# Patient-facing digital tools for delivering genetic services: a systematic review

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## ABSTRACT

This study systematically reviewed the literature on the impact of digital genetics tools on patient care and system efficiencies. MEDLINE and Embase were searched for articles published between January 2010 and March 2021. Studies evaluating the use of patient-facing digital tools in the context of genetic service delivery were included. Two reviewers screened and extracted patient-reported and system-focused outcomes from each study. Data were synthesised using a descriptive approach. Of 3226 unique studies identified, 87 were included. A total of 70 unique digital tools were identified. As a result of using digital tools, 84% of studies reported a positive outcome in at least one of the following patient outcomes: knowledge, psychosocial well-being, behavioural/management changes, family communication, decision-making or level of engagement. Digital tools improved workflow and efficiency for providers and reduced the amount of time they needed to spend with patients. However, we identified a misalignment between study purpose and patient-reported outcomes measured and a lack of tools that encompass the entire genetic counselling and testing trajectory. Given increased demand for genetic services and the shift towards virtual care, this review provides evidence that digital tools can be used to efficiently deliver patient-centred care. Future research should prioritise development, evaluation and implementation of digital tools that can support the entire patient trajectory across a range of clinical settings. PROSPERO registration numberCRD42020202862.

## INTRODUCTION

Increased demand for genetic counselling and testing services has placed unsustainable pressure on traditional models of care.[1 2] The emergence of the COVID-19 pandemic in early 2020 further exacerbated the situation and forced genetics services to explore and expand the use of digital tools in patient care.[3–9] To increase access to and efficiency of genetic services, a variety of digital health technologies have been developed and implemented.[1 3–5] To date, digital tools have been integrated into various points in the genetic testing pathway, including clinical assessment, family history-taking, education, post-test counselling and follow-up.[10 11] Examples of digital tools in genetics include pedigree software,[12] hereditary cancer risk assessment tools,[13] online decision aids,[14 15] and computer-based facial dysmorphology analysis tools.[16]

To date, most studies evaluating the impact of digital technologies in genetic services have focused on the use of digital tools in the context of telemedicine and telegenetics.[17–21] These studies show that telegenetics is non-inferior to inperson consults in achieving similar patient outcomes, including patient knowledge, psychosocial and counselling measures.[17–21] Studies show that digital tools are well received by patients, with high levels of acceptability and satisfaction.[15 22] Tools have generally had a positive impact on patient outcomes, including increasing knowledge, reducing decisional conflict, initiating active decision-making for patients and overall enabling patient-centred care.[11 23] For clinicians and the healthcare system, digital tools have improved provider capacity and efficiency.[7 8] However, digital tools carry potential risks, including data security breaches, access challenges in remote locations and patient anxiety associated with accessing medical information outside of a face-to-face encounter.[24 25]

To inform efforts to scale the use of digital tools outside the context of telemedicine and telegenetics, we conducted a systematic review to synthesise existing evidence on the use of digital tools throughout the genetic counselling and testing trajectory.

## METHODS

### Study registration

The protocol for this review was submitted to PROSPERO in October 2020 (registration number CRD42020202862). Preparation of this paper was guided by the Preferred Reporting Items for Systematic Reviews and Meta-Analyses reporting checklist.[26]

### Study selection criteria

#### Population

Studies evaluating the use of patient-facing digital tools in the context of genetic service delivery were included. For the purposes of this study, a patient-facing digital tool was defined as any digital tool that was intended for use by patients. Studies were excluded if the users of the tool were limited to healthcare professionals (HCPs).

#### Intervention

The intervention of interest was any patient-facing tool that was used in genetic service delivery through electronic technology. This included, but was not limited to, web-based portals, chatbots,

software, videos and eBooks. Genetic service delivery included the following steps: clinical assessment, education, pretest genetic counselling, result reporting, post-test counselling and reanalysis. Studies were excluded if the tool was only used in somatic genetic testing or if its focus was on lifestyle-related education tools for polygenic conditions that did not incorporate genetic testing.

### Comparator

Comparator groups were patients who did not use a digital tool or received usual care. Studies without comparator groups were included.

### Outcomes

Outcomes were determined based on review of the relevant literature and discussion among the research team. Studies were included if they reported data on patient-reported usability or patient-reported outcomes. Specifically, data on users' acceptability, satisfaction or intention to recommend the digital tool to others were collected.[27] For patient-reported outcomes, data on the following were collected: knowledge/understanding, psychosocial well-being (eg, anxiety, distress), behavioural or management changes, family-related communication, facilitation of decision-making, patient engagement (eg, self-reported level of patients' involvement in their care) and quality of life.[28 29]

In addition, system-focused outcomes (eg, effect on wait times and time with HCP, workflow efficiency) and provider-reported outcomes (eg, satisfaction with the tool, efficiency in providers' daily practice) were included if reported.

