Rapid Technology Scan: Polygenic Risk Scores (PRS)

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Background: NIHR Innovation Observatory

- Horizon scanning centre based at Newcastle University

- Delivering horizon scanning and early awareness notification service to national healthcare bodies within the UK

- Identifying promising health innovations: medicine, medical devices, diagnostics and digital technologies

- Working closely with the Accelerated Access Collaborative (AAC), and national bodies such as NHS E&I and NICE, UK HSA to support accelerated access of new innovative health technologies and services to NHS patients

Objectives & Scope

- The Innovation Observatory (IO) sought to:

    1. Undertake a rapid technology scan for polygenic risk score (PRS) technologies and identify companies with implementation-ready PRS end-to-end service tools

    2. Summarise key findings regarding the outcomes of this scan and insights gained from business intelligence and academic sources

- To be considered as part of this scan, a company and its technology must appear 'implementation-ready' for use in the UK, meaning: geographically or commercially positioned to allow for the processing of samples, the performance of assay screens, and the carrying out data analysis in the UK.

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Methods

- For the purposes of this rapid technology scan:

  - IO developed a detailed dataset of technologies by formulating search strategies for PRS, based on a comprehensive list of terms with input from expert panels

  - Primary and secondary sources were systematically scanned using a combination of traditional scanning methods, automated and novel AI/machine learning techniques

  - Screened results and extracted intelligence was used in further data processing

- Information sources used as part of this scan included, *inter alia:*

  - Bibliographic databases, including PubMed, EMBASE and MEDLINE

  - MedTech news websites

  - Commercial websites, reports and press releases

  - Academic institution webpages

  - IO ScanMedicine trial database (11 clinical trial registries including UK, EU, USA)

  - Patent databases

  - Regulatory agency

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Limitations

1. This is a rapid technology scan with a limited scope, and results should be interpreted as such

2. IO are not subject matter experts in PRS technologies

3. The classification of technologies was complex and at times subjective due to limited or incomplete information available for the technology or complex aspects of the technology or application which require expert knowledge

Table of Identified Companies/Products

Developer Name	Product Name	Product Description	Country of Development
Genomics PLC	Integrated Risk Tool	Calculate risks for conditions that affect large numbers of patients - either Population Health Management or Clinical Decision Tool.	UK
23andMe	Health + Ancestry Service	Non-invasive DNA test detailing an individual's chance of developing certain health conditions, as broken down by genetic data and ancestry.	USA
Cytox	genoSCORE-LAB test	Non-invasive test that can predict the risk of an individual developing Alzheimer's disease from a saliva sample.	UK
Futura Genetics	Futura Genetics DNA test	Non-invasive DNA test designed to assess risk of developing each of the 28 most common conditions worldwide.	Israel
Predilife	ProstRisk	Multi-pathology prediction tool covering primary cancers (breast, lung, prostate, colorectal, and melanoma) as well as cardiovascular diseases.	France
	MammoRisk	First breast cancer risk prediction solution in France offering women a clear report, with a view to personalized screening.	France
AnteGenes	Ante	A genetic test that assesses a man's personalized risk of developing prostate cancer using a polygenic risk score. The risk model has been evaluated and validated using anonymous population data of the Estonian Biobank and UK Biobank.	Estonia
Myriad Genetics	myRisk® with riskScore	Combination risk score testing that blends genetic test status and personal cancer family history, representing a single hereditary cancer risk test.	USA

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Summary of Key Messages

Our rapid technology scan of PRS technologies found:

1. Significant **interest in the application of PRS** across healthcare –high number of academic and commercial publications on the subject.

2. However, most activity revolves around **early-stage research** and technologies that are **not yet commercialised** – indicating **a pipeline of potential services** that may reach maturity in the near-future.

3. There are **few companies with an end-to-end** commercial offering, especially those localised to Europe.

4. Conversely, geographic development and investment opportunities appear **centered to the American marketplace**.

5. Several companies with commercial offerings are **limited to relatively few disease** areas – though there are some (e.g., 23andMe) who have successfully **expanded the conditions** their products are said to evaluate.

Conclusions & Implications

- IO have identified immediately relevant data on PRS services and provided insights into the pipeline of development, to allow decision-makers and healthcare organisations to evaluate the potential impact of these technologies against their priorities

- There is evidence that a growing number of PRS services are in the pipeline, thus there may be value in considering how this will impact the commercial agreements necessary to implement current and future PRS technologies in the NHS

- The emerging number of technologies also presents an opportunity for the stakeholders to influence the development of early, non-commercialised PRS technologies within the UK

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