

Telegenetics: a systematic review of telemedicine in genetics services

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Purpose: Telemedicine is being increasingly used in many areas of health care, particularly to reduce the barriers that rural populations face in accessing health-care services. Telemedicine may also be effectively utilized in clinical genetics services—an application that has been termed “telegenetics.”

Methods: A systematic review of the literature was conducted to identify studies of genetic consultations carried out through video-conferencing so as to determine whether conclusions can be drawn about the value of telegenetics. A total of 14 articles reporting data from 12 separate studies met the inclusion criteria.

Results: In a majority of these studies, patients received their telegenetics consultation at a local clinic or outreach center, from where they communicated via a synchronous video link with a genetics practitioner. All the studies reported high levels of patient satisfaction

with telegenetics, and patients were generally more receptive to telegenetics than the genetics practitioners were. The studies had limitations of small sample sizes and lack of statistical analyses.

Conclusions: This review suggests that telegenetics may be a useful tool for providing routine counseling and has the potential to evaluate pediatric patients with suspected genetic conditions. Prospective, fully powered studies of telegenetics that explore the accuracy of diagnoses and patient outcomes are needed to allow informed decisions to be made about the appropriate use of telemedicine in genetics service delivery.

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Key Words: clinical genetics; genetic counseling; systematic review; telegenetics; telemedicine

Telemedicine is the use of electronic and communication technologies for medical diagnostic, monitoring, and therapeutic purposes when distance and/or time separate the participants.¹ The term telemedicine often refers to a real-time interaction between a patient and a health professional through video-conferencing rather than face to face. By enabling contact with health service providers in distant locations, telemedicine can benefit rural populations by reducing geographical barriers to accessing specialist health services.^{2–5} Other benefits of telemedicine include improved efficiency and reduced costs of health services. The large number of articles and reviews published in recent years about telemedicine demonstrates the increasing use of, and research into, telemedicine services.

In a review of reviews, Ekeland *et al.*⁶ identified 80 systematic reviews published between 2005 and 2009 on the impact and cost of telemedicine in various areas of health care, including psychiatry, dermatology, and diabetes. Ekeland *et al.* concluded that the results of these reviews were diverse; some suggested that telemedicine has positive therapeutic effects and increases efficiency within health services,^{7,8} while other reviews concluded that the evidence for the benefits of telemedicine was inconsistent and limited.^{9,10} The data relating to the cost-effectiveness of telemedicine are also mixed; a recent review found that synchronous video communication was cost effective for home

care and access to on-call hospital specialists but not for the local delivery of services between hospitals and primary care.¹¹ Despite the lack of consensus within the literature regarding the effectiveness of telemedicine in improving the quality of, or access to health care, its use remains widespread.¹²

Telemedicine may be particularly effective in medical specialties in which verbal interactions are a key part of the assessment process, such as in psychiatry and neurology. Research in these areas demonstrates that care via telemedicine produces outcomes comparable to those of face-to-face consultations.^{13–17} Genetics services are often based on counseling and may therefore also be a specialized field of health care in which telemedicine can be effectively utilized as an alternative to standard face-to-face interactions between health professionals and patients.

Referrals to genetics services are generally suggested if a patient is suspected of being at risk of, or affected by, a genetic disorder. For example, genetics specialists can diagnose a patient with symptoms of a disease or carry out a disease risk assessment or a reproductive risk assessment relating to a possible carrier status or pregnancy. This often involves several visits to a genetics clinic to undergo genetic counseling and physical evaluation, receive genetic test results, and attend follow-up appointments. In Europe and the United States, access to genetics services is often available only in urban centers,^{18,19}

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which means that remote populations may not have access to the information, treatment, and psychosocial support that genetics services can offer. The potential for using telemedicine in clinical genetics service delivery has been highlighted by a number of authors,^{19–21} who have suggested that it may enable more equitable access to genetics services for people who must otherwise travel long distances to specialist centers. Indeed, an increasing number of studies are exploring alternative models of service provision, including genetic counseling over the telephone,^{22,23} and genetics consultations via real-time videoconferencing,²⁴ so as to improve equality of access, improve cost efficiency, and help to meet the increasing demand for these services.

Our systematic review aims to synthesize the evidence available to date about genetic consultations carried out via videoconferencing, a process that has been termed “telegenetics”.^{24,25} Our aim was to determine whether conclusions can be drawn about the value of telegenetics, and to provide recommendations for further research and its adoption in clinical practice.

METHODS

Search strategy

The literature search focused on English-language articles pertaining to the use of telemedicine in clinical genetics services (see [Table 1](#) for search key words). The following electronic databases were searched: MEDLINE, EMBASE, PsychINFO, CINAHL, British Nursing Index, Cochrane Library, and the Web of Science. Each database search was conducted during the week of 28 November 2011 to identify articles published from January 1996 up to November 2011. The Science Citation Index was searched to find articles that cited the studies included in the database search results. The reference lists of all relevant articles and other reviews were used to identify additional studies.

Eligibility criteria

Studies were included in this review if they were published in a peer-reviewed journal in English and were aimed at evaluating the effectiveness, cost, or feasibility of synchronous telemedicine consultations in any area of clinical genetics. Studies that did not include genetics consultations were excluded. Randomized controlled trials and nonrandomized pilot studies were considered for inclusion. Both quantitative and qualitative studies were eligible, but reviews, letters with no primary data, case studies, and unpublished studies were excluded.