## Study design

Experimental studies, observational studies and case series were eligible for inclusion. Protocols, systematic reviews, commentaries, animal studies, conference abstracts and non-English articles were excluded.

## Search strategy

The search strategy was developed in consultation with a medical librarian. Search terms included the following: eHealth OR virtual OR digital AND genetic testing OR genome sequencing OR genetic counseling. The complete list of search terms is provided in online supplemental file S1. The reference lists of included studies were hand-searched to identify additional articles. Grey literature and trial registries were not searched.

## Information sources

A systematic, comprehensive search of Ovid MEDLINE, Ovid MEDLINE-in-Process, MEDLINE Epub Ahead of Print and Embase Classic+Embase databases (OvidSP) was initially run on 16 July 2020, with an updated search on 29 March 2021, to identify relevant articles published between January 2010 and March 2021. The year 2010 was selected as the starting point because multiple professional guidelines were published at this time on the use of more comprehensive genetic tests (eg, chromosomal microarray and genome-wide sequencing) for various indications, including developmental disorders and hereditary cancers, leading to an increased demand for genetic services.[30–34]

## Selection of studies

Search results were imported to Covidence software (http://covidence.org). All abstracts and full texts were reviewed by at least two independent reviewers (DA+WL or DA+SS). Conflicts were resolved by discussion and a third reviewer not involved in the initial screening was consulted where necessary (SL). The per cent agreement between reviewers for article inclusion at the abstract and full-text screening stages was calculated.

## Data extraction

To ensure data extraction from each publication was conducted by at least two independent reviewers, six team members (DA, WL, SL, SS, AT and CS) were divided evenly into reviewer pairs. Data extracted were bibliographic information, study characteristics (eg, methodology, country, year), participant characteristics (eg, clinical population), tool characteristics (eg, purpose, target audience, components) and data relevant to the aforementioned patient-reported and system-focused outcomes of interest. Any discrepancies in the extracted data were resolved by discussion among the six team members.

For study characteristics, the following items were extracted: publication year, country of data collection, study methodology (eg, quantitative, qualitative, multimethod and mixed method) and purpose of study. Studies that used a mixture of quantitative and qualitative methods without mixing of the quantitative and qualitative data were categorised as multimethod, while studies that involved mixing of methodologies were categorised as mixed method. Study purposes included acceptability, development, evaluation, feasibility, implementation, usability and validation. The primary purpose of the study was determined based on the study aims or objectives as described by the study authors. Secondary and tertiary purposes were extracted if described.

The following items were extracted for participant characteristics: type, size, clinical setting, and diversity, equity and inclusion (DEI) related outcomes (ie, ethnicity, socioeconomic level, education level, general literacy, health literacy, digital literacy, reading level of the content and whether fluency in English was required to participate in the study).

For studies that recruited two or more participant populations, we considered the digital tool's target population as the primary population of the study. Other populations that were included in the study were categorised as secondary (eg, healthcare providers). For example, in a study in which the main objective was to evaluate change in patient knowledge after using the digital tool and its secondary aim was to measure provider satisfaction with the tool, patients were considered the primary population and providers were considered the secondary population.

The data extraction form was built in Covidence and was piloted prior to data extraction (online supplemental file S2).

## Data synthesis

To synthesise the data, we described the general characteristics of the studies and primary outcomes (eg, patient-reported outcomes), as well as any secondary outcomes and the characteristics of the digital tools identified in the studies.

We summarised the six primary patient-reported outcomes and whether the tool resulted in an outcome that was favourable, unfavourable or had no effect based on the results as reported by the included studies. We also conducted additional analyses to understand the relationship between the purpose of a given tool and the patient-reported outcomes measured.

## Quality assessment

A quality appraisal was conducted for all included studies. Two independent reviewers scored the studies using the QualSyst quality assessment tool.[35] Disagreements were resolved through discussion or by input from a third team member. The QualSyst quality assessment criteria were used for qualitative, quantitative,

