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Reproductive Choices and Attitudes Toward Preimplantation Genetic Testing Among Individuals with Hereditary Cancer Predisposition

Joanna Kufel-Grabowska
Gdańsk Medical University
Mikolaj Bartoszkiewicz
m.bartoszkiewicz@ump.edu.pl
Poznan University of Medical Sciences
Daria Swiniuch
Poznan University of Medical Sciences
Izabela Izabela Laczmanska
Wroclaw Medical University

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Abstract

Purpose

Carriers of pathogenic variants in hereditary cancer-predisposition genes face complex reproductive decisions, including the risk of transmitting the familial variant to offspring. This study assessed awareness, acceptance, and determinants of willingness to use preimplantation genetic testing for monogenic disorders (PGT-M) combined with in vitro fertilisation (IVF) among individuals with hereditary cancer predisposition.

Methods

Two independent cross-sectional online surveys were conducted in 2023 (n = 72) and 2026 (n = 92) among respondents aged ≤ 45 years reporting hereditary cancer predisposition. The questionnaires included 22 items addressing demographic characteristics, genetic background, cancer history, reproductive status, and attitudes toward PGT-M + IVF. Associations between willingness to use PGT-M + IVF and predefined determinants were evaluated separately for each cohort.

Results

Awareness of PGT-M was low in both cohorts (28% in 2023 and 24% in 2026). Despite this, willingness to use PGT-M + IVF to avoid transmitting a pathogenic variant was high and stable (72% and 70%, respectively). In 2026, 51% of respondents were unsure whether they would self-fund the procedure. In 2023, willingness did not differ significantly by predefined determinants. In 2026, childless respondents were more willing to use PGT-M + IVF than respondents with children (90% vs 65%, p = 0.028); personal cancer history showed a borderline association (76% vs 52%, p = 0.056).

Conclusion

Acceptance of PGT-M + IVF among reproductive-age individuals with hereditary cancer predisposition is high despite limited awareness. These findings support routine integration of reproductive counselling into hereditary cancer care.

Introduction

Breast cancer (BC) is the most common cancer in women, with mortality rates second only to lung cancer. In 2022, 2.3 million women worldwide were diagnosed with breast cancer, 670,000 died, and 11.7 million had a breast cancer diagnosis within the previous five years (Bray et al., 2024).

The primary risk factors for BC are gender and age. The vast majority of breast cancers are detected after the age of 50, mostly in women, with younger individuals accounting for roughly 20% in developed countries. The lifetime risk for any given woman is about 12%. In addition to genetics and a family history of cancer, environmental factors include obesity in postmenopausal women, poor diet, alcohol and tobacco use, sedentary lifestyles, early menarche, late menopause, late childbirth, not breastfeeding, and radiation exposure to the chest area (Kashyap et al., 2022; Liu et al., 2021; Mahdavi et al., 2019).

Pathogenic variants (mutations) in genes that increase cancer risk, most commonly involved in DNA repair pathways, are responsible for approximately 2.5-5% of breast cancers. The reported frequency of identified variants may rise owing to increased public awareness, greater availability, and lower prices for genetic testing, particularly next-generation sequencing (NGS) (Doraczynska-Kowalik et al., 2022; Kowalik et al., 2018).

The *BRCA1/2* genes are tumour suppressor genes that repair double-stranded DNA breaks. Accumulated genetic damage causes chromosomal instability and increases the likelihood of carcinogenesis or apoptosis. Carrying pathogenic variants in the *BRCA1/2* may be associated (depending on the pathogenic variant) with an approximately 80% risk of developing breast cancer, a 40–60% risk of developing ovarian cancer, as well as an increased risk of developing pancreatic cancer, melanoma, and, in men, prostate cancer (Mahdavi et al., 2019).

The hereditary component of breast cancer goes beyond *BRCA1* and *BRCA2*, which account for roughly 50–60% of clinically relevant hereditary breast and ovarian cancer (HBOC) cases. Recent multigene sequencing studies have revealed several additional high- and moderate-penetrance susceptibility genes, including *PALB2, TP53, PTEN, CDH1, STK11, ATM, CHEK2, BARD1, BRIP1, RAD51C*, and *RAD51D*. These genes are largely responsible for homologous recombination repair (HRR), DNA damage response, cell cycle regulation, and genome stability. Among them, *PALB2* is considered the most important non-*BRCA* breast cancer susceptibility gene, with a lifetime breast cancer risk comparable to that of *BRCA2* carriers (Doddato et al., 2021). Whole-exome sequencing studies have revealed that pathogenic variants in less frequently mutated genes together may account for 40–50% of hereditary breast cancer cases previously classed as *BRCA1/2*-negative. As a result, high-throughput genetic testing methodologies increasingly focus on multigene next-generation sequencing (NGS) panels rather than just *BRCA1/2* analysis, allowing for better risk assessment, individualized surveillance, and targeted therapeutic decision-making (Concolino et al., 2019; Doddato et al., 2021; Gennari et al., 2021; Potrony et al., 2025).

In Poland, genetic testing for *BRCA1* and *BRCA2* is integrated into National Cancer Prevention and Treatment Programs, providing access to both targeted screening for 10 recurrent mutations in *BRCA1*, *PALB2*, *CHEK2* and comprehensive HBOC genes NGS analysis for patients with breast, ovarian, pancreatic, and prostate cancers and cancer family history, as well as (to a lesser extent) their relatives. Despite the expansion of genetic testing programs, access to genetic counselling services in Poland remains limited, resulting in long waiting times, especially for unaffected relatives of mutation carriers who require predictive testing and tailored cancer prevention plans (Doraczynska-Kowalik et al., 2022).

A bilateral mastectomy is the only surgical procedure that can reduce the risk of breast cancer by up to 90%. The effect of this procedure on overall survival has not been fully determined. Furthermore, bilateral oophorectomy reduces the incidence of breast and ovarian cancer while increasing survival in those who carry a deleterious variant in the *BRCA1.* Importantly, salpingo-oophorectomy with delayed oophorectomy is a non-standard treatment and should not be recommended (Bertozzi et al., 2023). A patient's age at which cancer-reducing surgery should be offered is determined by an individual's family history of cancer, fertility goals, and personal preferences. Bilateral salpingo-oophorectomy is often performed on women aged 35–40 who carry a pathogenic variant in the *BRCA1*, and around 5–8 years later on women who carry a pathogenic variant in the *BRCA2* gene (Bertozzi et al., 2023).

