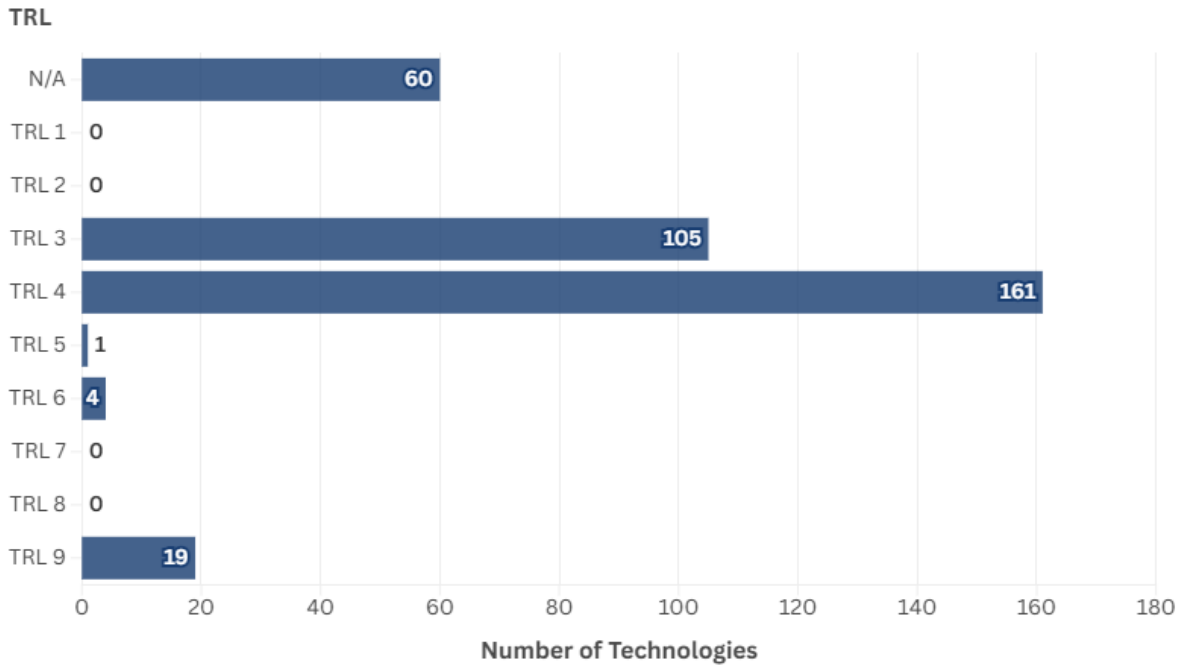

Figure 3. Number of technologies at each TRL.

Information regarding a UKCA mark was only found for three of the technologies: Genomadix CYP2C19 system, Genedrive CYP2C19 ID Kit, and Genedrive MT-RNR1 ID Kit. All three were identified as having UKCA marks.

A technology radar to visualise the number of technologies at each TRL for the different conditions is given by Figure 4.

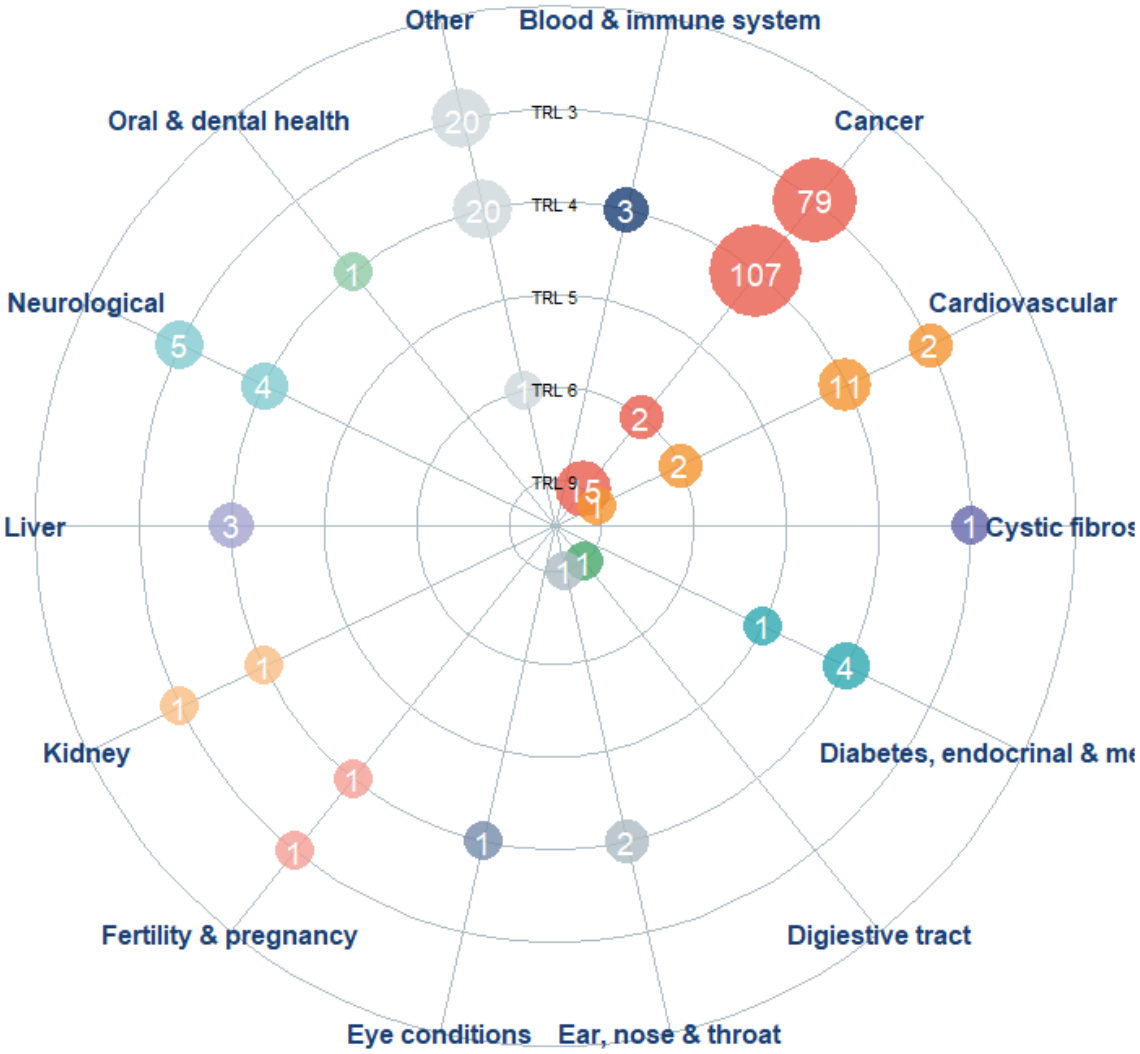

Figure 4. Technology radar showing TRL 3, TRL 4, TRL 5, TRL 6 and TRL 9. 'Other' encapsulates: N/A; N/R; various; multiple long-term conditions (multiple chronic diseases); injuries, accidents and wounds. Note that technologies for which the TRL could not be determined (N/A; n=60) are not included in this radar. Some technologies (n=8) are represented more than once due to having multiple indications. The colours represent the different indications as classified by the NICE conditions and diseases topics.

There were 24 technologies at TRL 5 or above: one at TRL 5, four at TRL 6, and 19 at TRL 9. The second technology radar (Figure 5) visualises these technologies, with the conditions also further broken down. Details of these technologies are also given in the following section (Sub-Analysis of TRL 5-9 Technologies)**Error! Reference source not found.**; full information is included in the spreadsheet of products.

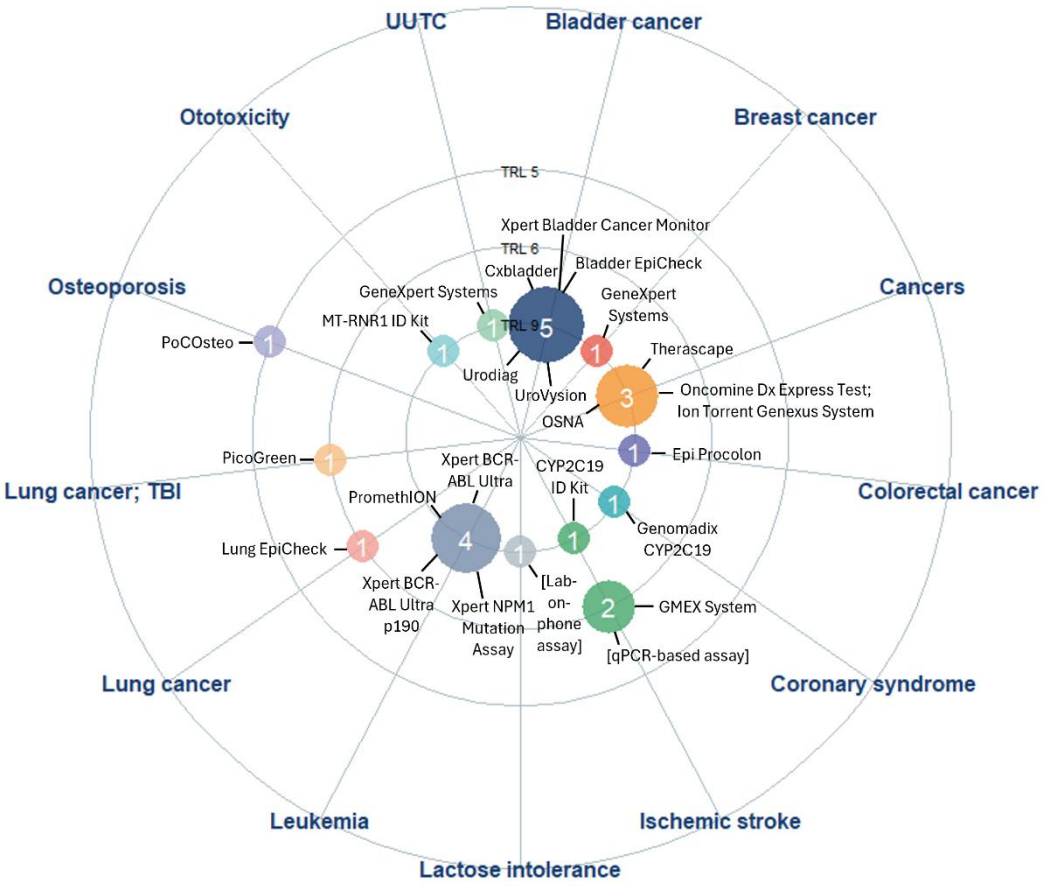

Figure 5. Technology radar showing detail of TRL5, TRL6 and TRL 9. TBI: traumatic brain injury. UUTC: upper urinary tract carcinoma.

Characteristics of Technologies

Sub-Analysis of TRL 5-9 Technologies

A sub-analysis of the technologies identified at the later stages of development (TRL 5, TRL 6 and TRL 9) was carried out in terms of purpose, setting, operator and conditions. There was one technology in TRL 5, four in TRL 6, and 19 in TRL 9, the details of which are given in Table 4.

Table 4. Details of the technologies identified at TRL 5, TRL 6 and TRL 9.

Product (Developer)	TRL	Condition	Target	Purpose	Time to Result	Setting	Operator	Sample Type	CE/ UKCA Mark
PoCOsteo (Tehran University of Medical Sciences)	5	Osteoporosis	WNT16 (rs2908007); RSPO3 (rs10457487); RNMT/FAM210 A (rs4635400); SOST (rs2741856); MCM6/LCT-13910 (rs4988235)	Diagnosis	N/R	POC (in office)	Non-specialist	Whole blood	N/R
GMEX System (Chongqing Jingyin Bioscience)	6	Ischaemic stroke	CYP2C19 (*2; *3; *17)	Treatment decisions	1.5 hours	POC	Non-specialist	Buccal	N/R
Lung EpiCheck (Nucleix)	6	Lung cancer	ctDNA; methylation patterns	Diagnosis	N/R	Send-off	Developer	Blood	N/R
PicoGreen (Invitrogren)	6	Lung cancer; traumatic brain injury	ccf-DNA	Diagnosis/ treatment decisions/ prognosis	N/R	Near-patient	Specialist	Serum	N/R

qPCR-based assay* (University of Virginia)	6	Acute ischemic stroke	CYP2C19	Treatment decisions	N/R	N/R	N/R	N/R	N/R
Genomadix CYP2C19 System (Genomadix)	9	Acute coronary syndrome	CYP2C19 (*2; *3; *17)	Treatment decisions	55 mins	POC	Non-specialist	Buccal	CE; UKCA
CYP2C19 ID Kit (Genedrive)	9	Ischaemic cerebrovascular events	CYP2C19 (*2, *3, *4, *8, *17, *35)	Treatment decisions	69 mins	POC	Non-specialist	Buccal	CE; UKCA
Xpert BCR-ABL Ultra (Cepheid)	9	Chronic myeloid leukaemia	BCR-ABL	Prognosis/ treatment decisions	<2.5 hours	Near-patient	Specialist	Blood	CE
Xpert BCR-ABL Ultra p190 (Cepheid)	9	Philadelphia chromosome-positive (Ph+) ALL	BCR-ABL	Diagnosis	<2.5 hours	Near-patient	Specialist	Blood	CE
Xpert Bladder Cancer Monitor (Cepheid)	9	Non-muscle invasive bladder cancer; bladder cancer in patients with haematuria	ABL1; CRH; IGF2; UPK1B; ANXA10	Prognosis/ treatment decisions/ diagnosis	90 mins	Near-patient	Specialist	Voided urine	CE
Xpert NPM1 Mutation	9	Acute myeloid leukemia	NPM1 mutation (A, B, D)	Prognosis	<3 hours	Near-patient	Specialist	Peripheral blood	CE