The outcomes of interest were clinical efficacy, knowledge, perceived risk, affective outcomes (e.g., distress, anxiety, and depression), behavioral outcomes (e.g., surveillance/surgery uptake and genetic testing uptake), satisfaction, and cost of the service. All titles and abstracts were screened in accordance with the inclusion criteria. Full-text articles were obtained whenever more information was required to make a decision about inclusion. Two reviewers independently reviewed the full-text studies to determine whether they met the inclusion

criteria. Any disagreement about a particular study was resolved through discussion and/or referred to a third person.

Information extraction

Two review authors independently extracted data from each of the studies included in the review. The data extracted included study design, method and setting; participant characteristics; and relevant outcomes. Any differences in opinion were resolved through discussion. Reviewers were not blinded as to authorship, journal, or institution.

Quality assessment

A formal quality assessment was not performed because a majority of the studies were nonrandomized studies with small sample sizes, whereas all well validated quality assessment tools are intended for use in the context of randomized controlled trials.

RESULTS

Study characteristics

A total of 14 articles reporting data from 12 separate studies met all the study selection criteria. Details of the studies included, published between the years 2000 and 2011, are provided in [Table 2](#). The studies were conducted in the United States, Canada, the UK, and Australia. All the studies utilized real-time videoconferencing via multiple high-speed ISDN lines (between 128 and 384 kbps) to provide clinical genetics services to remote or outreach areas. ISDN lines are considered to be effective in transmitting high-resolution images and are reasonably fast and secure. In a majority of the studies, patients received their telegenetics consultations at a local clinic or outreach center. Exceptions to this were Meropol *et al.*,²⁶ a study in which the participants were counseled in their own homes, and Gattas *et al.*,²⁷ in which both the telegenetics group and the face-to-face group had their consultations in the same hospital in Brisbane, Australia, so as to remove the advantages of travel time and cost.

The studies included in our review related to the use of telegenetics in counseling for hereditary cancer,^{26,28–33} prenatal counseling,³⁴ pediatric services,^{35,36} and services for a range of genetic disorders.^{24,27,37} Of the 125 patients who were evaluated and/or counseled via telemedicine in the study by Lea *et al.*,³⁷ 64% received pediatric and/or neurological genetics consultations and the rest received consultations for cancer or reproductive genetics counseling. Pediatric telegenetics services often included physical examinations, whereas consultations for cancer generally only involved a review of the patients' family history, the provision of information, and psychosocial support. The type of genetics practitioner involved in the telemedicine consultations varied between studies. In several studies, patients had a genetics counselor or nurse present with them at the outreach clinic while they communicated via a synchronous video link with a geneticist or a genetics counselor.^{24,27,30–33,38} In a study by Stalker *et al.*,³⁶ a genetics counselor obtained information on patients' family and medical history via teleconferencing, and a local pediatrician conducted a physical examination. The

Table 1 Review search strategy

MEDLINE (OVID)	EMBASE (OVID)	PsychINFO (OVID)	CINAHL
1. Telemedicine/ 2. Remote consultation/ 3. Videoconferencing/ 4. Internet/ 5. Telecommunications/ 6. (Telehealth\$ or tele-health\$ or telemed\$ or tele-med\$ or telecommunication\$ or telemanag\$ or tele-manag\$ or telecare or tele-care or telesupport\$ or tele-support\$ or telemonitor\$ or tele-monitor\$ or mobile health\$ or ehealth\$ or e-health\$ or mhealth or m-health or mobile device\$).tw. 7. Computer mediated.tw. 8. ((Tele\$ or remote or video\$) adj2 (care\$ or consult\$ or clinic\$ or counsel\$)).tw. 9. Or/1-8 10. Genetics/ 11. Genetics medical/ 12. Genetic counseling/ 13. Exp genetic services/ 14. Genetic predisposition to disease/ 15. (Genetic\$ adj3 (service\$ or risk\$ or counsel\$ or clinic\$ or test\$ or screen\$)).tw. 16. Or/10-15 17. 9 and 16 18. (Telegenetic\$ or tele-genetic\$).tw. 19. 17 or 18 20. Limit 19 to English language	1. Exp telemedicine/ 2. Exp teleconsultation/ 3. Exp videoconferencing/ 4. Internet/ 5. Exp telecommunication/ 6. (Telehealth\$ or tele-health\$ or telemed\$ or tele-med\$ or telecommunication\$ or telemanag\$ or tele-manag\$ or telecare or tele-care or telesupport\$ or tele-support\$ or telemonitor\$ or tele-monitor\$ or mobile health\$ or ehealth\$ or e-health\$ or mhealth or m-health or mobile device\$).tw. 7. Computer mediated.tw. 8. ((Tele\$ or remote or video\$) adj2 (care\$ or consult\$ or clinic\$ or counsel\$)).tw. 9. Or/1-8 10. Exp medical genetics/ 11. Genetic counseling/ 12. Exp genetic service/ 13. Exp genetic predisposition/ 14. (Genetic\$ adj3 (service\$ or risk\$ or counsel\$ or clinic\$ or test\$ or screen\$)).tw. 15. Or/10-14 16. 9 and 15 17. (Telegenetic\$ or tele-genetic\$).tw. 18. 16 or 17 19. Limit 18 to (human and English language)	1. Telemedicine/ 2. Internet/ 3. Telecommunications/ 4. (Telehealth\$ or tele-health\$ or telemed\$ or tele-med\$ or telecommunication\$ or telemanag\$ or tele-manag\$ or telecare or tele-care or telesupport\$ or tele-support\$ or telemonitor\$ or tele-monitor\$ or mobile health\$ or ehealth\$ or e-health\$ or mhealth or m-health or mobile device\$).tw. 5. Computer mediated.tw. 6. ((Tele\$ or remote or video\$) adj2 (care\$ or consult\$ or clinic\$ or counsel\$)).tw. 7. Genetics/ 8. Genetic counseling/ 9. (Genetic\$ adj3 (service\$ or risk\$ or counsel\$ or clinic\$ or test\$ or screen\$)).tw. 10. (Telegenetic\$ or tele-genetic\$).tw. 11. 1 or 2 or 3 or 4 or 5 or 6 12. 7 or 8 or 9 13. 11 and 12 14. 10 or 13	1. (MH "Telemedicine") 2. (MH "Telehealth+") 3.(MH "Telenursing") 4. (MH "Remote Consultation") 5. (MH "Videoconferencing") 6. TX telehealth* or tele-health* or telemed* or tele-med* or telecommunication* or telemanag* or tele-manag* or telecare or tele-care or telesupport* or tele-support* or telemonitor* or tele-monitor* or mobile health* or ehealth* or e-health* or mhealth or m-health or mobile device* 7. Telegenetic* or tele-genetic 8. 1 or 2 or 3 or 4 or 5 or 6 9. (MH "Genetics, Medical+") 10. (MH "Genetic Counseling") 11. (Genetic*) NE (service* or risk* or counsel* or clinic* or test* or screen*) 12. 9 or 10 or 11 13. 8 and 12 14. 13