Infertility is defined as the failure to conceive within 12 months despite regular sexual intercourse (2–4 times per week) without the use of contraception. Globally, infertility affects approximately 8–12% of couples, and in vitro fertilisation (IVF) is one of the most effective treatment options currently available (Mascarenhas et al., 2012). Women carrying pathogenic variants in HBOC-associated genes face additional challenges that extend beyond an increased risk of cancer. Several studies have demonstrated that these women may have a diminished ovarian reserve due to accelerated oocyte depletion, which can impair fertility and increase the need for assisted reproductive technologies (ART) (Maletta et al., 2026; Silvestris et al., 2024). Moreover, carriers are either more likely to be diagnosed with cancer at an early age or have a 50% chance of passing on a genetic cancer predisposition to their children (Finch et al., 2013). When analysing their condition holistically, it is worthwhile to consider preimplantation genetic testing (PGT-M) for embryo selection alongside IVF. Taken together, these reproductive, oncological, and hereditary challenges place a substantial psychological burden on individuals carrying pathogenic variants in HBOC-associated genes (M. Lambertini et al., 2024; M. M. P. Lambertini et al., 2020a; Maksimenko et al., 2022).

The survey described in this paper was administered twice — first in 2023 and again in 2026. Since 1 June 2024, IVF, including fertility preservation procedures for oncology patients have been reimbursed under the Ministry of Health Program for sexually mature women up to 40 years of age and sexually mature men up to 45 years of age in Poland. With the increased availability of gamete cryopreservation for oncology patients, awareness of the role of IVF has evolved, as reflected in the growing interest among reproductive-age patients in participating in the program. According to official data published on the Polish government portal, by 31 May 2026, 2,311 oncology patients had enrolled in the national fertility preservation program, and 2,003 had undergone oocyte or sperm cryopreservation, demonstrating substantial uptake of fertility preservation services following the introduction of public funding (*Program wsparcia in vitro | Pacjent*, 2026).

Although the clinical and ethical aspects of preimplantation genetic testing for monogenic disorders (PGT-M) in hereditary cancer syndromes have been widely discussed, relatively little is known about mutation carriers' attitudes toward its use in routine clinical practice. Moreover, limited data evaluate whether the introduction of publicly funded oncofertility services influences patients' awareness and acceptance of reproductive technologies. Addressing these knowledge gaps is essential for developing

patient-centred genetic counselling and reproductive care strategies for individuals with hereditary cancer predisposition.

The present study aimed to evaluate the individual and family circumstances of carriers of pathogenic variants in HBOC-associated genes, their awareness of hereditary cancer risk and reproductive options, and their attitudes toward the use of PGT-M combined with IVF. To describe attitudes in the period before and after the implementation of publicly funded fertility preservation services in Poland, two independent questionnaire surveys were conducted before and after the introduction of the national reimbursement programme.

Ethics

The Poznan University of Medical Sciences Ethics Committee evaluated and approved the study (No. 511/2026). An anonymous survey was submitted to the Bioethics Committee at Poznan University of Medical Sciences. The Committee determined that the study was not a research experiment and might be used for scientific purposes. Participation was voluntary, and informed consent to participate was obtained from all participants through their voluntary completion and submission of the anonymous online questionnaire.

Materials and methods

This report presents two separately conducted cross-sectional questionnaire surveys examining the opinions of individuals reporting confirmed or suspected hereditary cancer predisposition regarding the use of PGT-M alongside IVF to avoid transmitting a cancer-predisposing variant to their offspring. By design, participants were enrolled irrespective of their current childbearing intentions; their opinions were nonetheless considered informative. The 2023 and 2026 cohorts are independent convenience samples and are analysed and reported independently below.

Study population

A total of 164 respondents met the eligibility criteria and were included in the final analysis, comprising 72 participants in the 2023 cohort and 92 in the 2026 cohort. The baseline characteristics of both study groups are presented in Table 1. The majority of participants were women of reproductive age carrying pathogenic variants in BRCA1 or BRCA2 and were enrolled in oncogenetic surveillance programs. Most respondents reported a family history of cancer, and a substantial proportion had a personal history of malignancy and had already completed family planning. Compared with the 2023 cohort, participants surveyed in 2026 more frequently reported a personal history of cancer and included a broader spectrum of hereditary cancer–predisposition genes. In the 2026 cohort, a small subgroup reported no genetic test or did not report the affected gene; these respondents were retained in the descriptive analyses because they reported hereditary cancer risk or participation in oncogenetic care, and this should be considered when interpreting the findings.

Table 1
Baseline characteristics of study participants

Characteristic	2023 cohort (n = 72)	2026 cohort (n = 92)
Age, years		
Mean ± SD	36.6 ± 4.6	37.4 ± 4.9
Median (range)	37 (26–45)	38 (24–45)
Sex, n (%)		
Female	71 (99%)	92 (100%)
Male	1 (1%)	0 (0%)
Affected gene, n (%)		
BRCA1	55 (76%)	44 (48%)
BRCA2	13 (18%)	13 (14%)
CHEK2	1 (1%)	8 (9%)
PALB2	1 (1%)	7 (8%)
TP53	1 (1%)	2 (2%)
Other (*RAD51*, *NBN*, multiple genes)	1 (1%)	4 (4%)
None / not tested	0	10 (11%)
Not reported	0	4 (4%)
Personal history of cancer, n (%)		
Yes	30 (42%)	71 (77%)
Has children, n (%)		
Yes	52 (72%)	69 / 90 (77%)
Risk-reducing surgery, n (%)		
Risk-reducing mastectomy	44 (61%)	50 (54%)
Risk-reducing salpingo-oophorectomy	25 (35%)	31 / 91 (34%)
Any prophylactic surgery	50 (69%)	58 (63%)
Regular genetic surveillance, n (%)		
Yes	67 (93%)	58 / 91 (64%)

To focus on the population for whom PGT-M + IVF is a realistic option, the analyses were restricted to respondents aged 45 years or younger at the date of the survey. This excluded 12 of 84 respondents in the 2023 cohort (analytic n = 72) and 37 of 129 in the 2026 cohort (analytic n = 92); respondents with a missing or unparseable age were also excluded.

