Enabling and Enhancing Telehealth for Rare Diseases Across the Globe

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On behalf of the IRDiRC Telehealth Task Force*

OBJECTIVES

The IRDiRC Telehealth (TH) Task Force was formed in October 2021 to explore the use of telehealth (TH) to improve access to diagnosis, care, and research experiences for rare disease (RD) patients worldwide, to identify existing models of TH, and to develop “best practices” for introducing TH services into RD communities.

1. Conduct a systematic review of published literature and interview key opinion leaders about existing models of telehealth, their uptake and usage by the rare disease community, and their specific value and effectiveness to identify the factors that enhance or limit their adoptability, sustained use, ease of access, and effectiveness in the rare disease community.

2. Identify barriers to and opportunities for the use of telehealth to improve access to diagnosis, care, prevention, and research experiences for rare disease patients as well as continuing education and peer mentoring for healthcare providers—including technological, legal, cultural, linguistic, healthcare system, and patient/provider factors.

3. Develop “best practices” for introducing telehealth services into are disease communities.

RESULTS

The group modified a definition of TH from the World Health Organization (2010) specifically for RD: “The delivery of health care services, where distance is a critical factor, by all health care professionals using information and communication technologies for the exchange of valid information for diagnosis, treatment and prevention of RD, research and evaluation, and for the continuing education of health care providers, all in the interests of advancing the health of individuals with RD and their communities.”

Identified 440 articles in PubMed with TH and RD keywords
Reduced to 356 based on screening of abstract for relevance

Main TH beneficiaries are Patients and Families
Majority of papers were published by European (159) and North American (78) authors

Topic 1 (180) Topic 2 (136) Topic 3 (40)
Peak of publication occurred in 2021 at the time of COVID-19 pandemic

Key questions used in the KOL interviews:
- Model of TH used and strategies for implementation
- Strengths, Weaknesses, Opportunities, and Threats (SWOT) Analysis of the TH model and its implementation for rare diseases for patients/families, healthcare providers, private and public payers, regulatory bodies, and researchers
- Aspects specific or unique to RD

METHODS

The IRDiRC TH Task Force is composed of 22 members, from 14 countries, covering the EU, North America, Asia, Africa and Oceania. The members include diverse expertise, representing RD patients, patient advocates, physicians, industry leaders, academicians, and government representatives. The groups were divided into 3 working groups to focus on three topics, based on the WHO TH definition:

Topic 1: Diagnosis, Treatment, and Prevention

Topic 2: Research and Evaluation

Topic 3: Continuing Education of Healthcare Providers

The group has conducted a systematic review of literature on the topic, and is now interviewing ~25 Key Opinion Leaders (KOL) about existing models of TH.

CONCLUSION

While the analyses of the literature and the KOL interviews are ongoing, there are some emerging conclusions.

1. Use of TH for those with pre-existing medical conditions or wishing to avoid COVID exposures at hospitals and clinics has led to increased utilization by RD patients.

2. TH can improve accessibility and delivery of medical/clinical services and fulfill individual clinical needs for RD patients, with reported high patient satisfaction.

3. TH affords greater opportunity to conduct research, train a specialized workforce, and enhance primary care physicians’ awareness and knowledge of RD to build an interconnected RD community.

4. Different regions with varying resources and technology platforms may require different approaches to TH for RD patients.

5. Resources for building infrastructure, access to, and familiarity with TH technologies, and concerns for privacy and data security are among some of the factors limiting the use of TH.

6. From KOL interviews, different countries have different regulations and laws that may facilitate or limit the use of TH for RD patients, most notably insurance coverage of TH and medical licensure requirements.

There are few reports that have directly investigated the quantitative impact of TH on RD clinical outcomes as compared to in-person visits in rigorously conducted studies.

8. In spite of regulatory, legal, and privacy challenges, TH will likely grow in use, at least for RD patients with limited access to personalized therapies.

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