



Telegenetics and COVID-19: Through the pandemic and beyond

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Introduction: *The COVID-19 pandemic led to restrictions on the conventional delivery of health care. Telemedicine provided a viable solution that was in line with the social distancing policies aimed to minimize disease transmission. This demanded physicians adapt to new ways of healthcare delivery. We surveyed geneticists across the country to determine their experience and to ascertain if telegenetics will be a lasting change.*

Materials and methods: *A 23-item standardized survey was distributed to various US-based geneticists via email and other social media platforms focusing on their experience of providing care via telemedicine.*

Results: *We received 69 responses from 120 geneticists across 26 states in the United States. Of these, 91% practiced in academia. The majority, 70%, responded that pediatric genetics*

takes up more than half of their practice, and 68% had over half of their practice switch to telemedicine. Most (77%) felt they could provide adequate care via telemedicine, and 94% of providers would like to continue telemedicine post-pandemic.

Conclusion: *The future of telemedicine looks promising as the majority of clinicians would like to use telemedicine routinely post-pandemic. Uniform guidelines for the use of telemedicine in genetics may need to be proposed by professional societies and supported by federal laws.*

The COVID-19 pandemic has been of historic proportions and has disrupted health care across the globe. On March 11, 2020, the World Health Organization (WHO) declared COVID-19 a pandemic, at that point various social distancing policies were put in

place to reduce disease transmission. These changes necessitated a change in healthcare delivery.^{1,2} Telemedicine had been in use prior to this pandemic, most notably in the areas of stroke neurology, psychiatry, and dermatology, but COVID-19 has accelerated its development and use across all medical specialties.^{3,4} Some experts even argue that telemedicine is now an emerging new specialty with its unique challenges.⁵

The ‘stay at home’ restrictions imposed across the United States limited the face-to-face encounters of providers and patients, but telemedicine provided a solution to maintain continuity of care via telephone and video visits, which also helped reduce occupational exposure for providers.⁶ Telehealth has been adopted widely across various specialties including genomic practices and is called telegenetics when specifically used to provide genetics-related care to the patient.⁷ Telegenetics has previously been used with success to provide genetic counseling, which is mostly all verbal interactions.⁸

BACKGROUND

The COVID-19 pandemic has had a unique impact on the care of patients with rare genetic disorders. To recognize the impact of telemedicine/telegenetics in the field of genomics, we surveyed geneticists across the country to learn the differences that they may have experienced in providing patient care virtually in an outpatient setting, the challenges they encountered, and level of satisfaction with the change in practice. We also wanted to ascertain if telemedicine will be a more permanent change which will remain in use post-pandemic.

MATERIALS AND METHODS

Data collection

A 23-item standardized survey was distributed to 120 US-based geneticists. The survey was filled

out between October 24 and November 26, 2020. The questionnaire was in English, which was the primary language for all participants, and was developed by the authors on issues addressing the impact of telehealth in providing patient care. It was sent out to geneticists both in academic teaching hospitals and in private practice via e-mail and was shared on various social media platforms like Twitter and ResearchGate. The questionnaire focused on the experience of providing patient care via telemedicine during the COVID-19 pandemic in the outpatient setting. Six questions inquired about the participants’ basic demographics – gender, state, type of practice, and type of institution – and the other 17 questions inquired about the changes in practice that physicians dealt with while caring for patients virtually.

The survey was filled out voluntarily and anonymously. We received a total of 69 responses out of 120 physicians across 26 states in the United States corresponding to a response rate of 57.5%.

Ethics approval

This study was exempted from ethical approval by the Institutional Review Board since it is an online self-administered survey that was voluntary and anonymous.

RESULTS

Demographics

A total of 69 responses were received from geneticists in 26 states across the United States. The demographics are shown in Table 1. Most physicians who responded were practicing at academic teaching institutes (91%), 67% of responders were geneticists practicing independently, and 33% were geneticists doing their fellowships and were in training. Pediatric genetic practice in an outpatient setting made up more than 50% of the practice of most of the surveyed individuals (70%).