The survey contained 22 questions. The first two questions concerned demographic data: age and gender; the following 10 questions concerned respondents' genetic burden: whether they carried mutations in genes that increase the risk of cancer, the age at which the mutation was diagnosed, its type, and preventive measures taken (age when cancer-reducing procedures were performed, participation in genetic surveillance programs). Three issues arose: whether the respondents had family mutations, whether their relatives had cancer, and whether they had received oncological treatment. The next 6 questions inquired about the respondents' offspring, preimplantation genetic diagnosis, and their opinions on using Preimplantation Genetic Testing for Monogenic Disorders (PGT-M) to avoid passing on the pathogenic variant to their offspring, including whether they would have to cover the costs themselves. IBM SPSS Statistics 26 was used to conduct the analysis. Preliminary results from the 2023 survey were presented as a conference abstract in Cancer Research (2023) 83 (5_Supplement): P6-05-47. The questionnaires were developed by the study authors specifically for the purposes of this research and were not adapted from a previously published instrument. An English-language version of the questionnaire is provided as Supplementary Material 1.

Statistical analysis

Two independent cross-sectional online surveys were conducted in 2023 and 2026. As almost none of the participants in the 2026 survey reported completing the earlier questionnaire, the datasets were analysed independently rather than as a longitudinal follow-up. Although the questionnaires were largely comparable, minor differences in question wording and response options precluded pooling of the datasets; therefore, results are presented separately for each survey.

To restrict the analysis to individuals for whom preimplantation genetic testing for monogenic disorders (PGT-M) combined with in vitro fertilisation (IVF) represents a realistic reproductive option, only respondents aged ≤ 45 years at the time of the survey were included. Age was calculated from the year of birth in the 2023 survey and recorded directly in the 2026 survey. After applying the eligibility criteria, 72 of 84 respondents from the 2023 survey and 92 of 129 respondents from the 2026 survey were included in the final analysis.

Questionnaire responses were reviewed and harmonised before analysis. Binary variables were standardised to three categories (Yes, No, and Don't know). Free-text responses concerning offspring status and the identified pathogenic variant were categorised into predefined groups. Missing responses were excluded on an item-by-item basis; therefore, the number of analysed observations varied slightly between variables.

Categorical variables are presented as frequencies and percentages calculated from non-missing responses. Continuous variables are reported as mean ± standard deviation, median, and range. The primary outcome was willingness to use PGT-M in combination with IVF (Yes vs No). Associations between willingness to use PGT-M and four predefined variables (previous awareness of PGT-M, personal history of cancer, parenthood status, and age group [< 40 vs 40–45 years]) were analysed separately for each survey. Fisher's exact test was used for 2 × 2 contingency tables because of small expected cell counts, whereas the chi-square test was applied to larger contingency tables. Participants responding "Don't know" to the primary outcome were excluded from these analyses. All statistical tests were two-sided, and a p-value < 0.05 was considered statistically significant. Owing to the relatively small subgroup sizes, no adjustment for multiple testing was performed, and subgroup analyses should be considered exploratory.

All statistical analyses were performed using Python (Python Software Foundation, Wilmington, DE, USA) with standard scientific libraries for data processing, statistical analyses, and graphical visualisation.

Results

Part I — 2023 cohort (n = 72, age ≤ 45)

Awareness of the technology was low — only 28% (20/72) of respondents knew what preimplantation genetic testing is, while almost three-quarters had never heard of it. Despite this, acceptance was high: 72% (52/72) reported being willing to use PGT-M + IVF to prevent transmission of the variant, and 28% were not. Among the 58 women who answered the conditional cost question, 69% (40) were willing to self-fund the IVF + PGT-M procedure. Carrier status had influenced past reproductive decisions in only 35% of respondents — most had not let their genetic risk drive family planning. Yet, a clear majority endorsed the technology in principle (Fig. 1).

The cohort represented a selected high-risk population with substantial engagement in oncogenetic care. A personal history of malignancy was reported by 42%, while risk-reducing surgery was already common — 61% had undergone risk-reducing mastectomy and 35% risk-reducing salpingo-oophorectomy. Almost all (93%) were under regular genetic surveillance, and 89% reported a family history of cancer. Just under three-quarters (72%) already had children, meaning that for many, the question of reproductive prevention was at least partly retrospective (Fig. 2).

The genetic background was overwhelmingly *BRCA*-driven: *BRCA1* accounted for 76% and *BRCA2* for 18% of respondents (~ 94% *BRCA1/2* combined), with only isolated *TP53*, *CHEK2, PALB2* or multi-gene carriers. After the age restriction, the cohort was tightly clustered in the younger reproductive age range — mean 36.6 ± 4.6 years (median 37, range 26–45) — i.e., precisely the window in which decisions about PGT-M + IVF are clinically actionable (Fig. 3).

Willingness to use PGT-M + IVF was uniformly high (65–80%) across every subgroup, and none of the four pre-specified determinants reached statistical significance: prior awareness of PGT-M (75% vs 65%,

p = 0.40), personal cancer history (76% vs 67%, p = 0.43), already having children (80% vs 69%, p = 0.56) and age group (70% for < 40 vs 79% for 40–45 years, p = 0.56). Acceptance in the 2023 cohort was therefore broad and essentially independent of clinical or demographic background (Fig. 4).

Part II — 2026 cohort (n = 92, age ≤ 45)

Awareness of PGT-M remained low and essentially unchanged from 2023 — only 24% knew what the technique is. A clear majority, 70% (64/91), would be willing to use PGT-M + IVF, while 30% would not. The cost question, which in this instrument offered an explicit “don’t know” option, was dominated by uncertainty — 29% yes, 20% no and 51% undecided — showing that financial readiness, rather than acceptance of the method itself, may be an important area of uncertainty. Carrier status had influenced past reproductive decisions among 30% of respondents (Fig. 5).

The 2026 cohort was more heavily affected by cancer than the 2023 cohort: 77% reported a personal history of malignancy (versus 42%). Risk-reducing mastectomy was performed in 54% and, salpingo-oophorectomy in 34%, 64% were under regular genetic surveillance, and 74% reported a family history of cancer. Most respondents (77%) already had children, again indicating a substantially post-reproductive sample (Fig. 6).

