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Italian Melanoma Intergroup tele-genetic counseling project for melanoma predisposition syndromes: five-year evaluation of effectiveness and outcomes

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Ethics approval and consent to participate: the study was conducted in accordance with the Declaration of Helsinki. Patient data were collected anonymously via a self-administered questionnaire that was not traceable to individual identities, ensuring confidentiality. Given the non-interventional design and full anonymization, ethics committee approval was waived.

Consent for publication: not applicable

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Abstract

This study assessed the implementation of a tele-genetic counseling (TGC) pathway for hereditary melanoma risk using the culturally adapted Italian Melanoma Intergroup Tele-Genomics Outcome Scale (IMI-TGOS) questionnaire. Among 278 eligible patients, 177 (64%) completed the survey across three Italian centers. Responses showed high comprehension and perceived utility: 88% of participants reported understanding the information received, 96% recognized familial implications, and over 85% felt confident in managing risk and making decisions. While emotional responses were more varied, most respondents expressed confidence in future planning. These findings confirm the feasibility, acceptability, and informativeness of the IMI-TGOS in a remote counseling setting. The study supports the comparability of tele-genetic and in-person counseling in terms of understanding, retention, and decision-making. Moreover, the IMI-TGOS emerged as a promising tool for evaluating real-world genetic services. Broader adoption may help monitor outcomes and improve quality and equity in the expanding landscape of digital genetic care.

Introduction

Genetic counseling and associated testing services are increasingly acknowledged for their pivotal role in delivering critical information and meaningful clinical benefits to individuals and families affected by conditions with a suspected genetic basis. A growing body of literature has consistently demonstrated that genetic counseling improves patient outcomes by enhancing knowledge, refining risk perception, and reducing anxiety and decisional conflict. Moreover, it promotes positive health behaviors – such as increased adherence to surveillance protocols – and reinforces individuals' sense of personal control over their health trajectory.[1] Given these multifaceted effects, the comprehensive evaluation of genetic counseling services must encompass a broad array of variables across all stages of the process, including both pre-test and post-test phases. Key outcomes to assess include knowledge acquisition, information retention, reproductive intentions, decisional clarity, levels of psychological distress, patient satisfaction, perceived risk, empowerment, behavior change, and decisional conflict.[2] To support and standardize such evaluations, the Genetic Counseling Outcome Scale (GCOS-24) was developed in 2011 as a patient-reported outcome measure (PROM) specifically designed for clinical genetics services.[2] The GCOS-24 is grounded in the theoretical construct of empowerment – a multidimensional concept comprising five interconnected domains: cognitive control, decisional control, behavioral control, emotional regulation, and hope. The scale has been rigorously validated, demonstrating strong psychometric properties and broad applicability in assessing the quality and effectiveness of genetic counseling.[2] To enhance feasibility in routine clinical practice and reduce respondent burden, a shortened 6-item version of the GCOS was

introduced in 2019 (The Genomics Outcome Scale [GOS]). This abbreviated form retains the core theoretical construct of empowerment and offers a practical, scalable tool for clinical audits and service evaluations.[3] Despite these advancements, the integration of standardized outcome measures into genetic counseling practice remains limited in Italy. At the European level, comprehensive evaluation frameworks are still under development, with efforts to promote broader adoption ongoing but incomplete.[4] The application of validated tools such as the GCOS in real-world outpatient settings thus represents a crucial step toward improving the quality, accountability, and patient-centeredness of genetic services.

This study builds upon the initiative of the Italian Melanoma Intergroup (IMI), which, since June 1, 2019, has implemented a telemedicine-based approach to deliver tele-genetic counseling (TGC) services to selected patients. The primary aim of the program is to identify individuals eligible for genetic testing for hereditary melanoma syndromes or other cancer predisposition syndromes in which melanoma is a significant associated malignancy.[5-7] To underscore the scale and impact of this initiative, which has expanded access to modern genetic services in regions historically underserved, data as of December 31, 2023, indicate that over thirty IMI-affiliated specialists submitted 640 TGC requests across nine Italian regions (Lombardy, Tuscany, Lazio, Veneto, Liguria, Marche, Apulia, Campania, and Sicily). Of these, 559 genetic tests were proposed, and 422 post-test consultations were completed. These five-year outcomes confirm the effectiveness of the program in enhancing equitable access to genetic counseling and multigene testing through telemedicine. The initiative has supported nationally recognized referral centers, reduced patient mobility, and fostered continuity of care through collaboration with local healthcare professionals.[6]