Assay (Cepheid)									
GeneXpert Systems (Cepheid)	9	Breast cancer	DNA methylation in ten tumor-specific gene markers	Diagnosis	5 hours	POC	Non-specialist	Fine needle aspirate of lymph nodes	Ce
GeneXpert Systems (Cepheid)	9	Upper urinary tract carcinoma	5 target miRNAs	Diagnosis	90 mins	Near-patient	Specialist	Urine	CE
MT-RNR1 ID Kit (Genedrive)	9	Antibiotic-induced ototoxicity in neonates	MT-RNR1 m.1555A>G	Treatment decisions	26 mins	POC	Non-specialist	Buccal swab	CE; UKCA
Oncomine Dx Express Test; Ion Torrent Genexus System (Thermo Fisher)	9	Various cancers	49 genes	Diagnosis/ treatment decisions/ prognosis	~24 hours (20 mins hands-on time)	Near-patient	Specialist	Plasma	CE
UroVysion (Abbott)	9	Bladder cancer	Aneuploidy in chromosomes 3, 7, 17 and loss of 9p21	Diagnosis	N/R	Near-patient	Specialist	Urine	CE
Cxbladder (Cxbladder)	9	Bladder cancer	mRNA: MDK, HOXA13, CDC2, IGFBP5, CXCR2	Diagnosis	5 business days	Send-off	Developer	Urine	CE

Urodiag (OncoDiag)	9	Non-muscle invasive bladder cancer	FGFR3; HS3ST2; SEPTIN9; SLIT2	Prognosis/ treatment decisions	15 days	Send-off	Developer	Urine	CE
Therascape (aiGENE)	9	Cancers	ctDNA	Treatment decisions	N/R	POC	Non-specialist	Blood	CE
PromethION (Nanopore)	9	Acute lymphoblastic leukemia	High hyperdiploidy, translocation (9;22) or Ph-like leukemia	Diagnosis/ treatment decisions	N/R	Near-patient	Specialist	Bone marrow	CE
OSNA (Sysmex)	9	Prostate cancer (in development); breast cancer; colon cancer; gastric cancer	Cytokeratin19 (CK19) mRNA	Diagnosis	30 mins	Near-patient (intraoperative)	Specialist	Sentinel lymph nodes	CE*
Lab-on-phone assay* (STAB VIDA)	9	Lactose intolerance	MCM6 c.C13910T	Diagnosis	1.5 hours	POC	Non-specialist	Buccal swab; capillary blood	CE
Bladder EpiCheck (Nucleix)	9	Non-muscle invasive bladder cancer	15 methylation markers	Diagnosis	N/R	Send-off	Developer	Blood	CE
Epi Procolon (Bosch)	9	Colorectal cancer	SEPT9 methylation	Diagnosis	N/R	Near-patient	Specialist	Blood plasma	CE

* *OSNA is CE-marked for breast cancer, colon cancer, and gastric cancer*

Purpose

Diagnosis was the most reported purpose (n=11) in technologies identified as TRL 5-9. Six technologies were used for more than one purpose (diagnosis, prognosis and treatment decisions (n=3); prognosis and treatment decisions (n=2); and diagnosis and treatment decisions (n=1)). Treatment decisions were reported as the purpose for six technologies in this category and one was used for prognosis.

Setting and Operator

The technology in TRL 5 (PoCOsteo) is being developed as a POC device and does not require a specialist for its operation. In the TRL 6 category, one has a POC setting and does not require a specialist operator, one of the devices was sent to the developer for analysis, one is being developed for near patient use and requires a specialist, while the setting and operator required for the fourth technology was not reported. Of the 19 technologies that were in the TRL 9 category, 6 had a POC setting and could be operated by a non-specialist. Ten were developed for use in a near-patient setting requiring specialists, and for the other three technologies, the samples are sent to the developer/laboratory for analysis (send-off).

Condition

The majority of the 24 technologies in TRL 5-9, according to NICE classifications, target cancer (n=17), followed by cardiovascular conditions (n=4); ear, nose and throat conditions (n=1); digestive tract conditions (n=1); injuries, accidents and wounds (n=1); and diabetes and other endocrinal, nutritional and metabolic conditions (n=1). Notably, one of these technologies targets both cancer and traumatic brain injury through analysing levels of circulating cell free DNA, which is upregulated in both conditions.

Of the 17 TRL 5-9 technologies that target cancer, 20 cancers are named as target conditions. The One Step Amplification Assay (OSNA) names four individual cancer types. Bladder cancer is the most prevalent (n=6), followed by blood and bone marrow cancer (n=4); colorectal cancer (n=2); lung cancer (n=2); prostate cancer (n=1); and stomach cancer (n=1). Two technologies did not specify the specific cancer in the available literature and only reported that they targeted multiple cancers.

Timeline

The approximate time to market of technologies may be given in terms of three horizons:

- Emerging horizon: Unknown innovations (approximately 5-15 years from market)
- Transitional horizon: Innovations entering late-stage clinical trials (up to approximately 7 years from market)
- Imminent horizon: Current status quo of innovation (approved or undergoing approval)

Figure 6 represents a timeline of the identified technologies by mapping the TRLs onto a timeline of the three horizons.

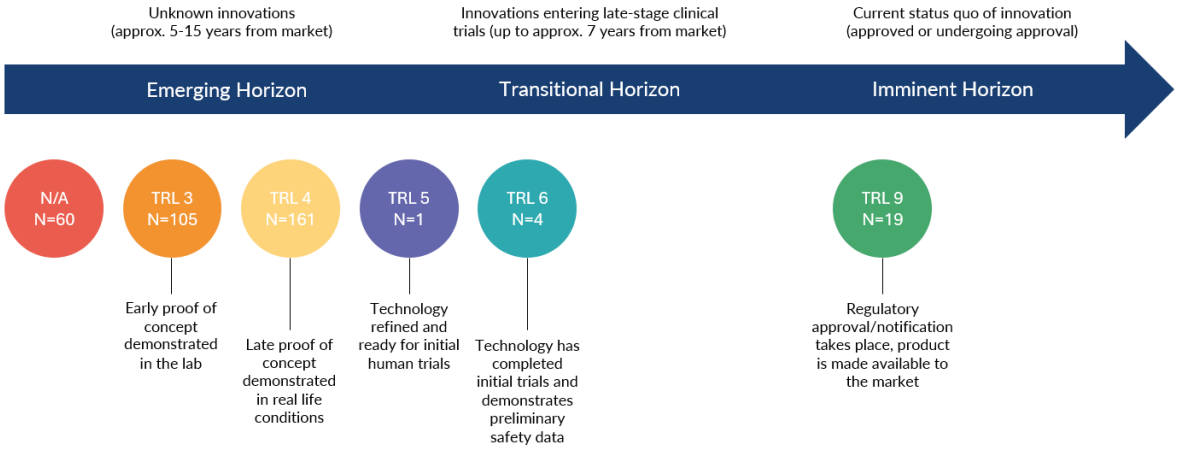

Figure 6. Timeline showing estimated time to market of the technologies.

As shown above, the TRL framework categorises technologies based on their maturity. TRL 9 represents technologies that are currently available on the market, while TRL 6 encompasses technologies undergoing clinical trials. Additionally, there are technologies at earlier stages, such as TRL 4, which may have demonstrated clinical validation outside of formal trials. Consequently, the estimated time to market for these technologies is approximate and may involve some overlap. Although technologies in clinical trials are typically more advanced, the estimated timelines acknowledge the possibility that technologies at earlier stages could achieve market readiness within a five-year period.

Purpose

Figure 7 depicts the purpose of the 350 included technologies. Most of these technologies are in development for use in diagnosis (n=266). This is followed by technologies in development for multiple purposes (n=55). Of the 55 technologies classed as multiple, 17 were in development for diagnosis and prognosis, followed by diagnosis, prognosis and treatment decisions (n=15) and diagnosis and treatment decisions (n=15). The least common in the multiple category was prognosis and treatment decisions (n=8). There were 22 technologies in development for use in treatment decisions. There were the fewest number of technologies in development for prognosis (n=3). The purpose of four technologies was classed as not reported.

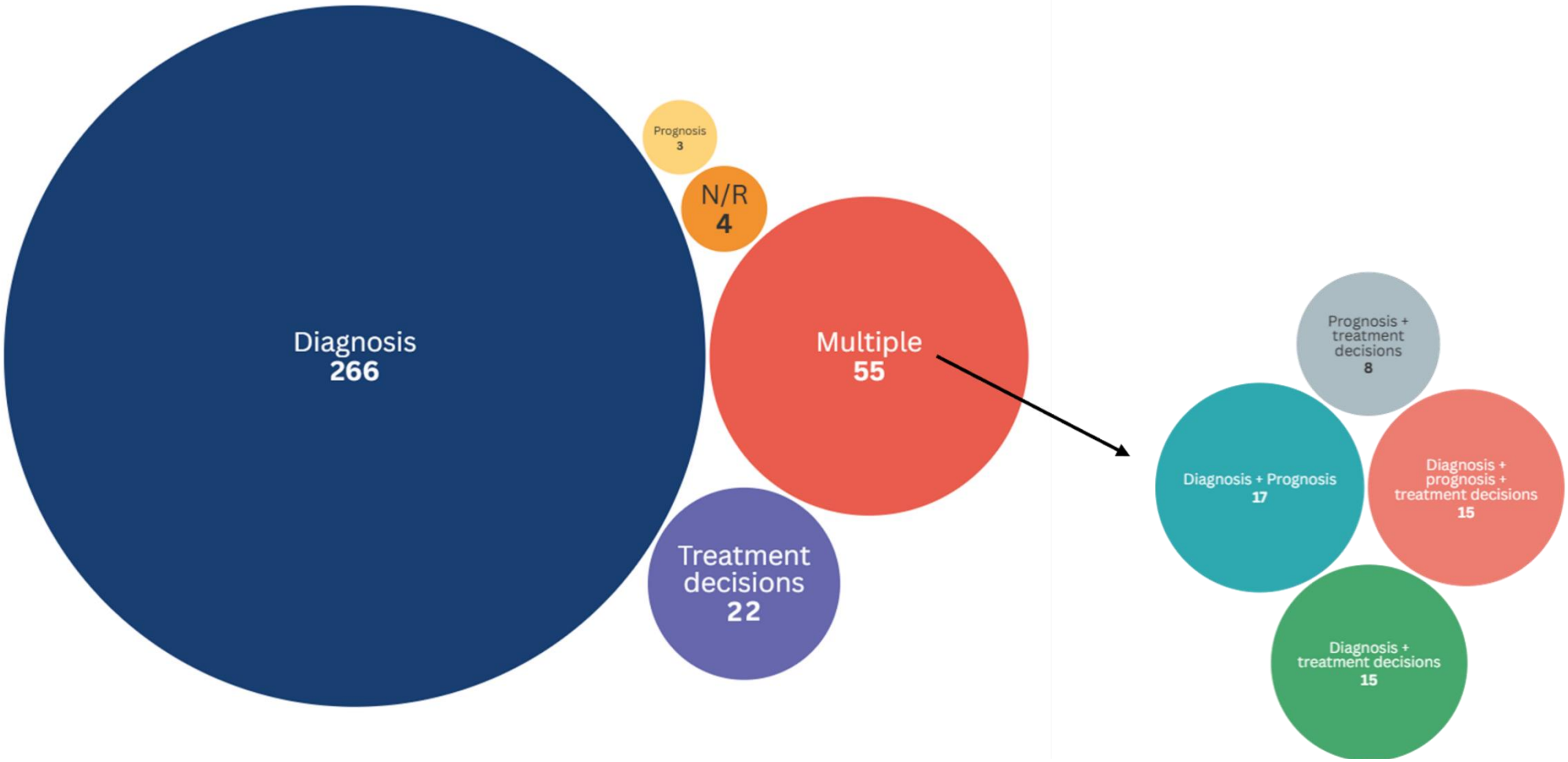

Figure 7. Number of technologies identified by purpose. Those classed as multiple have been separated into their multiple uses.

Setting

The technologies were classified based on the settings where they could be deployed (Figure 8). These include near-patient settings (n=307) and POC settings (n=36). Some of the samples were sent for analysis in another location and were categorised as send-off (n=4). Setting was not reported in three of the included articles.

