

BMJ Open Defining precision health: a scoping review protocol

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ABSTRACT

Introduction Precision health is a nascent field of research that would benefit from clearer operationalisation and distinction from adjacent fields like precision medicine. This clarification is necessary to enable precision health science to tackle some of the most complex and significant health problems that are faced globally. There is a pressing need to examine the progress in human precision health research in the past 10 years and analyse this data to first, find similarities and determine discordances in how precision health is operationalised in the literature and second, identify gaps and future directions for precision health research.

Methods and analysis To define precision health and map research in this field, a scoping review will be undertaken and reported according to Preferred Reporting Items for Systematic Reviews and Meta-Analyses - Scoping Review Extension guidelines. Systematic searches of scientific databases (Medline, Embase, Scopus, Web of Science and PsycINFO) and grey literature sources (Google Scholar, Google Patents) identified 8053 potentially eligible articles published from 1 January 2010 to 30 June 2020. Following removal of duplicates, a total of 3190 articles were imported for screening. Article data will be extracted using a customised extraction template on Covidence and analysed descriptively using narrative synthesis.

Ethics and dissemination Ethics approval is not required. Findings will be disseminated through professional networks, conference presentations and publication in a scientific journal.

BACKGROUND

Supporting the health and well-being of a growing population, in the context of ageing populations and increasingly prevalent chronic diseases, remains one of the most complex and pressing global challenges.¹ Chronic diseases, such as cardiovascular disease, chronic respiratory disease, type 2 diabetes and cancers, carry enormous social, medical and economic burdens.² Furthermore, the aetiology of these non-communicable diseases is largely rooted in lifestyle and environmental factors that are challenging to modify, given the nature of modern environments and occupations.³ In many countries, overnutrition (as a primary driver of non-communicable disease) sits

Strengths and limitations of this study

- This study will be the first to systematically identify and review research in the nascent field of precision health. Relevant research will be identified using a comprehensive and systematic search strategy with a broad operationalisation of precision health.
- The review will take a rigorous approach, adhering to Preferred Reporting Items for Systematic Reviews and Meta-Analyses-Scoping Review guidelines and conducting abstract screening, full-text screening and data extraction in independent duplicate to facilitate greater inter-reviewer reliability.
- The analysis is expected to pinpoint significant concentrations and gaps in precision health research in relation to studied populations, health parameters, disease states and research methods. Insights from these can highlight promising future directions for this rapidly growing field.
- A limitation is that only articles in English will be reviewed.

alongside undernutrition and malnutrition within the same population,⁴ highlighting the complexity and ‘super-wicked’ nature of the global health challenges that we face.⁵

Precision health is a nascent scientific discipline that seeks to develop proactive and personalised solutions to health problems, integrating interindividual variability in genetic, behavioural and environmental determinants of health.⁶ In this way, precision health is not only about enhanced risk profiling and population stratification, but it must also encompass how that information is used to improve people’s health. As an illustration of this mission, a precision health system might see health co-managed proactively by patients and healthcare providers through the synchronous integration of information—starting with genotyping at birth, regular screening throughout the lifespan and combined continuous health monitoring and provision of actionable advice and early intervention at the precise moment when the individual needs it.⁶ A current example is the Integrated Human Microbiome Project



that seeks to improve health outcomes for preterm babies and for people with inflammatory bowel disease and/or type 2 diabetes through the collection and integration of longitudinal multi-omic and functional data about the host.⁷ In another example, scientists, through the application of machine-learning techniques to data from more than 500 000 pregnant women, developed a new predictive model for gestational diabetes mellitus that had just nine questions, representing a cost-effective condition screening tool that can significantly advance women's health.⁷

Aside from these early examples, fulfilling the vision of precision health and well-being for all is still far from realisation. Precision health is in its infancy, having been formalised as a progeny of precision medicine only in the last decade. It is also highly multidisciplinary, attempting to incorporate behavioural, environmental and multi-omic information into cohesive interventions and products that are effective and person centred. As such, one of the current challenges within precision health is its rapid growth combined with a lack of consensus on how it is defined and operationalised across multiple scientific and medical domains.⁸ Addressing this challenge is the focus of this scoping review.

