Trends and gaps in precision health research: a scoping review

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ABSTRACT

Objective To determine progress and gaps in global precision health research, examining whether precision health studies integrate multiple types of information for health promotion or restoration.

Design Scoping review.

Data sources Searches in Medline (OVID), PsycINFO (OVID), Embase, Scopus, Web of Science and grey literature (Google Scholar) were carried out in June 2020.

Eligibility criteria Studies should describe original precision health research; involve human participants, datasets or samples; and collect health-related information. Reviews, editorial articles, conference abstracts or posters, dissertations and articles not published in English were excluded.

Data extraction and synthesis The following data were extracted in independent duplicate: author details, study objectives, technology developed, study design, health conditions addressed, precision health focus, data collected for personalisation, participant characteristics and sentence defining ‘precision health’. Quantitative and qualitative data were summarised narratively in text and presented in tables and graphs.

Results After screening 8053 articles, 225 studies were reviewed. Almost half (105/225, 46.7%) of the studies focused on developing an intervention, primarily digital health promotion tools (80/225, 35.6%). Only 28.9% (65/225) of the studies used at least four types of participant data for tailoring, with personalisation usually based on behavioural (108/225, 48%), sociodemographic (100/225, 44.4%) and/or clinical (98/225, 43.6%) information. Participant median age was 48 years old (IQR 28–61), and the top three health conditions addressed were metabolic disorders (35/225, 15.6%), cardiovascular disease (29/225, 12.9%) and cancer (26/225, 11.6%). Only 68% of the studies (153/225) reported participants’ gender, 38.7% (87/225) provided participants’ race/ethnicity, and 20.4% (46/225) included people from socioeconomically disadvantaged backgrounds. More than 57% of the articles (130/225) have authors from only one discipline.

Conclusions Although there is a growing number of precision health studies that test or develop interventions, there is a significant gap in the integration of multiple data types, systematic intervention assessment using randomised controlled trials and reporting of participant gender and ethnicity. Greater interdisciplinary collaboration is needed to gather multiple data types; collectively analyse big and complex data; and provide interventions that restore, maintain and/or promote good health for all, from birth to old age.

INTRODUCTION

Precision health is a nascent field that seeks to maximise population health and wellbeing while minimising premature disability and death through the continuous monitoring of key health data, generation of actionable health discoveries and recommendation of personalised interventions.1,2

It is derived from precision medicine, which similarly considers individual variation in biological, environmental and behavioural data to inform the diagnosis and treatment of disease.3 Distinct from precision medicine, precision health takes a lifespan perspective in health monitoring, identifying actionable risks and intervening early. Interventions may include continuous health screening, early diagnostic testing and support to improve behaviour and lifestyle. Interventions are also personalised, with each piece of information considered in context, different to the typical ‘one-size-fits-all’ approach of modern medicine.

While precision health is in its infancy, early work has produced promising findings. In
the Integrated Personal Omics Profiling (iPOP) study, longitudinal health monitoring was undertaken using a comprehensive array of multiomic (eg, genome, transcriptome and microbiome), lifestyle and clinical measures to identify actionable health discoveries for people with type 2 diabetes. Integration of these measures led to accurate disease diagnosis, which can enable personalised treatment plans. Majority of participants took action as a result of study participation, modifying their lifestyle and discussing findings with medical practitioners. The iPoP study highlights the potential value of precision health, using comprehensive, highly specific and integrated data for health management. Other promising signs for precision health include establishment of substantive research groups and collaborative efforts, formative work to develop technologies and algorithms, and proof-of-concept studies with limited populations. However, there is still limited knowledge on the characteristics of studies categorised as precision health, and the extent to which they fulfill the vision of a precision healthcare future.

The early stages of precision health offer a unique opportunity to determine key research agendas, identify promising research trajectories, and highlight knowledge gaps, which can then help shape future directions for research and implementation. As a first step towards achieving these goals, this scoping review maps precision health research in the past 10 years and provides an overview of research progress, trends and gaps.

