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Evaluating the Feasibility of Genetic Counselling as a Telehealth Service

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Abstract: The COVID-19 pandemic profoundly impacted healthcare delivery, including the traditionally face-to-face practice of genetic counselling. Telehealth emerged as an innovative solution, offering remote genetic counselling services while minimizing infection risks. This study evaluates the feasibility of genetic counselling as a telehealth service during the pandemic; focusing on patient satisfaction, session outcomes, and the broader implications for accessibility and efficiency in healthcare delivery. Five cases spanning various specialties were analyzed, highlighting high patient satisfaction and a preference for online counselling. This research underscores the transformative potential of telehealth in genetic counselling and identifies areas for future research, such as privacy concerns and long-term outcomes.

Keywords*: Telehealth Services, Genetic Counselling, Covid Pandemic, Genetic Disorders, Healthcare Accessibility.*

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I. INTRODUCTION

Genetic counselling, a critical component of personalized medicine, assists individuals and families in understanding and managing the implications of genetic conditions. (Middleton *et al*., 2017) Its scope includes interpreting genetic risks, facilitating decision-making about genetic testing, and offering psychological support. Genetic Counselling is a nascent branch in the field of Human Genetics and medicine (Seller, 1982). The term genetic counselling was coined by Sheldon Clark Reed in 1947 (Anderson, 2003). Since then the field of Genetic Counselling has grown exponentially. The first course of Genetic Counselling was started at Sarah Lawrence College in New York in 1969 and in 1979 National Society of Genetic Counselors (NSGC), New York, USA was founded (Heimler, 1997). Traditionally conducted in clinical settings, genetic counselling relies heavily on personal interactions to establish trust and deliver sensitive information effectively.

The advent of the COVID-19 pandemic disrupted healthcare systems globally, creating challenges for in-person consultations. Social distancing measures and the risk of viral transmission necessitated alternative approaches to healthcare delivery. In 1974, at a workshop by National Genetics Foundation, Inc., the following definition of Genetic Counselling was adopted "Genetic Counselling is a communication process which deals with human problems associated with the occurrence, or the risk of occurrence, of a genetic disorder in a family" (Fraser, 1974). The currently accepted definition is "Genetic Counselling is the process of helping people understand and adapt to the medical, psychological and familial implications of genetic contributions to the disease. (Mohanty and Das, 2011). This process integrates the following: interpretation of medical and familial histories to assess the chance of disease occurrence or recurrence; Education about inheritance, testing, management, prevention, resources and research; Counselling to promote informed choices and adaptation to the risk or condition" (Resta *et al*., 2006).

In India, the Board of Genetic Counselling India (BGCI) established in 2014 certifies Genetic

Counsellors and maintains professional standards (Ygo *et al*., 2018; Abacan *et al*., 2019). Till date the number of genetic counsellors around the world is much less than the demand but it is consoling to know that every year new members are added to the fraternity.

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Telehealth, defined as the use of communication technologies to provide remote healthcare services, became a cornerstone of medical practice during this period (Tuckson *et al*., 2017). Telehealth's application in genetic counselling addressed key barriers to care, such as geographical inaccessibility and limited availability of trained genetic counselors. (Dorsey and Topol, 2016). By leveraging video conferencing and digital tools, telehealth allowed patients to receive genetic counselling from the safety and convenience of their homes. (Bennett *et al.*, 2008; Human Genetics Society of Australasia, 2008; Meiser *et al*., 2008). This transition not only preserved the continuity of care during the pandemic but also opened new avenues for enhancing accessibility and efficiency in genetic counselling. (Deldar, 2016; Wessels *et al*., 2021).

Despite its potential, the shift to telehealth posed challenges, including technological limitations, privacy concerns, and the inability to perform physical examinations. (Rao *et al*., 2021). This study aims to evaluate the impact of telehealth on genetic counselling during the pandemic, focusing on patient experiences, session outcomes, and the feasibility of remote counselling as a sustainable model for the future.

II. METHODOLOGY STUDY DESIGN

This study employed a descriptive design to evaluate patient outcomes and experiences with telehealth-based genetic counselling during the COVID-19 pandemic. The research was conducted at a genetic counselling centre that transitioned to telehealth to maintain service continuity.

A. Patient Recruitment

Patients were referred to the genetic counselling department by healthcare providers across India. Referrals were received through email or phone, and eligible patients were contacted to confirm their participation. Inclusion criteria included:

- Diagnosis or suspicion of a genetic condition.
- Desire for information about genetic testing, family history, or genetic risks.
- Concerns about prenatal screening results, reproductive risks, or hereditary conditions.

