

Quality indicators in identification and care of patients with genetic predisposition to cancer: An AIFET proposal

Tumori Journal
1–7
© Fondazione IRCCS Istituto
Nazionale dei Tumori 2026
Article reuse guidelines:
sagepub.com/journals-permissions
DOI: 10.1177/03008916261451688
journals.sagepub.com/home/tmj



**Andrea Remo^{1,2†} , Maria Grazia Tibiletti^{1,3†},
Giovan Battista Rossi¹, Ileana Carnevali^{1,4} , and
Cristina Oliani¹ on behalf of AIFET**

Abstract

Hereditary cancer genetics is increasingly recognized as an integral component of cancer care, contributing to treatment, early detection, and risk reduction strategies for patients and their relatives. In this context, several hereditary cancer programs, databases, recommendations, and guidelines have been implemented within the Italian National Health System and at hospital level. However, independent and specific quality indicators for hereditary tumor management are still lacking, limiting the ability to evaluate activities across individual centers. To address this gap, AIFET (Associazione Italiana Familiarità ed Ereditarietà Tumori) has established a dedicated scientific committee for the development of such quality indicators. Here, for the first time to our knowledge, we present a set of 37 quality indicators derived from a national Italian survey. The indicators were organised into four sections: General indicators (applicable to all syndromes), specific to hereditary breast and ovarian cancer, specific to colorectal cancer and polyposis, specific to melanoma predisposition. By promoting consistency across centres, this model may not only enhance the quality of care nationally but also foster collaboration with European institutions toward shared standards in hereditary cancer management.

Keywords

quality indicator, inherited cancer, genetic predisposition, certification, Lynch syndrome

Date received: 21 January 2026; revised: 20 April 2026; accepted: 4 May 2026

Introduction

The genetics of hereditary tumors are increasingly recognized as an integral component of cancer care, playing a fundamental role in treatment, early detection, and risk reduction for patients and their families. Identifying hereditary cancer predisposition can provide substantial benefits for the community by detecting individuals at increased risk. Specifically, such benefits include reducing the likelihood of cancer development, diagnosing tumors at an earlier and more treatable stage, informing the optimal choice of targeted therapies when cancer develops, and guiding reproductive decisions.

Genetic testing of probands—usually individuals already affected by cancer—together with cascade testing of relatives and the provision of comprehensive downstream services, can effectively reduce both cancer incidence and mortality in the population. Achieving this goal requires the establishment of dedicated services that

are adequately resourced and subject to regular quality monitoring.

In Italy, many centers currently identify genetic cancer predisposition, but some have expertise limited to specific syndromes depending on hospital activity (e.g., breast/

¹AIFET Scientific Committee for Developing Quality Indicator in Management of Patients with Genetic Predisposition to Cancer, Rome, Italy

²Pathology Unit, Service Department ULSS9 “Scaligera”, Verona, Italy

³Centro di Ricerche Tumori Eredo Familiari, Università dell’Insubria, Varese, Italy

⁴Servizio di Genetica Medica ASST Settelaghi, Varese, Italy

[†]Equal contributors.

Corresponding author:

Andrea Remo, Pathology Unit (Eusoma's certification n° 1030/00), Service Department, ULSS9 “Scaligera”, via Valverde 2, 37144 Verona, Italy.

Email: andrea.remo@aulss9.veneto.it

ovarian or colorectal cancer). In such a fragmented scenario, certain steps in the diagnostic–therapeutic pathway for inherited tumor identification and carrier management are often outsourced or lacking. This heterogeneity is generally due to two main reasons:

1. The highly specialized role of certain centers acting as tertiary referral hospitals (e.g., Breast, Prostate, or Lung Units).
2. The management of tumors requiring less specialized or more widespread care (e.g., cutaneous tumors or intestinal adenomas).

Currently, most centers operate independently, without a coordinated national framework. There is no standardized model of care for patients with hereditary cancer predisposition in Italy, nor are there multidisciplinary national guidelines for genetic assessment, testing, or management. For this reason, the definition of quality indicators (QIs) specific to hereditary cancers is essential to evaluate both the quality and the completeness of each step in the care pathway, from diagnosis to the management of at-risk relatives.