METHODS
This scoping review is based on a published protocol developed in accordance with the Preferred Reporting Items for Systematic Reviews and Meta-Analyses-Scoping Review Extension (online supplemental appendix 1) and Joanna Briggs Institute guidelines. Information sources
Searches were undertaken in five electronic bibliographic databases including Medline, PsycINFO (through OVID), Embase, Scopus, Web of Science and grey literature databases including Google Scholar on 30 June 2020. Grey literature searches were considered important in the context of this review due to the likelihood of innovations published outside conventional academic databases. Hand searching of the reference lists of reviews or discussion papers and the publication lists of precision health research groups on their websites was also undertaken.

Search strategy
Preliminary searches established the final search terms and eligibility criteria, which were designed to capture a range of precision health research. Search terms included ‘precision’ or its synonyms ‘personalised’, ‘individualised’, ‘stratified’ or ‘tailored’, as identified by Ali-Khan et al. and with ‘health’ immediately adjacent (eg, ‘precision health’). The search strings used for Medline (OVID), PsycINFO (OVID), Embase, Scopus, Web of Science and grey literature (Google Scholar) are presented in online supplemental appendix 2.

Eligibility criteria
Eligible articles included those published between 1 January 2010 and 30 June 2020, with the term ‘precision health’ being established as independent from ‘precision medicine’ after 2010. Articles were included if they described original research or the protocol for original research, involved human participants or samples including historical datasets and collected health-related data. Articles were required to make a reference to ‘precision health’ or its derivatives in the title and/or abstract and the introduction, methods and/or results. Reviews, editorial articles, conference abstracts or posters and dissertations were excluded, along with articles not published in English and those where the full-text article could not be retrieved using resources from three different institutions. No limitations were placed on the study population, context or setting.

Screening
All stages of the screening process were conducted in Covidence (Veritas Health Innovation), an online tool for systematic reviews. Duplicates were removed by Covidence, with the authors manually checking for missed duplicates. Title, abstract and full-text screening against inclusion and exclusion criteria were performed independently in duplicate by JNV, JCR, SE, CM, HS and SG. Discrepancies at each screening stage were discussed until consensus was reached.

Data extraction
The Covidence extraction form was modified by all team members using an iterative process. Data extraction was conducted independently in duplicate using Covidence. A third, independent reviewer undertook consensus on the extracted data. Data extracted included funding sources, number of authors, conflicts of interest, the country of author affiliations, author disciplinary association, study purpose, technology being developed or application of the findings, study design and setting, sample size, focal health condition being addressed, precision health focus, the type of data collected for the purpose of personalisation/tailoring, and participant characteristics (ie, age, sex, health status, ethnicity and socioeconomic status). The sentence in the body of the paper defining the term ‘precision health’ or its derivatives was also extracted.

Data synthesis and analysis
Data were analysed and summarised narratively in text and presented in tables and graphs where appropriate.

Content analysis on free-text data defining precision health and describing the objectives of each study was performed using Leximancer. Data were imported into Leximancer, and standard parameters were used to process and clean the data. Specifically, text was analysed...
in maximum two-sentence segments. Data were processed with standard stop words removed from text automatically. In addition, the search concepts (precision, health, personalised, stratified, tailored and individualised) were included as additional stop words, while analysis of the study aims included ‘aim’ and ‘aims’ as additional stop words. Similar concepts were also merged (eg, intervention and interventions, method and methods).

**Patient and public involvement**

No patients were involved in this scoping review.

**RESULTS**

**Article screening**

Searches retrieved 8053 articles, with 225 included after removal of duplicates and title, abstract and full-text screening ([figure 1](#) online supplemental appendices 3 and 4A,B). The reference lists of 30 relevant reviews or discussion papers were also searched; however, no additional primary studies were retrieved. Almost half of the articles (104/225, 46.2%) were published between 2017 and 2020, and only three articles (1.3%) in 2010.

**Context and characteristics of included studies**

A summary of study characteristics is presented in [table 1](#). Majority of the studies were in North America (97/225, 43.1%), led by the USA. There were few studies conducted in African (12/225, 5.3%) and South American countries (2/225, 0.9%). Authors were mostly based in health and medical research departments and in universities/academic institutions. Over half of the articles had authors from just one discipline (130/225, 57.8%) and one type of institution (132/225, 58.7%). More than half of the studies (120/225, 53.3%) were directly funded by governments, including national research funding agencies and initiatives.