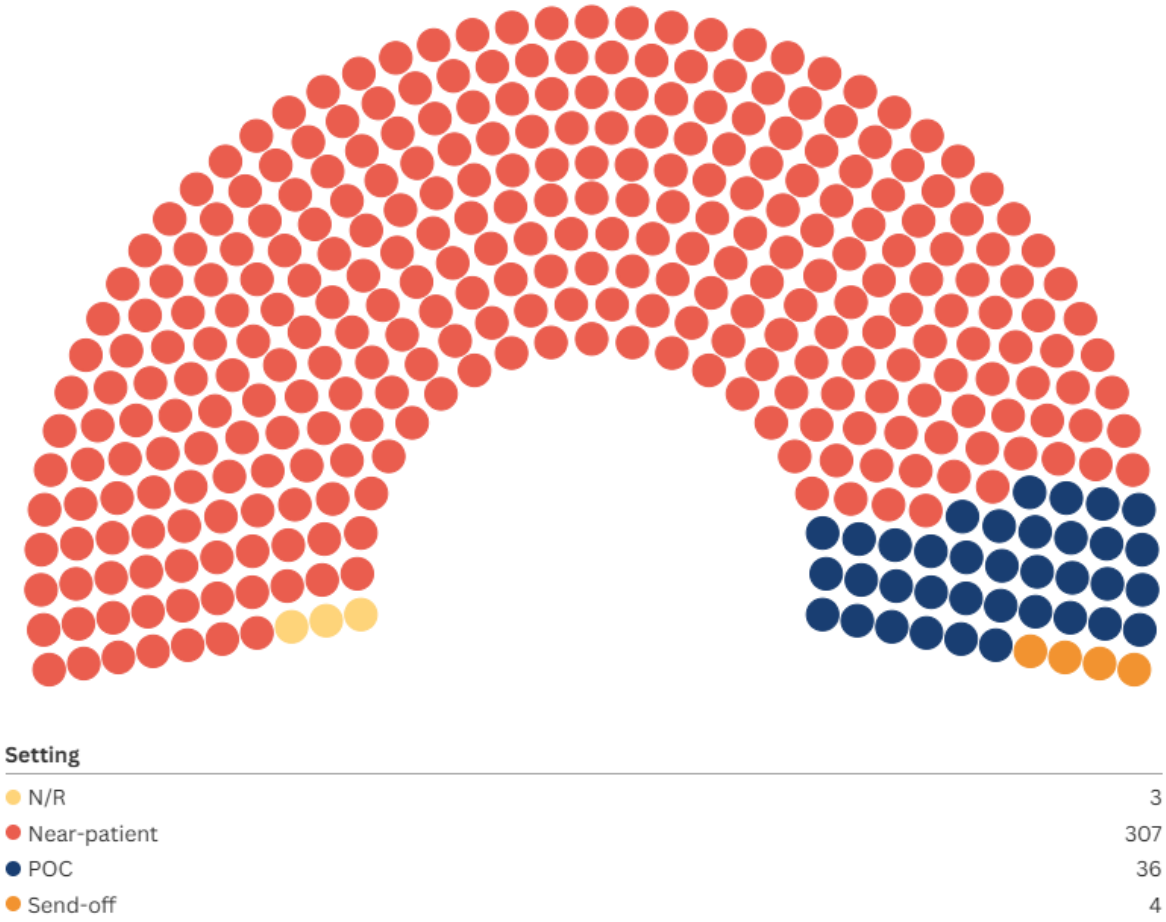

Figure 8. Number of unique technologies categorised by setting.

Operator

The identified technologies were categorised based on the level of specialist knowledge required to operate them (Figure 9). Most of the technologies required a specialist (n=306). However, 37 did not require specialists; these can conceivably be carried out by a healthcare practitioner (e.g., nurse) at the bedside who possess no specialised genomics knowledge or honed laboratory technique. Four technologies were sent to the developing company for analysis (e.g. requires input from "developer"); whilst this contradicts the POC nature of the target technologies, these tests still possess POCT qualities, and the nature of the testing

process and location may be a commercial decision rather than a scientific necessity. Finally, three technologies did not report the skillset required to conduct the test (e.g. “N/R”).

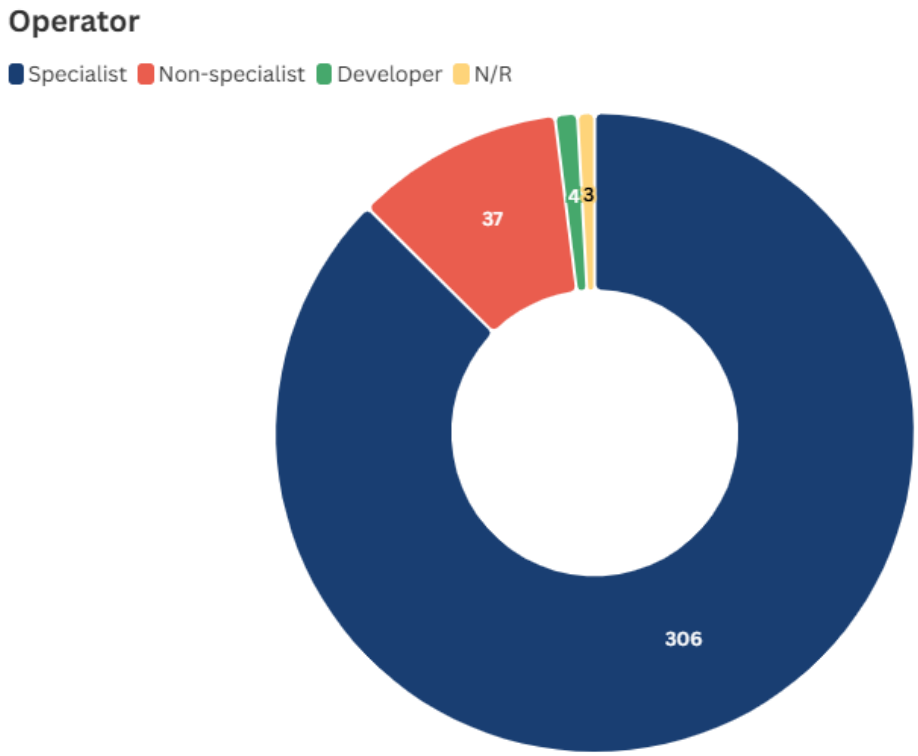

Figure 9. Number of technologies classed by operator.

Sample Type

The technologies identified used varying samples for analysis, and some devices were found to use more than one type of sample (Figure 10). These samples include: serum (n=94); blood (n=65); DNA (n=50); plasma (n=43); RNA (n=41); urine (n=21); buccal swab (n=13); saliva (n=11); body fluids (n=9); cell-based sample (n=7); tumour specimen (n=7); extracellular vesicles (n=3); lymph nodes (n=2); bone marrow (n=2); tissue sample (n=2); gestational products (n=1); and ribonucleoproteins (n=1). Cell-based samples refer to samples like cell lines, cell lysates and cancer cells. The body fluids category included samples like amniotic fluid, synovial fluid and extracellular body fluids. The most frequently used samples in these technologies were serum, blood, DNA, plasms and RNA samples. The sample types used nine of the identified devices were not reported.

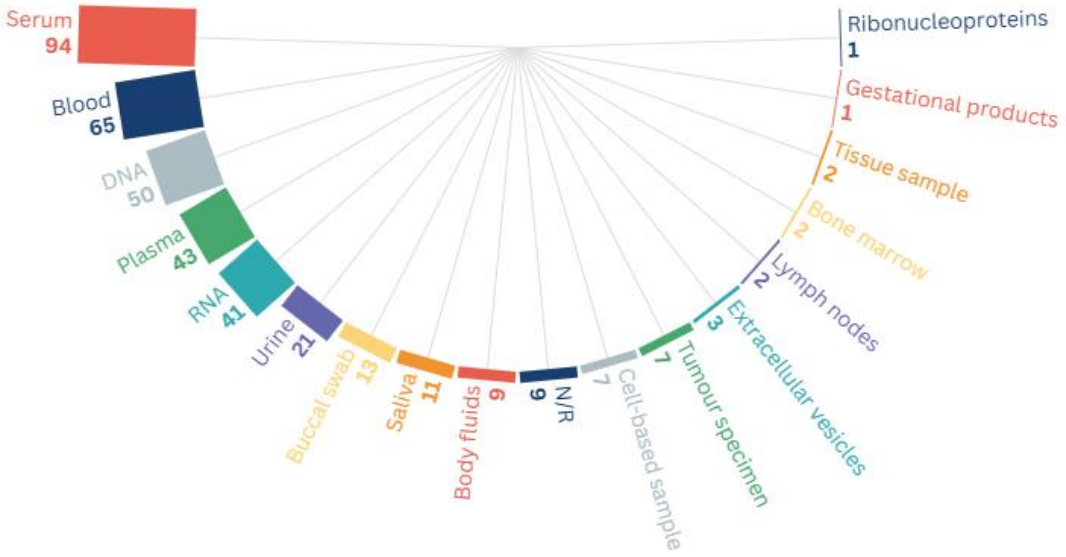

Figure 10. Number of sample types used by the included technologies. Some technologies reported use of more than one sample type.

Genomic Target

The genomic target for each of the technologies are given in Table 5 and Figure 11 in terms of the number of targets at each tier. Gene targeting technologies and miRNA targeting technologies represented 40.6% (n=142) and 40.3% (n=141) of the total technologies, respectively. Very few technologies (n=9, 2.6%) analyse both miRNA and genes; these are predominantly at the early stages of development; none being above TRL 4.

Table 5. Comprehensive overview of each type of technology in each tier (outlined in Table 2). Proof-of-concept (green) technologies are not counted towards totals.

	Tier A	Tier B	Tier C	Tier D	Tier E	Tier F	Tier G	Tier H	Tier I	Total
Inexplicit	3	1	3		3	3	5	5	20	43
Epigenetic	3	1		1		2	1		7	15
miRNA			3		2	18	31	31	56	141
Gene	13	2	24	3	4	8	34	16	38	142
Gene + miRNA							2		7	9
Total	19	4	30	4	9	31	73	52	128	350
Proof-of-concept			*4*		*3*	*3*	*7*	*9*	*19*	*45*

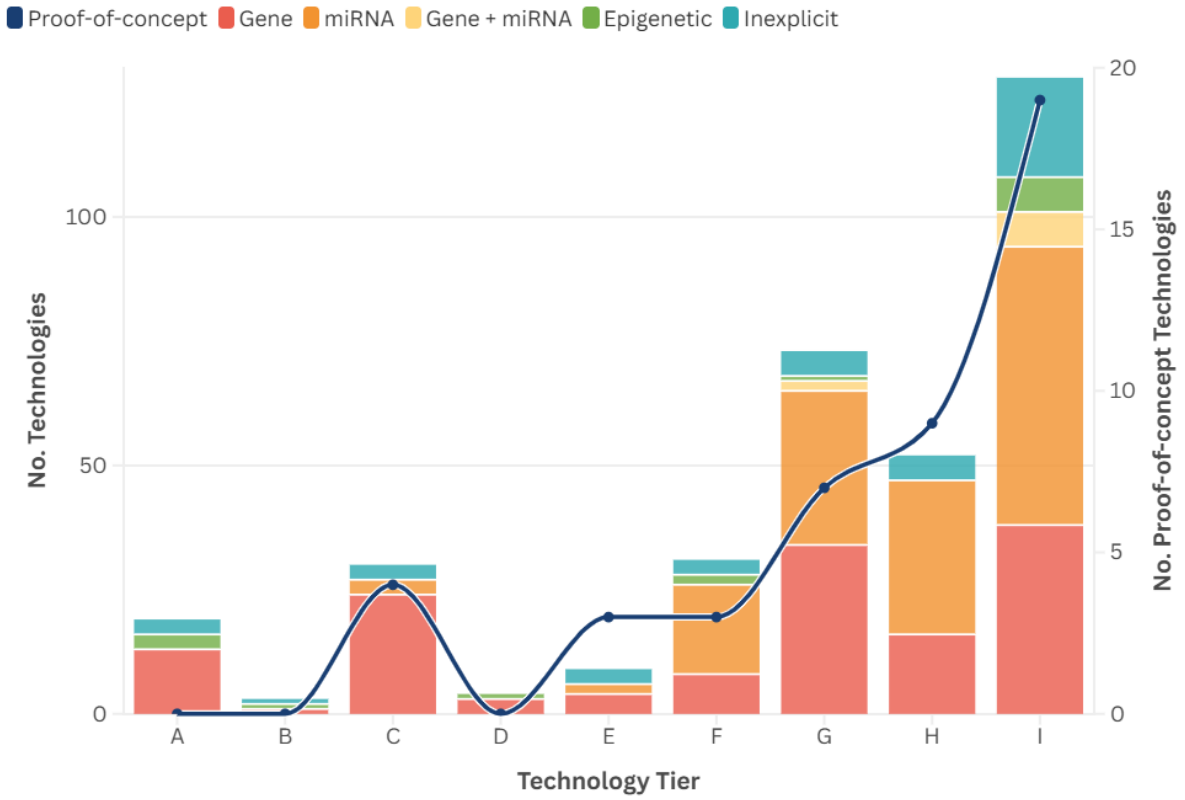

Figure 11. Visualisation of Table 5. Left Y axis represents number of total technologies in each targeting category. Right Y axis indicated how many technologies in each tier are proof-of-concept. X axis represents tier of POCT technologies.

