

**SPECIAL ISSUE**

# Genetic counseling practice for inherited eye diseases in an Israeli medical center during the COVID-19 pandemic

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**Abstract**

Inherited eye diseases (IED) are among the most common causes for childhood and young adulthood blindness in developed countries. Genetic counseling and testing have become an essential part of caregiving for families affected by one of these severe ocular pathologies. The objective of our study is to describe our experience during the 2020 (COVID-19) pandemic, following a practice protocol of safe genetic counseling for inherited ophthalmic diseases. We conducted a review of the genetic counseling practices from January until December 2020 in a multidisciplinary clinic for patients with visual impairment, in a tertiary hospital. The new protocol covered patient screening, required personal protective equipment, and the implementation of telemedicine.

One hundred and eighty-three counseling sessions were done in this period of time; 33/183 were telemedicine counseling. The results of this study indicate that the practice of genetic counseling in regard to inherited eye diseases in the era of COVID-19 is effective and safe. Despite the high risk of infectivity that threatened healthcare professionals, safety measures adopted to reduce the risk of infection allowed us to prevent the cancelation of routine counseling, while keeping patient care our priority. The use of telemedicine was a very useful tool for providing counseling during lockdown periods in 2020.

**KEYWORDS**

COVID-19, genetic counseling, inherited eye disease, telemedicine

## 1 | INTRODUCTION

Genetic counseling and testing have become an integral part of patient care in ophthalmology, playing an important role in helping patients to understand the transmission of inherited eye disease (IED), means of prevention, and possible genetic therapy in some cases (Stone et al., 2012). Genetic testing has significantly advanced in the field of ophthalmology over the past decades, moving from mainly 'research' to an important clinical service for patients. The number of known disease-causing genes has increased significantly, and their screening in parallel and at a low cost is readily available. A molecular diagnosis is essential for patient eligibility for new therapies such as

voretigene neparvovec for RPE65-associated retinal degeneration, and other new clinical trials investigating the use of approaches such as gene replacement, small molecule drugs for nonsense mutation suppression, and antisense oligonucleotides for splice modulation (Russell et al., 2017; Havens & Hastings, 2016; Rodrigues et al., 2019; Richardson et al., 2020).

A clear molecular diagnosis assists in providing the affected family with data on the disease's course and expected prognosis, provides comfort to the patient and/or their family, and also aids their family planning decisions (Ménéjéca et al., 2020).

Due to the COVID-19 pandemic, most clinics and hospitals have decreased patient care of nonessential health services, including

genetic counseling, to slow the spread of the virus (Lai et al., 2020). However, delaying genetic counseling can be problematic, for example, in situations where genetic testing would affect decisions during a pregnancy or candidacy for gene replacement therapy (Stone, 2007).

Genetic counseling practice can involve several visits to undergo anamnesis and physical evaluation, receive genetic test results, and attend follow-up appointments. Telemedicine can be effectively utilized in this area as an alternative to standard clinic visits, even after this pandemic has ended (Hilgart et al., 2012; Saleem et al., 2020; Shannon et al., 2020).

During the first lockdown in our country in March–April 2020, the ministry of health in Israel instructed to close all elective medical practice in the country. In our center, most visits and genetic counseling sessions were canceled. In May, elective medical services were re-established; in our clinic, patients were contacted and offered to choose between telemedicine (to try to reduce direct patient–healthcare professional encounter) or in-person counseling.

Hereby we present the genetic counseling experience during the 2020 COVID-19 pandemic in our multidisciplinary clinic, treating children and families with an IED leading to visual impairment or blindness. The ophthalmic genetic counseling and testing practice is an integral part of this center, which together with our social worker provides emotional support for families in distress when learning the cause of an IED and updating patients and their relatives regarding different available related resources.

## 2 | METHODS

Retrospective review of genetic counseling practice for ocular diseases was conducted, during the 2020 (COVID-19) pandemic, from January until December 2020, in a multidisciplinary clinic for patients with low vision in a tertiary hospital in Israel. Our team includes two senior pediatric ophthalmologists, a social worker, two licensed genetic counselors specialized in ophthalmic diseases, four low vision optometrists, and a secretary. Our multidisciplinary center takes care of our visually impaired patients and families in many life aspects, such as medical diagnosis and treatment, social needs, governmental support, and assessing need as well as provision of special glasses and visual aids. Patients with suspected inherited eye diseases are referred for genetic counseling and testing with our specialized counselors in IED taking care of around 200 new patients per year. A protocol for safe counseling during pandemic times was used including measures such as patient screening for fever and COVID-19 symptoms, the use of personal protective equipment (wearing a surgical mask full time) for patients and personnel, prohibition of eating/drinking inside the clinic (including patients waiting room, common tea room for personnel), and the option of telemedicine offered to patients seeking genetic counseling.