In November 2021, the Italian Association for Familial and Hereditary Tumours (AIFET, Associazione Italiana Familiarità ed Ereditarietà Tumori) was founded, building on the Italian Association for Familial and Hereditary Gastrointestinal Cancers (AIFEG), established in 2002.¹ AIFET is a multidisciplinary national society with the mission of improving the identification and management of hereditary tumors in Italy. To coordinate and strengthen familial cancer services across the country, AIFET has developed a set of specific QIs designed to measure adherence of clinical practice in this field to international standards.

The proposed QIs focus on hereditary syndromes involving breast cancer, colorectal cancer, and melanoma. These areas were selected by AIFET expert sub-panels because they are potentially relevant in every hospital, regardless of the level of specialization, and are therefore considered “Universal.”

To test the proposed QI set, all Italian institutions affiliated with AIFET were invited to participate in a survey assessing adherence to these indicators. This pilot study aims to provide the foundation for developing a robust set of indicators for future accreditation programs, designed to measure and monitor the quality of care pathways for individuals with hereditary cancer predisposition.

Methods

To identify appropriate indicators for hereditary cancers, the AIFET Committee conducted a literature review of evidence-based recommendations. Specifically, guidelines

from NCCN,^{2–3} ERN Genturis,⁴ NICE,⁵ and recommendations from the Italian Societies of Medical Oncology (AIOM), Pathology (SIAPEC),⁶ and Human Genetics (SIGU)⁷ were considered. However, no publications directly addressing this specific topic were identified. All documents were shared with experts invited to contribute to this consensus paper. The indicators identified were derived using national and international guidelines, shared by all commission members and are in agreement with therapeutic diagnostic pathways for hereditary tumors published by the Italian Ministry of Health (AGENAS).⁸

The AIFET Committee, which includes specialists in genetics, oncology, surgery, gastroenterology, pathology, and psychology, met in 2023 and agreed to develop a set of quality indicators for the following cancer predisposition syndromes⁹:

- Lynch syndrome (ORPHA 144)
- Familial polyposis syndromes (ORPHA 733, 247798)
- Hereditary breast and ovarian cancer syndrome (ORPHA 145)
- Melanoma predisposition syndrome (ORPHA 618)

According to the Agency for Healthcare Research and Quality (AHRQ) Evidence Report No. 105 (04-E030-2, 2004), the key properties of a quality measure are *reliability* and *validity*. Validity means that the indicator truly measures what it is intended to, while reliability implies that the observation is consistently reproducible—either by the same observer over time or by different observers.

Two additional properties are also relevant:

- Usability, meaning the results generated are easy to interpret and can inform actions to improve health-care delivery.
- Feasibility, meaning data collection should be straightforward during routine clinical practice, with limited costs.

The expert panel was therefore asked, whenever possible, to select indicators that were simple, clearly defined, and covered both process and outcome measures. In the context of certification, the number of QIs was deliberately kept to a minimum, with the goal of reflecting the entire diagnostic and therapeutic process while relying on readily available, systematically collected variables.

For each indicator, both its definition and assigned numerical values were reported. These values were determined based on the clinical relevance of the recommendation. The sum of all values produced a score for each participating institution.

The indicators were organized into four sections:

1. General indicators (applicable to all syndromes)
2. Indicators specific to hereditary breast and ovarian cancer
3. Indicators specific to colorectal cancer and polyposis
4. Indicators specific to melanoma predisposition

These indicators are qualitative, systemic, and syndrome-oriented. No quantitative indicators were included.

The indicators were tested across Italian centers affiliated with AIFET. To this end, each center completed a structured questionnaire, which was subsequently approved by the competent administrative authority of the respective healthcare provider.

The study was conducted in accordance with the procedures for observational studies and in compliance with the ethical principles of the Declaration of Helsinki (59th WMA General Assembly, Seoul, Korea, October 2008). The study protocol was submitted to the Ethics Committee of Verona (CESC).