mixed and multimethod studies (online supplemental file S3). Studies that used a mixture of quantitative and qualitative methods without mixing of the quantitative and qualitative data were categorised as multimethod. For the quantitative studies, 14 items (eg, assessing study objective, methodology, analysis, conclusions) were scored depending on the degree to which specific quality criteria were met ('criteria met'=2, 'criteria partially met'=1, 'criteria not met'=0). Items not applicable to a particular study methodology were marked 'n/a' and were excluded from the calculation of the summary score. A score was calculated for each article by summing the scores obtained across applicable items and dividing by the total possible score. Scores for the qualitative studies were calculated in a similar fashion based on the scoring of 10 items. Since assigning 'n/a' was not permitted for qualitative studies,[36] the total possible score for each qualitative study was 20. For multimethod studies, both the quantitative and qualitative criteria were completed, and the total possible score was calculated based on the number of items marked. For mixed methods studies, both the quantitative and qualitative criteria were completed and an additional five criteria from the Mixed Methods Appraisal Tool (MMAT) were used,[36] assessing the integration of the qualitative and quantitative methods and conclusions. The five MMAT criteria items were rated on the same 0–2 scale. Final scores for mixed methods studies were determined by adding the scores from the qualitative, quantitative and mixed methods appraisal tools divided by the total possible score, as done in previous reviews.[37]

### Patient and public involvement

A patient advisory board consisting of individuals with experience receiving genetic services was established. Specifically, the advisory board consisted of four adult patients with genetic conditions and two parents of children with genetic conditions. The members of the advisory board were identified and recruited through the research team's professional networks and prior genetics research projects. The advisory board was consulted on the scope of the systematic review and its main outcomes based on their experience with receiving genetic services. The advisory board subsequently provided feedback on the relevance of the review's findings on patients and the public.

**Figure 1: PRISMA Flow Diagram**

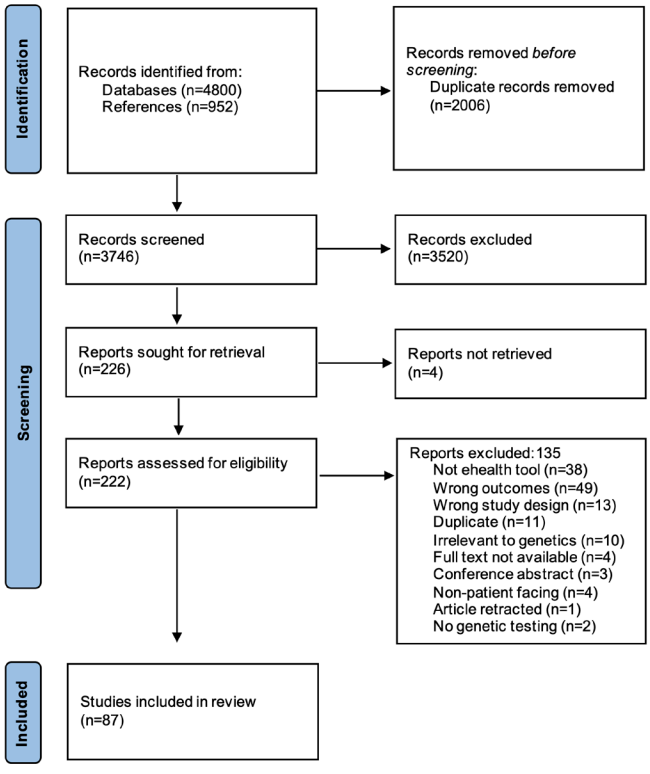


**Figure 1** PRISMA flow diagram. PRISMA, Preferred Reporting Items for Systematic Reviews and Meta-Analyses.

## RESULTS

The literature search identified 4800 records from databases plus an additional 952 articles from hand-searching of reference lists. Of these, 2006 were duplicates, leaving 3746 studies to be screened. Following title and abstract screening, 226 full-text articles were assessed for eligibility. Of these, 87 studies met the inclusion criteria and were included in this review (figure 1; online supplemental files 4 and 5).

### Characteristics of included studies

The characteristics of the included studies are summarised in tables 1 and 2. About half of the included studies were published between 2018 and March 2021 (n=44, 50.6%), with the remaining published between 2014 and 2017 (n=26, 29.9%) and between 2010 and 2013 (n=17, 19.5%). The majority of the 87 studies were conducted in the USA (n=58, 66.7%). Sixteen studies were conducted in Europe (18.4%), six in Canada (6.9%), four in the UK (4.6%), one in Australia

**Table 1** Study characteristics

| N=87 | n | % |
|---|---|---|
| Years | | |
| 2018–2021 | 44 | 50.6 |
| 2014–2017 | 26 | 29.9 |
| 2010–2013 | 17 | 19.5 |
| Country of data collection | | |
| USA | 58 | 66.7 |
| Europe (excluding UK) | 16 | 18.4 |
| Canada | 6 | 6.9 |
| UK | 4 | 4.6 |
| Australia | 1 | 1.1 |
| Other | 2 | 2.3 |
| Study type | | |
| Quantitative | 63 | 72.4 |
| Randomised controlled trials | 34 | 54.0 |
| Observational | 29 | 46.0 |
| Qualitative | 9 | 10.3 |
| Mixed method | 9 | 10.3 |
| Multimethod* | 6 | 6.9 |
| Study aim | n=144† | % out of 87 studies |
| Evaluation | 71 | 81.6 |
| Usability | 24 | 27.6 |
| Development | 16 | 18.4 |
| Feasibility | 11 | 12.6 |
| Implementation | 11 | 12.6 |
| Acceptability | 4 | 4.6 |
| Validation | 2 | 2.3 |
| Other | 5 | 5.7 |

*Studies that used a mixture of quantitative and qualitative methods without mixing of the quantitative and qualitative data were categorised as multimethod.
†41 of 87 studies reported two or more aims.