One of the difficulties with defining or operationalising precision health is its entanglement with adjacent disciplines, particularly precision medicine, which uses advanced -omics testing to customise medical treatments based on an individual's unique biomarker profile.⁹ Juengst *et al* (10, p. 883) referred to 'three critical rebranding episodes in contemporary genomics¹: the recent transition from personalised to precision medicine²; the ongoing transition from precision medicine to precision health and³ the incipient transition from precision health to wellness genomics'. This ongoing transition started with efforts to conceptualise the application of human genomics to healthcare and the unconventional idea of individualising treatment and empowering patients to take more responsibility for their own health. Then, interests shifted towards giving clinicians better tools and more authority to use genomic information under the rubric of precision medicine, which in turn led to the movement's importing public health goals and expanding its scope to precision prevention at the population level.¹⁰ Reviews of early precision medicine research indicate progress towards significant advancements in medical science, such as metabolomics (particularly for drug discovery)¹¹ and targeted immunotherapies to treat cancers.¹² Today, the process of merging precision medicine and precision prevention in precision health is giving incipient evidence of another game-changing vision—wellness genomics, which is predicted to achieve goals beyond healthcare.¹¹ Establishing a shared understanding of precision health and its aims is needed to realise the full potentials of precision health and wellness genomics.

Study rationale

Over the past decade, precision health has grown quickly as a scientific discipline. However, it is still a long way away from realising its promise, which will require interdisciplinary collaboration on an unprecedented scale.^{8 13} The lack of a clear definition of precision health and the interchangeable use of terms such as 'precision medicine', 'personalised health', 'precision health' and even 'precision wellness' present a major obstacle to progress in the field.^{13 14} Reviewing how precision health is currently operationalised in existing literature is also needed to facilitate more responsible innovation¹⁵ through better identification of ethical, legal and societal concerns arising from studies tagged as precision health research.^{16 17} Although precision and personalised medicine research have been extensively reviewed,^{18–21} to date, no systematic or scoping reviews of precision health research exist. In order to address this challenge, a scoping review of peer-reviewed and grey literature academic articles focusing on or mentioning 'precision health' and its derivatives will be conducted.

Review objectives

The primary objective of this scoping review is to map the current state of human precision health research, specifically to:

1. Identify how precision health is operationalised in scientific manuscripts and patents.
2. Summarise precision health research study characteristics (eg, study settings, sample characteristics and research designs).
3. Identify gaps in the sociodemographic and health status of participants included in precision health research and determine how these differ across various health domains and contexts.

METHODS

This article outlines the protocol for a scoping review that is currently being conducted. The Preferred Reporting Items for Systematic Reviews and Meta-Analyses-Scoping Review Extension²² and the guidelines set by the Joanna Briggs Institute²³ are used to ensure rigour and facilitate replicability of the scoping review.

Formative work: development of the search strategy

The search strategy was developed using an iterative approach, with two rounds of preliminary searches and refinement of the search strategy based on initial search results. Each preliminary search was performed in two databases, Scopus and Medline, applying database-specific Boolean operators. To maintain feasibility of the preliminary searches, each search was constrained to research published in 2020.

Preliminary searches were undertaken in March and April 2020. The initial search strategy was guided by synonyms for 'personalised medicine' identified by Brooks,²⁴ including the following: precision, personalisation, individualisation, stratification, integrated and

tailored. Two group leaders of precision health research were also consulted to gauge the suitability of the search terms. The term ‘precision’ or its permutations were used with ‘health’ to search for relevant articles on Scopus and PubMed. Only the titles and abstracts were searched, and only articles that were published in 2020 were included for screening. We retrieved a total of 253 unique abstracts. Two authors screened each abstract in independent duplicate, with the inclusion criterion being an empirical study and the exclusion criteria being a review, editorial or perspective piece. Initial abstract screening resulted in more than 50 disagreements, which were resolved through discussion. From this initial preliminary search, we found that the term ‘integrated’ mostly captured articles that refer to integrated health systems rather than current conceptions of precision or personalised health,^{7 25} and this term was, therefore, excluded from the next iteration of the search strategy. Furthermore, many of the irrelevant articles that were captured in earlier searches included the concepts ‘precision’ and ‘health’ but not necessarily in combination. Therefore, a further refinement to the search strategy was made such that the two search concepts are adjacent in the text.