Table 1. Study population demographics

Sex	Female	Male
	41 (59%)	28 (41%)
Type of institution	Academic	Private
	63 (91%)	6 (9%)
Type of practice	Independent[†]	Training
	46 (67%)	23 (33%)
Time spent practicing pediatric genetics	<50%	50–90%
	20 (29%)	22 (32%)
		>90%
		27 (39%)

[†]Average time spent in independent practice is 13.6 years.

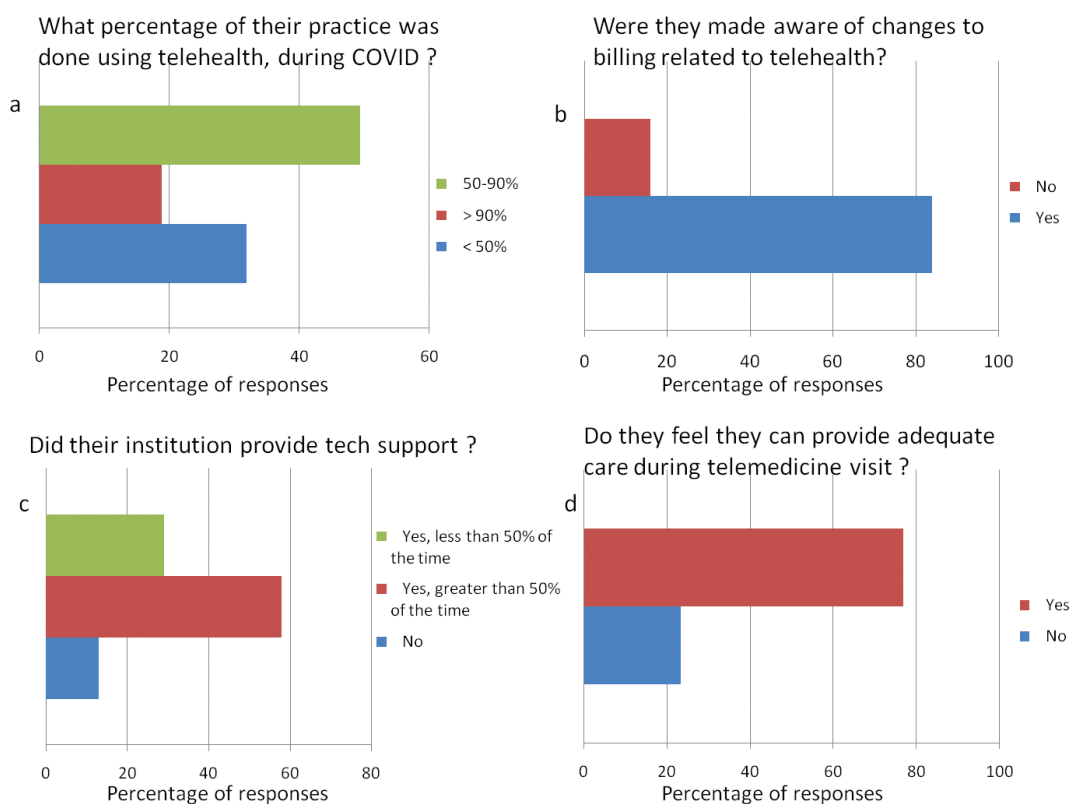


Figure 1— Changing role of telehealth as a result of COVID-19.

Practice of telemedicine during COVID-19

According to the information gathered in our survey, almost half of the respondents (49%) made use of telehealth, both audio and video calls, in 50–90% of their practice. One-fifth (19%) made use of telehealth more than 90% of

the time, and 32% made use of telehealth less than 50% of the time (Fig. 1a).

In total, 84% of responders were aware of changes to billing related to telehealth (Fig. 1b). Over half of the responders were given technical

support by their institution for telehealth more than half of the time, while a few (13%) of the responders were not provided any technical support by their institution (Fig. 1c). Three-quarters (77%) of the responders felt that they were able to provide adequate care during telemedicine visits (Fig. 1d).