Results

A total of 37 benchmark QIs that could be adopted by Italian hereditary cancer services were identified. Table 1 shows the general QIs (A) and the syndrome-specific QIs for breast and ovarian cancer (B), colorectal cancers and polyposis (C), and melanoma (D), along with the assigned values.

All Italian centers affiliated with AIFET were included in the survey through a questionnaire requiring adherence to the identified indicator. Twenty-two centers (Figure 1) submitted the completed questionnaire along with a self-declaration from the applicant. These included academic institutions, cancer care and research centres (IRCCS, Istituti di Ricovero e Cura a Carattere Scientifico), and hospitals organized within the Hub-and-Spoke model. The 37 indicators and their corresponding scores, as defined by the AIFET Committee, are listed in Table 1. Thirty-one indicators related to the identification and management of patients and their relatives affected by hereditary cancer, while six indicators focused on aspects such as exemption from co-payment (“ticket”) and access to clinical cancer genetic services (Figure 2).

Among syndrome-specific indicators, eight referred to HBOC (ORPHA 145), 11 to inherited colorectal cancers and polyposis (ORPHA 144, 733, 247798), and five to familial melanoma (ORPHA 618). For each QI, the degree of implementation of specific activities was linked to assigned scores. The sum of these scores generated a final value indicative of the quality of cancer genetic services, including diagnosis, treatment, and follow-up for both affected individuals and their healthy relatives.

The survey was conducted as a self-assessment exercise (each center evaluating itself). No external verification was performed, and the results were analyzed anonymously.

Figure 3 illustrates the national survey results. These reveal a stronger focus on breast cancer predisposition syndromes, and comparatively less attention to familial melanoma (see QIs A2, A3, A4, A5). We found that multi-disciplinary disease-related groups, where care strategies for patients are discussed, are quite widespread (95%, 72%, 41% for QIs B1, C1, and D1, respectively). However, this activity was not consistently associated with the existence of specific care pathways for the three familial cancer categories (77%, 50%, 54%, 32% for QIs A2, A3, A4, A5). This finding highlights the limited advocacy among Italian healthcare providers in making hereditary pathways visible and accessible (QIs A2–A5; A11, A12), likely reflecting difficulties in managing these patients—especially unaffected relatives—outside the immediate cancer treatment pathway (QIs B3, C6, D4).

Nevertheless, prophylactic surgery was available in all centers (100%; QIs B5, B6, C9), regardless of the type or level of the institution.

The availability of genetic testing was also very high (100%; QI A11), suggesting particular attention is paid to the diagnosis of inherited cancers. Multigene panel tests were widely used for all cancer types considered, although they were slightly less common for familial melanoma (QI B7, C11, D2: 100%, 90%, and 81%, respectively).

Interestingly, Universal Screening for MMR defects¹⁰ was more frequently available than reflex testing for *BRAF* mutations and *MLH1* hypermethylation (90% vs 81%, QIs C3 and C4). This gap indicates that in some cases the hereditary diagnostic pathway remains incomplete, as MMR deficiency (*MLH1*/*PMS2* loss) requires reflex testing (C4) to exclude sporadic carcinoma. This finding underlines the need for a new indicator to capture the rate of incomplete Lynch syndrome screening. As with other screening programs (e.g., breast cancer), detection rates are proportional to patient adherence; incomplete pathways decrease the identification rate of Lynch syndrome.

The survey also revealed limited availability of dedicated software for evaluating polyp history (QI C10; 50%) and, notably, underutilization of pathology department software that could track cumulative polyp counts (>10), which is essential for polyposis identification.

Care pathways for high-risk individuals were present in 81% of centers (QI A12). Risk-reducing surgery was available for breast and ovarian cancer in most centers, and to a lesser extent for colorectal cancer (QIs B5, B6, C8, C9). Psychological support was available in 81% of centers (QI A13). However, dedicated clinical appointment slots were available in only 54–68% of centers, depending on the cancer type and procedure (QIs B3, C6, D4).