Within this framework, the IMI tele-genetic counseling project also aimed to pilot the implementation of structured evaluation tools to assess both the effectiveness and acceptability of TGC among patients. The goal was to obtain measurable feedback on the utility of TGC, its impact on patient comprehension, perceived clinical benefit, acceptance of surveillance recommendations, and increased awareness among patients and their families. Importantly, outcome assessment tools were also intended to help identify potential weaknesses in the counseling process, thereby enabling the development of targeted quality improvement strategies.

Materials and Methods

A rigorously translated and culturally adapted version of the GOS,[3] the Italian Melanoma Intergroup Tele-Genomics Outcome Scale (IMI-TGOS) was used to ensure conceptual and linguistic equivalence for the Italian population. A pilot study was conducted at the IRCCS Ospedale Policlinico San Martino in Genoa, drawing upon successful methodologies developed by other

research groups.[8] The instrument was tested on a heterogeneous case series, encompassing a broad range of diagnostic indications, to assess its applicability across diverse clinical scenarios and real-world outpatient settings. Although no formal validation study was conducted, the aim was to implement a previously validated instrument in a clinical context, in order to collect structured feedback from Italian-speaking patients receiving tele-genetic counseling. For this survey, a total of 278 patients were enrolled from the three IMI-affiliated centers most actively involved in delivering the TGC service. These patients were invited by their referring clinicians to independently complete the culturally and linguistically adapted Italian version of the Genomics Outcome Scale (IMI-TGOS), following their post-test consultation. Patient privacy and data protection were ensured through anonymization of all responses. The anonymized data were subsequently centralized and processed at the Cancer Genetics Unit of IRCCS Ospedale Policlinico San Martino in Genoa, Italy. Participation in the survey was entirely voluntary. Patients were explicitly informed that their responses should reflect their honest personal experience, that completing the questionnaire would not influence their clinical care, and that they were welcome to add open-ended comments to complement the quantitative items. The IMI-TGOS questionnaire consisted of six items, each rated on a 5-point Likert scale (ranging from *Absolutely no* = 1 to *Absolutely yes* = 5). The items were designed to evaluate key domains of patient empowerment and understanding within the context of genetic counseling. Specifically:

- **Item 1** assessed the patient's perceived ability to understand and process the information received during the counseling session, including the potential to communicate it effectively to others.
- **Item 2** evaluated the level of comprehension regarding the hereditary nature and transmission of the identified genetic risk.
- **Item 3** explored the presence or absence of emotional distress or concern related to the possible clinical implications of the test results.
- **Item 4** measured the clarity and understanding of the surveillance program recommended following the genetic test.
- **Item 5** gauged the patient's attention to personal and familial health implications, highlighting the extension of awareness to relatives.
- **Item 6** focused on the perceived impact of the genetic information on the patient's ability to plan for the future.

Results

Out of 278 eligible patients who received TGC across the three most actively involved IMI centers, 177 completed the IMI-TGOS survey, yielding a response rate ranging from 31% to 80% across centers. Among respondents, 15 (8.5%) had either positive or uncertain genetic test results (*i.e.*, pathogenic variants or variants of uncertain significance). Responses to each of the six IMI-TGOS items are summarized in Figure 1.