**Table 2** Tool characteristics

| | Total (n) | % out of 87 studies |
|---|---|---|
| Tool administration | N=87 | |
| Web-based | 67 | 77.0 |
| Mobile application | 7 | 8.0 |
| Non-web-based: computerised/CD | 8 | 9.2 |
| Other | 3 | 3.4 |
| Unreported/unclear | 2 | 2.3 |
| Intended user | n=91* | |
| Adult patient | 46 | 52.9 |
| General population | 22 | 25.3 |
| Prospective parent | 16 | 18.4 |
| Caregiver of a minor patient | 5 | 5.7 |
| Healthcare professional | 2 | 2.3 |
| Intended clinical setting | n=79† | % out of 70 tools |
| Oncology | 34 | 48.6 |
| Adult (non-oncology) | 15 | 21.4 |
| Reproductive | 17 | 24.3 |
| Paediatric | 6 | 8.6 |
| Primary care | 3 | 4.3 |
| All settings | 2 | 2.9 |
| Direct-to-consumer testing | 1 | 1.4 |
| Pharmacogenomics | 1 | 1.4 |
| Purpose of tool | n=153‡ | |
| Education | 59 | 84.3 |
| Decision-making | 32 | 45.7 |
| Psychosocial/needs or value assessment | 20 | 28.6 |
| Clinical assessment | 17 | 24.3 |
| Return of results | 12 | 17.1 |
| Post-test counselling and management | 11 | 15.7 |
| Consent | 1 | 1.4 |
| Sequencing, analysis and interpretation | 1 | 1.4 |

*Four of 87 studies reported two types of intended users.
†Nine of 70 tools reported two or more intended clinical settings.
‡Fifty-one of 70 tools had two or more purposes.

(1.1%), one in Singapore (1.1%) and one study across the USA and Canada (1.1%).

Of the 87 studies, 63 (72.4%) were quantitative, 9 were qualitative, 9 were mixed methods and 6 were multimethod. Of the 63 quantitative studies, 34 (54.0%) were randomised controlled trials.

The primary aim of over half of the included studies was evaluation (n=52, 59.8%), while the remaining studies focused on development (n=12, 13.8%), usability (n=12, 13.8%), feasibility (n=8, 9.2%) and implementation (n=3, 3.4%) of digital tools.

## Characteristics of included digital tools

A total of 70 unique digital tools were identified across 87 studies reviewed (table 2). The intended users of the tools were adult patients (n=46), general population (n=22), prospective parents (n=16), caregivers of a minor patient (n=5) and HCPs (n=2). The tools were intended for the following clinical settings: oncology (n=34), adult (non-oncology) (n=15), reproductive (n=17), paediatric (n=6), primary care (n=3), direct-to-consumer testing (n=1), pharmacogenomics (n=2) and all settings (n=2). Of note, some tools had multiple intended users and clinical settings.

The tools were targeted for use at various points in the patient trajectory, including clinical assessment and family history collection (n=17), education (n=59), psychosocial/needs or values assessment (n=20), decision-making about pursuing genetic testing and/or types of results to receive (n=32), consent (n=1), sequencing, analysis and interpretation (n=1), return of results (n=12), and post-test counselling and management (n=11) (table 3).

In 68 studies, the digital tool provided information on one or more of the following genetic tests: gene panels (n=28), prenatal screening and diagnosis (n=11), genome-wide sequencing (n=14), chromosomal microarray (n=3), single gene test/targeted variant analysis (n=5), paired germline/tumour testing (n=3), direct-to-consumer testing (n=2), newborn screening (n=1), and preimplantation genetic diagnosis (n=1).

Most tools were administered through a web-based application (n=67). The remainder were a combination of mobile applications (n=7), non-web-based modalities (ie, computerised/CD) (n=8), a kiosk (n=1), film (n=1) or a direct link to a PowerPoint recording sent via email (n=1). The mode of tool administration was unreported or unclear in two studies. Most tools did not require the involvement of an HCP (n=65) and over two-thirds of the tools were tailored to the user, meaning that the information provided to the user changed based on the user's input (n=47). An example of such tools is a breast cancer risk assessment tool that generates cancer risk calculation based on the user's medical and family history.