Table I. Quality indicators (QIs), along with the corresponding numerical values, developed in this study: general QIs for cancer genetics clinics (A), and specific QIs for breast and ovarian cancer (B), colorectal cancer and polyposis (C) and melanoma (D).

Section A	Quality indicators for familial/hereditary cancer clinics: general items	Evaluation
A.1	Identification of the representative for the certification process	Name
A.2	Presence of a specific care pathway for inherited breast cancer	10
A.3	Presence of a specific care pathway for inherited colorectal cancer	10
A.4	Presence of a specific care pathway for inherited gynaecological cancers	10
A.5	Presence of a specific care pathway for inherited melanoma	10
A.6	Visibility on the website of pathways for cancer predisposition syndromes	5
A.7	Direct access to cancer predisposition syndrome services (<i>by email, telephone . . .</i>)	10
A.8	Availability on-site or on convention for Cancer Genetic Counselling service	20
A.9	Availability on-site or on convention for genetic tests to identify cancer predisposition syndromes of breast, ovarian, colorectal cancers and melanoma	20
A.10	Quality certified genetic testing laboratory	20
A.11	Availability on-site or on convention for genetic tests to identify specific genetic variants in family members	20
A.12	Healthcare pathway for subjects at high cancer risk	20
A.13	Availability of Psycho-oncology support specific to cancer predisposition syndromes	10
Section B	Quality indicators for familial/hereditary breast and ovarian cancers	Evaluation
B.1	Multidisciplinary team for inherited cancers discussion	20
B.2	Availability of procedures for exemption from co-payment for services inherited -related for carriers of pathogenic variants at high cancer risk	10
B.3	Dedicated radiology slots for screening in carriers of pathogenic variants	20
B.4	Appropriate follow-up for carriers of pathogenic variants of cancer genes	20
B.5	Availability of gynaecological risk-reduction surgery	10
B.6	Availability of breast risk-reduction surgery	10
B.7	Availability of multigene panel testing for hereditary breast/cancer, including genes other than BRCA1 and BRCA2	5
B.8	Availability of follow-up for organs (ie, pancreas, prostate) other than breast and ovary in carriers of pathogenic variants	10
Section C	Quality indicators for familial/hereditary colorectal cancer and polyposes	Evaluation
C.1	Multidisciplinary team for inherited cancers and polyposis discussion	20
C.2	Synergic cooperation between inherited cancer team and polyposis team	10
C.3	Availability of universal screening for MMR defects on colorectal and endometrial cancers and <i>alert</i> for Genetic counselling in the presence of MMR defect	20
C.4	Availability of reflex test for <i>BRAF</i> mutation and <i>MLH1</i> promoter methylation analyses	5
C.5	Availability of procedures for exemption from co-payment for services inherited -related for carriers of pathogenic variants at high cancer risk	10
C.6	Dedicated endoscopy slots for carriers of pathogenic variants of cancer genes	10
C.7	Appropriate gynaecologic follow-up for carriers of pathogenic variants	10
C.8	Availability of specific surgery procedures for ileo-rectal anastomosis and procto-colectomy with pouch	10
C.9	Availability of gynaecological risk-reduction surgery	10
C.10	Availability of dedicated software for clinical history of polyps	5
C.11	Availability of multigene panel testing for hereditary colorectal cancers and polyposis syndromes	20
Section D	Quality indicators for familial/hereditary melanoma	Evaluation
D.1	Multidisciplinary team for inherited melanoma discussion	20
D.2	Availability of multigene panel testing for hereditary melanoma	20
D.3	Availability of procedures for exemption from co-payment for services inherited -related for carriers of pathogenic variants at high cancer risk	10
D.4	Dedicated video-dermatoscopy slots for carriers of pathogenic variants	10
D.5	Availability of surveillance for organs other than the skin (ie, pancreas)	5