The gene spectrum was broader than in 2023: *BRCA1* 48% and *BRCA2* 14% (~ 62% *BRCA1/2*), with a more visible contribution of moderate-penetrance genes — *CHEK2* 9% and *PALB2* 8% — plus single *RAD51, NBN, TP53* and multi-gene carriers. About 11% reported no genetic test or no confirmed variant, consistent with the inclusive enrolment criterion. After the age restriction, the cohort spanned a similar reproductive band to 2023 — mean 37.4 ± 4.9 years (median 38, range 24–45) (Fig. 7).

Non-significant differences were observed for prior awareness of PGT-M and personal cancer history. Willingness was higher among women already aware of PGT-M (82% vs 67%, p = 0.28) and, more markedly, among those with a personal cancer history (76% vs 52%, p = 0.056 — borderline). The only determinant to reach significance was parenthood: participants without children were more willing to use PGT-M + IVF than those who already had children (90% vs 65%, Fisher’s exact test p = 0.028) — a pattern consistent with completed family planning. Willingness did not differ significantly by age group (66% for < 40 vs 77% for 40–45 years, p = 0.35) (Fig. 8).

Summary of key observations (both cohorts, age ≤ 45)

- Acceptance of PGT-M + IVF is high and stable — about 72% (2023) and 70% (2026) of carriers of reproductive age would use it to avoid transmitting a cancer-predisposing variant.
- Awareness of the technology is consistently low (~ 24–28%) despite this high willingness, indicating a clear unmet information and counselling need.
- Cost is the principal hesitation: when an explicit “don’t know” option was offered (2026), about half of respondents (51%) were unsure whether they could self-fund the procedure.

- Already having children was the only factor significantly associated with lower willingness (2026: 65% vs 90%, p = 0.028); a personal cancer history showed a borderline positive association (76% vs 52%, p = 0.056).
- Restricting to age ≤ 45 removed 12/84 (2023) and 37/129 (2026) respondents and tightened both cohorts to a comparable reproductive-age band (mean ≈ 37 years), with the headline proportions changing only modestly.

Family history of cancer

A positive family history of cancer was reported by 89% (64/72) of respondents in the 2023 cohort and 74% (68/92) in the 2026 cohort. Similarly, at least one cancer-affected relative was reported by 88% and 75% of participants, respectively. Among respondents reporting a positive family history, the mean number of affected relatives was 2.4 in the 2023 cohort and 1.8 in the 2026 cohort.

First-degree relatives were the most frequently affected family members, being reported by 69% of participants in the 2023 cohort and 48% in the 2026 cohort. Mothers represented the largest subgroup (63% and 33%, respectively), followed by fathers and siblings. Second-degree relatives with a history of cancer were reported by 54% and 41% of respondents, most commonly grandmothers and aunts. Approximately 11% of participants in both cohorts also reported cancer occurring in third-degree or more distant relatives (Table 2).

Table 2
Distribution of cancer-affected relatives according to degree of kinship.

Characteristic	2023 cohort (n = 72)	2026 cohort (n = 92)
Family history of cancer (positive), n (%)	64 (89%)	68 (74%)
Reported ≥ 1 cancer-affected relative, n (%)	63 (88%)	69 (75%)
Affected relatives reported per respondent[1]	2.4 (median 2, max 11)	1.8 (median 1, max 8)
Any first-degree relative[2], n (%)	50 (69%)	44 (48%)
Mother	45 (63%)	30 (33%)
Father	4 (6%)	8 (9%)
Sister	7 (10%)	6 (7%)
Brother	1 (1%)	1 (1%)
Daughter	4 (6%)	0 (0%)
Son	1 (1%)	0 (0%)
Sibling (unspecified)	0 (0%)	2 (2%)
Any second-degree relative[2], n (%)	39 (54%)	38 (41%)
Grandmother	27 (38%)	22 (24%)
Grandfather	6 (8%)	12 (13%)
Aunt	20 (28%)	18 (20%)
Uncle	3 (4%)	4 (4%)
Any third-degree / more distant relative[2], n (%)	8 (11%)	10 (11%)
Great-grandparent	2 (3%)	2 (2%)
Great-aunt / great-uncle	5 (7%)	1 (1%)
Cousin / more distant	2 (3%)	7 (8%)

Values are n (% of cohort) unless stated. Analyses are restricted to participants aged ≤ 45 years at the survey date; the 2023 and 2026 cohorts are independent samples. Affected relatives were coded from a free-text field in which respondents listed family members with cancer, their relationship and age at diagnosis.

[1] Mean number of cancer-affected relatives reported, among respondents who reported at least one; median and maximum in parentheses.

[2] “Any … relative” counts respondents reporting at least one affected relative of that degree of kinship; because a respondent may report several relatives, the degree subtotal is smaller than the sum of the specific relationships listed beneath it, and a respondent may contribute to more than one degree.

Degrees of kinship

first-degree = mother, father, sister, brother, child; second-degree = grandparent, aunt, uncle (parent’s sibling), niece/nephew; third-degree / more distant = great-grandparent, great-aunt/great-uncle (grandparent’s sibling), first cousin and remoter relatives.

Important limitation

the questionnaires did not record whether relatives underwent genetic testing or carried a pathogenic variant; this table therefore describes cancer occurrence in relatives only, not their mutation status.

Discussion

Diagnosis of the hereditary breast and ovarian cancer (HBOC) risk has far-reaching consequences beyond the affected patient. The identification of a pathogenic variant enables cascade testing of at-risk relatives, thereby allowing unaffected carriers to benefit from individualised surveillance, risk-reduction programs, and early cancer detection strategies. Genetic counselling should be complemented by appropriate psychological assistance, as disclosing genetic cancer risk can have a major impact on quality of life, family relationships, and reproductive decision-making. In young women, fertility preservation and reproductive planning are critical components of care. BRCA1 mutation carriers, in particular, appear to be at a higher risk of premature ovarian insufficiency and menopause than the general population, emphasising the importance of seeking appropriate reproductive counselling before undergoing risk-reducing salpingo-oophorectomy. Recent studies have also shown that pregnancy after breast cancer is achievable and does not appear to adversely affect oncological outcomes in BRCA1/2 mutation carriers, highlighting the necessity of considering fertility preservation options and future motherhood at the time of diagnosis. Furthermore, for carriers of pathogenic variants who wish to avoid transmission of the familial mutation to their offspring, PGT-M should be discussed as a reproductive option within specialised genetic and reproductive medicine services. As a result, comprehensive management of HBOC families should include genetic testing, cascade screening, oncological prevention, psychological care, and psychiatric and reproductive counselling in a multidisciplinary setting .