Regarding item 1, which assesses understanding and the ability to reprocess and communicate information about their condition, 91 patients (51.4%) answered "*5 - Absolutely yes*", and 64 (36.2%) selected "*4 - More yes than no*". Overall, nearly 88% of respondents reported a high level of comprehension. Only 4 patients (2.3%) selected "*1 - Absolutely no*", and 2 (1.1%) chose "*2 - More no than yes*". For item 2, which evaluates awareness of the possible hereditary nature of the condition, 149 patients (84.2%) selected "*5 - Absolutely yes*", and 21 (11.9%) chose "*4 - More yes than no*". This indicates that 96% of respondents understood the implications for family members. Very few (3, 1.7%) selected "*1 - Absolutely no*", and none chose "*2 - More no than yes*". Item 3, which assesses emotional distress related to the condition, received more variable responses. A total of 72 patients (41%) expressed concern: 33 (18.6%) selecting "*5 - Absolutely yes*" and 39 (22.0%) "*4 - More yes than no*". In contrast, 71 patients (40.1%) reported low or no worry: 39 (22.0%) chose "*2 - More no than yes*" and 32 (18.1%) "*1 - Absolutely no*". Additionally, 34 (19.2%) selected the neutral option "*3 - Neither yes nor no*". For item 4, which measures understanding of the proposed surveillance measures, 118 patients (66.7%) responded "*5 - Absolutely yes*", and 35 (19.8%) "*4 - More yes than no*", totaling 86% reporting strong awareness. Only 5 (2.8%) answered "*1 - Absolutely no*". For item 5, which assesses the broader psychological impact of the information received, 132 patients (74.6%) selected "*5 - Absolutely yes*" and 34 (19.2%) "*4 - More yes than no*". Only 2 patients (1.1%) selected "*1 - Absolutely no*", and one (0.6%) did not answer. Finally, item 6, which reflects perceived decisional autonomy, received "*5 - Absolutely yes*" from 114 patients (64.4%) and "*4 - More yes than no*" from 42 (23.7%). Only 3 (1.7%) reported "*1 - Absolutely no*".

Discussion

This study, which utilizes the translated and culturally adapted GOS[3] (IMI-TGOS) within a TGC setting, offers valuable insights into patient empowerment and understanding following remote genetic counseling. The methodology, based on clinician-administered surveys, strict anonymization, and centralized data processing, ensures the validity and reliability of the findings. Patients were explicitly assured of the anonymity and non-influence of their responses on care, a crucial factor for honest and unbiased reporting. High rates of positive responses regarding understanding and re-

elaboration of information, as well as awareness of familial risk, indicate a solid transmission of knowledge. The findings suggest that the TGC service is effective in conveying complex genetic information. These aspects are central to the construct of empowerment and reflect positively on the ability of remote counseling to achieve core objectives typically associated with in-person genetic services. The results are consistent with prior literature showing that TGC can match in-person counseling in effectiveness, supporting its broader adoption.[9] Notably, emotional responses – such as worry about personal or familial health risks – were more heterogeneous. While a substantial proportion of patients reported concern, others expressed neutral or minimal emotional reactions. This variability likely reflects differences in personal and family histories, emotional resilience, or stage in the diagnostic process. It highlights the importance of further research, particularly on larger samples and stratified by clinical presentation, to explore the psychological dimensions of genetic counseling and their determinants. Strong results were also observed in areas related to behavioral control and future planning. The large majority of patients reported understanding the proposed surveillance strategies and feeling empowered to make health-related decisions. These findings underscore the success of the counseling model not only in delivering information but also in fostering a sense of agency. This essential outcome translates into proactive health behaviors and improved family communication. Importantly, the use of the adapted GOS (IMI-TGOS) proved feasible and informative within the real-world context of an Italian telemedicine service. Its application provides a replicable framework for evaluating genetic services and facilitates international comparisons. The data presented here offers a valuable baseline for future evaluations and service optimization.

The IMI initiative[7] has effectively broadened access to genetic expertise across multiple regions, mitigating traditional barriers of geography and availability. High rates of test uptake and follow-up consultations, combined with favorable patient-reported outcomes, support the clinical and organizational feasibility of this approach. Moreover, the detection of pathogenic variants in 8.1% of patients confirms the utility of TGC in identifying individuals eligible for targeted prevention and monitoring programs. Nonetheless, this analysis is not without limitations. The sample size and the relatively small proportion of patients with positive or uncertain results constrain the generalizability of the findings. Furthermore, although the GOS questionnaire was carefully translated and culturally adapted, the Italian version used in this study was not subjected to a formal psychometric validation process, which may limit the interpretability and comparability of the results. However, the cohort was homogeneous in terms of diagnostic suspicion and followed a consistent counseling pathway, which supports the internal validity of the results. A formal validation study of the Italian version of the GOS could prove valuable in future research, enabling broader adoption and ensuring

psychometric robustness in the Italian-speaking population. Future studies should aim to explore subgroup differences more deeply, particularly those related to psychological impact and behavioral outcomes.