In 28 studies, tools were available in other languages, including Dutch (n=10), Spanish (n=7), French (n=2), Swedish (n=2), Danish (n=1), Italian (n=1), American Sign Language (n=1) and multiple languages (n=4). The reading level of the tool was reported by 14 studies. The levels were sixth grade level (n=2), seventh grade (n=2), eighth grade (n=6), ninth grade (n=2) and tenth grade (n=2).

## Characteristics of study participants

Over half of the 87 studies included adult patient participants (n=46), followed by general population (n=22) and prospective parents (n=14). Six studies included caregivers of minor patients. Thirty studies included a secondary population, with HCPs as the secondary population in 60% of these studies.

Of the 87 studies, 83 reported at least one of ethnicity/ancestry, income or education level. However, only 30 studies reported all three variables. Forty-five studies reported more than two ethnic backgrounds for their study participants. Most studies (n=55) also reported more than two education levels. Only 25 studies reported more than two income levels of study participants. Of 87 studies, 71 did not report the general, health or digital literacy levels of the study participants. Of the 16 studies that reported literacy levels, the most common forms were digital (n=9) and health (n=9) literacy levels.

## Primary outcomes

### Patient-reported outcomes

Of the studies included in this review (N=87), 60 measured participant acceptability, 46 measured participant satisfaction and 23 measured participants' intention to recommend the tool to others, with all these studies reporting favourable outcomes.

Table 4 summarises the number of studies that measured each type of patient-reported outcome and whether the outcome was favourable, unfavourable or had no effect. The patient-reported outcomes that were most frequently measured by the included studies were participant knowledge/understanding,

**Table 3** Digital tools and their intended use in the patient trajectory

| Author name and year | Clinical setting | | Tool name | Clinical assessment | Education | Psychosocial/needs assessment | Decision-making | Consent | Sequencing, analysis and interpretation | Return of results | Post-test counselling/ management |
|---|---|---|---|---|---|---|---|---|---|---|---|
| Kaplan (2014) | Primary care | | BreastCARE (Breast Cancer Assessment of Risk and Education) | ✓ | | | | | | | |
| Rupert (2013) | | Oncology | Cancer in the Family | ✓ | ✓ | | ✓ | | | | |
| Wu (2013), Buchanan (2015), Wu (2019), Fung (2021) | | | MeTree | ✓ | ✓ | | ✓ | | | | |
| Bowen (2017) | | | Not reported | ✓ | ✓ | ✓ | | | | | |
| Wolfe (2015) | | | BRCA Gist (BReast CAncer Genetics Intelligent Semantic Tutoring) | | ✓ | | ✓ | | | | |
| Grimmett *et al* (2019)[22] | | | Breast Cancer Choices | | ✓ | | ✓ | | | | |
| Sherman (2017) | | | Breast RECONstruction Decision Aid (BRECONDA) | | ✓ | ✓ | ✓ | | | | |
| Boudreault (2018) | | | Cancer Genetics Education Module (CGEM) | | ✓ | | | | | | |
| Joseph (2010) | | | Cancer Risk Education Intervention Tool (CREdIT) | | ✓ | | | | | | |
| Pritzlaff (2014) | | | CancerGene Connect (CGC) | ✓ | | ✓ | | | | ✓ | ✓ |
| Cragun (2020) | | | Not reported | | ✓ | | | | | | |
| Culver (2011) | | | Not reported | | ✓ | | ✓ | | | | |
| Schackmann (2013) | | | Not reported | ✓ | | | ✓ | | | | ✓ |
| Sie (2016) | | | DNA Direct | | ✓ | | | | | | |
| Albada (2011, 2012a, 2012b, 2015) | | | E-info gene | | ✓ | ✓ | | | | | |
| Makhnoon (2021) | | | FamilyTalk | | ✓ | ✓ | | | | | ✓ |
| Gaba (2020) | | | Not reported | ✓ | ✓ | ✓ | | | | | |
| Solomon (2019) | | | Helping Oncology Patients Explore Genomics (HOPE-Genomics) | | ✓ | | | | | ✓ | |
| Hooker (2011) | | | Not reported | | ✓ | ✓ | ✓ | | | | |
| Gornick (2018) | | | iCanDecide | | ✓ | | ✓ | | | | |
| Eden (2020) | | | MammoScreen | ✓ | ✓ | | ✓ | | | | |
| Manchanda (2016) | | | Not reported | | ✓ | | | | | | |
| Manne (2010) | | | Not reported | | ✓ | ✓ | ✓ | | | | |
| McCuaig (2019) | | | Not reported | | ✓ | | | | | | |
| Vogel (2019) | | | mobile Application for Genetic Information on Cancer (mAGIC) | | ✓ | ✓ | ✓ | | | | |
| Hall (2014) | | | MyFoxChase ePHR | | ✓ | | | | | | |
| van Erkelens (2017, 2018) | | | Online self-test | ✓ | | | | | | | |
| Luba (2018) | | | PREMM1,2,6 model | ✓ | ✓ | | ✓ | | | | |
| Kukafka (2015) | | | RealRisks | ✓ | ✓ | | ✓ | | | | |
| Sussner (2010) | | | Not reported | | ✓ | | | | | | |
| Stefansdottir (2020) | | | www.arfgerd.is | | ✓ | | | | | ✓ | ✓ |