Importantly, 43 technologies (12.3%) are deemed '*Inexplicit*', as they do not contain sufficient information to record which specific gene or miRNA they target e.g., '8-oxo-dG', 'cell-free DNA', '49 genes'. Finally, 45 (12.9%) technologies are claimed by the developer/author as 'proof-of-concept'; these represent a variety of genomic targets across genes (n=24), miRNAs (n=16), both genes and miRNA (n=3), and finally two technologies were methylation targeting. For these technologies, though the type of genomic target (e.g., miRNA) may stay consistent through continued development, the final genomic target may alter.

miRNA

miRNAs are small noncoding transcripts that regulate gene expression posttranscription.[20] The most prevalent miRNA targeted by POCT in these results is miRNA-21, investigated through a variety of methods in 90 (25.7%) of all technologies (Figure 12). This is significantly more than the next most prevalent miRNA targets miRNA-155 and miRNA-let7a, both of which are investigated by 4.3% and 2.6% of the total technologies, respectively. miRNA-21 is one of the most abundant and highly conserved miRNAs, expressed in almost all cells, and is a biomarker of conditions such as cancer and cardiovascular disease.[21]

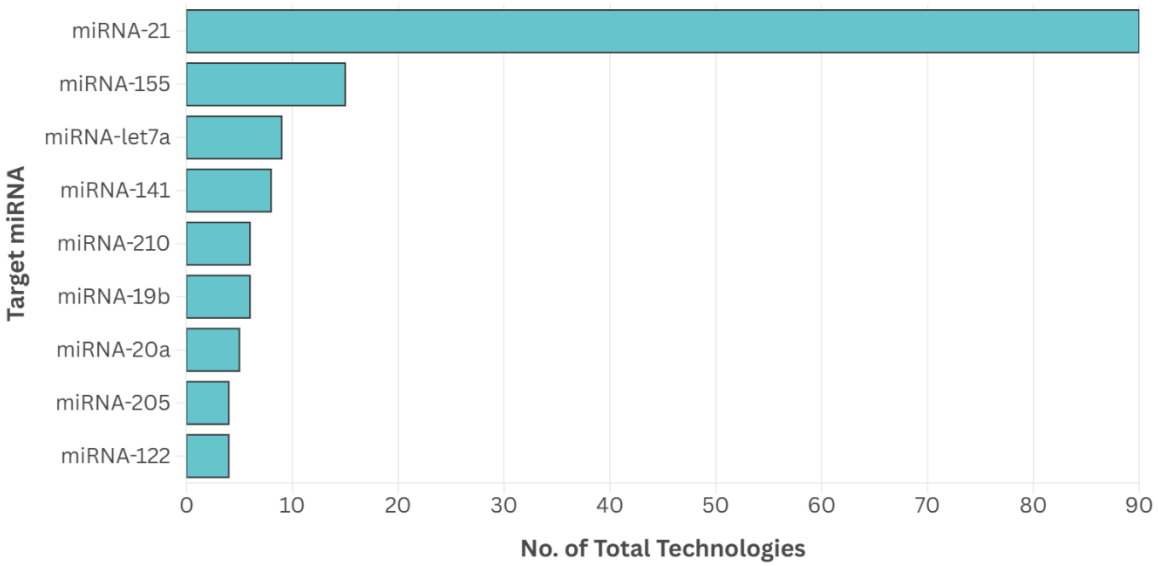

Figure 12. Every miRNA investigated by more than three unique technologies, represented by the number of total technologies targeting them.

Genes

There were 108 unique named genes targeted between the 350 technologies. The top ten most frequently investigated genes (Figure 13) are mainly associated with cancer development; *EGFR* for example, was targeted by 19 (5.4%) of all POCT technologies. Exceptions include *CYP2C19* polymorphisms associated with clopidogrel resistance, and *MTHFR* polymorphisms, particularly p.C677T, which are involved in homocystinuria pathogenesis.

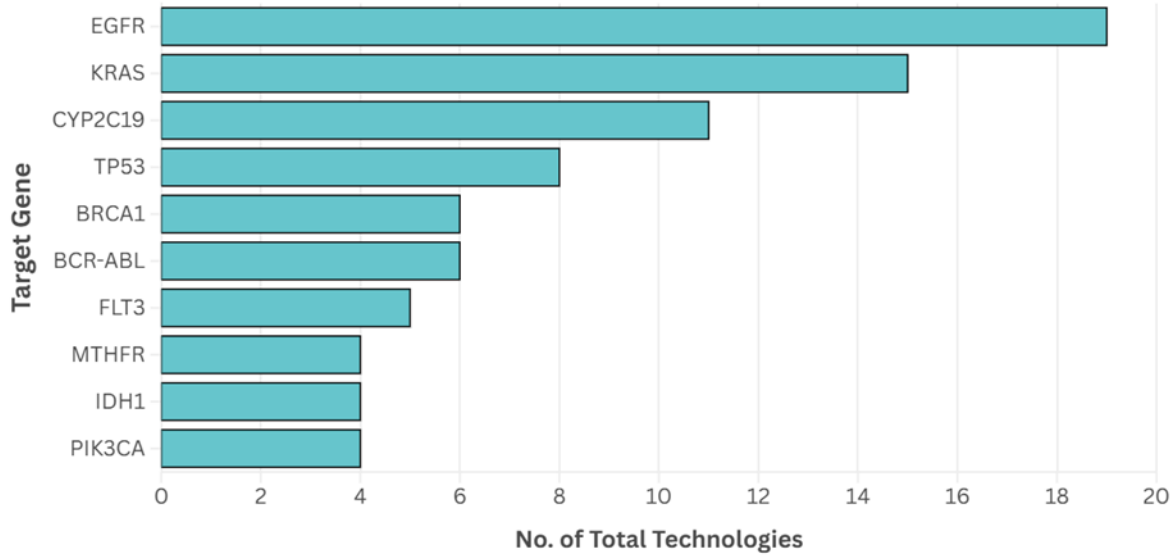

Figure 13. Top ten most frequently investigated genes.

Methylation

Methylation directly affects gene expression levels. Technologies analysing methylation (n=16, 4.6%) can target either genes or microRNAs. To avoid duplication of results, despite the overarching genomic target, if the technology analyses the epigenome, it has only been counted as an epigenetic technology. Of the 16 explicitly stated epigenetic analysis technologies, 4 (25%) analyse the *SEPTIN9* gene, for which abnormal expression caused by methylation has been associated with various cancers.

Condition

To properly standardise the therapeutic areas for which each technology is being developed for, the NICE 'Conditions and diseases' topics were utilised (available at: https://www.nice.org.uk/guidance/conditions-and-diseases) and visualised in Figure 14. Cancer (n=241, 68.9%) is the most prevalent condition for which the POCT technologies are being developed for a variety of diagnosis, prognosis, and treatment decisions.

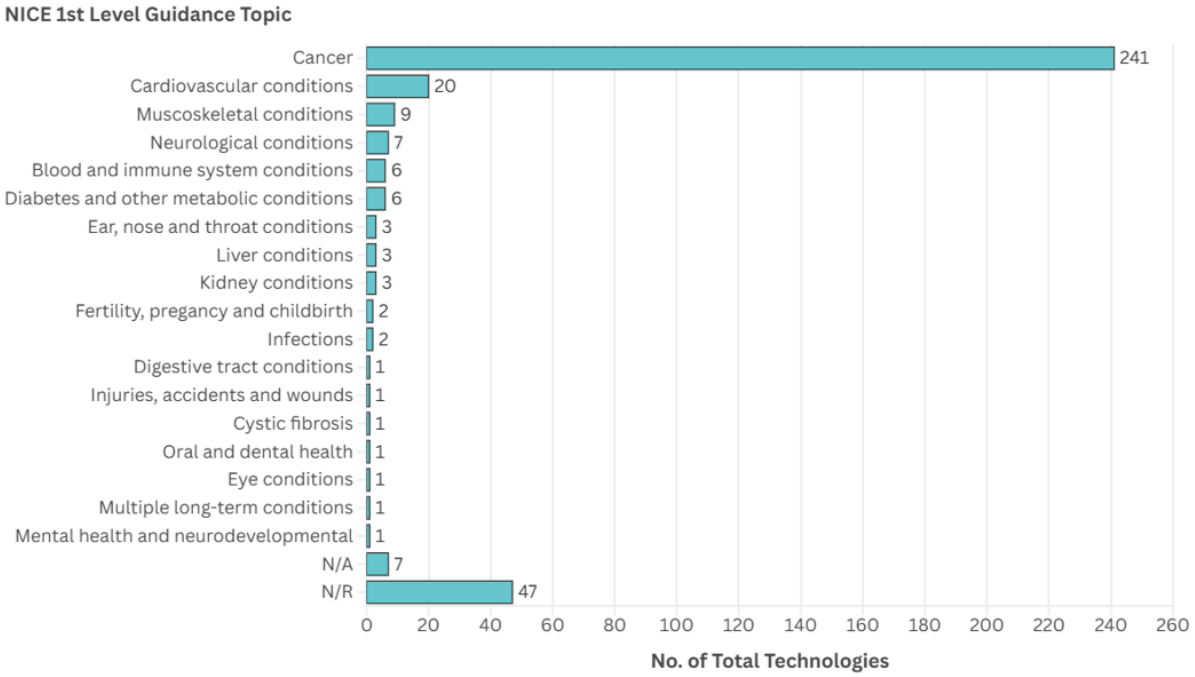

Figure 14. Each NICE 'Condition and disease' guidance topic represented as the number of technologies that are investigating it. N/R (not reported). N/A (not applicable).

There were 47 (13.4%) technologies where the condition was not explicitly reported (N/R), as well as 7 (2.0%) technologies that were not applicable (N/A) to NICE guidance topics. The N/A conditions are as follows:

- Pain sensitivity and perioperative opioid use
- Genetically-induced hair loss
- Radiation exposure assessment
- Oxidative damage
- Sex chromosomal abnormalities
- Non-communicable diseases
- Drug hypersensitivity

Further analysis of cancer subtype according to NICE 'Cancer' guidance topics (available at: https://www.nice.org.uk/guidance/conditions-and-diseases/cancer) yielded 17 broad cancer subtypes that are being investigated by the technologies found in this report (Figure 15). The most common condition investigated by POCT technologies in this search was lung cancer (n=32, 9.2%). However, many technologies (n=88, 35.9% of all cancer-targeting results) do not report a specific type of cancer being investigated.

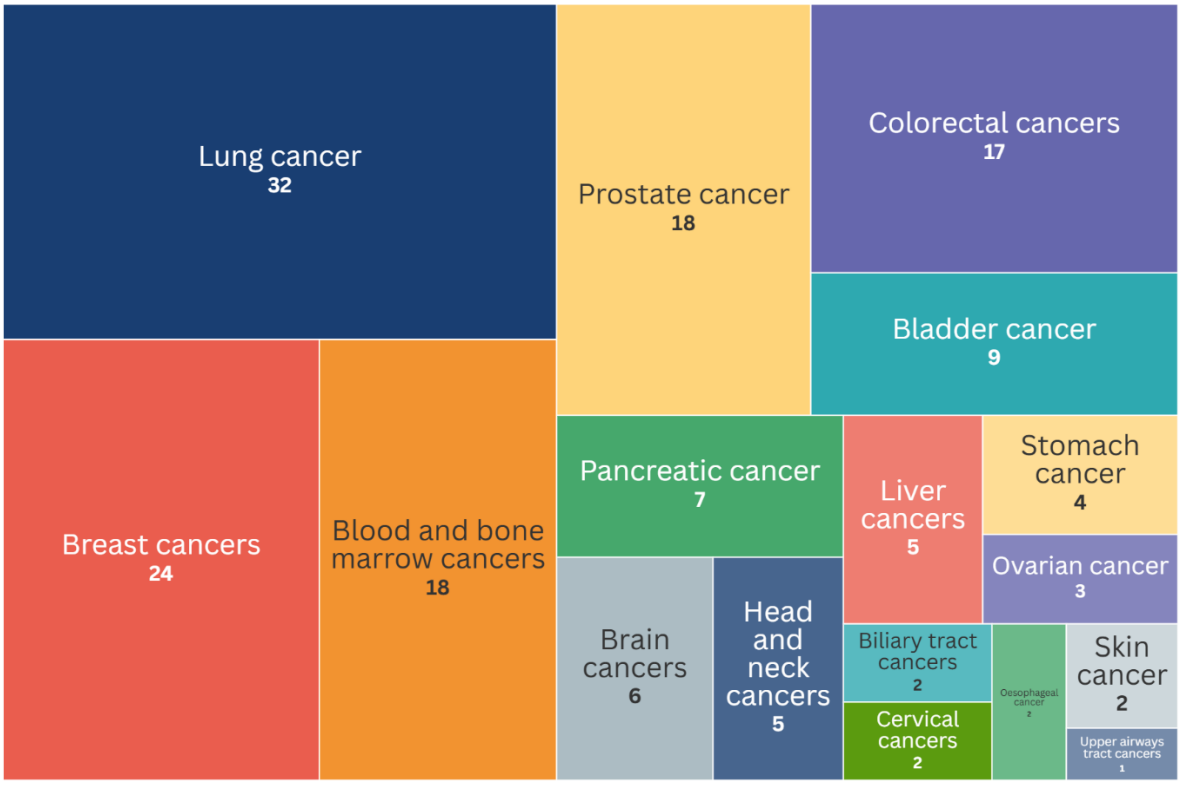

Figure 15. NICE 2nd Level classification 'Cancer' subtypes. Does not include any cancer technologies that did not specify subtype of cancer.