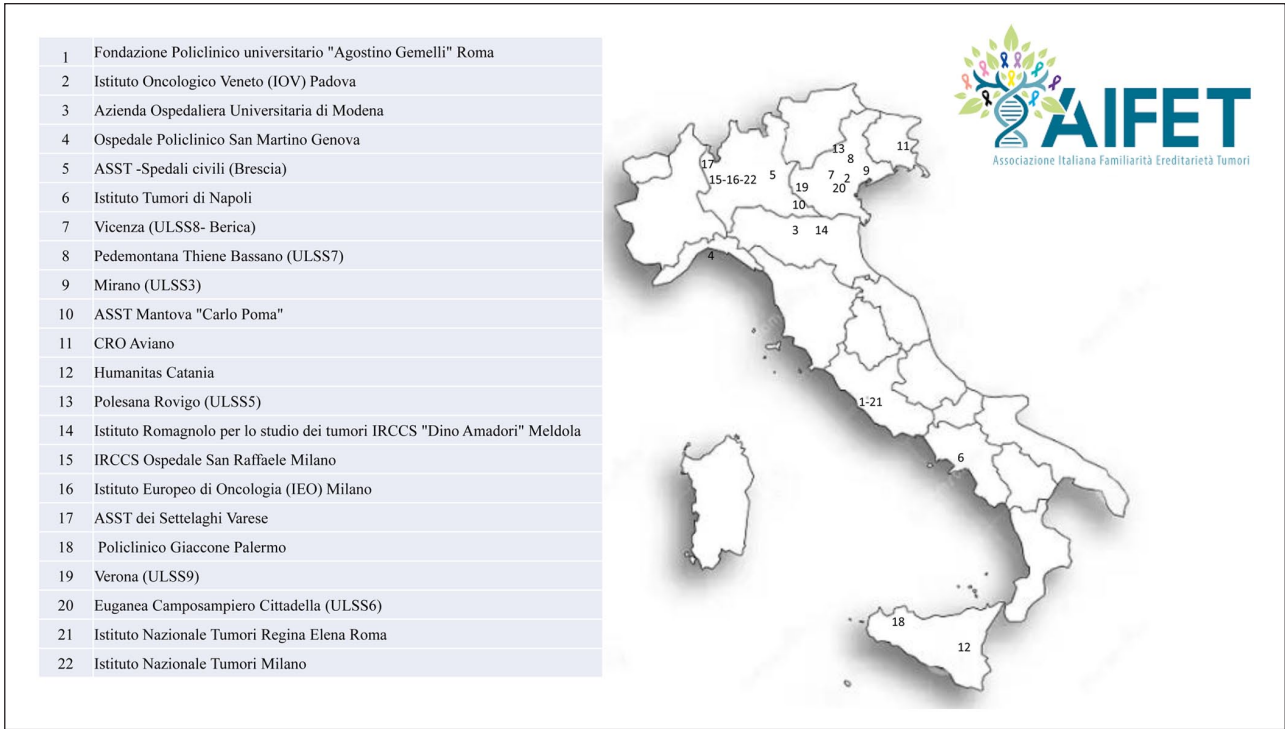


Figure 1. Geographical location of the 22 centers participating in the survey.

Italian National Health System (SSN)

The Servizio Sanitario Nazionale (SSN) is Italy’s national, universal healthcare system, established in 1978. It is guided by the principles of universality, solidarity, and equality. The SSN provides coverage for all Italian citizens and legal foreign residents, with a small co-payment for services known as the “ticket.”

Healthcare services for people with specific pathological conditions, as defined by the Livelli Essenziali di Assistenza (LEAs) and certified through exemption codes, are provided free of charge. While national-level administration ensures that the general objectives and fundamental principles are upheld, the Italian healthcare system is largely regionally based. As a result, the quality of healthcare facilities may vary across the 21 regions of Italy.

Figure 2. Definition of “ticket” within Italian National Health system (SSN).

Regarding patient accessibility to specialized centers and exemption from co-payments for genetic services, these features were consistently present across AIFET centers—an expected finding given their institutional role.

In 2024, the AIFET Steering Committee analyzed the data from the 21 centers and calculated overall scores. The mean institutional score was 362.0, ranging from 130 to 460.

