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Five Years of Genetic Counseling for Hereditary Cancer Syndromes: Insights from a Bulgarian Center

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ABSTRACT

This investigation delivers a 5-year review, looking backward, at the provision of genetic counseling (GC) focused on hereditary cancer syndromes (HCS) within a single Bulgarian center. The intention was to map the demographic and epidemiological contours of attendees, gauge willingness to proceed to genetic testing, and catalog the array of disease-linked mutations detected. The trends point toward progressively greater engagement with GC. Prominent observations include divergent characteristics between those evaluated for overarching HCS risk and those whose workup began with tumor biomarker assays, the degree to which out-of-pocket costs shaped testing decisions, and the identification of a harmful mutation in 28% of those screened. Hereditary Breast and Ovarian Cancer Syndrome, along with Lynch Syndrome, surfaced most often, with culpable alterations pinpointed in loci such as BRCA1, MSH2, PALB2, and STK11. What emerges from these data is a clear call to broaden awareness, remove financial hurdles to testing, and put in place formal cascade testing pathways across Bulgaria.

Keywords: Hereditary cancer, Tumor predisposition syndromes, Genetic counseling, Genetic testing, HBOC, Lynch syndrome

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Introduction

When a person inherits a gene fault, the resulting condition—termed a hereditary cancer syndrome (HCS)—can sharply raise the lifetime odds of specific malignancies. Genetic counseling (GC) serves as the cornerstone for identifying those who carry such risk, weighing that risk, outlining which tests can and cannot reveal, and empowering sound choices regarding surveillance and prophylactic steps. The journey is multi-step; a key element is pre-analytical GC, the consultative phase that unfolds before a test is authorized, serving as the bedrock for genuine informed consent. Getting to grips with who seeks out GC and what emerges from subsequent genetic investigations, particularly in national settings, is pivotal for sharpening how services operate and raising the standard of care. The primary purpose of this paper was to chronicle, over a half-decade at one Bulgarian institution, the demographic and epidemiological patterns of patients who entered the GC pathway for HCS and later underwent genetic analysis.

Materials and Methods

The design was a retrospective chart audit anchored at a single clinical genetics service in Bulgaria. Case records were retrieved for all individuals who had a genetic counseling encounter during 2020-2024. Two distinct streams of patients made up the study: Stream A comprised those counseled expressly to explore a possible underlying hereditary tumor predisposition syndrome (HTPS) (n = 154); Stream B involved those who first had molecular dissection of a tumor sample and were subsequently counseled regarding what the findings might imply for their germline (n = 157). Details abstracted from the files covered age at consultation, sex, geographic origin, how the

person came to the service (referral pathway), contributory history (own cancer story and that of blood relatives), tumor site, test breadth (e.g., focused gene panels, exome-level sequencing), and the constitutional variants ultimately identified. The analytic approach was descriptive, boiling the cohort's makeup and GC plus testing endpoints down to raw counts and proportional breakdowns.

Results and Discussion

For ease of comparison, the two patient streams and their core demographic and clinical features are laid out side by side in **Table 1.**

Table 1. Demographic and clinical characteristics of patient groups.

Characteristic	Group 1: Direct GC for HTPS (n = 154)	Group 2: GC after biomarker testing (n = 157)	Subgroup: Completed DNA analysis (n = 76)
Average age (years)	50	58	47
Sex distribution	66% female	39% female	85% female
Main indication	67% had a personal history of cancer or polyposis	100% had a personal history of cancer	52% had a personal history of cancer or polyposis
	18% had only a family history	57% also reported a family history	31% had only a family history
	5% had no clear indication (proactive)	–	10% had no clear indication (proactive)
Most common cancer types	Breast, ovarian, and gastrointestinal/polyposis	Colorectal and lung	Breast, ovarian, and gastrointestinal/polyposis
Uptake of DNA testing	49% (75/154)	<1% (1/157)	100%

Profile of patients counseled for potential HTPS

Overall numbers

Even as the price of services jumped twofold after October 2022, and 2024 brought a slight pullback, the general direction from 2020 to 2024 was one of increasing attendance for HTPS-focused GC. Individuals from Varna city accounted for roughly four out of every five, the remaining fifth hailing from locales up to 130 km distant. The slice of the total GC workload devoted to cancer genetics ranged from 4.3% to 7.5% in any given year.

Age and sex split

Broken down by decade, 56% (n = 86) fell at or below the 50-year mark, and 44% (n = 68) lay above it, the midpoint age being 50 years. A clear female skew was evident, with women accounting for 66% (n = 102) and men for 34% (n = 52).