There are three main groups of applicants for genetic counseling at our clinic: (a) patients suspected to have IED, mostly retinal

### What is known about this topic:

Telemedicine has been used in genetic counseling in previous years and has become a useful tool during the COVID-19 pandemic.

### What this paper adds to the topic:

Genetic counseling in ophthalmology has become of critical importance in preventing future incidences of blindness in a family. Furthermore, genetic therapy is available for some inherited eye diseases and can prevent visual deterioration in children and young adults. Using a safe protocol and telemedicine allowed continuance of this essential healthcare service during the COVID-19 pandemic.

disease or albinism, with no genetic diagnosis, (b) individuals without IED seek genetic counseling due to family history of IED with or without genetic diagnosis, and (c) patients affected with IED genetically tested in a research setting and referred to genetic counseling upon learning the cause of their IED.

The pretest counseling session for patients from the first two groups includes explanations about the benefits and limitations of genetic tests, different options for genetic tests (e.g., targeted test for founder mutations, gene panel, or whole-exome/genome sequencing), explanations of possible results and potential implications of results for patients and their relatives, and cost of testing and expectation regarding insurance coverage. The first counseling session in our clinic for IED patients who participated in genetic testing in a research setting is naturally different, with less focus on different options for genetic testing, as they were already tested. Nevertheless, it is highly recommended for these patients to be tested again, in a clinical setting, to validate the research results. Post-test counseling sessions are usually held one or two months later, upon receiving the genetic test results. The genetic test results are explained, and if necessary, additional genetic tests are offered to the patient, his or her reproductive partner, or other family members.

## 3 | RESULTS

During the studied period, 183 genetic counseling sessions were done, and 33/183 were done using telemedicine (Table 1). Genetic testing was completed as advised in 105/150 (70%) of face-to-face visits and 21/33 (63%) of telephone counseling sessions. In the pre-pandemic year of 2019, genetic testing was completed in 103/123 (84%) of cases.

During the first lockdown in March and April, there was a marked decrease in number of patients using the clinic's services. Starting in April, telephonic genetic counseling sessions were offered and in May, most counseling sessions were telephonic (16/22) (Table 1).

**TABLE 1** Genetic counseling practice in inherited eye diseases during 2020 COVID-19 pandemic

| 2020/Month | In-person counseling | Genetic testing done (%) | Telephone counseling | Genetic testing done (%) | Total counseling |
|------------|----------------------|--------------------------|----------------------|--------------------------|------------------|
| January    | 13                   | 8 (61%)                  | 0                    | -                        | 13               |
| February   | 25                   | 20 (80%)                 | 0                    | -                        | 25               |
| March      | 5                    | 4 (80%)                  | 0                    | -                        | 5                |
| April      | 2                    | 2 (100%)                 | 4                    | 2 (50%)                  | 6                |
| May        | 6                    | 5 (83%)                  | 16                   | 12 (75%)                 | 22               |
| June       | 11                   | 9 (81%)                  | 8                    | 5 (62%)                  | 19               |
| July       | 9                    | 6 (66%)                  | 0                    | -                        | 9                |
| August     | 11                   | 8 (73%)                  | 3                    | 1 (33%)                  | 14               |
| September  | 10                   | 6 (60%)                  | 0                    | -                        | 10               |
| October    | 16                   | 9 (56%)                  | 1                    | 1 (100%)                 | 17               |
| November   | 25                   | 17 (68%)                 | 0                    | -                        | 25               |
| December   | 17                   | 11 (65%)                 | 1                    | 0                        | 18               |
| Total      | 150                  | 105 (70%)                | 33                   | 21 (63%)                 | 183              |

The use of telephonic counseling was very high and all patients were happy to take this option and avoid reaching the hospital where a possible expose to COVID-19 could occur. Some patients were asked to come to an in-person counseling session due to special situations in which the rest of the multidisciplinary team was needed as part of the diagnosis/emotional support and others. Regarding presumed COVID-19-free individuals who opted to come to clinics, a protocol was followed to define risk levels of each patient regarding COVID-19. Assessment of body temperature and a basic questionnaire to identify patients with possible exposure to COVID-19 were administered. The questionnaire included questions about symptoms (fever, dry cough, sore throat, diarrhea, headache, loss of taste/smell) and proximity to a confirmed COVID-19 case as well as travel abroad history. Face-to-face visits for patients with suspicious symptoms were swapped with telemedicine, based largely on telephone calls.

Personal protective equipment included the use of surgical masks and hand hygiene measures both for patients and healthcare personnel. Academic and administrative meetings were conducted over video chatting. Clinical rotations for students of genetic counseling were reduced during national lockdown based on the level of infections that the country was presently experiencing.