Other Data

In addition to the main data fields for which the results have been reported in the prior sections, other data was also extracted and included in the spreadsheet of products. This included diagnostic accuracy, amplification type, analysis type, and preparation. For each of these fields, there was substantial variation in the data extracted; the main summative points are given in this section.

For diagnostic accuracy, the sensitivity, specificity, and reference standard were extracted where this information was reported. Where specific values were reported (i.e., a percentage), these were used; otherwise, a standardised set of terminology was used (good or moderate) to correspond with that used by the study. For the technologies which had specific sensitivity and/or specificity values reported, the reference standard was also extracted (if this was reported). Fifteen technologies had an identified reference standard, of which seven reported the sensitivity and specificity of the reference standard, two reported only the sensitivity of the reference standard, and four did not report either the sensitivity or sensitivity of the reference standard. For sensitivity, 22 technologies had specific values reported, ranging from 46% to 100%. Forty-six did not report the sensitivity, seven were classed as moderate, and 276 were classed as good. For specificity, 19 technologies had specific values reported, ranging

from 75.53% to 100%. Seventy-one did not report the specificity, nine were classed as moderate, and 252 were classed as good.

Most (n=288, 82.3%) of the technologies had clinical validation reported, though there was variation in this, with some using real patient samples, whilst others used samples such as spiked serum samples or animal samples. There was also variation in the level of detail reported, for example whether studies specified the number of samples that the clinical validation was performed on.

There were various types of amplification for the identified technologies, including PCR and different subtypes (e.g., reverse transcription PCR (RT-PCR), quantitative PCR (qPCR) etc.), which was represented by 69 (19.7%) of the technologies. Other types of amplification included loop-mediated isothermal amplification (LAMP), which was represented by 25 (7.1%), recombinase polymerase amplification (RPA), which was represented by 20 (5.7%), and hybridisation chain reaction (HCR), which was represented by 12 (3.4%). The amplification type was either not reported or not applicable for 102 (29.1%).

There were also various types of analysis for the technologies. This included fluorescence, which was represented by 133 (38.0%), colorimetric, which was represented by 57 (16.3%), electrochemical, which was represented by 52 (14.9%), and surface-enhanced Raman spectroscopy (SERS), which was represented by 14 (4.0%). The analysis type was not reported for 25 technologies (7.1%).

Most (n=307, 87.7%) of the technologies had some amount of required preparation, though the level varied. Examples of required preparation included DNA/RNA extraction and mixing of reagents. For 124 (35.4%) technologies, 'multiple specialised steps' was recorded. For 25 technologies (7.1%), there was no preparation required. For 18 (5.1%), this information was not reported.

Discussion & Conclusions

In this horizon scan, we identified 350 POCTs in use or development. Most (n=307, 87.7%) of these were at earlier stages of development and could not yet be classified as 'true POCTs' according to our definition of being highly automated, capable of processing complex biological samples, and able to be operated by a non-specialist. This means that many of the identified technologies were instead classed as 'near patient' but are presumed to be in development as POCTs.

In terms of the original research question, to identify genomics POCTs with the potential to reach market in five years or under, a focus was put on technologies determined as being at TRL 5-9 due to these likely being the most developed of those identified. There were 24 such technologies: one at TRL 5, four at TRL 6, and 19 at TRL 9. TRL 5 and TRL 6 corresponded to technologies at the clinical trial stage, whereas TRL 9 corresponded to technologies already on the market.

Thirteen specific conditions were represented in the technologies classed as TRL 5 and higher; the majority of these were cancers (n=17, 70.8%). The majority were for diagnosis (n=11, 45.8%), and most required a specialist operator as well as being classed as near-patient (n=11, 45.8%). Of the technologies at clinical trial stage (TRL 5 and TRL 6), two (GMEX System at TRL 6 and PoCOsteo at TRL 5) were classed as being POC and operated by a non-specialist.

While a focus was put on technologies at TRL 5 and above, the search for this horizon scan was wider to understand the longer-term pipeline, as well as to capture technologies which may not be in clinical trials. Looking at the group of technologies identified as a whole, the characteristics were similar to those for the TRL 5-9 technologies specifically. Cancer again accounted for the majority of the conditions (n=241, 68.9%), with this consisting of 17 different types of cancers. Again, the majority also were for diagnosis (n=266, 76.0%), required a specialist operator (n=306, 87.4%), and were classed as near-patient (n=307, 87.7%).

Overall, cancer seems to be a major area in terms of future development of POCTs. For the technologies at TRL 5 or above, bladder cancer was the most prevalent type of cancer targeted; when looking at all identified technologies it was lung cancer, though several other types of cancers were represented in both lower and higher TRL groups. Diagnosis was also identified as a main area for future POCT development, as this was the purpose for most of the identified technologies, including for technologies at TRL 5 and above specifically.

A main challenge of estimating market readiness of the technologies was the limited clinical trial records identified. The TRL guidance relies on clinical trial data to monitor the development of a technology. Clinical validation reported in studies was used as a primary source of information to determine TRLs where clinical trials were not available, however this limited the TRL of a technology to TRL 3 or TRL 4. In terms of the number of products by TRL, there were 105 at TRL 3, 161 at TRL 4, one at TRL 5, four at TRL 6, 19 at TRL 9, and 60 for which a TRL was not applicable due to a lack of information.

We also used a tier system to group the technologies in order to differentiate based on principles such as CE mark, clinical trials, high automation, use of complex samples, and clinical

validation. Nineteen technologies were identified as having CE marks, a further four were identified as being in clinical trials, and 30 as being highly automated, using complex samples and having clinical validation (as reported in the studies).

Overall, a wide range of genomic POCTs in development were identified, though relatively few were at later stages of development and/or not yet to be classed as being 'true POCTs'.

Appendix

Appendix A. Searches

Searches were conducted 19th December 2024

Embase <1974 to 2024 December 18>

1	("point of care testing"/ or rapid on-site evaluation/) and ("point of care" or POC or POCT or "on-site" or onsite or laboratory independent or bedside or "near patient" or (rapid not (rapid adj2 review)) or fast or ultrafast or ultrarapid or quick* or simpler or simplif* or easier or easy or user-friendly or convenien* or cost effective* or full* automat* or (automat* adj4 (extract* or prepar*))).ti,ab,kf.	19106
2	((laboratory independent or bedside or "near patient") adj5 (test* or analy* or sequenc* or genotyp* or pharmacogenom* or genomic* or diagn* or detect* or sensor? or biosensor? or assay* or instrument? or device?)).ti,kf,ab.	9592
3	((onsite or "on-site" or "point of care" or portable) adj5 (test* or analy* or sequenc* or genotyp* or pharmacogenom* or genomic* or diagn* or detect* or sensor? or biosensor? or assay* or instrument? or device?)).ti,kf.	18676
4	((onsite or "on-site" or "point of care" or portable) adj5 (test* or analy* or sequenc* or genotyp* or pharmacogenom* or genomic* or diagn* or detect* or sensor? or biosensor? or assay* or instrument? or device?)).ab. and ((rapid not (rapid adj2 review)) or fast or ultrafast or ultrarapid or quick* or simpler or simplif* or easier or easy or user-friendly or convenien* or cost effective* or full* automat* or (automat* adj4 (extract* or prepar*))).ti,ab,kf.	21629
5	("point of care" adj3 (poc or poct)).ab.	9206
6	sample-to-answer*.mp.	823
7	pharmacogenetic testing/ or genetic screening/ or genetic service/ or diagnosis/ or early diagnosis/ or diagnostic test/ or cancer rapid test/ or rapid test/ or tumor marker rapid test/ or exp nucleic acid/an or testing.ti. or screening.ti. or sequencing.ti. or detection.ti. or diagnos*.ti. or assay*.ti. or test*.ab. /freq=3 or screen*.ab. /freq=3 or detect*.ab. /freq=3 or diagnos*.ab. /freq=3	4938981
8	((point-of-care or poct).ab. and ((rapid not (rapid adj2 review)) or fast or ultrafast or ultrarapid or quick* or simpler or simplif* or easier or easy or user-friendly or convenien* or cost effective* or full* automat* or (automat* adj4 (extract* or prepar*))).ti,ab,kf.) or (point-of-care or poct).ti. or "point of care system"/ or "point of care".ab. /freq=2	34635
9	7 and 8	21914
10	7 and (automat* and ((rapid not (rapid adj2 review)) or fast or ultrafast or ultrarapid or quick* or simpler or simplif* or easier or easy or user-friendly or convenien* or cost effective*)).mp. and ((full* automat* or (automat* adj4 (extract* or prepar*))).mp. or ((rapid not (rapid adj2 review)) or fast or ultrafast or ultrarapid or quick* or simpler or simplif* or easier or easy or user-friendly or convenien* or cost effective*).ti.)	6965
11	7 and ((rapid not (rapid adj2 review)) or fast or ultrafast or ultrarapid or quick*).ti,ab,kf. and (simpler or simplif* or easier or easy or user-friendly or convenien*).ti,ab,kf. and ((rapid not (rapid adj2 review)) or fast or ultrafast or ultrarapid or quick* or (simpler or simplif* or easier or easy or user-friendly or convenien*)).ti.	6844
12	7 and (direct* detect* or detect* direct* or direct* identif* or identify* direct* or non-specialist? or non-expert? or ((without or avoid*) adj4 (expert* or pretreatment or pre-treatment or amplif* or extract*))).ti,ab. and ((rapid not (rapid adj2 review)) or fast	5454

	or ultrafast or ultrarapid or quick* or simpler or simplif* or easier or easy or user-friendly or convenien* or cost effective* or full* automat* or (automat* adj4 (extract* or prepar*))).ti,ab,kf.	
13	7 and ((direct* detect* or detect* direct* or direct* identif* or identify* direct*) and (non-specialist? or non-expert? or ((without or avoid*) adj4 (expert* or pretreatment or pre-treatment or amplif* or extract*))))).ti,ab.	245
14	7 and ((integrated adj3 microfluid*) or fully integrated).mp.	1885
15	or/1-6,9-14	75533
16	pharmacogenetic testing/ or genetic screening/ or genetic service/	134848
17	genomics/ or comparative genomics/ or functional genomics/ or imaging genomics/ or immunogenomics/ or methylomics/ or neurogenomics/ or oncogenomics/ or exp pharmacogenomics/ or physiological genomics/ or population genomics/ or proteogenomics/ or radiogenomics/ or structural genomics/ or transcriptomics/	202361
18	exp pharmacogenetics/	37964
19	exp high throughput sequencing/	162645
20	genetic analyzer/	61791
21	exp gene expression profiling/	237550
22	exp medical genetics/	15084
23	genetic association/	268553
24	genetic predisposition/ or genetic resistance/ or genetic susceptibility/	216962
25	human genome/	26307
26	epigenetics/	118010
27	genetic risk/	77722
28	genetic risk score/	12826
29	genetic analysis/ or genome analysis/	245714
30	allele/ or allotype/	230486
31	exp genotype/ or genotyping/	574055
32	genomic DNA/	115642
33	wild type/	207331
34	genetic polymorphism/ or single nucleotide polymorphism/	387653
35	allele specific polymerase chain reaction/ or allele specific real time polymerase chain reaction/	1841
36	cystic fibrosis test kit/ or genetic disease test kit/ or cystic fibrosis elisa kit/	73
37	exp tumor-related gene/	270062
38	exp genetic marker/	133126
39	DNA methylation age/ or methylation/ or DNA methylation/ or DNA methylation assay/	175799
40	RNA methylation/	5350
41	exp isothermal amplification/	9373
42	microRNA/	218458
43	exp cytochrome/	249676
44	microchip electrophoresis/	1379
45	real time polymerase chain reaction/ or allele specific real time polymerase chain reaction/ or fluorescence quantitative polymerase chain reaction/ or multiplex real	507413

	time polymerase chain reaction/ or quantitative methylation specific polymerase chain reaction/ or real time reverse transcription polymerase chain reaction/	
46	exp gene/	1316962
47	exp crispr cas system/	51224
48	exp nucleic acid/	2542862
49	exp genetic procedures/	3883473
50	next generation sequenc*.mp.	117353
51	(optical adj5 mapping).mp.	4464
52	long-read.mp.	7710
53	(isothermal adj5 amplification).mp.	9978
54	oncogene*.mp.	247595
55	crispr*.mp.	85557
56	single nucleotide polymorphism?.mp.	278654
57	(cyp2* or MT-RNR1*).mp.	47417
58	(precision medicine or personali* medicine).mp.	114822
59	personalized medicine/ or personalized cancer therapy/	91232
60	microfluidic device/ or lab on a chip/	12249
61	((lab adj3 chip) or (microfluidic adj3 (digital or device or assay*))).mp.	21927
62	cancer diagnosis/ or cancer screening/ or cancer test/ or early cancer diagnosis/ or cancer classification/ or tumor classification/ or disease classification/ or exp *neoplasm/di	1149013
63	(or/58-62) and (pharmacogen* or gene or genetic* or genomic* or genotyp* or allele* or sequencing or polymorphism* or methylat* or transcriptom* or epigenetic* or mRNA or miRNA or microRNA or DNA or RNA or mutation?).ti,ab,kf.	206726
64	(pharmacogen* or gene or genetic* or genomic* or genotyp* or allele* or sequencing or polymorphism* or methylat* or transcriptom* or epigenetic* or mRNA or miRNA or microRNA or ((DNA or RNA) adj6 (sequenc* or test* or analy* or assay* or detect* or biosensor* or sensor* or liquid biops*)) or exosome* or mutation?).ti.	1927136
65	15 and (or/16-57,63-64)	19622
66	genedrive*.af. and (MT-RNR1* or CYP2*).mp.	5
67	Genomadix*.af. and (cube* or CYP2*).mp.	6
68	(GMEX or GMEXtm or GMEXr or (GMEX* and CYP2*)).mp.	3
69	(spartan* rx* or spartan* frx* or (spartan* and CYP2*)).mp.	56
70	((xpert* or genexpert*) adj4 (BCR* or bladder or NPM1 or FII or FV)).mp.	166
71	or/66-70	232
72	(65 or 71) not ((nonhuman/ or exp animal/ or exp animal disease/ or exp plant/ or exp plant disease/) not ((human or humans).hw,ti. or (animal experiment/ and model*.mp.) or animal model/))	15959
73	limit 72 to yr="2019 -Current"	8568
74	73 not ((exp *virus examination/ or exp *virus/ or exp *virus infection/ or exp *bacterium examination/ or exp *bacterium/ or exp *parasitosis/ or exp *bacterial infection/ or exp *mycosis/ or exp *fungus/ or exp *sepsis/ or exp *microorganism detection/ or exp *chromalveolata/ or exp *excavata/ or *virus dna/ or *bacterial dna/ or *fungal dna/ or *environmental dna/ or *coronavirus disease 2019/ or *food safety/ or exp *environmental monitoring/ or exp *environmental chemical/ or exp	4023