patients then returned to the clinic for a face-to-face consultation with the clinical geneticist. Three of the studies explored genetics counseling via teleconference with a genetics counselor only; these comprised one study involving prenatal counseling³⁴ and two studies involving hereditary cancer counseling.^{26,29} Coelho *et al.*,²⁸ described a study of genetics counseling for cancer via teleconference with a genetics consultant.

Only one of the studies in our review involved randomization of participants to receive either a telegenetics consultation or a face-to-face consultation, with both groups attending the same hospital for their appointments.²⁷ Patients in the telegenetics group communicated via a video link with the consultant, who was located only 10 meters away. The majority of the studies used a retrospective or cross-sectional design, with evaluations and assessments occurring after the telegenetics consultation. Three studies were prospective studies with assessments both before and after telegenetics counseling.^{24,28,33} Two studies involved conducting qualitative interviews, one with patients³² and the other with genetics professionals,^{30,31} to explore their experiences of telegenetics. The other studies involved the gathering of data through surveys and/or telephone interviews, and

many did not involve any statistical analyses on outcomes measures because of small sample sizes. Only one study indicated that validated measurement tools with acceptable reliability had been used to assess outcomes.³³ That study was also the only one to mention that power calculations were conducted to ensure that any significant between-group differences were detected. Eight of the studies did not have a comparison group, and reported data solely on those who received genetics services via a telemedicine.^{24,26,29–32,35–38} All the studies measured patient satisfaction with the telegenetics consultation; in some cases, the health professionals' satisfaction was also assessed. The methods of measuring satisfaction varied between studies, with many researchers developing their own scales of satisfaction, whereas others based their scales on previous research. Three studies reported on affective outcomes such as cancer-specific distress, general anxiety, and depression.^{24,28,33} There were no reports on clinical or behavioral outcomes such as uptake of genetics testing or cancer screening after telegenetics counseling for hereditary cancer. However, Lea *et al.*,³⁷ Hopper *et al.*,³⁵ and Stalker *et al.*³⁶ reported on the clinical efficacy of diagnosing pediatric genetic disorders through the use of telegenetics.