Most articles were leading to the development of an individual digital health promotion tool or community/public health programme, while there were few studies on implanted medical devices. Digital tools for health management included web-based programmes, mobile phone apps or text messages and wearables. Community or face-to-face health programmes have been developed for or implemented in churches, rural communities, youth healthcare centres, adult day-care centres, paediatric practices, assisted living facilities, hospitals, mobile health counselling units, schools and workplaces.

We used the model of Gambhir **et al.** to determine the stage in the precision health ecosystem that our reviewed articles focus on. Their cyclical model involves four key components, (1) risk assessment at all life stages, (2) customised personal and environmental monitoring, and an integrated health portal where (3) data is analysed and (4) personalised interventions are provided. In our review, 105 articles (46.7%) developed or tested
an intervention, 53 (23.6%) focused on risk assessment, 42 (18.7%) primarily involved data analytics, and only 25 (11.1%) were dedicated to customised monitoring.

Risk assessment articles include studies that determined risk for hospital readmission for people with cardiovascular disease, human papillomavirus infection, 10-year survival of people with myotonic dystrophy and diabetes based on lifestyle information. Data analytics studies developed algorithms or analysed big datasets to monitor air pollution, forecasted wellness from ECG signals or categorised diseases. Finally, studies on customised monitoring have continuously obtained data on home...
The diseases, behaviours and/or conditions targeted by each study, based on their aims, are presented in the top panel of Table 2. Chronic diseases (103/225 articles, 45.8%), including type 2 diabetes, cardiovascular disease and cancer, are among the 10 most common conditions targeted. There were also studies focused on modifying environments; metabolites, vital signs, or neural activation during physical activity; blood glucose and uric acid, or bioimpedance for respiratory assessment.

In terms of study design, only 36 articles conducted or planned to conduct a randomised clinical trial, whereas cross-sectional studies made up a quarter of the articles.

### Conditions targeted or addressed

The diseases, behaviours and/or conditions targeted by each study, based on their aims, are presented in the top panel of Table 2. Chronic diseases (103/225 articles, 45.8%), including type 2 diabetes, cardiovascular disease and cancer, are among the 10 most common conditions targeted. There were also studies focused on modifying behaviours, such as physical activity and substance use (28/225, 12.4%). Several studies addressed sexual health issues, including HIV and HPV. In terms of actual participants recruited, 87 articles (38.7%) did not mention the health condition of their participants, and 81 articles (36%) indicated that they recruited participants who have a chronic illness. There were 43 articles (19.1%) that recruited participants at-risk for certain conditions or in a predisease stage, 11 articles (4.89%) that recruited participants with an acute illness and 22 articles (9.78%) that explicitly mentioned the recruitment of ‘healthy’ or non-diseased populations. The 10 most common conditions of participants in articles that involved individuals at-risk or have acute or chronic diseases are presented in the bottom panel of Table 2.

### Types of data for personalisation

In terms of data gathered for or that had implications for health personalisation (Figure 2), 102 studies (45.3%) collected one or two types of data, whereas only 6 studies (2.7%) used seven or more types of data for personalisation. Almost half of the studies used behavioural or lifestyle information (108/225, 48%) and more than 40% used sociodemographic (100/225, 44.4%) or clinical (98/225, 43.6%) information; however, genetic data were rarely gathered (19/225, 8.4%). Among the behavioural information that studies have used for personalising health interventions are cigarette or alcohol consumption, physical activity, sleep, sexual behaviour, television viewing and computer use and/or drug use. Sociodemographic and clinical data frequently used for intervention tailoring encompassed age, gender, blood glucose, blood pressure, cholesterol, body fat, medical history, comorbidities and/or disease severity.

### Participant demographics

Sample sizes varied between one and 7 995 048, with the median being 120 (IQR 28–600). There were 20 studies (8.9%) that included 10 or fewer participants, whereas 12 studies (5.3%) included more than 10 000 participants. The median age of participants was 48 years (IQR 28.4–60.8), with the youngest participant being <1 year old and the oldest, 119 years. Based on the mean or median age of participants recruited in 123 studies providing these information, 100 studies (44.4%) focused on recruiting people from 20 to 69 years old, whereas only 11 studies (4.9%) primarily recruited people below 20 years old and 12 studies (5.3%) primarily recruited people older than 69 years old.