	*toxic substance/ or exp environmental chemical/an or exp toxic substance/an or exp *plant/ or exp *plant disease/ or exp *animal disease/ or virus*.ti. or viral*.ti. or bacteri*.ti. or microbi*.ti. or infect*.ti. or pathogen?.ti. or covid.ti. or covid19.ti. or sars-cov*.ti. or coronavirus*.ti. or (food not "food and drug").ti. or agricultur*.ti. or (agric* or plant or food or veterin*).jw.) not (16 or 18 or 23 or 24 or 25 or (62 not hpv.mp.) or human gen*.mp. or human dna.mp. or long-covid*.mp. or post-covid*.mp.))	

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Ovid MEDLINE(R) ALL <1946 to December 18, 2024>

1	(point-of-care testing/ or rapid diagnostic tests/ or rapid on-site evaluation/) and ("point of care" or POC or POCT or "on-site" or onsite or laboratory independent or bedside or "near patient" or (rapid not (rapid adj2 review)) or fast or ultrafast or ultrarapid or quick* or simpler or simplif* or easier or easy or user-friendly or convenien* or cost effective* or full* automat* or (automat* adj4 (extract* or prepar*))).ti,ab,kf.	4510
2	((laboratory independent or bedside or "near patient") adj5 (test* or analy* or sequenc* or genotyp* or pharmacogenom* or genomic* or diagn* or detect* or sensor? or biosensor? or assay* or instrument? or device?)).ti,kf,ab.	6344
3	((onsite or "on-site" or "point of care" or portable) adj5 (test* or analy* or sequenc* or genotyp* or pharmacogenom* or genomic* or diagn* or detect* or sensor? or biosensor? or assay* or instrument? or device?)).ti,kf.	15302
4	((onsite or "on-site" or "point of care" or portable) adj5 (test* or analy* or sequenc* or genotyp* or pharmacogenom* or genomic* or diagn* or detect* or sensor? or biosensor? or assay* or instrument? or device?)).ab. and ((rapid not (rapid adj2 review)) or fast or ultrafast or ultrarapid or quick* or simpler or simplif* or easier or easy or user-friendly or convenien* or cost effective* or full* automat* or (automat* adj4 (extract* or prepar*))).ti,ab,kf.	18526
5	("point of care" adj3 (poc or poct)).ab.	6890
6	sample-to-answer*.mp.	646
7	genetic services/ or genetic carrier screening/ or genetic testing/ or pharmacogenomic testing/ or diagnosis/ or early diagnosis/ or "diagnostic techniques and procedures"/ or clinical laboratory techniques/ or exp Nucleic Acids/an or testing.ti. or screening.ti. or sequencing.ti. or detection.ti. or diagnos*.ti. or assay*.ti. or test*.ab. /freq=3 or screen*.ab. /freq=3 or detect*.ab. /freq=3 or diagnos*.ab. /freq=3	2949959
8	((point-of-care or poct).ab. and ((rapid not (rapid adj2 review)) or fast or ultrafast or ultrarapid or quick* or simpler or simplif* or easier or easy or user-friendly or convenien* or cost effective* or full* automat* or (automat* adj4 (extract* or prepar*))).ti,ab,kf.) or (point-of-care or poct).ti. or point-of-care systems/ or "point of care".ab. /freq=2	32989
9	7 and 8	18683
10	7 and (automat* and ((rapid not (rapid adj2 review)) or fast or ultrafast or ultrarapid or quick* or simpler or simplif* or easier or easy or user-friendly or convenien* or cost effective*)).mp. and ((full* automat* or (automat* adj4 (extract* or prepar*))).mp. or ((rapid not (rapid adj2 review)) or fast or ultrafast or ultrarapid or quick* or simpler or simplif* or easier or easy or user-friendly or convenien* or cost effective*).ti.)	4573

11	7 and ((rapid not (rapid adj2 review)) or fast or ultrafast or ultrarapid or quick*).ti,ab,kf. and (simpler or simplif* or easier or easy or user-friendly or convenien*).ti,ab,kf. and ((rapid not (rapid adj2 review)) or fast or ultrafast or ultrarapid or quick* or (simpler or simplif* or easier or easy or user-friendly or convenien*)).ti.	5663
12	7 and (direct* detect* or detect* direct* or direct* identif* or identify* direct* or non-specialist? or non-expert? or ((without or avoid*) adj4 (expert* or pretreatment or pre-treatment or amplif* or extract*))).ti,ab. and ((rapid not (rapid adj2 review)) or fast or ultrafast or ultrarapid or quick* or simpler or simplif* or easier or easy or user-friendly or convenien* or cost effective* or full* automat* or (automat* adj4 (extract* or prepar*))).ti,ab,kf.	4697
13	7 and ((direct* detect* or detect* direct* or direct* identif* or identify* direct*) and (non-specialist? or non-expert? or ((without or avoid*) adj4 (expert* or pretreatment or pre-treatment or amplif* or extract*))))).ti,ab.	254
14	7 and ((integrated adj3 microfluid*) or fully integrated).mp.	1603
15	or/1-6,9-14	56398
16	genetic services/ or genetic carrier screening/ or genetic testing/ or pharmacogenomic testing/	56513
17	genomics/ or epigenomics/ or human genome project/ or proteogenomics/ or imaging genomics/ or radiation genomics/	85048
18	Pharmacogenetics/	14009
19	exp High-Throughput Nucleotide Sequencing/	57576
20	exp Gene Expression Profiling/	171758
21	Genetics, Medical/	14165
22	genetic predisposition to disease/ or anticipation, genetic/ or genetic risk score/	164527
23	Genome, Human/	32005
24	Alleles/	122754
25	Genotyping Techniques/	8423
26	exp Genotype/	475256
27	exp Polymorphism, Genetic/	314702
28	Genetic Markers/	59138
29	methylation/ or dna methylation/ or rna methylation/	100732
30	micrornas/ or circulating microrna/	138544
31	exp Cytochromes/	132750
32	exp Polymerase Chain Reaction/	470216
33	exp genes/	841101
34	CRISPR-Cas Systems/	21113
35	exp Nucleic Acids/	1681592
36	exp Genetic Techniques/	2014145
37	next generation sequenc*.mp.	67576
38	(optical adj5 mapping).mp.	2892
39	long-read.mp.	6138
40	(isothermal adj5 amplification).mp.	7629
41	oncogene*.mp.	344503

42	crispr*.mp.	51972
43	single nucleotide polymorphism?.mp.	102766
44	(cyp2* or MT-RNR1*).mp.	37472
45	Precision Medicine/	30000
46	(precision medicine or personali* medicine).mp.	65422
47	lab-on-a-chip devices/ or microphysiological systems/	8550
48	((lab adj3 chip) or (microfluidic adj3 (digital or device or assay*))).mp.	19060
49	"Early Detection of Cancer"/ or exp *Neoplasms/di	333483
50	(or/45-49) and (pharmacogen* or gene or genetic* or genomic* or genotyp* or allele* or sequencing or polymorphism* or methylat* or transcriptom* or epigenetic* or mRNA or miRNA or microRNA or DNA or RNA or mutation?).ti,ab,kf.	66523
51	(pharmacogen* or gene or genetic* or genomic* or genotyp* or allele* or sequencing or polymorphism* or methylat* or transcriptom* or epigenetic* or mRNA or miRNA or microRNA or ((DNA or RNA) adj6 (sequenc* or test* or analy* or assay* or detect* or biosensor* or sensor* or liquid biops*)) or exosome* or mutation?).ti.	1582786
52	15 and (or/16-44,50-51)	11579
53	genedrive*.af. and (MT-RNR1* or CYP2*).mp.	5
54	Genomadix*.af. and (cube* or CYP2*).mp.	4
55	(GMEX or GMEXtm or GMEXr or (GMEX* and CYP2*)).mp.	3
56	(spartan* rx* or spartan* frx* or (spartan* and CYP2*)).mp.	22
57	((xpert* or genexpert*) adj4 (BCR* or bladder or NPM1 or FII or FV)).mp.	46
58	or/53-57	76
59	(52 or 58) not ((exp Animals/ or exp Animal Diseases/ or exp Plants/ or exp Plant Diseases/) not ((human or humans).hw,ti. or (Animal Experimentation/ and model*.mp.) or models, animal/))	10286
60	limit 59 to yr="2019 -Current"	5281
61	60 not ((exp *Viruses/ or exp *Virus Diseases/ or exp *Bacteria/ or exp *Parasitic Diseases/ or exp *Bacterial Infections/ or exp *Mycoses/ or exp *Fungi/ or exp *Sepsis/ or exp *Viridiplantae/ or exp *alveolata/ or exp *Euglenozoa/ or exp *stramenopiles/ or exp *Viridiplantae/ or exp *alveolata/ or exp *Euglenozoa/ or exp *stramenopiles/ or *dna, bacterial/ or *dna, fungal/ or *dna, viral/ or *dna, environmental/ or *Covid-19/ or exp *Food Safety/ or exp *Environmental Monitoring/ or exp *Environmental Pollutants/ or exp Environmental Pollutants/an or exp *Animal Diseases/ or exp *Plants/ or exp *Plant Diseases/ or virus*.ti. or viral*.ti. or bacteri*.ti. or microbi*.ti. or infect*.ti. or pathogen?.ti. or covid.ti. or covid19.ti. or sars-cov*.ti. or coronavirus*.ti. or (food not "food and drug").ti. or agricultur*.ti. or (agric* or plant or food or veterin*).jw.) not (16 or 18 or 22 or 23 or (49 not hpv.mp.) or human gen*.mp. or human dna.mp. or long-covid*.mp. or post-covid*.mp. or Post-Acute COVID-19 Syndrome/))	2101

https://ovidsp.ovid.com/ovidweb.cgi?T=JS&NEWS=N&PAGE=main&SHAREDSEARCHID=4dXtEbxNkSUNYvj7CeJ0yNRNoZlnHeIcVZV4ypnSf13spBJkeqH4U2SLZmGsfopfI

Appendix B. Screening Guide

Quick process:

1. Ensure the **target** being analysed **is genetic** material
2. Ensure the genetic **target is** a **human** genetic target
3. Ensure **it is a device** that is designed to be POCT
4. Ensure the starting sample is **not FFPE**
5. If sample-to-answer is a matter of **hours** --> **INCLUDE**

EXCLUDES

- 'FFPE'
- 'Tissue sample'
- Almost anything haemoglobin related (sickle cell disease, HemoTypeSC etc. **unless** it refers specifically to mutation analysis of the *HBB* gene, the genotype, **not** the phenotype)
- 'Idylla' (diagnostic platform for FFPE tissues, **note:** some use plasma samples, although these are rare, make sure you differentiate)
- Anything pathogen related (bacteria, fungi, viruses – common ones are tuberculosis, HIV, monkeypox, strep A-C)
- Anything POCT that is analysing a phenotype i.e., protein levels, glucose levels, blood pressure etc.

INCLUDES

We want a POCT genomic device, however a lot of articles will include POCT to get their citation number up, saying 'this has the potential to be POCT down the line'.

Lots will also refer to genomic testing at the point-of-care, this can mean genetic counselling, getting whole genome sequencing done, family history questionnaires, or other processes that aren't **actually** POCT genomic testing, but because they start the process early, they call it point-of-care.