The present study extends the existing literature by providing contemporary data on attitudes toward preimplantation genetic testing for monogenic disorders (PGT-M) combined with IVF among individuals carrying hereditary cancer–predisposition variants in Poland. Unlike previous reports, which have primarily focused on the clinical, ethical, or technical aspects of PGT-M, our study evaluates patients' perspectives before and after the introduction of publicly funded oncofertility services. The findings demonstrate that acceptance of PGT-M is consistently high despite limited awareness of the technology,

suggesting that insufficient knowledge rather than lack of willingness may currently represent the principal barrier to wider implementation of reproductive genetic counselling in hereditary cancer care.

In this study, the majority of the participants were found to carry pathogenic variants in the *BRCA1* or *BRCA2* genes, which are linked to a significantly higher lifetime cancer risk of up to 80%, whereas the lifetime risk of ovarian cancer is estimated to be between 40% and 60%, respectively. However, as NGS technologies become more widely available and genetic testing costs continue to fall, the identification and diagnosis of individuals carrying hereditary cancer predisposition variants in other cancer-susceptibility genes are expected to improve (Kiser et al., 2024a).

An important finding of our study is the extensive family burden of cancer observed among mutation carriers. Nearly nine out of ten respondents in the 2023 cohort and three-quarters of those surveyed in 2026 reported a positive family history of cancer, with affected first-degree relatives identified in 69% and 48% of participants, respectively. Moreover, respondents with a positive family history reported an average of 2.4 and 1.8 affected relatives, highlighting the clustering of cancer within HBOC families. These findings reinforce the importance of cascade genetic testing and a family-centred model of care. Identification of a pathogenic variant should be regarded not as the endpoint of the diagnostic process but as the beginning of systematic identification, counselling, and surveillance of at-risk relatives. Early predictive testing allows unaffected family members to benefit from personalised cancer prevention strategies, appropriate surveillance, reproductive counselling, and informed decision-making, thereby extending the clinical benefits of genetic testing beyond the individual patient to the entire family. The high prevalence of cancer-affected first- and second-degree relatives observed in our study is consistent with previous reports describing the familial clustering of HBOC-associated malignancies and further supports the implementation of systematic cascade genetic testing among at-risk relatives (Doraczynska-Kowalik et al., 2022; Kiser et al., 2024b; Sarki et al., 2022).

In most countries, the indications for PGT-M are restricted and highly controlled. The primary goal of PGT-M is to prevent the transmission of serious genetic diseases for which no effective treatment exists (Carvalho et al., 2020). Across Europe, the legal framework for ART and PGT remains heterogeneous, with country-specific restrictions and variation in the scope of permitted PGT applications (Gliozheni et al., 2025). Strict regulation of PGT, particularly for non-medical purposes, reflects ethical concerns regarding eugenics and selective reproduction based on genetic features. Besides cystic fibrosis, hemoglobinopathies, muscular dystrophies, neurofibromatosis, Huntington's disease, hemophilia, and fragile X syndrome, autosomal dominant hereditary cancer predisposition syndromes are among the most common indications for PGT-M (Calhaz-Jorge et al., 2020).

Preparing a couple for hormonal stimulation before IVF and PGT-M is identical to what is necessary for traditional IVF procedures. In addition to karyotype analysis of both partners and carrier screening for pathogenic variants associated with conditions such as cystic fibrosis, hemoglobinopathies, and spinal muscular atrophy, most couples require comprehensive counselling, including family history assessment, during consultations with a clinical geneticist. Unlike in many other countries, the

profession of genetic counsellor is not formally recognized in Poland. Genetic counselling is provided exclusively by physicians specialized in clinical genetics, who are authorized to deliver medical genetic counselling and discuss the clinical implications of genetic test results. An important component of preparation for IVF is psychological counselling. (Nagy et al., 2024; Takeuchi & Park, 2025).

Genetic testing of an embryo can be performed after harvesting a single blastomere on day 3 or a group of trophectoderm cells on days 5–6 after *in vitro* fertilization. The former strategy allows embryo transfer during the same treatment cycle once genetic analysis is completed by days 5–6. In the latter strategy, however, embryo cryopreservation is often required, and transfer is postponed until a later cycle. The clinical pregnancy rate among patients who underwent IVF combined with PGT-M is projected to be 39.7% and 41.0% after fresh and frozen embryo transfer, respectively (de Geyter et al., 2020).

As the primary objective of PGT-M is to prevent the transmission of pathogenic variants to future offspring and thereby reduce the risk of severe hereditary disorders, long-term follow-up of children conceived through ART combined with PGT-M is essential to evaluate the safety and long-term outcomes of these procedures. Although IVF has been associated with a statistically significant increase in the prevalence of congenital malformations compared with natural conception, the absolute risk remains relatively low, with reported rates of 8.3% and 5.8%, respectively (Davies et al., 2012).

Data on the long-term health effects of infants born after IVF with PGT-M are sparse. However, existing evidence does not confirm that children conceived using these technologies have a higher prevalence of congenital malformations than those conceived via conventional IVF or in the general population (Bay et al., 2016; Desmyttere et al., 2012).

Numerous factors influence decisions about genetic testing, participation in surveillance systems, use of risk-reducing procedures, and communication of genetic risk to family members. These include family relationships, personal and family cancer histories, treatment outcomes, experiences supporting relatives throughout their illness, medical decision-making processes, personal beliefs, and reproductive considerations. Carrying a pathogenic variant not only influences an individual's perception of health, lifestyle choices, and participation in preventive measures, but it can also strengthen family bonds and motivate efforts to break the cycle of cancer morbidity and mortality within affected families (Douglas et al., 2009).