Reason for attendance and entry route

A cancer or advanced polyposis diagnosis in the patient's own history stood as the principal driver (67%, n = 103). A further slice (8%, n = 12) had faced multiple primary tumors. Clinically unaffected people presenting solely because cancer ran in the family composed 18% (n = 28). Only a small pocket (5%, n = 8)—among them three members of a single kindred—reached out independently, not triggered by clinical red flags but by a personal preventive-health motivation. As for the door through which they entered, the largest share (80%, n = 123) arrived on the advice of a hospital clinic, office-based physician, or tumor board. Walk-ins who approached on their own initiative made up the remaining 20% (n = 31). A clear uptick occurred in individuals opting for outpatient GC paid directly from personal funds (n = 100); meanwhile, the stream referred while admitted to hospital remained roughly level (n = 54).

Profile of patients counseled after tumor biomarker workup

Overall numbers

Year on year, the count of patients having focused biomarker assays on neoplastic tissue climbed steeply. From within that expanding pool, just over a quarter (26%, n = 157) were routed for additional GC to rule in or out a

background HTPS, and this subset grew threefold when comparing the final two years of the study (rising from 39 in 2023 to 118 in 2024).

Age and sex split

The age curve shifted decisively upward here: eight in ten had celebrated a 50th birthday or beyond, and the cohort's average age came to 58 years. Men outnumbered women (61% vs. 39%), a distribution mirroring the preponderance of colorectal cancer and non-small cell lung cancer in this arm.

Contributory features

Well over half (57%, n = 90) bore, alongside their own tumor, a positive family cancer narrative. A tumor debuting before age 50 accounted for 35% (n = 54). A history encompassing more than one primary malignancy was volunteered by 8% (n = 13).

Profile of patients moving forward with DNA analysis

Overall numbers

From the combined counseled population of 311, just under a quarter (24%, n = 76) ultimately had germline DNA analyzed, and this group was drawn almost exclusively from the first clinical stream—only a single individual came from the tumor-biomarker-first pathway. Varna residents were heavily overrepresented among the tested, at 83%.

Age and sex split

The step toward actual testing was statistically far more common among women (85%, n = 65) than men. Those who proceeded had a lower mean age—47 years—than those who did not —53 years. A strong and statistically meaningful link emerged between the funding model and the decision to test: consultations settled out-of-pocket by the patient (termed paid consultations) led to DNA testing in 93% of cases (n = 71), whereas for inpatient visits absorbed by the hospital budget, the corresponding figure was a mere 7% (n = 5 out of 54 inpatients).

Nature of the tests carried out, and the pathology uncovered

Regarding the breadth of investigation, a narrowly focused gene panel aligned with the patient's presenting features was chosen for just 18% (n = 14). In contrast, 78% (n = 59) elected to undergo the most extensive multi-gene panel available within a comparable cost bracket. Whole-exome sequencing (WES) was pursued by three individuals (4%).

A pathogenic or likely pathogenic (P/LP) variant corresponding to the presenting condition was identified in 28% (n = 21) of the 76 subjects screened. The rate at which a molecular diagnosis was established fluctuated according to the reason for referral: it reached 32% among those with breast and/or ovarian malignancy, 28% in the gastrointestinal cancer and/or polyposis group, 25% for other tumor types, and 25% for asymptomatic individuals. The variety of disease-causing mutations detected, along with the syndromes they are associated with, is presented in **Table 2**. Abnormalities tied to Hereditary Breast and Ovarian Cancer (HBOC) and Lynch Syndrome constituted the most frequent findings. A variant of uncertain significance (VUS) was returned for 25% (n = 19) of those tested.

Table 2. Spectrum of pathogenic/likely pathogenic (P/LP) variants identified.

Gene	Identified (P/LP) variant	Related syndrome	Clinical presentation of the patient
SMAD4	Partial deletion of exons 11–12	Juvenile Polyposis / HHT	Gastric and gastrointestinal polyposis, liver hemangioma
STK11	Deletion of exon 1	Peutz–Jeghers syndrome	Suspected Peutz–Jeghers syndrome with polyposis
MSH2	c.1386 + 1G>A	Lynch syndrome	Synchronous colon adenocarcinomas
APC	c.4564_4565del, p.(Leu1522Lysfs*10)	Familial adenomatous polyposis	Numerous colorectal polyps
MLH1	c.67del, p.(Glu23LusfsTer13)	Lynch syndrome	Colon cancer with a positive family history