For pediatric outpatient visits, most responders stated that parents did not understand that children had to be present during televisits (76%) (Fig. 2a). Examinations done using ‘video conferencing’ as a mode of telehealth were considered inadequate by more than half of the responders (51%) (Fig. 2b).

The vast majority of responders (94%) were sending DNA testing kits to patients’ homes, and

60% of them said that the turnaround time for testing did not increase. Most responders (81%) felt that their patient satisfaction score stayed the same while using telemedicine.

Use of telehealth post-pandemic

Almost all of the geneticists (94%) would like to continue to use some telemedicine in their outpatient practice even after the pandemic, if allowed by their institution and insurance (Fig. 3).

DISCUSSION

Our study aimed to survey the experience of geneticists providing health care for patients with rare genetic disorders via telemedicine, using modalities such as video conferencing and audio calling, across the country.

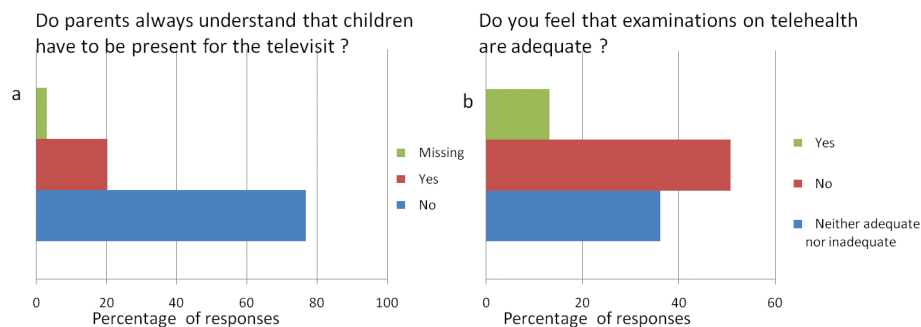


Figure 2— Challenges in the use of telehealth reported in pediatric practices.

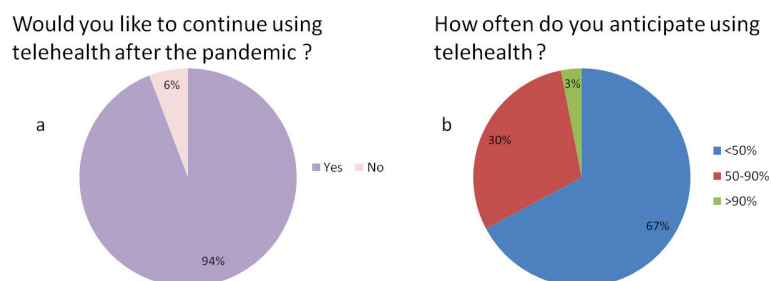


Figure 3— Most geneticists (94%) would like to continue using some telemedicine in their outpatient practice even after the pandemic.

Telehealth has evolved significantly since the 1879 article in the *Lancet*, which discussed using the telephone to reduce unnecessary office visits for patients.⁹ There has been tremendous innovation in the field of telehealth in the last decade, and even prior to the pandemic, efforts were being made to expand its use.¹⁰ Telemedicine has proven not to be inferior to traditional medicine, especially for follow-up visits.¹¹ Studies have also shown its cost-effectiveness as it saves travel time and time away from work for patients and families.¹²

The COVID-19 pandemic fueled the expansion and use of telehealth to minimize disease transmission,¹³ and besides, being an effective tool for social distancing, it also proved effective in providing care to patients in a safe environment. Almost half of the responders in our survey confirmed that they used telehealth in 50–90% of their practice, and almost one-fifth of the responders used telemedicine in more than 90% of their practice.