Staff members were routinely tested every 14 days to rule out COVID-19 and quarantined in case of contact with a patient confirmed positive for COVID-19 infection. Two separate negative PCR tests were required to be performed prior to returning to the clinic. No workers in this multidisciplinary center were infected by COVID-19 during 2020, thanks in part to this protocol used for safe practice.

## 4 | DISCUSSION

The practice of genetic counseling in regard to IED in the era of COVID-19 can be effective and safe. Despite the high risk of

infectivity that threatens health workers, safety measures adopted to reduce the risk of infection among healthcare providers allowed us to prevent the cancelation of ophthalmic genetic counseling, while prioritizing patient care.

The use of telemedicine was a very useful tool for providing counseling. Before COVID-19, we did not have any experience with telemedicine. When telephone counseling was offered, most of our patients were open to telephone call encounters.

Visually impaired patients did not report to have any challenges with telemedicine. In special cases, where learning the cause and implications of an IED could possibly cause emotional stress, patients were encouraged to come in person for genetic counseling so that emotional support could be provided to the family during this difficult moment, if needed. Other situations in which we requested patients to physically come in are when a new low vision patient with an uncertain diagnosis or in need of support in their daily activities needed the multidisciplinary assistance we provide at our center.

Since June 2020, we went back to almost all normal activities in our center, despite the new partial lockdowns that did not include medical services. The service of in-person counseling was offered relatively more frequently as a full multidisciplinary team, compared to telephonic counseling. The use of telephonic counseling was high when offered, but since June 2020 we only offered it for special cases.

In the first months since 2021, we find ourselves more organized in terms of offering telephonic counseling in certain cases where the full multidisciplinary team is not needed, such as in cases where patients just need confirmation of genetic testing results which was done in a research setting, or a known IED in other family members.

In the near future, we expect to improve our telemedicine services by using videoconference services on top of telephonic consultations. The younger generation, accustomed to smartphones

use and teleconferencing, is largely comfortable with such interactions (da Luz, 2019; Newton, 2014). In addition, the current work environment is highly advanced in technology, with video-equipped computers, high-resolution cell phone cameras, and fast broadband internet service (Lurie & Carr, 2018).

Despite the COVID-19 pandemic, we managed to maintain 91.5% (183 genetic counseling sessions were done from 200 annual available spots) of our total annual capacity during 2020, reaching even a higher number than in 2019 (123 genetic counseling sessions). This fact might imply that the need for genetic counseling for IED is on the rise. Patient agreement to proceed with genetic testing stayed about the same when comparing telemedicine (63%) with in-person counseling (70%), but somewhat lower when compared to 2019 (84%). Cost for telemedicine genetic counseling was completely covered by the public health system in Israel in the same manner as in-patient consultation.

Even though we have a relatively small patient volume in our ophthalmic genetic counseling service, this is a unique service in our country that might make a difference in terms of prevention and/or treatment in some cases, in families affected by a blinding genetic eye disease. Moreover, because many of our patients needing genetic counseling are visually impaired or blind, the use of telemedicine might be especially useful and convenient by helping these patients receive care from their homes. Looking ahead to better times, we plan to continue offering this service to our patients. A limitation of our work was the retrospective nature of the study, leaving us with some missing interesting information such as comparing 'in-person visits' to a 'telemedicine encounters' in terms of satisfaction level among patients.

## 5 | PRACTICE IMPLICATIONS

The long-term impact of COVID-19 pandemic on public health is yet to be seen. Re-structuration of health care in general, and of the genetic counseling service in particular, is needed in order to continue to provide patients with the assistance needed. We may find that among our low-vision patients, widespread acceptance of telephone genetic counseling continues even though the pandemic declines.

### AUTHOR CONTRIBUTIONS

Claudia Yahalom was involved in conception, design, writing, interpretation, and revising. Michal Macarov was involved in data acquisition, analysis, interpretation, and revising. C. Yahalom and M. Macarov confirm full access to all the data in the study. Nina Schneider and Avital Eilat were involved in acquisition of data, drafting, analysis, and revising. All of the authors gave final approval of this version to be published and agree to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved.

## COMPLIANCE WITH ETHICAL STANDARDS CONFLICT OF INTEREST

C. Yahalom, M. Macarov, N. Schneider, and A. Eilat declare that they have no conflicts of interest.

## HUMAN STUDIES AND INFORMED CONSENT

The data reported here was collected as part of quality improvement measures and does not constitute research requiring IRB review. No human studies were carried out by the authors for this article.

## ANIMAL STUDIES

No nonhuman animal studies were carried out by the authors for this article.

## DATA AVAILABILITY STATEMENT

Data available within the article.

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