Table 2 Summary of studies included in the review

Author and location	Reason(s) for genetics consultation and service(s) provided	Study design and methods	Outcome measures	Key findings	Conclusions
Abrams and Geier ³⁴ USA	Prenatal genetics counseling with genetics counselor via telemedicine Videoconferencing between the patient in an obstetrician's office in a satellite clinic and the genetics counselor at the clinical genetics clinic. All counseling sessions included discussion of testing options, acquisition of family history, and informed consent.	Quantitative cross-sectional survey completed post-counseling. Comparison between prenatal genetics counseling via telemedicine (n = 7) and face-to-face counseling (n = 14).	Postcounseling measures: -Satisfaction with the counseling process -Patient's experience of videoconferencing.	For both groups, satisfaction with counseling scores averaged between 4 and 5 (5 = highest satisfaction possible), including helpfulness of genetics counseling and satisfaction with amount of time given. Both groups felt session was confidential; 100% of telegenetics group gave positive responses about counseling process and a majority of in-person counseling responses were positive. All telegenetics patients reported that it was an advantage, mainly because of reduced travel time. One telegenetics patient would have preferred an in-person consultation in the clinic.	Patient satisfaction with prenatal genetic counseling was similar among those receiving the counseling via videoconferencing and those receiving it face-to-face in a clinic. Patients counseled via telegenetics reported positive experiences with using the technology, and stated that it reduced travel time. Confidentiality was not a concern for those receiving telegenetics counseling.
Coelho <i>et al.</i> ²⁸ UK	Family history of breast/ovarian or colon cancer Patients received genetics counseling for cancer with a genetics consultant. (Contents of counseling sessions are not described.)	Prospective study comparing telegenetics counseling (n = 16) and face-to-face counseling (n = 21) for cancer. Quantitative pre- and postcounseling questionnaire, including true/false questions, Likert scale questions, and open-ended questions for free-text responses. Participants assigned to either telegenetics group or face-to-face group depending on their geographical location.	-Knowledge of cancer genetics -Cancer risk-related anxiety -Satisfaction.	Increase in knowledge of genetics of cancer in both telegenetics and face-to-face groups. Statistically significant increase in knowledge in the combined (telemedicine plus face-to-face) group (P = 0.02). Significant decrease in anxiety after counseling in telegenetics group (P = 0.00), face-to-face group (P = 0.01), and combined group (P = 0.00). Total satisfaction score significantly higher in telegenetics group (mean = 23.12) than in face-to-face group (mean = 22; P = 0.08). Face-to-face group did not feel as satisfied as telegenetics group in regard to their emotional needs having been met (P = 0.02).	No significant difference in the quality of genetics counseling for cancer via telegenetics as compared to face-to-face. In both groups, knowledge of the genetics of cancer improved, and anxiety levels decreased. Overall, the patients were satisfied with telegenetics, which suggests that it is a useful alternative to face-to-face counseling when geographical distance is an issue.

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Table 2 Continued.

Author and location	Reason(s) for genetics consultation and service(s) provided	Study design and methods	Outcome measures	Key findings	Conclusions
d'Agincourt-Canning <i>et al.</i> ²⁹ Canada	Family history of breast/ovarian or colorectal cancer, or melanoma. Telegenetics counseling between Hereditary Cancer Program in Vancouver/Victoria and hospitals in six rural communities. Counseling provided by four genetics counselors. Counseling involved review of family history, discussion of cancer risk, risk management strategies, and psychosocial support.	Pilot study to assess the effectiveness and acceptability of cancer genetics counseling with a genetic counselor present via telemedicine (<i>n</i> = 48). Cross-sectional quantitative survey completed postcounseling, including Likert scale questions and open-ended questions.	Post-counseling measures: -General satisfaction, comfort level, comprehension of information, quality of service, time- and cost-savings for patient. -Genetics counselor's satisfaction with telemedicine.	High levels of satisfaction with the telegenetics counseling (average score 4.68 of 5). No participant reported that the telegenetics format made it difficult to communicate or understand the information. Cost savings averaged \$1,000 per person. Patients reported high levels of convenience with telemedicine and preferred consultations to take place in their local communities. Technical difficulties encountered were minor. Genetics counselors were less satisfied with telemedicine than patients (average score 3.97 out of 5) and reported difficulties in establishing rapport.	Telemedicine is an effective means of providing genetics counseling for cancer to rural and underserved populations, for whom economic or personal situations would have been a barrier to access. Telegenetics services were convenient, and resulted in time- and cost-savings to patients. A support person may be needed to assist patients at the remote site and act as a liaison between sites.
Gattas <i>et al.</i> ²⁷ Australia	Most of the patients were referred for counseling because of a family history of cancer. Pre-clinic telephone contact made by genetics counselor to gather family history information. Genetics counselor was in the room with the patient during the telegenetics session with the genetics consultant.	Randomized trial comparing telegenetics consultation (<i>n</i> = 16) and face-to-face consultation (<i>n</i> = 8). Patients were randomly allocated to the two groups. The consultations for both groups took place in the same hospital, with a genetic counselor and a genetics consultant. Cross-sectional quantitative telephone questionnaire: four Likert-scale questions completed postconsultation.	Postconsultation measures: -Ease of communication, maintaining eye contact, comfort of room, and satisfaction with clinic format.	Patients, counselors and consultants reported high satisfaction with telegenetics consultation. A majority indicated that they would be happy to use telemedicine in the future and would recommend it to others. Satisfaction scores for telegenetics consultation were comparable with those for face-to-face consultation.	Telegenetics is an acceptable alternative to face-to-face consultations and reduces travel cost and time. It is not suitable for family consultations because multiple patients are difficult to see on-screen.
Gray <i>et al.</i> ^{24,38} UK	Six patients had family histories of cancer. Two patients were referred for other genetic conditions (not specified by the authors). The patient and the genetics nurse communicated with geneticist via telemedicine from a remote clinic.	Prospective pilot study of telegenetics consultation (<i>n</i> = 8; six cancer-related and two for other genetic conditions). Quantitative questionnaire completed pre- and postconsultation.	Pre- and post-consultation measures: -Knowledge of genetics -Anxiety -Illness-related worries -Satisfaction (patients and health professionals).	Data showed a trend toward reductions in anxiety and worry and improvement in knowledge of genetics after telegenetics consultation. Patients reported higher satisfaction levels with telegenetics than health professionals did. Patients and genetic consultants reported that telegenetics consultation was no different from, or was	Telegenetics may be an acceptable method of communication between genetics specialists and patients, and may help meet the increasing demand for genetics services.