BRCA1	Exon 10 deletion, c.3756_3759del (p.Ser1253Argfs*10)	HBOC	Ovarian cancer with family history
BRCA1	c.5266dup, p.(Gln1756Profs*74)	HBOC	Breast cancer with a strong family history
CHEK2	c.592 + 3A>T	HBOC / HCS	Breast cancer
PALB2	c.2257C>T, p.(Arg753*)	HBOC	Breast cancer
BRCA2	c.682-1G>A	HBOC	Two primary breast cancers
NBN	c.657_661del, p.(Lys219Asnfs*16)	NBN-related HCS (HBOC spectrum)	Breast cancer with a positive family history
PMS2	c.904-2A>G, p.(Gly757Arg)	Lynch syndrome	Ovarian cancer
RET	c.2671T>G (p.Ser891Ala)	Multiple Endocrine Neoplasia type 2	Medullary thyroid cancer with family history
NF1	c.5749 + 332A>G	Neurofibromatosis type 1	Clinical features of NF1 with family history
PTPN11	c.922A>G, p.(Asn308Asp)	Noonan syndrome	Thyroid cancer with suspected Noonan features
NBN	c.657_661del, p.(Lys219Asnfs*16)	NBN-related HCS (HBOC spectrum)	Healthy individual with a family history
CHEK2	c.470T>C, p.(Ile157Thr)	HBOC / HCS	Healthy individual with a family history
CHEK2	c.444 + 1G>A	HBOC / HCS	Proactive testing in a healthy individual
CHEK2	c.470T>C, p.(Ile157Thr)	HBOC / HCS	Proactive testing in a healthy individual
ATM	c.3886_3889del, p.(Pro1296Ilefs*52)	Ataxia–telangiectasia (HCS)	Healthy individual with an affected child
BLM	c.1544dup, p.(Asn515Lysfs*2)	Bloom syndrome (HCS)	Healthy individual with a family history

Working outward from the 21 index patients in whom a P/LP variant was confirmed, 36 first-degree relatives were flagged as potentially carrying the same risk. Of this group, a scant 19% (n = 7) ultimately attended GC and had the targeted familial variant checked, representing a cascade screening effort. This process identified 4 additional asymptomatic carriers of harmful variants—specifically in the BRCA1, CHEK2, MSH2, and NBN genes—thereby opening the door to preventive interventions.

This single-institution investigation conducted in Bulgaria sheds meaningful light on the uptake patterns and the defining traits of people pursuing GC for HCS. Although the mounting number of attendees signals a rise in consciousness, the proportion of all counseled patients who come for oncogenetics remains small, with expansion occurring almost exclusively in the self-pay segment. This exposes a stark access gap, almost certainly shaped by economic hurdles.

The observation that the typical GC seeker is a younger woman is consistent with earlier reports [1]. On the flip side, the fact that older males dominate the tumor-to-germline testing pathway reveals a critical entry point for engaging a demographic that is otherwise seldom reached.

A pivotal result is the dramatically higher rate of actual DNA testing among those who covered their own counseling costs (93%) compared with those treated within the inpatient institutional framework (7%). This sharp divergence powerfully indicates that expense acts as a formidable barrier. Such an imbalance calls out for policies that actively promote fairness in access.

A 28% pathogenic variant yield stands in line with figures from comparable cohorts [2]. Pinpointing mutations in medically actionable loci such as BRCA1, BRCA2, MSH2, MLH1, APC, and STK11 reaffirms the tangible clinical value of genetic testing—both for identifying high-risk individuals and for guiding tailored management strategies [3-5]. Nevertheless, the heavy clustering of tested individuals within the city of Varna (83%) highlights how profoundly geography and accessibility shape uptake.

The marked tendency to opt for broader gene panels reflects a patient-driven desire for exhaustive genetic knowledge [6, 7]. The VUS return rate, which at 25% nearly mirrors the P/LP detection rate, reinforces the lasting importance of periodic variant reinterpretation—a foundational pillar of conscientious genomic practice.

The extremely modest fraction (5%) of counseling encounters that can be labeled as proactive—meaning consultations by healthy persons lacking a particularly striking family cancer history, undertaken purely for preventive insight—points to a deep need for amplified public education efforts [8, 9]. A noteworthy limitation of this work is its backward-looking design, which prevented systematic capture of the downstream clinical consequences of the genetic findings, such as adherence to intensified surveillance protocols or uptake of risk-reducing operations. Furthermore, the limited numbers in certain subsets limit the applicability of some conclusions. These gaps mark prime targets for subsequent forward-looking investigations.

Conclusion

This half-decade portrait of a single center's activity delivers a valuable window into the HCS genetic counseling environment in Bulgaria. The growing appetite for these services is heartening, yet obstacles rooted in awareness, the financial burden of testing, and the complexities of result interpretation persist. A diagnostic success rate of 28% and the specific array of detected mutations—most notably those within genes driving HBOC and Lynch Syndrome—affirm the pressing requirement for a nationwide game plan. Moving forward, priorities must center on elevating literacy among both the public and healthcare professionals, making the price of genetic testing less prohibitive, and, above all else, rolling out a coordinated cascade screening infrastructure. Only then can the full preventive promise of cancer genetics be realized for the Bulgarian population.

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Ethics Statement: Approval for this research, including the use of anonymized patient data, was granted by the Medical University Varna Ethics Committee (Ethics Approval Number 9/30.01.2025), and patient consent has been secured. The studies were conducted in accordance with the local legislation and institutional requirements. The participants provided their written informed consent to participate in this study.

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