Telehealth using audio and visual conference calls can pose difficulties when it comes to physical examinations and rapport building with the patient; however, a review was done in 2012 that concludes that patients are generally highly satisfied when cared for virtually by geneticists.¹⁴ Traditionally, physical examination is an invaluable tool in the assessment and diagnosis of many clinical conditions which a virtual examination cannot mimic. More than half of our responders felt that they were not able to perform an adequate examination; despite that, 77% of responders felt that they were able to provide adequate overall care via telehealth. There have been studies showing that telehealth is an effective tool to diagnose and assess genetics-related conditions in pediatric patients.¹⁴ Telegenetics has proven successful in confirming the absence of a clinical syndrome and in triaging

patients who were in need of a more hands-on dysmorphology review in an efficient and accurate way.¹⁵ Telegenetics has certainly made access to genetic professionals easier across state lines.⁷

Even though telemedicine is accepted by both clinicians and patients, there have been concerns about the lack of technical knowledge among practitioners and patients. This has been rapidly changing as more and more people have been getting familiar and comfortable with the use of video conferencing during this pandemic. Technology is, thus, not a limitation as was previously believed, and the learning curve for both patients and providers has been steep.¹⁶ Technical challenges can be avoided by educating the entire care team upfront, identifying pitfalls, and working around them to enable the smooth execution of telemedicine practices.² Availability of technical support by the institution is desired and was available to most providers in our survey.

The bigger problem seems to be a concern for privacy and safety of private information. Inadvertent leaks of information via mobile apps or home monitoring devices make issues of privacy a concern that may undermine the potential that telehealth offers and measures to protect patients need to be enforced.¹⁷

Telehealth works well for specialties where verbal interaction is a key part of the assessment process. It has been proven that genetic counseling can be effectively provided via telehealth,⁸ and in our survey, 94% of responders responded ‘Yes’ when asked if they would like to use telehealth post-pandemic. Telehealth promotes ease of access to genetic services to people who may live in remote areas, making specialist care available to them in a cost-effective way and enabling their healthcare needs

to be met.^{18–20} These factors mean it is likely to stay around after the pandemic is over.²¹

This study did have some limitations, including that it used a self-authored questionnaire, and that the survey did not include all 50 states and had a small sample size. Regardless, we believe it sheds light on this important topic.

CONCLUSIONS

Telemedicine has seen increasing use during the COVID-19 pandemic to reduce disease transmission. We surveyed various geneticists practicing in the United States who had sufficiently utilized telehealth. Several lessons have been learned at individual and institutional levels. Even with limitations that telehealth poses, such as inadequate physical examination and geneticists mostly felt that they were able to provide adequate care via telehealth and with support from their institution/insurance, they anticipate using some telehealth routinely as a way of providing care to patients. Uniform guidelines for the use of telemedicine in genetics may need to be proposed by professional societies and be supported by federal laws.

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REFERENCES

1. WHO. Director General's opening remarks at the media briefing on COVID-19. 2020 Mar 11 [cited May 2021]. Available from: https://www.who.int/docs/default-source/coronaviruse/transcripts/who-audio-emergencies-coronavirus-press-conference-full-and-final-11mar2020.pdf?sfvrsn=cb432bb3_2
2. Shur N, Atabaki SM, Kisling MS, Tabarani A, Williams C, Fraser JL, et al. Rapid deployment of a telemedicine care model for genetics and metabolism during COVID-19. *Am J Med Genet A*. 2021; 185(1):68–72. <https://doi.org/10.1002/ajmg.a.61911>
3. Wamsley CE, Kramer A, Kenkel JM, Amirlak B. Trends and challenges of telehealth in an academic institution: the unforeseen benefits of the COVID-19 global pandemic. *Aest Surg J*. 2020;41(1):109–18. <https://doi.org/10.1093/asj/sjaa212>
4. Trettel A, Eissing L, Augustin M. Telemedicine in dermatology: findings and experiences worldwide – a systematic literature review. *J Eur Acad Dermatol Venereol* 2018;32(2):215–24. <https://doi.org/10.1111/jdv.14341>
5. Waller M, Taylor L, Portnoy J. The medical virtualist: is pediatric patient care using telemedicine, a new specialty? *Pediatr Ann* 2019;48(6):e243–8. <https://doi.org/10.3928/19382359-20190520-01>
6. Wosik J, Fudim M, Cameron B, Gellad ZF, Cho A, Phinney D, et al. Telehealth transformation: COVID-19 and the rise of virtual care. *J Am Med Inform Assoc* 2020;27(6):957–62. <https://doi.org/10.1093/jamia/ocaa067>
7. Hilgart JS, Hayward JA, Coles B, Iredale R. Telegenetics: a systematic review of telemedicine in genetics services. *Genet Med* 2012;14(9):765–76. <https://doi.org/10.1038/gim.2012.40>
8. Zierhut HA, MacFarlane IM, Ahmed Z, Davies J. Genetic counselors' experiences and interest in telegenetics and remote counseling. *J Genet Couns* 2018;