For articles that mentioned participant sex (153/225, 68.0%), the median percentage of female participants is 53.5% (IQR 40%–72%). Of the articles that clearly reported participant race/ethnicity (87/225, 38.7%), the median percentage of non-Caucasian participants recruited per study is 45.5% (IQR 12.02%–100%). Several studies included participants from disadvantaged socioeconomic backgrounds (46/225, 20.4%), including

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**Table 2** Ten most common conditions and/or behaviours targeted (n=225 articles) and ten most common health issues of participants in 126 articles that recruited people who have preclinical, acute or chronic conditions

| Articles | % |
|----------|---|
| Metabolic disorder | 35 | 15.6 |
| Cardiovascular disease | 29 | 12.9 |
| General/preventive health, chronic diseases, wellness | 26 | 11.6 |
| Cancer | 26 | 11.6 |
| Physical activity and weight loss | 16 | 7.1 |
| Neurological disorders | 13 | 5.8 |
| Smoking, alcohol and substance use | 12 | 5.3 |
| Sexually transmitted infections and sexual health | 12 | 5.3 |
| Mental health and psychiatric disorders | 11 | 4.9 |
| Overweight, obesity | 11 | 4.9 |

*Articles can target more than one condition and also recruit participants with multiple conditions.*
people who have low income or are unemployed, low literacy, limited education, limited internet or technology access and/or no insurance; live in a rural area; and/or are migrants.

Conceptualising precision health
Results of the Leximancer text analysis are presented in figure 3. Mapping the conceptualisation of precision health in the body of the paper (figure 3A,B) reveals that the primary themes relate to patient care, intervention, information or data, monitoring and behaviour. For the study objectives (figure 3C,D), which reflect how precision health is operationalised, primary themes relate to patients, programmes and systems, monitoring, development and effects.

DISCUSSION
This scoping review systematically and rigorously mapped progress, trends and gaps in precision health over the past decade. Various reviews and perspective articles have introduced precision health as an emerging area of research, yet it is unknown whether aspirations align with research being conducted and identified as precision health. Although precision health aspires to combine multiple types of information, our review demonstrates that most studies only used one or two categories of information for the personalisation of interventions, with behavioural data the most common data type gathered. Most precision health studies also aim to deliver or test an intervention, and individual digital health promotion tools are the most common study outcome.
The vision for precision health, as proposed by Hickey et al., is the integration of phenotype, lifestyle and environmental factors, and genotype and other biomarkers to discover, design and deliver interventions for the prevention or management of disease symptoms. This review demonstrated limited integration of multiple types of data, with 45.3% of the reviewed articles collecting or using only one or two types of data for personalisation applications. Only two studies gathered eight different categories of data, using them to develop a child functional profile or community interventions for diabetes and hypertension. Our review also showed that behavioural, sociodemographic and clinical data were commonly gathered for personalising interventions or other precision health applications, and limited studies used genomic or socioenvironmental factors. Although this deviates from the focus of precision medicine on genomics, the increasing use of behavioural and environmental information suggests increasing acknowledgement of the importance of social and environmental determinants of health. Reviewing authors’ disciplinary and institutional affiliations also revealed 34 articles with authors from the behavioural and social sciences, further supporting the important role that these disciplines may play in developing precision health interventions that are attuned to personal needs, relationships and environments and that account for ethical and equity issues. Overall, our findings on data used for personalisation indicate that the vision for precision health is yet to be fulfilled, with the need to integrate a broader and more diverse array of measures for health maintenance, disease prevention and disease management or treatment.