Continued

**Table 3** Continued

| Author name and year | Clinical setting | | Tool name | Clinical assessment | Education | Psychosocial/needs assessment | Decision-making | Consent | Sequencing, analysis and interpretation | Return of results | Post-test counselling/ management |
|---|---|---|---|---|---|---|---|---|---|---|---|
| Sturm (2018) | Adult (non-oncology) | | Not reported* | | ✓ | | | | | ✓ | ✓ |
| Bombard (2018),[15] Shickh (2020), Shickh *et al* (2021)[8] | | | Genomics ADvISER | | ✓ | ✓ | ✓ | | | | |
| Jujjavarapu (2021) | | | ShareDNA | | ✓ | | | | | | ✓ |
| Cohn (2010) | | | Health Heritage | ✓ | | | | | | | |
| Biesecker (2018) | | | Not reported | | ✓ | | ✓ | | | ✓ | |
| Ekstract (2017) | | | Not reported | | ✓ | ✓ | ✓ | | | | |
| Schmidlen (2019) | | | Genetic Information Assistant (Gia) | | | | | ✓ | | | ✓ |
| Harris (2019) | | | Not reported | | ✓ | | | | | | ✓ |
| Hendershot (2010) | | | Not reported | ✓ | | ✓ | | | | ✓ | |
| Mills (2015) | | | Not reported | | ✓ | | | | | ✓ | |
| Kaphingst (2010) | | | Multiplex Initiative Web module | | ✓ | | ✓ | | | | |
| Arar (2011) | | | Surgeon General's family health history tool (SG-FHH) | ✓ | | | | | | | |
| Ponathil (2020) | | | Virtual Conversational Agent (VCA) | ✓ | | | | | | | |
| Truong (2020) | | | YourPGx Portal | | ✓ | | | | | ✓ | |
| Gallo (2014, 2016), Hershberger (2016) | | Reproductive | CHOICES | | ✓ | | ✓ | | | | |
| Fan (2018) | | | Not reported | ✓ | ✓ | | | | | | ✓ |
| Woodard (2018) | | | Pathways | | ✓ | ✓ | ✓ | | | | |
| Dugger (2020) | | | An Introduction to Carrier Screening | | ✓ | | ✓ | | | | |
| Yee (2014) | | | ArchieMD Interactive Informed Consent and Education Program for Pregnant Women | | ✓ | | | | | | |
| Beulen (2016) | | | Not reported | | ✓ | ✓ | ✓ | | | | |
| Björklund (2012) | | | Not reported | | ✓ | | ✓ | | | | |
| Carlson (2019) | | | Not reported | | ✓ | | ✓ | | | | |
| Castellani (2011) | | | Not reported | | ✓ | | | | | | |
| Arjunan (2020) | | | Counsyl Complete | | ✓ | | | | | ✓ | ✓ |
| Kalejta (2019) | | | Genetics Maven | | ✓ | | | | | ✓ | |
| Ahman (2016) | | | Not reported | | ✓ | ✓ | ✓ | | | | |
| Hardy (2018) | | | JScreen | | ✓ | | | | | | |
| Kuppermann (2014) | | | Prenatal Testing: Exploring Your Options | | ✓ | | ✓ | | | | |
| Skjøth (2015) | | | Not reported | | ✓ | | ✓ | | | | |