Conclusions

This AIFET initiative represents the first national effort to define quality indicators (QIs) for hereditary cancer services in Italy. The proposed QIs provide a practical framework to assess and improve diagnostic and care pathways, ensuring that genetic testing, surveillance, and risk-reducing strategies are systematically implemented.

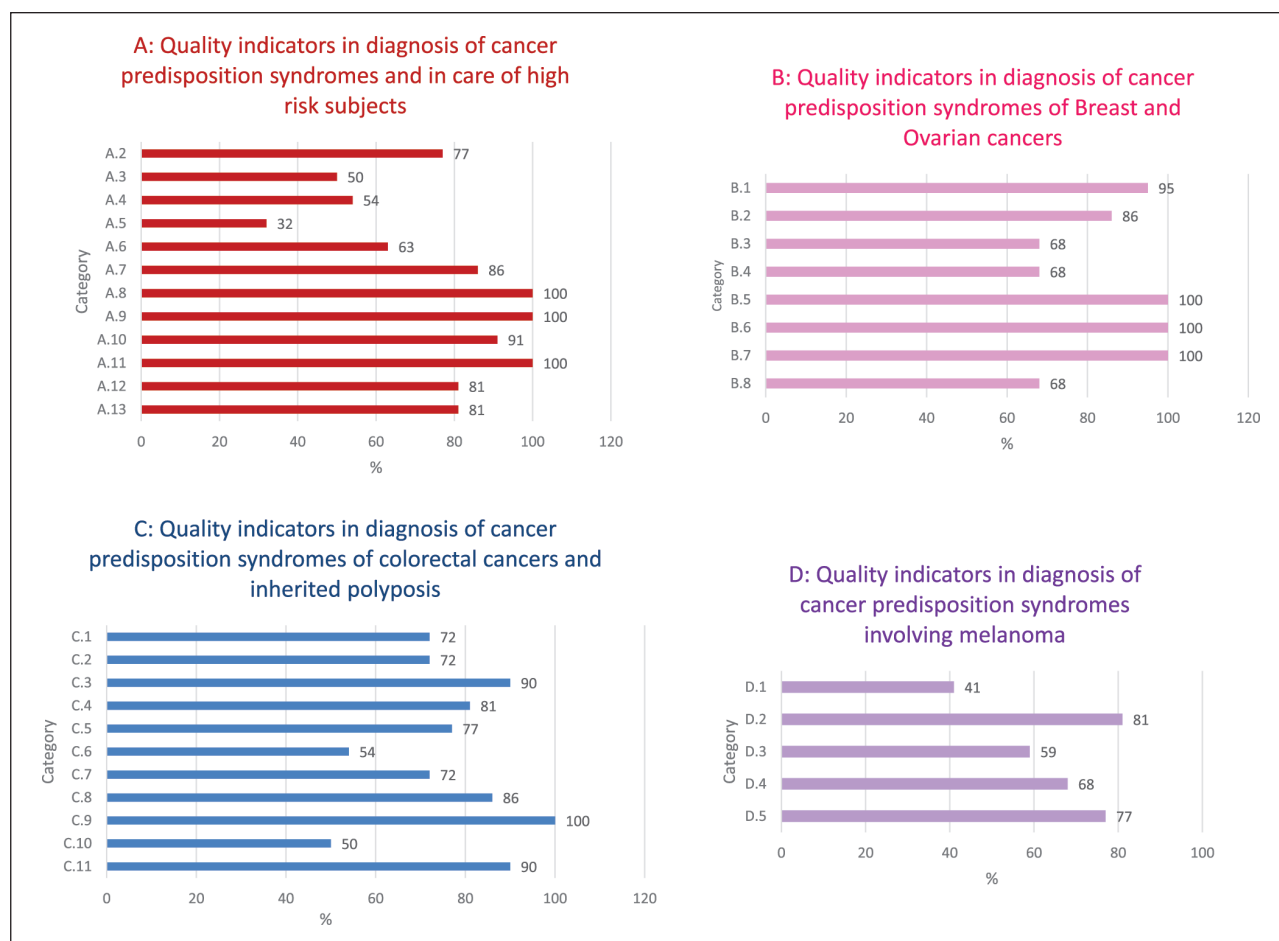


Figure 3. Results of the survey on the 37 QIs shown in Table I across national hereditary cancer centers.