Determining the stage in the precision healthcare ecosystem using the model of Gambhir et al. revealed that most studies focused on the development of interventions. Examining the outcomes of each study showed that majority developed digital health tools and community programmes. The prevalence of studies that focused on developing digital health tools underscores the key role of computer science in precision health, not just in developing web platforms and mobile health applications, but also in creating algorithms for the analysis of large datasets containing multiple types of information. The high percentage of studies developing community programmes illustrates that precision health also encompasses public health and face-to-face interventions, rather than simply using digital technologies and genetic data to assess and/or promote health. This finding also addresses concerns on the dehumanisation or depersonalisation of healthcare brought about by precision medicine and health information technologies. Reviewing study designs, there were only 36 randomised controlled...
Precision health is a rapidly growing field driven by a vision of integrating biological, psychological, lifestyle, social and environmental information to assess health status and provide interventions for maintaining or restoring health. To fulfil this aspiration and to address existing gaps, additional interdisciplinary work is needed in integrating different types of information and in determining the safety and efficacy of interventions using randomised controlled trials. Precision health studies can take advantage of ongoing precision medicine programs\textsuperscript{111,112} to include genomic information in health monitoring and management. Precision health teams also need to include computer scientists and social scientists to better understand social and environmental determinants of health and integrate them with individual biological and behavioural data. Finally, future studies should include data from children and the elderly, report participant gender and race/ethnicity, and be conducted in South America and Africa. Fostering diversity and inclusion will allow analyses of larger and more diverse datasets, which can then facilitate more accurate assessment of the disease risk and health status of individuals from a wide range of populations and contexts. For precision health to truly fulfil its promise, crucial steps must be taken to facilitate interdisciplinary, ethical, responsible and inclusive research, paving the way for quality healthcare regardless of age, gender, class, ethnicity and nationality.

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**Contributors** JNV conceptualised and managed the project; collected, curated, analysed and visualised the data; and wrote the original and revised draft. JNV is also the guarantor; who accepts full responsibility for the work and/or the conduct of the study, had access to the data, and controlled the decision to publish. JCR conceptualised and managed the project; collected, curated, analysed and visualised the data; and wrote and reviewed the draft. SE and CM collected, curated, analysed and visualised the data; and wrote and reviewed the draft. SG and HS collected and curated the data and reviewed the draft. N0'C conceptualised the project, reviewed the draft and provided funding for the publication of the scoping review protocol.

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**Trials conducted or planned, suggesting that most technologies or programmes are still in the early stages of development and that their efficacy and safety still need to be properly tested in comparison to a control group and outside the laboratory. Taken together, these results demonstrate promising progress in digital technologies and community programmes for precision health and set the challenge for translating technologies to the market or implementing programmes at a much larger scale, in addition to ensuring that they are effective, safe, and applicable to multiple populations and contexts.**

Our review has uncovered gaps in recruited participants of certain ages, reporting of participant gender and race/ethnicity, inclusion of people from disadvantaged socioeconomic backgrounds, and countries where precision health research is conducted. Few studies recruited children and adolescents or older adults. Although there are ethical and practical challenges in recruiting children, adolescents and the elderly,\textsuperscript{89} there is a need to obtain more data on and test monitoring systems, diagnostics and interventions for health issues experienced by these populations, following precision health’s goal of health maintenance across the lifespan.\textsuperscript{92} Second, more than one-third of the studies did not report their participants’ gender, less than half reported the race/ethnicity of their participants, and only 21% included individuals from disadvantaged socioeconomic backgrounds. These highlight significant gaps in ensuring equitable precision health research and application. Given gender\textsuperscript{92} and racial disparities\textsuperscript{94–96} in health and medical research, it is crucial to ensure representation of women and racial/ethnic minorities in precision health research, in addition to adequate reporting of outcomes for various participant subpopulations. Third, there are only limited studies conducted in African\textsuperscript{49} 97–103 or South American\textsuperscript{104} 105 countries, underscoring persistent global disparities in health technology innovation\textsuperscript{106} and medical research.\textsuperscript{107}

The strength of this scoping review lies in the breadth of the search, using synonyms of ‘precision health’\textsuperscript{11} to capture relevant studies; and the review’s rigour, where each step was conducted in independent duplicate. This review can serve as basis for future synthesis of precision health advances, which can expand the current review’s scope and fill in its limitations. Our review excluded dissertations and unpublished clinical trials, which could highlight preliminary findings and emerging advances in the field.\textsuperscript{108} We no longer included patents since their structure does not align with our data extraction sheet, limiting the information that can be extracted and compared with academic publications. Future reviews should include non-English publications and should expand the search strategy to better capture emerging subdisciplines such as ‘precision public health’\textsuperscript{4} and ‘precision mental health’.\textsuperscript{109} Although including stakeholder voices to re-examine findings\textsuperscript{110} is not required for scoping reviews,\textsuperscript{5} this step could be vital in understanding how precision health researchers perceive progress in the field and how they can address research gaps.
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