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Table 2 Continued.

Author and location	Reason(s) for genetics consultation and service(s) provided	Study design and methods	Outcome measures	Key findings	Conclusions
Hopper <i>et al.</i> ³⁵ Australia	Sessions involved examination of family history and discussion of hereditary cancer. Children with developmental delay. The genetics counselor took physical measurements and still images of the patient while a camera transmitted a live feed to a geneticist. A face-to-face session was arranged 3 weeks after the telemedicine session.	Evaluation of parents' satisfaction with a telegenetics consultation for children with developmental delay (<i>n</i> = 10). Quantitative satisfaction survey for parents of pediatric patients completed at 3 months (<i>n</i> = 4). Identical telephone survey at 12 months (<i>n</i> = 8). Referring pediatrician completed a survey at 3 months.	Postconsultation measures: -Satisfaction at 3 months post-consultation (outcomes not specified) -Satisfaction at 12 months postconsultation -Clinical efficacy of telegenetics consultation.	even preferable to, face-to-face interaction. Genetics nurses felt less at ease and less able to observe patients' nonverbal behavior. Clinically indicated tests were requested for several patients. Physical measurements taken by counselors and geneticists were sufficiently similar. All parents reported high levels of satisfaction with the telegenetics consultation, and 50% felt it was effective in genetic diagnosis for their child. At 12 months, all the respondents were satisfied with the telegenetics experience. One referring pediatrician noted that some dysmorphism features were missed through telemedicine.	Overall satisfaction with the technology was high from parents, pediatricians, and geneticists. The large camera was sometimes intrusive, but this can be overcome by using newer, smaller cameras. Genetics practitioners need to be familiar with the technology and know how to solve problems. With more experience, physical assessment of children via telemedicine will improve.
Lea <i>et al.</i> ³⁷ USA	Sixty-four percent of the patients were pediatric patients, 14% were adult patients referred for cancer genetics counseling, and 7% were referred for reproductive genetics counseling. During the 3-year project 105 patients and ~250 family members had genetics consultation via telemedicine. When necessary, the outreach site nurse performed physical measurements, and it was then determined whether a follow-up session in person with a consultant was required.	Evaluation of a telegenetics pilot project. Cross-sectional quantitative satisfaction questionnaire completed postconsultation by patients and primary-care providers. Telephone interviews to assess qualitative parameters conducted with six patients and eight providers.	Postconsultation measures: - Satisfaction (patients and primary-care providers).	Eighteen percent of the care providers completed the evaluation, mean satisfaction score was 3.83 of 4. Minor difficulties reported with connections and the equipment. Twenty-five percent of the patients completed the evaluation, mean satisfaction score was 3.56 of 4. All patients reported convenience in terms of reduced travel. Two patients would rather have seen the doctor in person. Providers felt that patients' problems were understood and addressed correctly. Drawbacks included lack of hands-on examination, but this was deemed not critical. The consulting geneticist and neurologist felt confident in evaluating patients via telemedicine.	Telemedicine offers an acceptable and accessible solution to meet the growing demand for genetics consultations from individuals who live far from genetics services centers. Telegenetics is suitable for physical examination, dysmorphism evaluations, and prenatal and cancer-related genetics counseling. Funding may be a barrier to sustaining telegenetics services, although cost savings are made through reductions in staff travel to remote sites.

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Table 2 Continued.

Author and location	Reason(s) for genetics consultation and service(s) provided	Study design and methods	Outcome measures	Key findings	Conclusions
Meropol <i>et al.</i> ²⁶ USA	Family history of colorectal or breast/ovarian cancer. Consenting participants were sent a Web-camera and videoconferencing software along with instructions. Participants were shown a 20-min educational video about cancer genetics followed by a counseling session with the genetic counselor.	Feasibility study of telegenetic counseling for cancer with genetic counselor in patients' homes, using videoconferencing software ($n = 31$; 18 probands, 13 family members). Quantitative cross-sectional questionnaire completed postcounseling.	<ul style="list-style-type: none"> -Postcounseling measures: -Technical feasibility -Education and information -Communication -Psychosocial comfort of the proband during counseling -Satisfaction with counseling session and preference for telegenetics. 	Twenty-six (84%) of the participants received counseling at a remote location. Participants were satisfied with the technology (mean score 4.3 of 5) although most experienced some technical problems. High satisfaction with education and information (mean score 4.7). Participants learned about their risk for cancer and received sufficient information about genetic testing (mean = 4.7). Patients were satisfied with communication (mean = 4.8). Fifty-three percent agreed that the session made them feel less anxious about their cancer risk. Twenty-two percent felt uncomfortable with being seen on the computer screen and 10% felt the session was too impersonal. All the patients would recommend the session to others. Nine patients would have preferred to have face-to-face counseling.	Patient satisfaction with telegenetics counseling for cancer is high, and it may therefore be a feasible alternative to face-to-face counseling sessions. Patients receiving telegenetics counseling may require technical support that can be accessed from their homes through the Internet. This requires high-speed Internet access, and this may potentially place a limitation on uptake.
Stalker <i>et al.</i> ³⁶ USA	Pediatric genetics referrals requiring dysmorphology examination. Patients seen by genetic counselor via videoconferencing to gather family and medical history. Pediatrician then performed physical examination and requested appropriate testing. Complex cases were scheduled for face-to-face consultation with clinical geneticist.	An evaluation of pediatric telegenetics consultations ($n = 40$). Patients with simple complaints such as cleft lip were selected by the geneticist and genetic counselor for inclusion in the telegenetics clinic. Postconsultation cross-sectional quantitative survey completed by parents.	<ul style="list-style-type: none"> -Satisfaction with telegenetics evaluation -Diagnostic effectiveness assessed by comparing diagnoses made via telegenetics and those made when patients returned for a follow-up face-to-face appointment. 	All parents agreed that the telemedicine evaluation of their child was appropriate and that their child's privacy was protected. One patient would have preferred a face-to-face appointment. Waiting times were reduced from 16.9 months to 3.0 months ($P < 0.0001$). Diagnoses were made for seven children, and these were confirmed in the traditional genetics clinic. No telegenetics diagnoses were subsequently judged to be incorrect. All the clinicians indicated a high level of comfort with the security and efficiency of the telegenetics clinic.	Telemedicine is an effective medium for the diagnosis of dysmorphic syndromes and can identify patients who are in need of further "hands on" review. Telegenetics reduces waiting times and travel costs for patients, and allows a quicker evaluation and diagnosis.