A second preliminary search took place in the second half of April 2020. Excluding ‘integrated’ from the search string increased the relevance of the results and resulted in 69 unique abstracts that were then screened. Additional details were added to the inclusion and exclusion criteria to increase agreement between the reviewers (see [table 1](#)). Following these criteria resulted in just nine disagreements, which were resolved through discussion. At this stage, the authors were satisfied that the search strategy was appropriate for the actual review.

Eligibility criteria

Eligibility criteria were established using insights from the preliminary searches. Since the primary aim of the review is to provide a wide overview of precision health research, we placed as few restrictions as possible on eligibility. Eligible studies included those that are published between 1 January 2010 (since precision health only formally distinguished itself from precision medicine after this date) and the search date (30 June 2020); describe original research or a protocol for original research that focuses on a health or medical outcome; include the terms ‘precision health’ or its synonyms in the title or abstract; and involve human participants, samples and/or datasets (see [table 1](#)). Eligible studies were not limited to a particular population or setting.

Information sources

The search was performed across seven databases to retrieve a wide range of potentially relevant peer-reviewed scientific reports, grey literature and patents. All searches were conducted on 30 June 2020. In particular, we searched for:

Peer-Reviewed scientific reports

Consistent with evidence regarding optimal coverage for health and medical topics,²⁶ Medline (through OVID), Embase, Scopus and Web of Science databases were searched. In addition, we also searched PsycINFO (through OVID) to capture broader applications of precision health that may pertain to well-being and mental health.

Table 1 Review eligibility criteria based on study population, concept, context and types of evidence

	Inclusion	Exclusion
Population	<ul style="list-style-type: none"> ▶ Human participants of any description (eg, adults, children, adolescents, older adults, populations with a particular health or medical condition, healthy people) ▶ Human samples (eg, tissue samples, genetic material). ▶ Analysis of historical datasets (eg, health records, epidemiological datasets). 	<ul style="list-style-type: none"> ▶ Evaluations of new technologies that do not include human participants (eg, evaluating tensile properties of a new fabric, developing a new medical diagnostic kit/device without in-human testing)
Concept	<ul style="list-style-type: none"> ▶ Studies that refer to the concept of precision health or its derivatives (eg, personalised health, individualised health, stratified health, tailored health) ▶ Any study collecting health-related clinical, psychosocial, or behavioural information (eg, weight loss, disease prevalence/risk, physical activity, mental health) 	<ul style="list-style-type: none"> ▶ Animal studies, in silico studies, and testing of materials for precision health applications ▶ Non-health outcomes including economic outcomes (eg, cost-effectiveness studies), human/sports performance outcomes (eg, physical conditioning programmes for healthy athletes)
Context	<ul style="list-style-type: none"> ▶ Any geographical location or setting of any nature (including online studies) 	<ul style="list-style-type: none"> ▶ None
Types of evidence	<ul style="list-style-type: none"> ▶ Primary empirical research studies, (eg, randomised controlled trials, cohort studies, cross-sectional studies, and case reports). ▶ Protocols for planned studies ▶ Full-text articles ▶ Full-text conference proceedings ▶ Articles written in English ▶ Patents 	<ul style="list-style-type: none"> ▶ Reviews (eg, systematic reviews, narrative reviews) ▶ Editorial articles (eg, perspective pieces, position statements) ▶ Abstracts or posters ▶ Articles for which we cannot obtain the full text or that are not written in English ▶ Dissertations

Grey literature and patents

Grey literature includes resources published outside traditional academic journals, and thus, encompasses government or commercial research reports, dissertations and conference reports²⁷; and patents or applications for exclusive rights to intellectual property. The inclusion of grey literature in systematic searches is considered important for establishing a balanced and complete picture of the available evidence.²⁷ In addition, including patents is crucial in the context of precision health because innovations are occurring in the private sector, in addition to traditional academic and scientific institutions. Consistent with previous research, we searched Google Scholar to identify any relevant grey literature research²⁸ and Google Patents to identify any patents or registered intellectual property. Google Patents has broad international coverage and indexes more than 100 patent offices around the world and more than 120 million patent applications.