Conclusions

The use of the culturally adapted GOS (IMI-TGOS) within the IMI tele-genetic counseling framework proved to be both feasible and informative. The high response rate and the patients' engagement with the survey suggest that administering a validated questionnaire for evaluating the counseling and testing pathway is acceptable and effective, even in a remote setting. This allowed for an initial assessment of the impact of a tele-genetic service that has been operating for five years across multiple centers in Italy. Although the current sample size does not yet allow for meaningful subgroup analyses – particularly regarding psychological outcomes among patients with positive *versus* inconclusive test results – the overall findings indicate that patients are generally able to understand, retain, and manage the information conveyed through TGC. This supports existing evidence on the comparability between tele-genetic and in-person counseling, especially in terms of information acquisition, comprehension, and decision-making capacity.[9] Furthermore, the study highlights the potential of the IMI-TGOS as a practical tool for outcome evaluation in real-world clinical genetics services. Its successful application in the Italian context provides a foundation for future use in larger populations and for comparisons across different care models. Continued assessment and refinement of such tools and services will be essential to ensure the quality, accessibility, and equity of genetic care in a progressively digital healthcare landscape.

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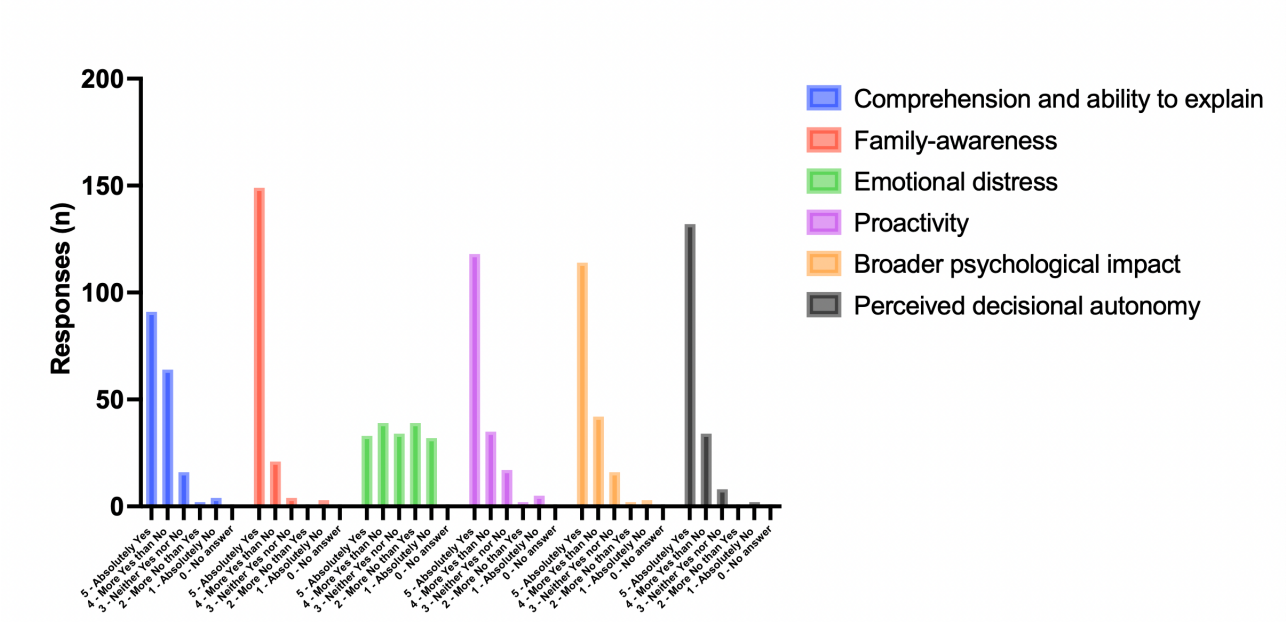

Figure 1. Patient responses to IMI-TGOS questionnaire, grouped by answer type and distributed across individual items (Items 1-6). Each color represents a specific item. The x-axis shows the response categories, ranging from "*Absolutely yes*" to "*Absolutely no*" and "*No answer*", while the y-axis indicates the number of responses (n). The items respectively assess: (1) Comprehension and ability to explain, (2) Family-awareness, (3) Emotional distress, (4) Proactivity (understanding of surveillance measures), (5) Broader psychological impact, and (6) Perceived decisional autonomy.