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Table 2 Continued.

Author and location	Reason(s) for genetics consultation and service(s) provided	Study design and methods	Outcome measures	Key findings	Conclusions
Zilliaccus <i>et al.</i> ³² Australia	Women with a family history of breast/ovarian cancer. The women attended an outreach clinic with a genetic counselor and communicated with the geneticist via videoconferencing.	Qualitative study of women's experience of telegenetics counseling for hereditary breast and/or ovarian cancer (n = 12). Semi-structured interviews conducted after the telegenetics consultation.	Interviews explored patient satisfaction with the technology and their interaction with genetics professionals.	All the women felt positive about the technology and the majority (n = 11) were satisfied with the interaction with their genetics clinician. Nine of the women would be happy to have a telegenetics consultation again, whereas 7 preferred to meet the clinician in person. All the women reported feeling a subtle difference between telegenetics and face-to-face interactions. Eleven of the women described a high degree of 'social presence' that led to increased comfort with technology. All the women felt at ease in communicating with the clinician and counselor. The advantage most commonly cited was reduction in travel time.	Women were highly satisfied with telemedicine consultations for hereditary breast/ovarian cancer because of reduced travel and associated costs. Telegenetics counseling may be particularly appropriate for those whose need for psychosocial support is low.
Zilliaccus <i>et al.</i> ³³ Australia	Women with a family history of breast/ovarian cancer. Women attending genetic counseling for the first time were invited to participate in the telegenetics consultation. The women attended an outreach clinic with a genetic counselor and communicated with a geneticist via videoconferencing.	Prospective study of women's experience of telegenetics counseling for hereditary breast vs. face-to-face counseling (n = 89). Quantitative questionnaire completed pre- and postcounseling.	Pre- and postcounseling measures: -Knowledge about the genetics of breast cancer -Perceived personal control -Impact of Events Scale (cancer-specific anxiety) -Hospital Anxiety and Depression Scale -Medical Interview Satisfaction Scale -Consultation and relational empathy -Telegenetics satisfaction.	Knowledge of hereditary breast and ovarian cancer increased (P < 0.001) and perceived control decreased (P < 0.001) over time in both telegenetics and face-to-face groups. No changes found in cancer-specific anxiety, general anxiety, and depression in either group. In terms of cancer-specific anxiety, face-to-face and telegenetics counseling were found to have equivalent outcomes (P = 0.13). No between-group differences were found with respect to change scores for knowledge, general anxiety, and depression. No between-group differences as regards satisfaction and perceived clinician and counselor empathy. Seven percent of telemedicine group would prefer a face-to-face appointment. Thirty-three percent would prefer telegenetics again.	Telegenetics counseling was at least as effective as face-to-face genetics counseling across all outcomes measured. It performed better in increasing perceived personal control. It is therefore a viable alternative method of service delivery to rural and outreach areas.

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Table 2 Continued.

Author and location	Reason(s) for genetics consultation and service(s) provided	Study design and methods	Outcome measures	Key findings	Conclusions
Zilliacus <i>et al.</i> ^{30,31} Australia	Women with a family history of breast/ovarian cancer. Women with a family history of breast and/or ovarian cancer attended an outreach clinic with a genetic counselor and communicated with a geneticist via videoconferencing.	Qualitative study of genetics practitioners' experiences of delivering genetics counseling via telemedicine ($n = 15$; six genetics clinicians, nine genetics counselors). Semi-structured interviews conducted postcounseling.	Interviews explored practitioners' experiences, satisfaction, advantages and disadvantages of the technology, and their perceived role within telehealth.	All the practitioners were highly satisfied with telegenetics consultations. Advantages of telegenetics included increased efficiency and convenience for geneticists, reduced travel and associated costs for the patient, and increased access for those in rural areas. Disadvantages included difficulty in establishing rapport and in detecting nonverbal cues. Telegenetics consultations were seen as allowing less time for emotional exploration, and as altering the genetic clinician's role to one of a "visiting specialist". Counselors reported taking on multiple roles including facilitator, administrative assistant, and counselor, which they regarded as similar to their roles in face-to-face sessions.	Practitioners were satisfied with telegenetics consultations and perceived the advantages as outweighing the disadvantages. The counselors felt themselves to be integral to the overall telegenetics process.