By promoting consistency across centers, this model may not only enhance the quality of care nationally but also foster collaboration with European institutions toward shared standards in hereditary cancer management.

Members of Associazione Italiana Familiarità ed Ereditarietà Tumori (AIFET) participating at this survey

Genuardi M., Grillo F., Ghiorzo P., Peccatori F., Urso EDL., Puccini A., Panfili A., Turchetti D., Caligo F., Terribile D., Vitellaro M., Lucci-Cordisco E., Roncucci L., Ghiorzo P., Barana D., Romeo S., Mammi I., Adami F., Viel A., Rizzo A., Zampiga V., Cavestro M., Bonanni B., Sessa F., Russo A., Greco F., Morabito A., Stigliano V., Zovato S. Giacchè M., D'Angelo V.

Contributions

A.R. and M.G.T. conceptualized, wrote, reviewed and edited the manuscript.

G.B.R., I.C. and C.O. wrote and reviewed the manuscript.

Declaration of conflicting interests

The authors declared no potential conflicts of interest with respect to the research, authorship, and/or publication of this article.

Funding

The authors received no financial support for the research, authorship, and/or publication of this article.

Ethical approval

This study was conducted in accordance with the ethical standards of the institutional and/or national research committee and with the 1964 Helsinki Declaration and its later amendments.

ORCID iDs

Andrea Remo  <https://orcid.org/0000-0002-2564-2988>

Ileana Carnevali  <https://orcid.org/0000-0002-0511-5427>

References

1. AIFET. Home page, 2025. <https://www.aifet.it/> (accessed 18 May 2026).
2. National Comprehensive Cancer Network (NCCN). Home page, 2025. Clinical practice guidelines in Oncology for genetic/familial high risk assessment breast and ovarian V2, <https://www.nccn.org/guidelines/guidelines-detail?category=2&id=1545> (2025, accessed 18 May 2026).

3. National Comprehensive Cancer Network (NCCN). Homepage, 2025. Clinical practice guidelines in Oncology for genetic/familial high risk assessment colorectal cancers V3, <https://www.nccn.org/guidelines/guidelines-detail?category=2&id=1544> (2024, accessed 18 May 2026)
4. ERN. Home page, 2025. Genturis Guidelines: Lynch Syndrome and polyposis, Hereditary breast and ovarian cancer syndrome, <https://www.genturis.eu/1=por/guidelines-and-pathways/clinical-practice-guidelines/endorsed-by-ern-genturis.html#sec434> (accessed 18 May 2026).
5. NICE. Home page, 2025. <https://www.nice.org.uk/guidance> (accessed 18 May 2026).
6. AIOM. Home page, 2025. <https://www.aiom.it/en/linee-guida-aiom/> (accessed 18 May 2026).
7. SIGU. Home page, 2025. <https://sigu.net/> (accessed 18 May 2026).
8. AGENAS. *Linee di indirizzo per la definizione del Percorso Diagnostico Terapeutico Assistenziale (PDTA) per i tumori su base ereditaria: colon, endometrio, mammella, ovaio, pancreas, prostata*. Agenzia Nazionale per i Servizi Sanitari Regionali, 2024.
9. ORPHA.net Home page, 2025. <https://www.orpha.net/> (accessed 18 May 2026).
10. Tibiletti MG, Carnevali I, Calò V, et al. Universal testing for MSI/MMR status in colorectal and endometrial cancers to identify Lynch syndrome cases: state of the art in Italy and consensus recommendations from the Italian Association for the Study of Familial Gastrointestinal Tumors (A.I.F.E.G.). *Eur J Cancer Prev* 2022; 31: 44-49.