B. Telehealth Procedure

Genetic counselling sessions were conducted via Zoom, with links shared one day before the appointment. Patients were provided with guidelines on preparing for the session, including gathering medical reports and family history details. The sessions followed a structured format:

➢ *Pre-session Preparation:*

- Collection of demographic information and referral details.
- Verification of eligibility based on medical history and referral criteria.

➢ *Counselling Session:*

- Discussion of the session's agenda and patient expectations.
- Collection and analysis of genetic and medical information.
- Explanation of genetic risks, inheritance patterns, and testing options.
- Addressing patient concerns in a supportive and non-coercive manner.

➢ *Post-session Documentation:*

- Summary of session outcomes and recommendations shared with patients.
- Scheduling follow-up appointments as needed.

C. Data Collection

Feedback was collected using a structured questionnaire, which included:

- Ratings of the online session (1 = poor, 5 = excellent).
- Preferences for online versus in-person counselling.
- Open-ended questions about session convenience and satisfaction.

D. Ethical Considerations

Informed consent was obtained from all participants. Data was anonymized and stored securely to ensure patient confidentiality.

III. DATA ANALYSIS

A. Patient Demographics and Case Distribution

Five cases were analyzed, spanning specialties such as pediatrics, prenatal counselling, oncology, and pre-conception genetic counselling.

Table 1 Summarizes the Patient Demographics and Session Details.

Patient ID	Age	Reason for Referral	Session Type	Specialty
GC-2022-001	27	High-risk maternal marker report	Pre-test	Prenatal
GC-2022-002	32	Newborn screening indicating G6PD deficiency	Post-test	Pediatric
GC-2022-003	45	Pathogenic BRCA2 mutation	Post-test	Oncology
GC-2022-004	30	History of medical termination for triploid fetus	Pre-test	Pre-conception
GC-2022-005	40	Child with hypoplastic nose and café-au-lait spots	Pre-test	Pediatric

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B. Session Outcomes

Outcomes were analyzed based on patient feedback, follow-up requirements, and session types.

Table 2 Outlines Session Recommendations and Follow-Up Needs.

Patient ID	Recommendations	Follow-Up Needed
GC-2022-001	Non-invasive prenatal testing (NIPT)	Yes
GC-2022-002	Dietary restrictions for G6PD deficiency	No
GC-2022-003	Family-wide BRCA testing	Yes
GC-2022-004	Couple's karyotype	Yes
GC-2022-005	Next-generation sequencing (NGS)	Yes

C. Patient Feedback and Preferences

Patients rated their online counselling experiences on a scale of 1 (poor) to 5 (excellent). All participants reported ratings of 4 or 5, indicating high satisfaction. Additionally, all five patients expressed a preference for online sessions over in-person consultations.

IV. RESULTS AND DISCUSSION KEY FINDINGS

- *High Patient Satisfaction:*

All participants rated their telehealth sessions positively (**Table 3**), with 60% giving the highest possible rating (**Figure 3**).

- *Preference for Telehealth:*

Despite the availability of in-person counselling, all patients expressed a preference for telehealth due to convenience and accessibility.

- *Diverse Case Distribution:*

Pediatric cases constituted the majority, possibly reflecting parental concerns about exposing children to public spaces during the pandemic (**Figure 1).** 60% of the cases referred were pre-test counselling cases showing that there was no bias to obtain in-person consultation when there might be a need to give sample. **(Figure 2**).

- *Effective Follow-Up:*

Four out of five cases required follow-up, highlighting the importance of continuity in genetic counselling, which telehealth effectively facilitates.

Table 3: Patient Feedback

S. No.	Patient ID	Feedback for online sessions
1	GC-2022-001	Satisfactory experience
2	GC-2022-002	Helpful guidance
3	GC-2022-003	Excellent experience
4	GC-2022-004	Convenient, easy and helpful
5	GC-2022-005	Convenient, satisfactory and time saving