- 27(2):329–38. <https://doi.org/10.1007/s10897-017-0200-x>
9. Nesbitt TS. The evolution of telehealth: where have we been and where are we going? In: Board on Health Care Services, Institute of Medicine. *The role of telehealth in an evolving health care environment: workshop summary*. Washington, DC: National Academies Press; 2012, pp. 11–16. [cited October 2021]. Available from: <https://www.ncbi.nlm.nih.gov/books/NBK207141/>
 10. Sood S, Mbarika V, Jugoo S, Dookhy R, Doarn CR, Prakash N, et al. What is telemedicine? A collection of 104 peer-reviewed perspectives and theoretical underpinnings. *Telemed J E Health* 2007;13(5):573–90. <https://doi.org/10.1089/tmj.2006.0073>
 11. Muller KI, Alstadhaug KB, Bekkelund SI. Headache patients' satisfaction with telemedicine: a 12-month follow-up randomized non-inferiority trial. *Eur J Neurol* 2017;24(6):807–5. <https://doi.org/10.1111/ene.13294>
 12. Avidor D, Loewenstein A, Waisbourd M, Nutman A. Cost-effectiveness of diabetic retinopathy screening programs using telemedicine: a systematic review. *Cost Eff Resour Alloc* 2020;18:16. <https://doi.org/10.1186/s12962-020-00211-1>
 13. Ananthakrishnan AN, Singh S. The doctor will call you now! Telemedicine in the midst of a pandemic. *Clin Gastroenterol Hepatol* 2020;18(8):1688–90. <https://doi.org/10.1016%2Fj.cgh.2020.04.031>
 14. Hilgart JS, Hayward JA, Coles B, Iredale R. Telegenetics: a systematic review of telemedicine in genetics services. *Genet Med* 2012; 14(9):765–76. <https://doi.org/10.1038/gim.2012.40>
 15. Stalker HJ, Wilson R, McCune H, Gonzalez J, Moffett M, Zori RT. Telegenetic medicine: improved access to services in an underserved area. *J Telemed Telecare* 2006;12(4):182–5. <https://doi.org/10.1258/135763306777488762>
 16. Garattini L, Badinella Martini M, Zanetti M. More room for telemedicine after COVID-19: lessons for primary care? *Eur J Health Econ* 2021;22(2):183–6. <https://doi.org/10.1007/s10198-020-01248-y>
 17. Hall JL, McGraw D. For telehealth to succeed, privacy and security risks must be identified and addressed. *Health Affairs* 2014;33(2):216–21. <https://doi.org/10.1377/hlthaff.2013.0997>
 18. Lea DH. A new world view of genetics service models. *Online J Issues Nurs* 2000; 5(3):5.
 19. European Society of Human Genetics' PPPC. Provision of genetic services in Europe: current practices and issues. *Eur J Hum Genet* 2003;11(2):S13–48.
 20. Hawkins AK, Hayden MR. A grand challenge: providing benefits of clinical genetics to those in need. *Genet Med* 2011; 13(3):197–200. <https://doi.org/10.1097/gim.0b013e31820c056e>
 21. Tuckson RV, Edmunds M, Hodgkins ML. Telehealth. *N Engl J Med* 2017;377(16):1585–92. <https://doi.org/10.1056/nejmsr1503323>

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