Continued

**Table 3** Continued

| Author name and year | Clinical setting | Tool name | Clinical assessment | Education | Psychosocial/needs assessment | Decision-making | Consent | Sequencing, analysis and interpretation | Return of results | Post-test counselling/ management |
|---|---|---|---|---|---|---|---|---|---|---|
| Paquin (2018), Peinado (2020) | Paediatric | NC NEXUS Decision Aid | | ✓ | ✓ | ✓ | | | | |
| Birch *et al* (2016),[11] Adam *et al* (2018)[10] | | DECIDE (Decision-aid and E-Counselling for Inherited Disorder Evaluation) | | ✓ | ✓ | ✓ | | | | |
| Goehringer (2018), Williams (2018) | | GenomeCOMPASS Report | | ✓ | ✓ | ✓ | | | ✓ | |
| Lewis (2020) | | My Genome Sequence | | ✓ | | | | | ✓ | ✓ |
| Temme (2015) | | Understanding Your Child's Newborn Screen for Cystic Fibrosis | | ✓ | ✓ | | | | | |
| Xu (2018) | | Not reported | | ✓ | | ✓ | | | | |
| Wang (2015) | All settings | Virtual Counselor for Knowing your Family History (VICKY) | ✓ | | | | | | | |
| Sanderson (2016), Sabatello (2019) | All settings | Whole Genome Sequencing and You | | ✓ | | | | | | |
| Reumkens (2019a, 2019b, 2021) | Oncology, reproductive | Not reported | | ✓ | ✓ | ✓ | | | | |
| Nelson (2019) | Direct to consumer | Third-party interpretation tools | | | | | | ✓ | | |

*This tool is reported to be also intended for pharmacogenomics setting.

patient engagement, psychosocial well-being and facilitation of decision-making. The majority of the studies reported that the use of the digital tool was associated with favourable outcomes.

While behavioural/management changes and family-related communication were assessed in a smaller set of studies, most reported that the tool was associated with favourable outcomes for these domains. The impact of the tool on quality of life was assessed in only three studies, with one study reporting a favourable impact on quality of life and two studies reporting no effect.

### Misalignment between purpose of the tool and measured outcomes

The stated purpose of the tool did not always align with the patient-reported outcomes measured. For example, of the 75 studies indicating education as a purpose of the tool, only 56 studies measured change in patient knowledge/understanding. Furthermore, two additional studies evaluated knowledge and understanding but did not state education as a purpose of the tool. Similarly, of the 44 studies indicating decision-making as a purpose, only 27 measured facilitation of decision-making as an outcome. Additionally, there were six studies that assessed whether their tool facilitated patients' decision-making, without indicating decision-making as a purpose of the tool.

## Secondary outcomes

### Provider-reported outcomes

Eleven studies measured providers' satisfaction, all of which reported that providers were satisfied with the digital tool. Eleven studies assessed providers' willingness to integrate the digital tool in their workflow. All studies reported favourable responses on digital tool integration into practice by providers after using the tool.

### System-focused outcomes

Ten studies evaluated the effect of the digital tool on time spent with the HCP, with the majority (n=7, 70%) indicating a reduction in time spent with their provider and three (30%) indicating no effect. Seven studies measured the impact of the tool on workflow efficiency (eg, chart preparation time is reduced), with the majority (n=6, 86%) reporting improvements in efficiency and only one study reporting no effect. None of the studies included in this review reported on the impact of the tool on wait times.

## Quality appraisal

Most studies (n=75) had a quality score of above 75%, which is a threshold often used for assessing quality in systematic reviews.[35 37] The highest average quality scores were in qualitative studies (88.9%, range=65.0–100), followed closely by quantitative studies (86.6%, range=28.6–100). Mixed and multimethod studies scored lower, with mixed methods studies having an average score of 78.3% (range=64.0–94.0) and multimethod studies scoring an average of 72.6% (range=60.5–82.5). See online supplemental file S3 for a breakdown of the average quality scores by study type.

## DISCUSSION

To our knowledge, this is the most up-to-date and comprehensive systematic review on digital tools used in genetic service delivery and their impact on patient care and system efficiencies. This review found 70 unique digital tools reported in 87 studies. Most of these tools were intended for use by adult patients or the general population, half were in oncology settings and most were intended to facilitate education and decision-making in the pretest phase. Most studies found that digital tools lead

**Table 4** Summary of patient-reported outcomes

| | Knowledge/ understanding | Decision-making | Patient engagement | Psychosocial well-being | Behaviour and management changes | Family communication |
|---|---|---|---|---|---|---|
| Number of studies that measured the outcome | 58 | 33 | 42 | 35 | 24 | 19 |
| Favourable | 55 | 28 | 40 | 23 | 18 | 17 |
| No effect | 3 | 5 | 2 | 11 | 6 | 2 |
| Unfavourable | 0 | 0 | 0 | 1 | 0 | 0 |

to favourable patient outcomes, including improved knowledge, engagement, psychosocial well-being and facilitation of decision-making. In addition, most studies provided evidence that digital tools can be used to support the pretest components of the counselling and testing trajectory. Studies also found that digital tools improved workflow and efficiency for providers, in addition to reducing the amount of time they needed to spend with patients. These findings are consistent with the results of other recent studies that suggest widespread interest in virtual care across various specialties and patient populations, with positive outcomes reported for patients and providers.[38–45] Given the increased demand for genetic services and the shift towards virtual care as the new norm during the COVID-19 pandemic and beyond, this review provides timely evidence that digital tools can be used to efficiently deliver tailored and patient-centred virtual care.