Overall, the studies included in this review are heterogeneous and vary in quality. Most of the studies had limitations of small sample sizes that precluded the use of statistical analyses. The limitations also included retrospective study designs and the lack of comparison groups, and the data were therefore subject to bias.

Satisfaction with telegenetics services

All the studies reported high levels of patient satisfaction with the telegenetics services, as ascertained through questionnaires or interviews. Patients were generally satisfied with the technology used, the education and information provided, and the opportunity to communicate with genetics professionals without having to travel long distances. Those involved in the care of pediatric patients felt that telemedicine was an effective medium through which to evaluate genetic conditions in their children.^{35–37} Where a comparison group was utilized, the satisfaction levels of those receiving telegenetics services were generally no different from those in the group receiving face-to-face counseling. Indeed, in one study, the satisfaction level in the telegenetics group was higher than in the control group.²⁸ The studies reported several benefits of telegenetics services: convenience, reduced travel time and associated costs, and reduced waiting times to see a genetics specialist.³⁶ Patients tended to be more satisfied than the genetics practitioners with telegenetics counseling.^{24,29,37,38} Patients viewed the technical difficulties involved as being less problematic, and were more satisfied with the rapport established with the genetics professional.

In a study by Gray *et al.*,^{24,38} nursing staff expressed dissatisfaction with telegenetics because they felt excluded from the geneticist–patient interaction and less able to observe the nonverbal behavior of patients. Similarly, in a study by Zilliacus *et al.*,^{30,31} genetics practitioners reported that there were disadvantages to telegenetics counseling for patients with hereditary breast/ovarian cancer, including inhibition of rapport between the geneticist and the patient, and difficulty for the practitioner in detecting nonverbal cues from patients. Patients may have been more satisfied with telegenetics consultations because of the savings in travel time and costs; a majority of the patients preferred to receive genetic counseling services in a local setting. In many studies, patients who received telegenetics counseling were asked whether they would have preferred to have a face-to-face appointment. Women receiving genetic counseling for cancer would be happy to have another telegenetics consultation, but a majority ($n = 7$) wanted to meet the geneticist in person.³² A study by Meropol *et al.*,²⁶ reported that all the participants would recommend telegenetics counseling to other patients, although nine of them (29%) agreed that they would have preferred a face-to-face session. In Abrams and Grier's³⁴ study of prenatal telegenetics counseling, only one participant would have preferred an in-person counseling session instead.

Affective outcomes

The three studies that reported on affective outcomes of telegenetics consultations used prospective study designs. All three

studies reported that patients' knowledge of genetics increased after the telegenetics consultations. Gray *et al.*,²⁴ reported that general anxiety and cancer-specific anxiety decreased over time in those who received telegenetics counseling. Coelho *et al.*²⁸ found significant decreases in anxiety levels after cancer genetics counseling in the telegenetics group ($P = 0.00$), the face-to-face group ($P = 0.01$), and the combined (telemedicine and face-to-face) group ($P = 0.00$). Zilliacus *et al.*³³ reported no change in cancer-specific anxiety, general anxiety, or depression in either the telegenetics group or the face-to-face group in a cohort of women receiving genetics counseling for hereditary breast/ovarian cancer. In studies that compared telegenetics groups with those receiving face-to-face consultations, there were no significant between-group differences in any of the affective outcome variables.^{28,33}

Clinical efficacy

Three of the studies explored the effectiveness of telegenetics in diagnosing genetics-related conditions in pediatric patients.³⁵⁻³⁷ In a study by Stalker *et al.*,³⁶ a genetics counselor ascertained the family history and a pediatrician performed a physical examination to screen for potential genetic syndromes. The first eight patients evaluated via telemedicine were also assessed in person by a clinical geneticist. Diagnoses were made for seven of the children via telegenetics, and these were confirmed in the face-to-face genetics clinic. No new diagnoses were made in the face-to-face sessions that had not been identified in the telegenetics sessions, and none of the diagnoses made through telegenetics were deemed inaccurate. The authors concluded that telegenetics was successful in identifying individuals in need of further hands-on dysmorphic review and in providing quicker confirmation of the absence of a particular genetic syndrome. Lea *et al.*³⁷ also reported that telegenetics was effective as a dysmorphism screening tool, because patients could be referred for an in-person evaluation if a dysmorphic genetic syndrome was suspected. However, in a study by Hopper *et al.*,³⁵ one referring pediatrician was of the opinion that several physical characteristics could potentially be missed in a telegenetics consultation. In that study, the genetics counselor took physical measurements and still images of the patients while a camera transmitted a live feed to the clinical geneticist. Face-to-face sessions were arranged 3 weeks after the telegenetics sessions. The measurements taken in person by the genetics counselors and those ascertained by the clinical geneticist from the live feeds of images varied slightly but did not affect either the assessments or the diagnoses. The authors of the study concluded that, with an increase in shared telegenetics experience between the genetics counselor and the geneticist, dysmorphic assessment of children via telegenetics consultations will improve.