Parenthood following cancer diagnosis, particularly among carriers of pathogenic *BRCA1* and *BRCA2* alterations, is a source of continuous debate and concern. A 2024 publication reported data from nearly 4,700 breast cancer patients with pathogenic *BRCA1/2* variants, of whom over 700 conceived at least once within 10 years of their cancer diagnosis, accounting for roughly 20% of the group. The findings showed that childbearing did not raise the chance of cancer recurrence. Furthermore, women who became pregnant after treatment tended to have equivalent, if not better, outcomes than individuals who did not attempt conception after receiving a cancer diagnosis (M. Lambertini et al., 2024; M. M. P. Lambertini et al., 2020b).

Although oncofertility procedures have been funded in Poland since 2024, access to the treatment remains limited, and patient awareness is low. Despite these obstacles, a sizable majority of survey respondents said they would be willing to undergo IVF with PGT-M, while willingness to self-fund differed across surveys and substantial uncertainty was observed in the 2026 cohort. Approximately one-third of respondents reported that awareness of carrying a genetic predisposition associated with an increased risk of cancer influenced their decisions regarding parenthood. Many carriers of pathogenic variants in cancer susceptibility genes choose not to have children because of concerns about transmitting the genetic predisposition to their offspring and the associated risk of developing cancer at a young age. An important observation was the remarkable stability of attitudes toward PGT-M between the two independent surveys despite the introduction of publicly funded fertility preservation services in Poland. This suggests that improving financial access alone may be insufficient to increase awareness and that educational interventions should accompany organisational changes in reproductive healthcare.

Strengths and limitations

This study has several limitations. First, both surveys were based on convenience samples and relied on self-reported data, which may have introduced selection and reporting bias. Owing to the mode of survey dissemination, it cannot be excluded that some individuals may have completed the questionnaire more than once. Second, although the questionnaires were largely comparable, minor differences in wording and response options prevented direct statistical comparison between the two cohorts. Third, the analysis was restricted to respondents aged ≤ 45 years, limiting the generalisability of the findings to older carriers of pathogenic variants. Participants were enrolled irrespective of their current reproductive intentions or plans to have children at the time of completing the questionnaire. Nevertheless, their responses provide valuable insights into the ethical, social, and reproductive considerations associated with the use of PGT-M in the context of hereditary cancer syndromes. Finally, the relatively small sample size, particularly in subgroup analyses, limited the statistical power to detect modest associations; therefore, the findings should be considered exploratory and interpreted with caution.

Conclusion

Although the survey included people with a genetically determined high risk of developing cancer at various stages of life and whose personal circumstances may have influenced their attitudes toward the use of IVF combined with PGT-M, the results show a strong interest in this reproductive option. Our findings suggest that PGT-M is perceived as a potentially valuable intervention for people who inherit hereditary cancer risk variants.

Improving access to genetic counselling, genetic testing, and PGT-M, along with implementing comprehensive preventive strategies and educational programs, may help reduce the burden of hereditary cancer predispositions and increase affected individuals' chances of having healthy biological offspring. Our findings support the routine integration of reproductive counselling and discussion of PGT-M into the multidisciplinary management of individuals with hereditary cancer predisposition.

Abbreviations

ART
assisted reproductive technologies
BC
breast cancer
HBOC
hereditary breast and ovarian cancer
HRR
homologous recombination repair
IVF
in vitro fertilisation
NGS
next-generation sequencing
PGT-M
preimplantation genetic testing for monogenic disorders.

Declarations

Ethics approval and consent to participate

The Poznan University of Medical Sciences Ethics Committee reviewed and approved the study (No. 511/2026). The study was conducted in accordance with the Declaration of Helsinki. The Committee determined that the anonymous survey was not a research experiment and could be used for scientific purposes. Participation was voluntary, and informed consent to participate was obtained from all participants through their voluntary completion and submission of the anonymous online questionnaire.

Consent for publication

Not applicable.

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Author Contribution

JK-G: conceptualization, methodology, investigation, questionnaire design, data curation, data analysis, writing – original draft. MB: methodology, questionnaire design, data analysis, writing – review & editing.

DS: writing – review & editing. IL: data analysis, writing – review & editing, final manuscript preparation. All authors have read and approved the final version of the manuscript.

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Availability of data and materials

The datasets analysed during the current study are available from the corresponding author on reasonable request.

Competing Interests

The authors declare that they have no competing interests.