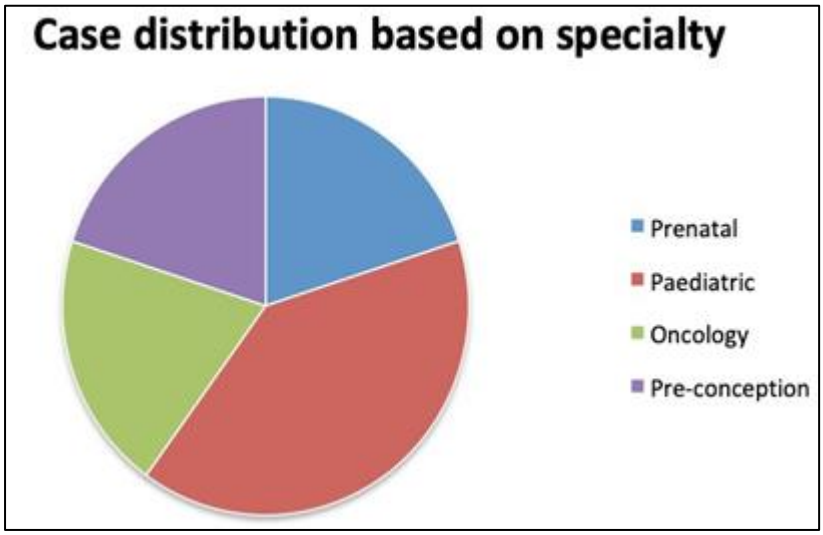

Fig 1: Distribution of Cases based on Specialty

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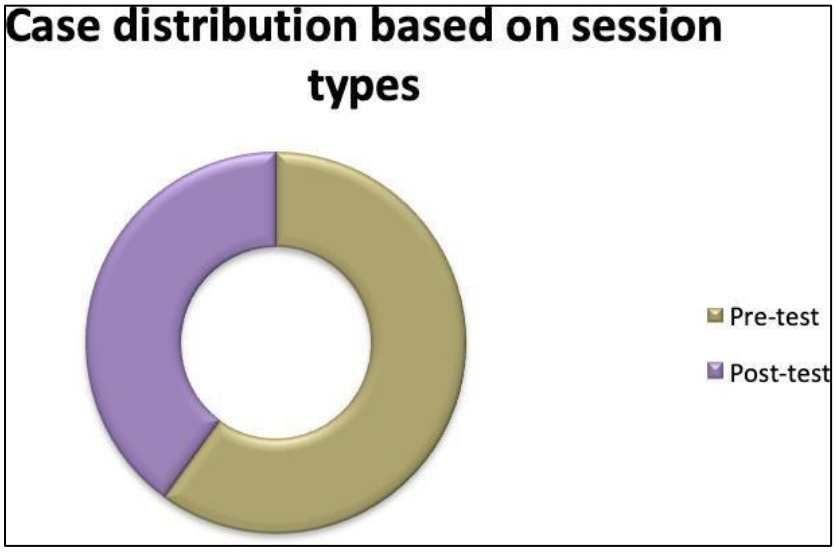

Fig 2: Distribution of Cases based on Session Types

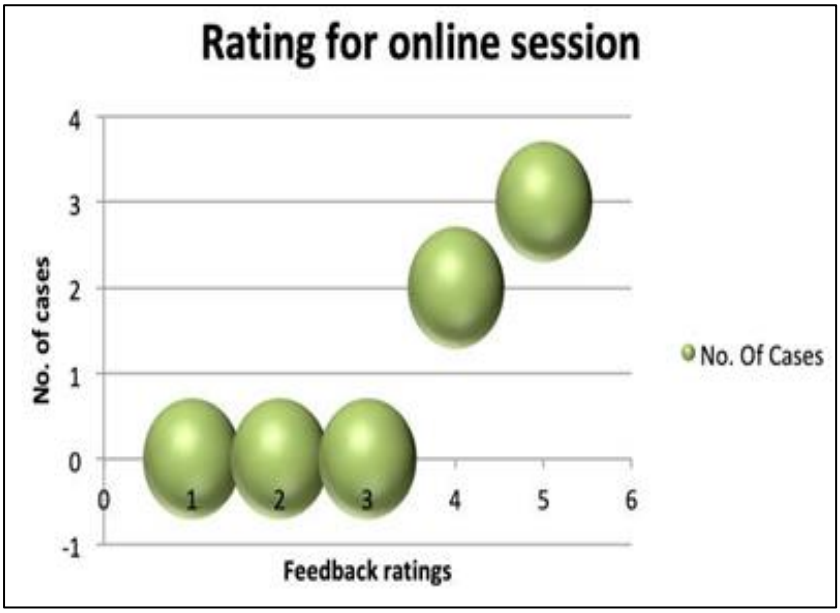

Fig 3: Ratings Given by Patients for Online Genetic Counselling Sessions

V. DISCUSSION

Telehealth has proven to be a valuable alternative to traditional counselling methods, particularly during the pandemic. Its benefits include enhanced accessibility, reduced travel time, and minimized exposure to infectious diseases. However, challenges such as technological barriers and the inability to perform physical examinations warrant consideration.

Future research should explore long-term outcomes of telehealth genetic counselling and its effectiveness compared to in-person sessions. Additionally, integrating telehealth with innovations such as remote sample collection could further enhance its utility.

VI. CONCLUSION

The COVID-19 pandemic accelerated the adoption of telehealth in genetic counselling, demonstrating its feasibility and acceptance among patients. This study highlights telehealth's potential to transform genetic counselling by improving accessibility and convenience without compromising the quality of care. Expanding this approach beyond the pandemic could bridge gaps in healthcare delivery, particularly in underserved regions.

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