Costs of telegenetics

None of the studies included in our review formally measured the costs of telegenetics services. Most of the studies reported that patients made savings in terms of travel time and costs by receiving telegenetics services rather than face-to-face genetics

services. In one study, telegenetics was estimated to confer an average saving of \$1,000 per person.²⁹ The same authors also concluded that videoconferencing is less expensive than outreach programs. They estimated that, in Canada, it would cost \$700 for a genetics counselor to travel to a specific site to provide outreach care for 12 patients at a 2-day clinic. In contrast, the costs associated with the same number of appointments through videoconferencing are negligible, provided the necessary equipment is already available and in place.²⁹

DISCUSSION

Our review shows that patients are generally highly satisfied with the use of telemedicine in genetics service delivery, including genetics counseling and diagnoses of pediatric genetic conditions. The studies included in the review also show that the affective outcomes for patients receiving telegenetics consultations are comparable to those for patients receiving face-to-face consultations. This suggests that telegenetics is acceptable to patients and does not appear to have any detrimental effects on patient anxiety. Neither the type of health professional involved in the telemedicine consultation nor the presence of a genetics nurse/counselor at the outreach site appeared to affect patient satisfaction. Comparable results were reported from the various studies conducted in different countries. The opinions of genetics practitioners regarding telegenetics services were not always as positive, and many expressed concerns about the difficulty in establishing a rapport with patients via telemedicine and difficulties associated with the use of the technology. Many of the studies concluded that, with appropriate training and growing experience with telegenetics services, genetics practitioners will become more accepting of the technology. In a study by Lea *et al.*,³⁷ practitioners rated telegenetics services higher after they had personally provided these services, suggesting that familiarity and confidence with using the technology will reduce practitioners' resistance to adopting telegenetics services. Although most patients appreciated the fact that telegenetics counseling had the benefits of reduced waiting times and/or shorter travel distances, there is a need to be cautious when interpreting data from patient surveys regarding telegenetics consultations. These patients have received a service they ordinarily would not have, and they may therefore feel obligated to endorse it.³⁹ This is especially relevant in studies wherein patients were allowed to choose whether to receive telegenetics services or face-to-face services (e.g., the studies by d'Agincourt-Canning *et al.*,²⁹ and Meropol *et al.*²⁶ Indeed, previous research has found that patients who were able to choose how they received their *BRCA1/2* genetic test result—whether by telephone or in person—reported higher levels of satisfaction than patients who did not have a choice.²² Studies with random assignment to telegenetics services or face-to-face services may help to reduce this response bias.

It could also be argued that the selection of new referrals for telegenetics consultations may introduce some bias into research results (e.g., the study by Stalker *et al.*³⁶). However, many studies have stated that telegenetics is not intended to replace existing services but may be a useful adjunct to

traditional service delivery for dysmorphic evaluation and/or genetics consultations. In the study by d'Agincourt-Canning *et al.*,²⁹ some participants reported that they would not have followed through with their referral to the Hereditary Cancer Program had they been unable to have an appointment in their local area. Telemedicine may therefore extend access to remote populations and reduce waiting times to consult with genetics specialists, and thereby increase the capacity to provide genetics services to those who require them and not just to those fortunate enough to live close to genetics centers.^{21,39}

The cost-utility assessment of providing telegenetics services, including costs of equipment, transmission lines, training, and personnel has not been formally carried out, and is a vital area for further research. High-quality evidence on the efficacy and cost-effectiveness of telegenetics will encourage the expansion of these services within the National Health Service in the UK. In the United States, reimbursement of telegenetics costs needs to be sufficient in order to provide sustainable telegenetics clinics. This involves ensuring that telegenetics has the support of health insurers, not only the private insurance companies but also Medicare and Medicaid, which are the federal health insurers for the elderly and poor. To date, Medicaid does not universally reimburse for telemedicine consultations in all 50 states, nor do private payers. Under Medicare programs there are also wide variations in service coverage, payment policies, and other stipulations,⁴⁰ and these are considered to be barriers to the long-term sustainability and expansion of telemedicine services.^{41–43} Whitten and Buis⁴¹ suggest that, in order to promote more consistent reimbursement for telemedicine services, it is essential that both consumer demand and provider acceptance for these services increase. They also suggest that demonstrating the clinical effectiveness of telemedicine will encourage reimbursement for these services in the long term. Our review has demonstrated that patients are generally satisfied with telegenetics, but that further research is needed to explore beyond self-reported measures of patient satisfaction and fully investigate patient outcomes so as to determine whether quality of care is affected by the type of consultation received.^{44,45}

Conclusions

Although the studies relating to the use of telegenetics are few in number to date and generally small in sample size, they suggest that telegenetics may be appropriate for providing additional genetics services to reduce inequalities in access to genetics specialists. Telemedicine technology is being utilized in many medical specialties; teledermatology, telepsychiatry, and teleradiology are already being used, and the delivery of genetics services through telegenetics could be a feasible approach. The studies included in this review suggest that telegenetics may not only be a useful tool to provide routine counseling and follow-up appointments, but that it also has the potential to evaluate and diagnose pediatric patients with suspected genetic conditions. Prospective, fully powered, and well designed studies of telegenetics services, assessing the accuracy of diagnoses, the impact of receiving a diagnosis this way, and patient outcomes are needed to make

informed decisions about the appropriate use of telemedicine in genetics service delivery.

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DISCLOSURE

The authors declare no conflict of interest.

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