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Figures

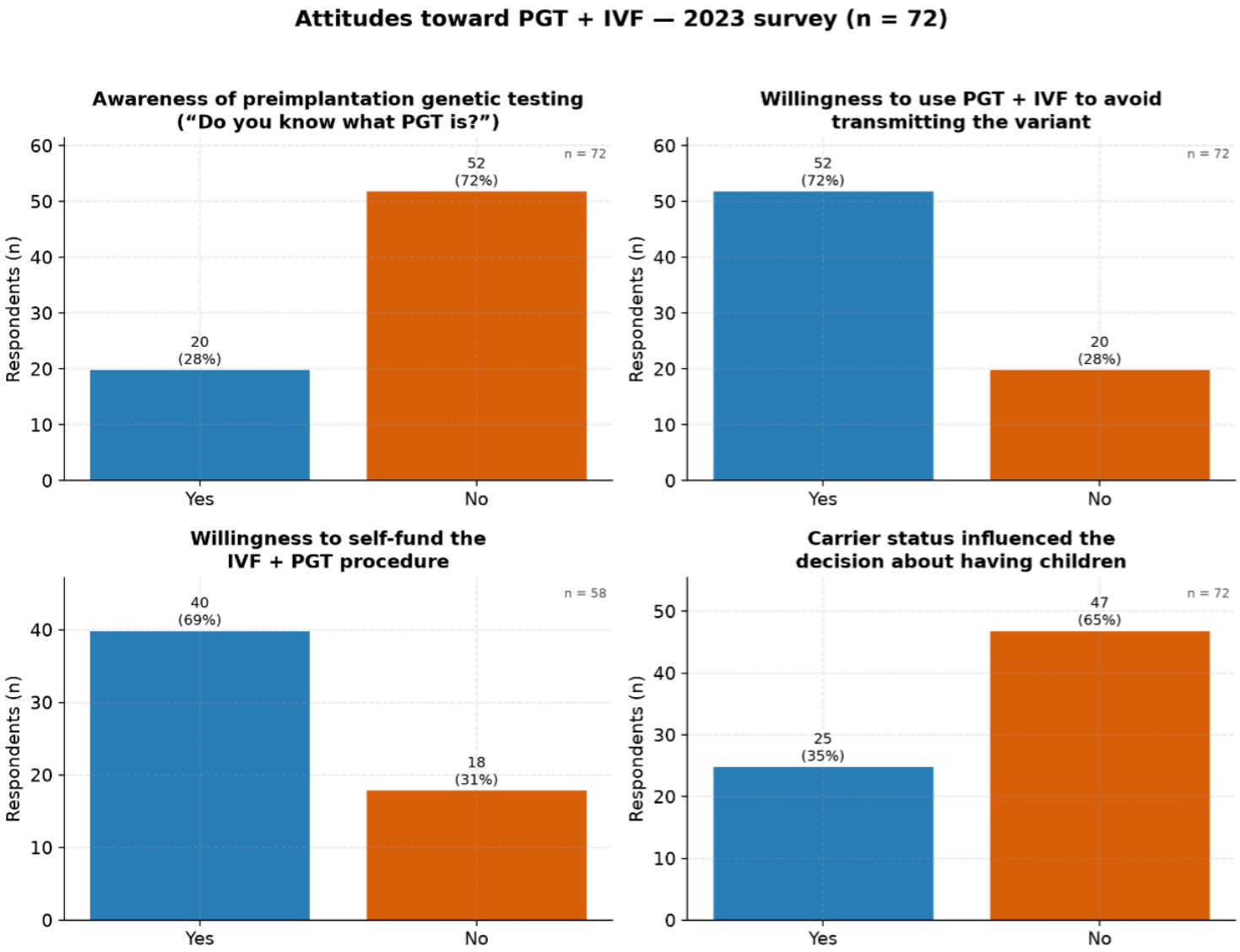


Figure 1

Core attitudes toward PGT-M + IVF (2023, n = 72).

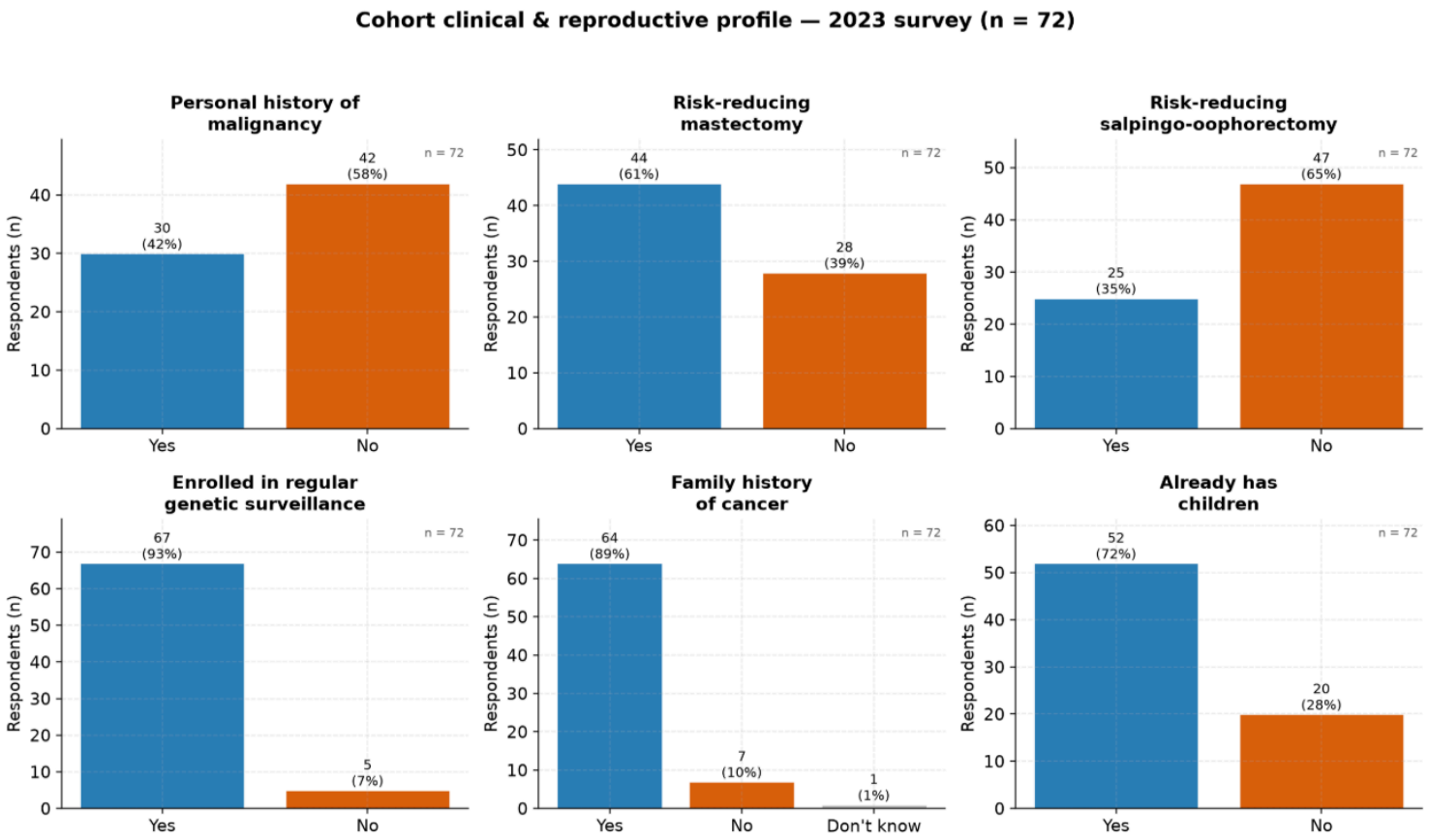


Figure 2

Clinical and reproductive profile of the cohort (2023, n = 72).