We also undertook hand searching of the reference lists of relevant primary studies and reviews, as well as publications on websites of precision health research groups.

Search strategy and terms

The search terms included two concepts:

1. “preci(sion)*” OR “personal(ised)*” OR “individual(ised)*” OR “stratif(ied)*” OR “tailor(ed)*” AND
2. “health”

Search terms were adapted for each database with proximity parameters applied such that the concepts ‘precision’ and ‘health’ are immediately adjacent. For example, the search string for Scopus was:

(TITLE ((precision OR personali* OR individuali* OR stratif* OR tailo*) PRE/0 health) OR ABS ((precision OR personali* OR individuali* OR stratif* OR tailo*) PRE/0 health)).

The search identified a total of 8053 articles. Covidence detected and removed 4863 duplicates, leaving 3190 studies for screening.

Screening procedure

The review is being conducted in Covidence, a web-based systematic review platform, in four stages. In stage 1, all articles obtained from the database searches were imported into Covidence, and the platform was used to detect and remove duplicates. In stage 2, we checked all the remaining article titles to remove duplicates that were not detected by Covidence. In stage 3, all article abstracts were screened in independent duplicate to gauge shared understanding of the selection criteria, discuss any disagreements and further specify the inclusion and exclusion parameters. Any abstract where it is not clear whether the inclusion criteria are met was automatically passed through to the full-text screening stage.

Stage 4, which involves independent duplicate screening of all full texts, is currently underway. In addition to the article eligibility criteria in [table 1](#), we have also decided to exclude articles that only use the search

terms in the abstract, discussion and/or conclusion. Only including articles that also mention the search terms in the introduction, methods and/or results allows us to focus on articles that are primarily positioned as precision, personalised, individualised, stratified or tailored health. In stage 4, discrepancies in the inclusion or exclusion decision are resolved through discussion.

Data charting

The next stage in the review process is data charting (to be commenced). Data from all included articles will be extracted in independent duplicate to ensure charting consistency and inter-reviewer reliability. We plan to extract information on the type of study, country or countries of affiliation of the authors, authors’ disciplinary affiliations, characteristics of the human participants recruited (age, gender, health status, socio-economic status, and race/ethnicity), purpose of the study, research design and setting, focal health issue being monitored or addressed, type of intervention (for interventional studies) and nature of the data gathered from participants. Data charting will be conducted using Covidence.

We will also extract as qualitative data the sentence in which the authors refer to the concept ‘precision health’ or its derivatives. If the concept is mentioned multiple times, we will extract the sentence that best defines the concept.

Data synthesis

Data will be analysed and summarised descriptively, with study characteristics presented in tabular and graphical forms and summarised in the text using a narrative approach. Research gaps will be identified through comparative analysis of study and participant characteristics.

Qualitative data that capture the authors’ reference to precision health will be subjected to thematic analysis according to Braun and Clarke’s inductive-deductive approach to identify themes and commonalities in how precision health is operationalised or co-opted as a theme or rationale for research studies.²⁹ In reporting our results, we will also highlight similarities and differences in how precision health is operationalised in the academic literature.

ETHICS AND DISSEMINATION

This study involves neither human participants nor unpublished secondary data. As such, approval from a human research ethics committee is not required. Findings of the scoping review will be disseminated through professional networks, conference presentations and publication in a scientific journal.

Patient and public involvement

This work analyses existing research studies, and therefore, involves no patients or members of the public.

Implications

This scoping review aims to provide a snapshot of the current state of human precision health research. Future directions for the field will be identified as part of the broader discussion of our results. In addition, implications of our findings for ensuring responsible innovation¹⁵ in this research domain will be explored. Particularly, our findings could help researchers reflect more on the extent of interdisciplinarity of their work and on the efforts they are making to realise an equitable precision health future for diverse populations, one that integrates individual genetic, behavioural and environmental information for health monitoring and maintenance across the lifespan.⁷

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