For what we want, we need:

- A genomic target (or at least a proof-of-concept genomic target), this can be DNA, RNA, mRNA, microRNA, extracellular vesicles (as long as the nucleic acid cargo is the target), and epigenetics (methylation, acetylation)
- An indication
- A sample type (complex biological matrices such as saliva, whole blood etc. are the best – but plasma, purified DNA/RNA are also acceptable)
- A time frame for completion

We'd like:

- A profile for the person conducting the test (as unskilled as we can get)
- Clinical relevance (tested on 68 patient samples etc.)

Appendix C. Data Extraction Guide

Data Column	Description
Type	Almost always 'Article'
Link	DOI
Developer	Available in the Funding, Acknowledgements, or other similar section
Test Description	Sum up the test, use the sentence in the abstract/intro where it sums up the article, i.e. 'In this work we introduce a XYZ'; use the XYZ as the test description. If it has a name, include here.
Cost	Usually 'N/A' but include if it's available
Genomic Target	Include the specific mutation if possible. If the target is proof-of-concept, put that in brackets next to the target For one target, e.g.: • miRNA-21 (proof-of-concept) • *KRAS* G12D ctDNA For multiple targets, e.g.: • *EGFR*; *SMN1* exon 7 deletion; *BRAF*
Condition/Indication	Be as specific as possible, sometimes, specifically with miRNAs, the indication is vague, e.g.: • Spinal muscular atrophy • Cardiovascular disease • Cancer • Dependent upon final product (this is only used for proof-of-concept POCTs)
Time to results	Be as accurate as you can, if it's not explicitly stated, add up from 'Methods' section, e.g.: • >3 hours
Setting	This is our differentiation between true POCT and near-POCT. If it can be used with a complex biological sample (e.g. blood, saliva), does not require specialised steps, and can be done next to the patient: • POC If it requires DNA/RNA extraction, any specialised steps i.e., PCR, centrifugation etc. but can still be done in a hospital setting: • Near-patient If it's neither of these, then flag it as it shouldn't be included.
Operator	Similar to the above, if a nurse could carry out the test at the POC: • Non-specialist If it requires fancy steps or expert analysis:

	• Specialist
Purpose	Can be a combination of these or just a single one: • Diagnosis • Prognosis • Treatment decisions • Intra-operative decision making
Sample Type	Include volume (e.g. '50uL') if available: Complex: • Buccal swab • Saliva • Whole blood • Plasma • Serum • Urine Non-complex: • DNA • RNA
Preparation	Keep it simple. Include time-frames if possible. If it's a sample-in-answer-out, fully automated device: • N/A For more complex tests, e.g.: • DNA/RNA purification • Mixing of reagents • PCR • Sample dilution • Assay washing (30 mins) • Multiple specialised steps
Clinical Validation	How has the device been tested, and on how many people? In the face of no human samples, please include cell lines, e.g.: • Clinical samples (N=) • Human cell lines (22Rv1, HEK293T) • N/R
Amplification Type	This is likely to be either PCR or isothermal amplification, which has various subtypes, only include the acronym in the table though, e.g.: • HCR (hybridisation chain reaction) • RPA (recombinase polymerase amplification) • LAMP (loop-mediated isothermal amplification) • PCR (polymerase chain reaction) • qPCR • RT-PCR

	If there's no amplification e.g. it's a lateral flow assay: • N/A
Analysis Type	• Electrochemical • Fluorescence • Colorimetric • SERS
Sensitivity	Colour coded due to variance of reporting techniques: • Green = Tested and it's good • Amber = Tested and it's okay • Red = Tested and it's poor **or** not mentioned/tested
Specificity	Colour coded due to variance of reporting techniques: • Green = Tested and it's good • Amber = Tested and it's okay • Red = Tested and it's poor **or** not mentioned/tested
Technology Readiness Level (TLR)	To be done after data extraction.

Appendix D. Project Protocol

Below is the content of the protocol (project plan) v11, dated 12/12/2024.

Background

Point-of-Care Testing (PoCT) in genomics allows clinicians to rapidly detect genetic alterations and mutations, as well as analyse the results of those tests. Genetic testing is essential for diagnosing and treating genetic diseases, as well as predicting a patient's risk for developing certain diseases. PoCT in genomics may also enable clinicians to identify potential targets for therapeutic drugs. Additionally, PoCT in genomics can help clinicians identify gene variants that may be associated with how the drug is metabolised or risk of adverse events, allowing them to determine the most appropriate course of treatment for their patients. Recent technological advancements such as high-throughput sequencing and microarray technologies have significantly enhanced capabilities in genomics, and alongside this PoCT has advanced. These innovations allow for rapid and accurate analysis of large genetic datasets, making it feasible to perform complex genomic analyses in a point-of-care setting. However, NHSE currently have no horizon scanning mechanism in place to assess the current and future landscape of PoCT in genomic medicine and therefore no means by which NHS commissioners can plan for developments in PoCT technology in this area. Thus, this is what this project aims to accomplish, by providing a one-off horizon scan on PoCT that could inform genomic strategy, planning and commissioning both current and in the near future.

Aims and Objectives

Aim: To give an overview of the pipeline of PoCT available in the next 5 years, to identify variation(s) in the genome that enable the use of genetic information to inform diagnosis, prognosis, and treatment decisions in any clinical area.

Objectives:

1. Identify PoCT technology (standalone tests) in genomic medicine with the potential to reach market in 5 years or under
2. Describe PoCT technology (standalone tests) in genomic medicine with the potential to reach market in 5 years or under

Definitions

A PoCT is defined by Public Health England as a “medical diagnostic test, performed at or near the site of patient care, undertaken by healthcare professionals who may not be trained laboratory staff. It is a test to support clinical decision making, and for which the results can be available in real time”.[2]

PoCT in the context of genomic medicine can be described using PoCT technology to analyse genetic polymorphisms that influence disease and/or the effectiveness and safety of medicines.[22]

Diagnosis is the process of identifying a disease, condition or injury from its signs and symptoms.[3]

Prognosis is the likely outcome or course of a disease; the chance of recovery of recurrence.[4]

Treatment decisions involve deciding what treatment a patient will receive. These decisions may be based on the degree of certainty of a diagnosis, the harms and benefits of a treatment, and the harms and benefits of further information-gathering activities, including the impact of delaying treatment.[5]

Project Management

Roles and Responsibilities

Role	Responsibility	Named individual (per project)
Project Lead (NHSE - policy programme lead)	Provides the vision and requirements for the project. Attends project meetings to steer, and corresponds directly with IO relationship manager.	Vicky Chaplin
Project Sponsor (NHSE Horizon Scanning team)	Supports to development of project vision and ensure the project is delivered to meet NHSE needs/requirements. Attends project meetings and is included in correspondence between the project lead and manager/relationship manager.	Claire Ball (CSU) (acting under guidance from the NHSE SRO)
Project Lead (IO)	Coordinates the delivery of the project, works with the IO SRM to determine the delivery schedule/timescales, driving project delivery through managing actions and meetings.	Shona Haston
Stakeholder Relationship Manager (IO)	Main point of communication between IO and NHSE. With the IO project lead determine the delivery schedule/timescales, driving project delivery through managing actions and meetings. Arranges and runs all meetings.	Ruth Wilson
Horizon Scanning project lead (IO Senior Research Associate)	Supports the development of the horizon scanning technical component of the project (horizon scan search/analysis).	Katie Thomson, Andrew Mkwashi

	Supports development of all IO project documentation (protocol, interim, final reports, dataset). Attends project meetings to support the scoping, presenting, and sharing horizon scan findings.	
Senior Responsible Owner - NHSE	Accountable for the project meeting the needs of the commissioning organisation.	Dr. Ilaria Mirabile
Senior Responsible Owner – NIHR IO	Accountable for the project meeting the needs of the commissioning organisation.	Professor Dawn Craig

The duration of the project will be approximately 6 months.

There will be 'touch points' where the IO and NHSE project teams will meet to review progress. These will be at key points in the project timeline following completion of the following stages: pilot, screening, data extraction, data analysis, and final report. In the event that the scientific clinicians need to be involved in this process, they will be consulted, and sufficient time will be allocated to gain their input.

| | | |

Piloting	Project update following completion of pilot Accompanied by: • Pilot dataset (excel doc) • Brief written summary of issues/challenges e.g. data fields which are proving tricky, classification or categorisations which need further refinement) and recommendations on next steps/changes needed from pilot.	w/c 2nd Dec 2024	w/c 9th Dec 2024	Agree any refinements or edits to the scope based on the pilot dataset/information.
Screening	Project update following completion of searches and progress of screening Accompanied by: • Dataset (excel doc) of examples from screening • Brief written summary of the size and scale of the data found	w/c 13th Jan 2025	w/c 13th Jan 2025	Understand the size and scale of the dataset/project and discuss if analysis plans (in protocol) are sufficient or if further work (e.g. shortlisting) is needed.
Screening	Project update following completion of screening: Accompanied by: • Dataset (excel doc) of examples from screening • Brief written summary of the size and scale of the data found	w/c 24th Jan	w/c 27th Jan	Discussion of the data set and answering any queries about the next steps.

Data extraction (interim report)	Project update following completion of data extraction Accompanied by: • Written narrative interim report (if relevant at this stage) on methods, data collection and data alignment. • Dataset (excel doc)	w/c 10th Feb 2025	w/c 17th Feb 2025	Understand the longlist of products which answer the policy question and agree the analysis (i.e. support identification of themes).
Data analysis	Project update following completion of data analysis Accompanied by: Written narrative final report (early draft) on project (methods, data collection, data alignment, data analysis). • Dataset (excel doc) with any relevant updates.	w/c 3rd Mar 2025	w/c 10th Mar 2025	Understand the final data set and themes. Project group to consider and agree implications/ take away messages from horizon scan. Feedback to be provided on the draft report by NHSE project team.
Output	Final report	w/c 7th Apr 2025	w/c 14th Apr 2025	Any further feedback on the report to be provided by NHSE project team. Agree on whether any further outputs will be done (i.e., publication).

Methods

The main question that will guide this project is: **What does the current landscape for genetic PoCT look like - what is in the pipeline, what is closer to market entry, and what key areas are being reviewed by researchers?**

An evidence synthesis approach will be used alongside the horizon scan to combine information from trial registries, news sites, and academic literature. Horizon scanning is an

umbrella term that is used to describe the systematic examination of information sources to detect early signs of important developments. By incorporating more traditional evidence synthesis approaches alongside (e.g., the searching of published literature and conference proceedings), a more complete assessment of the evidence ecosystem will be provided.

Eligibility Criteria

Inclusion criteria defined using the PICOS framework:

- **Population:** any (children and adults)
- **Intervention:** PoCT technology to identify variation(s) in gene expression (PoCT for human genome only (excluding those for viruses and bacteria), including epigenetic)
- **Comparator:** other lab testing or may be none
- **Outcomes:** overview of the technology, including diagnostic accuracy if available
- **Setting:** clinical setting outside of a specialist genomics lab (e.g., clinic, bedside OR at an on-site (non-genomics) lab), able to be caried out by a non-scientist operator (e.g., clinician) for results in real-time

PoCT included will be able to be performed at or near to the side of patient care. The main list of identified technologies will include only tests that require no sample manipulation or additional interpretation. However, additional technologies will be identified which do require sample manipulation, and differentiated from the main list.

Initially, all clinical trial phases and statuses will be included.

Pre-clinical trials may be included, depending on the number of trials identified. In the case that a large number are found, those of phase II onwards will be prioritised.

Examples of relevant PoCT technologies currently available are as follows:

Device	Assay
GeneDrive	*GeneDrive MT-RNR1*
	GeneDrive CYP2C19
Genomadix	*Genomadix Cube CYP2C19*
GeneXpert	Xpert BCR-ABL Ultra
	Xpert BCR-ABL Ultra p190
	Xpert Bladder Cancer Detection
	Xpert Bladder Cancer Monitor
	Xpert NPM1 Mutation
	Xpert FII & FV

Limits

Trial registries will be searched back 5 years.