Despite the improved care and workflow efficiencies offered by digital tools, this systematic review found three major gaps in the development, evaluation and implementation of digital tools for genetic service delivery. First, none of the tools included in this review was developed to encompass the entire genetic counselling and testing trajectory. Furthermore, most of the tools were developed for use in the pretest counselling phase (eg, education, decision-making), with a limited number of tools designed to support the post-test phase of patient care (eg, return of results, family communication). These gaps limit the understanding of the utility of these tools in the post-test phase, a time point when many patients require extensive counselling and support. As such, future studies should focus on the development, evaluation and implementation of digital tools that encompass not only the pretest phase but also the post-test phase of the patient's counselling and testing trajectory.

Second, this review found that while many tools had more than one intended clinical setting, half of these studies took place in the oncology setting. Genetic testing is becoming mainstream outside of oncology across various specialties, including neurology, cardiology and nephrology, where there is often a lack of genetic counselling support.[46 47] Patients and their caregivers in these specialties and those being tested outside of the traditional clinical genetics settings may have distinct needs from patients in oncology but their needs have been inadequately addressed by existing digital tools. Digital tools have the potential to play a critical role in streamlining the delivery of genetic testing outside of oncology and traditional clinical genetics settings. The development and implementation of digital tools for genetic service delivery in resource-constrained clinical settings may warrant prioritisation.

Finally, a lack of alignment was identified between the purposes of the tool and its measured outcomes. This was especially apparent in studies where the tools were designed for education or decision-making, such that some of these studies did not assess the impact of the tool on the outcomes they intended to achieve. Furthermore, there were studies that evaluated outcomes that did not match with their tool's intended purpose. Therefore, although digital tools generally improved knowledge and facilitated decision-making, the misalignment of the tool's purposes and the study outcomes among this subset of studies makes it challenging to determine whether these tools achieved their intended aims and whether the improved outcomes can be attributed to the tool. This is especially critical to highlight given that 71 of the 87 studies reported that evaluation of the digital tool was one of the purposes of their study. Future research should ensure alignment between the tool's purpose and the study outcomes to enable assessment of whether observed outcomes can be attributed to the intervention studied.

This review also revealed a significant lack of consideration for general, health and digital literacy in the included studies, with 82% of studies not providing any information on participants' literacy levels. This has important implications for the validity of the outcomes reported in the included studies. It is difficult to conclude that digital tools used within the genetic service pathway result in positive outcomes for patients of varying levels of literacy or if their effectiveness is limited to users with high literacy levels. A systematic review of health literacy in the context of eHealth services found that poor readability of content and poor usability of eHealth services can lead to limited access to and use of online health information by users.[48] As such, target users' health and digital literacy levels should be taken into account in the design and development of digital tools. Also, future studies evaluating the effectiveness of digital tools in genetic services should measure participants' literacy levels and examine their association with digital tool effectiveness.

## Limitations

This study has several limitations. First, given the variability in the measures of the primary outcomes, we did not conduct a meta-analysis. For both primary and secondary outcomes, the results were categorised (eg, decreased, increased, no effect) but not quantified, and as such effect sizes of these outcomes are not captured. Furthermore, the secondary outcomes (system and provider level) of this review were not reported across all the included studies; therefore, conclusions around these outcomes are less robust. Future studies should examine the impact of digital tools on system-level and provider-level outcomes. Additionally, this review only included English-language articles and articles published in and after January 2010, limiting the evidence available for the review. While the number of levels of DEI variables reported was extracted, the frequency of patient characteristics related to DEI variables (eg, the percentage of non-white participants) was not. As such, the study cannot further elaborate on the diversity of the participants included in the studies on digital tools within genomics. Lastly, the measurement tools used to assess the outcomes were not extracted, limiting the study in defining what measures represented each outcome.

## CONCLUSION

This systematic review provides a comprehensive overview of digital tools used in genetic service delivery and their impact on patient care and system efficiencies. Digital tools generally improved patient-reported outcomes related to knowledge and decision-making. However, most tools were focused on the pretest phase and none covered the complete genetic testing trajectory. Also, most studies were focused on the use of digital tools in oncology settings. Finally, within a subset of studies, there was misalignment between the tool's intended purposes and the outcomes measured. The findings of this study suggest that future research should prioritise development, implementation and robust evaluation of digital tools that can support the entire patient trajectory across diverse patient populations in a range of clinical settings.

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