Search Strategy

The provisional search strategy for Ovid EMBASE is as follows:

Embase <1974 to 2024 December 03>

1	("point of care testing"/ or rapid on-site evaluation/) and ("point of care" or POC or POCT or "on-site" or onsite or laboratory independent or bedside or "near patient" or (rapid not (rapid adj2 review)) or fast or ultrafast or ultrarapid or quick* or simpler or simplif* or easier or easy or user-friendly or convenien* or cost effective* or full* automat* or (automat* adj4 (extract* or prepar*))).ti,ab,kf.	19007
2	((laboratory independent or bedside or "near patient") adj5 (test* or analy* or sequenc* or genotyp* or pharmacogenom* or genomic* or diagn* or detect* or sensor? or biosensor? or assay* or instrument? or device?)).ti,kf,ab.	9569
3	((onsite or "on-site" or "point of care" or portable) adj5 (test* or analy* or sequenc* or genotyp* or pharmacogenom* or genomic* or diagn* or detect* or sensor? or biosensor? or assay* or instrument? or device?)).ti,kf.	18580
4	((onsite or "on-site" or "point of care" or portable) adj5 (test* or analy* or sequenc* or genotyp* or pharmacogenom* or genomic* or diagn* or detect* or sensor? or biosensor? or assay* or instrument? or device?)).ab. and ((rapid not (rapid adj2 review)) or fast or ultrafast or ultrarapid or quick* or simpler or simplif* or easier or easy or user-friendly or convenien* or cost effective* or full* automat* or (automat* adj4 (extract* or prepar*))).ti,ab,kf.	21493
5	("point of care" adj3 (poc or poct)).ab.	9141
6	sample-to-answer*.mp.	820
7	pharmacogenetic testing/ or genetic screening/ or genetic service/ or diagnosis/ or early diagnosis/ or exp nucleic acid/an or testing.ti. or screening.ti. or sequencing.ti. or detection.ti. or diagnos*.ti. or assay*.ti. or test*.ab. /freq=3 or screen*.ab. /freq=3 or detect*.ab. /freq=3 or diagnos*.ab. /freq=3	4872993
8	((point-of-care or poct).ab. and ((rapid not (rapid adj2 review)) or fast or ultrafast or ultrarapid or quick* or simpler or simplif* or easier or easy or user-friendly or convenien* or cost effective* or full* automat* or (automat* adj4 (extract* or prepar*))).ti,ab,kf.) or (point-of-care or poct).ti. or "point of care system"/ or "point of care".ab. /freq=2	34444
9	7 and 8	21636
10	7 and (automat* and ((rapid not (rapid adj2 review)) or fast or ultrafast or ultrarapid or quick* or simpler or simplif* or easier or easy or user-friendly or convenien* or cost effective*)).mp. and ((full* automat* or (automat* adj4 (extract* or prepar*))).mp. or ((rapid not (rapid adj2 review)) or fast or ultrafast or ultrarapid or quick* or simpler or simplif* or easier or easy or user-friendly or convenien* or cost effective*).ti.)	6896

11	7 and ((rapid not (rapid adj2 review)) or fast or ultrafast or ultrarapid or quick*).ti,ab,kf. and (simpler or simplif* or easier or easy or user-friendly or convenien*).ti,ab,kf. and ((rapid not (rapid adj2 review)) or fast or ultrafast or ultrarapid or quick* or (simpler or simplif* or easier or easy or user-friendly or convenien*)).ti.	6767
12	7 and (direct* detect* or detect* direct* or direct* identif* or identify* direct* or non-specialist? or non-expert? or ((without or avoid*) adj4 (expert* or pretreatment or pre-treatment or amplif* or extract*))).ti,ab. and ((rapid not (rapid adj2 review)) or fast or ultrafast or ultrarapid or quick* or simpler or simplif* or easier or easy or user-friendly or convenien* or cost effective* or full* automat* or (automat* adj4 (extract* or prepar*))).ti,ab,kf.	5429
13	7 and ((direct* detect* or detect* direct* or direct* identif* or identify* direct*) and ((non-specialist? or non-expert? or (without or avoid*)) adj4 (expert* or pretreatment or pre-treatment or amplif* or extract*))).ti,ab.	242
14	7 and integrated microfluid*.mp.	456
15	or/1-6,9-14	73990
16	pharmacogenetic testing/ or genetic screening/ or genetic service/	134170
17	genomics/ or comparative genomics/ or functional genomics/ or imaging genomics/ or immunogenomics/ or methylomics/ or neurogenomics/ or oncogenomics/ or exp pharmacogenomics/ or physiological genomics/ or population genomics/ or proteogenomics/ or radiogenomics/ or structural genomics/ or transcriptomics/	201177
18	exp pharmacogenetics/	37887
19	exp high throughput sequencing/	160637
20	genetic analyzer/	60958
21	exp gene expression profiling/	234991
22	exp medical genetics/	15053
23	genetic association/	268160
24	genetic predisposition/ or genetic resistance/ or genetic susceptibility/	216672
25	human genome/	26221
26	epigenetics/	117767
27	genetic risk/	77493
28	genetic risk score/	12669
29	genetic analysis/ or genome analysis/	244966
30	allele/ or allotype/	229557
31	genotype/ or genotyping/	497436
32	genomic DNA/	114997
33	wild type/	206933
34	allele specific polymerase chain reaction/ or allele specific real time polymerase chain reaction/	1805

35	cystic fibrosis test kit/ or genetic disease test kit/ or cystic fibrosis elisa kit/	72
36	exp tumor-related gene/	269240
37	transcriptomics/	80143
38	DNA methylation age/ or methylation/ or DNA methylation/ or DNA methylation assay/	175016
39	exp isothermal amplification/	9344
40	microRNA/	217513
41	microchip electrophoresis/	1379
42	real time polymerase chain reaction/ or allele specific real time polymerase chain reaction/ or fluorescence quantitative polymerase chain reaction/ or multiplex real time polymerase chain reaction/ or quantitative methylation specific polymerase chain reaction/ or real time reverse transcription polymerase chain reaction/	503756
43	exp nucleic acid/	2534722
44	exp genetic procedures/	3866911
45	next generation sequenc*.mp.	116530
46	(optical adj5 mapping).mp.	4447
47	long-read.mp.	7634
48	(precision medicine or personali* medicine).mp.	113945
49	personalized medicine/ or personalized cancer therapy/	90461
50	microfluidic device/ or lab on a chip/	12058
51	((lab adj3 chip) or (microfluidic adj3 (digital or device or assay*))).mp.	21717
52	cancer diagnosis/ or cancer screening/ or cancer test/ or early cancer diagnosis/ or cancer classification/ or tumor classification/ or disease classification/	494289
53	(or/48-52) and (pharmacogen* or gene or genetic* or genomic* or genotyp* or allele* or sequencing or polymorphism* or methylat* or transcriptom* or epigenetic* or mRNA or miRNA or microRNA or DNA or RNA or mutation? or single nucleotide polymorphism?).ti,ab,kf.	127377
54	(pharmacogen* or gene or genetic* or genomic* or genotyp* or allele* or sequencing or polymorphism* or methylat* or transcriptom* or epigenetic* or mRNA or miRNA or microRNA or ((DNA or RNA) adj6 (sequenc* or test* or analy* or assay* or detect* or biosensor* or sensor* or liquid biops*)) or exosome* or mutation? or single nucleotide polymorphism?).ti. or (cyp2* or MT-RNR1*).mp.	1951027
55	15 and (or/16-47,53-54)	18720
56	genedrive*.af. and (MT-RNR1* or CYP2*).mp.	5
57	Genomadix*.af. and (cube* or CYP2*).mp.	6
58	(GMEX or GMEXtm or GMEXr or (GMEX* and CYP2*)).mp.	3
59	(spartan* rx* or spartan* frx* or (spartan* and CYP2*)).mp.	56

60	((xpert* or genexpert*) adj4 (BCR* or bladder or NPM1 or FII or FV)).mp.	161
61	or/56-60	227
62	(55 or 61) not ((nonhuman/ or exp animal/ or exp animal disease/ or exp plant/ or exp plant disease/) not (human.hw,ti. or (animal experiment/ and model*.mp.) or animal model/))	15210
63	limit 62 to yr="2019 -Current"	8189
64	63 not ((exp *virus examination/ or exp *virus/ or exp *virus infection/ or exp *bacterium examination/ or exp *bacterium/ or exp *parasitosis/ or exp *bacterial infection/ or exp *mycosis/ or exp *fungus/ or exp *sepsis/ or exp *microorganism detection/ or exp *chromalveolata/ or exp *excavata/ or *virus dna/ or *bacterial dna/ or *fungal dna/ or *environmental dna/ or *coronavirus disease 2019/ or *food safety/ or exp *environmental monitoring/ or exp *environmental chemical/ or exp *toxic substance/ or exp environmental chemical/an or exp toxic substance/an or virus*.ti. or viral*.ti. or bacteri*.ti. or microbi*.ti. or infect*.ti. or pathogen*.ti. or covid.ti. or covid19.ti. or sars-cov*.ti.) not (16 or 18 or 23 or 24 or 25 or (52 not hpv.mp.) or human gen*.mp. or human dna.mp. or long-covid*.mp. or post-covid*.mp.))	3872
65	64 and ((clinical* or clinician* or physician* or hospital* or bedside or "near patient" or primary care or market* or commercial* or app? or smartphone?).mp. or (point of care and test*).ti. or diagnostic accuracy/ or *"point of care testing"/ or *rapid on-site evaluation/ or *"point of care system"/ or di.fs.)	2649

https://ovidsp.ovid.com/ovidweb.cgi?T=JS&NEWS=N&PAGE=main&SHAREDSEARCHID=6wuztN4tmq6032ZMaSxIeXDOUPyuJxUTZGhDNXR8E1mBmuU63cNvhhbDg8rJMTDYv

The search strategy will be translated to Medline (Ovid) to identify other academic literature. Once the search strategy is developed in Ovid, other databases will be examined to assess their relevance (e.g., Engineering databases). In addition, relevant systematic reviews and included studies will be citation chained and their contents assessed for relevance.

The ClinicalTrials.gov search will use the following terms:

Other terms: ("point of care" OR POCT OR onsite OR "on-site" OR "laboratory independent" OR bedside OR "near patient" OR (nanopore AND portable) OR (rapid AND portable))

AND Intervention/treatment: (gene OR genetic OR pharmacogenomic OR genomic OR genotyping OR genotype OR miRNA OR mRNA OR sequencing OR assay OR cyp2c19 OR "MT-RNR1") NOT (covid OR "sars-cov-2") |

AND Primary completion date: on or after 01/01/2018

See: link to clinicaltrials.gov search

Along with targeted searches for named technologies, e.g. *Other terms*: genedrive OR "gene drive" OR genomadix OR (("gene xpert" OR genexpert) AND (BCR OR bladder OR NPM1 OR mutation OR FII OR FV)) | *Primary completion* on or after 01/01/2018 Link

The following sources will be searched:

- Trial registries – ClinicalTrials.gov
- News – BioSpace
- Academic literature – Ovid EMBASE, Ovid Medline

The search process is challenging in multiple ways. First, it is difficult to distinguish (in search terms) between material about tests of the human genome from material about genetic tests of pathogens. Second, it is very difficult to distinguish between material about tests with the potential to be used at point-of-care from material about more general genomic testing. Numerous articles about relevant technologies do not use any terminology to directly refer to the potential for being used at point-of-care. There are significant risks of either missing relevant material or including large quantities of non-relevant results making sifting impractical.

Search strategies will be designed to mitigate both these risks. A range of ways of identifying the potential for point-of-care use will be used, but once relevant technologies have been identified, specific top-up searches for those named technologies and citation-chaining will be used to ensure comprehensiveness.

Data Extraction

An overview of each identified technology will be given by the following data fields:

- Name of device – *e.g., GeneXpert*
- Name of assay – *e.g., Xpert BCR-ABL Ultra*
- Developer – *e.g., Cepheid*
- Genomic target – *e.g., BCR-ABL chromosome translocation*
- Condition – *e.g., chronic myeloid leukaemia*
- Population
- Time to results
- Setting – split by at clinic/bedside and at on-site lab
- Operator
- Purpose (prognosis, diagnosis, treatment decisions)
- Cost (if known)
- Diagnostic accuracy (if known) – in relation to comparison with any lab-based tests
    - Sensitivity
    - Specificity
- Stage of development

- Trial phase
- Technology readiness level (TRL) – Measure the maturity of certain technologies. Technology readiness levels measure the maturity of certain technologies, *"However, the readiness of a technology does not necessarily indicate that the related products are ready to be manufactured in an affordable and operationally effective manner"* [Wang et al. (2017)]. To fill this gap, we will look at manufacturing readiness levels (only if the information is available).
- Manufacturing Readiness Level (MRL) - a method that has been applied to assess manufacturing maturity and to identify risks and gaps in technology-manufacturing transitions. (At the most mature stage, or at the highest readiness level, "All materials, tooling, inspection and test equipment, facilities and manpower are in place and have met full rate production requirements")

The data will be recorded in an Excel spreadsheet. Where possible, information for each field will be recorded (e.g., "Good" for specificity if stated in a record, where a value is not given). "Not reported" will be used if no information is available for a field.

Data Analysis

Data derived from the horizon scanning will be combined with results from the evidence synthesis and will be at reported at the level of individual technologies. Data sources will be combined (e.g., clinical trial records/published articles) where appropriate to maximise the information extracted, particularly for technologies which are closer to market and have published articles detailing their purpose, setting, and diagnostic accuracy.

Quality assurance (QA) will be conducted internally at screening and data extraction stage. A 10% sample of records will be used as a pilot screening, where the same records are blindly screened by two or more reviewers, and conflicts resolved prior to moving to single screening. At data extraction, a 10% sample will be double checked by a second reviewer. External QA will also be incorporated into the development process of the report by sending interim reports to the IRLSS team for review.

Data analysis will include:

- A descriptive overview of the results
  - Total number of records retrieved, excluded, and included
  - Total number of trials identified
  - Total number of unique technologies
- Trends in the data
  - Number of technologies for each prognosis, diagnosis, and treatment decisions
  - Stage of development of technologies
  - Healthcare areas being addressed

Deliverables

An Excel spreadsheet and/or written report will be produced, depending on the number of technologies found. A report would follow a 1:3:25 format – 1 page of key messages in plain language, a 3-page executive summary with more detail, and then a 25-page technical report. The length of the report may change based on the findings from the horizon scan. The report may be used as a basis for an academic publication.

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