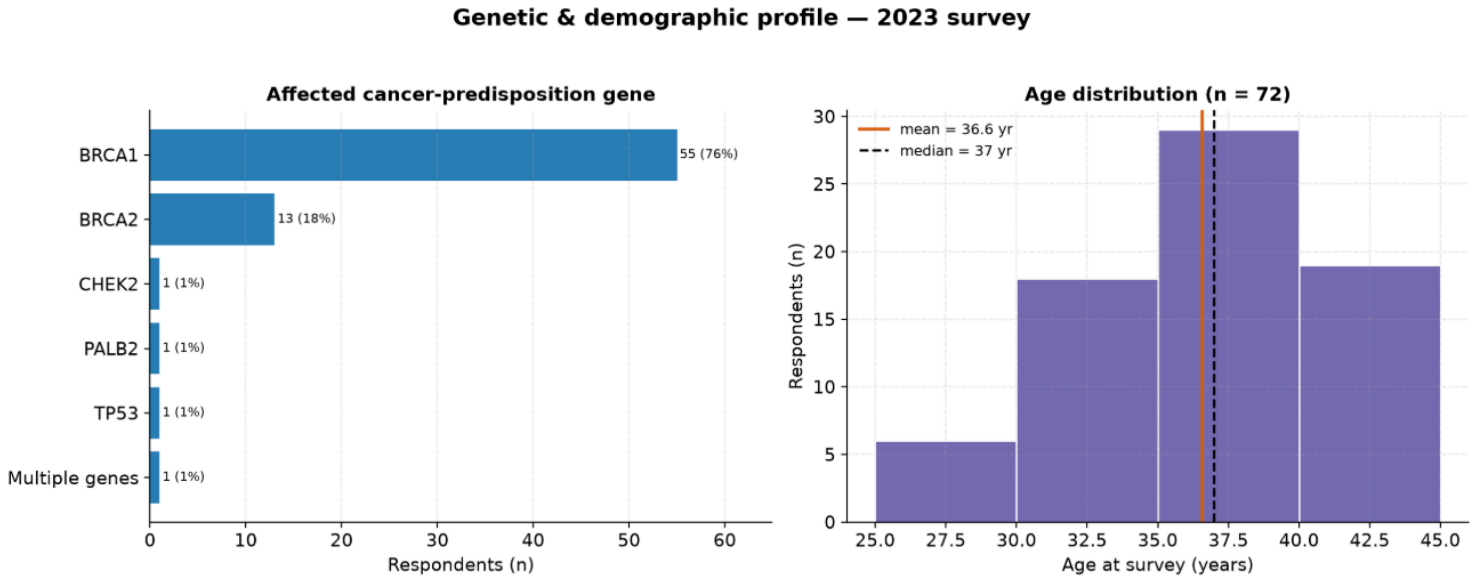


Figure 3

Affected cancer-predisposition gene and age distribution (2023, n = 72).

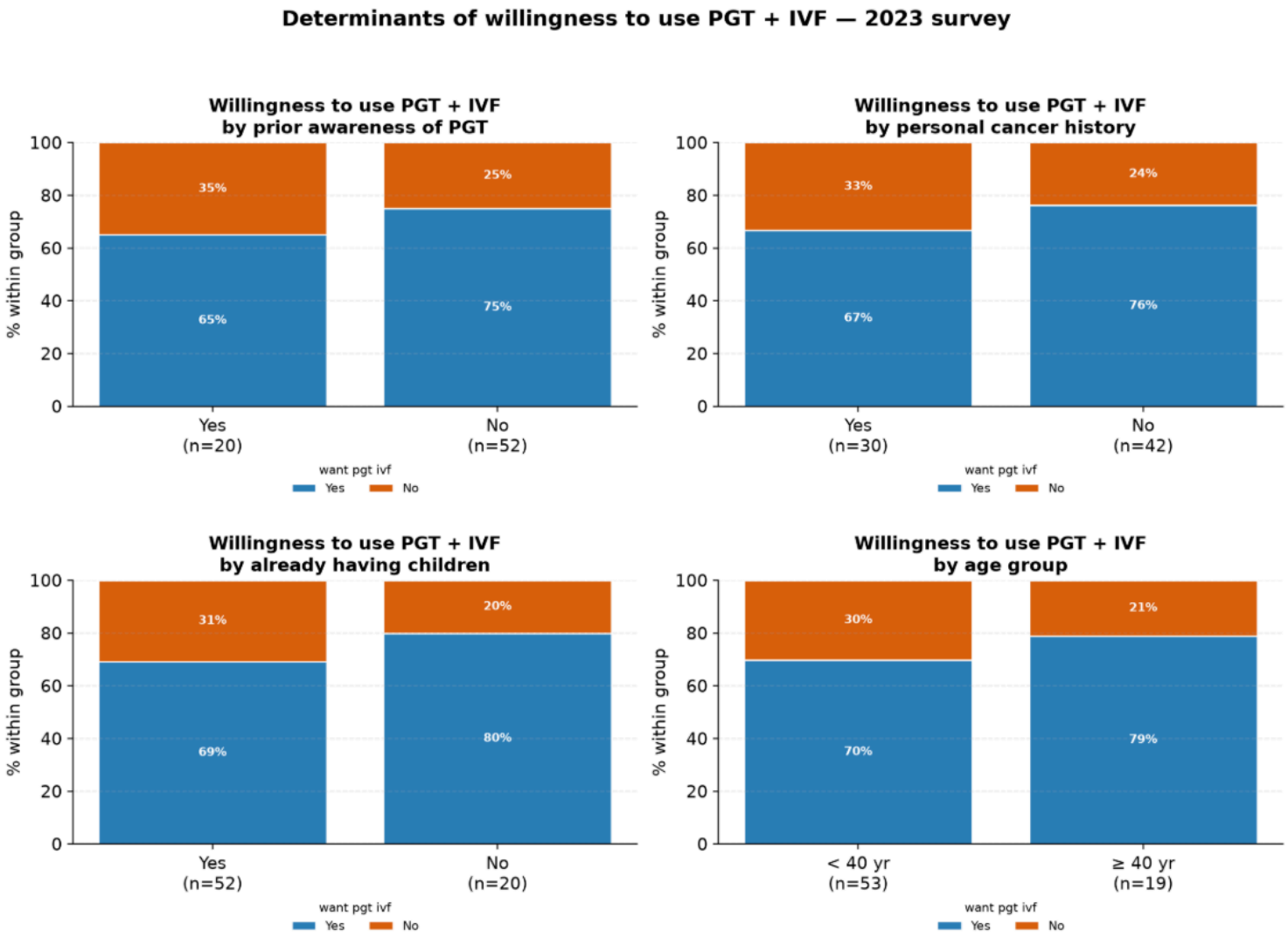


Figure 4

Willingness to use PGT-M + IVF stratified by candidate determinants (2023).

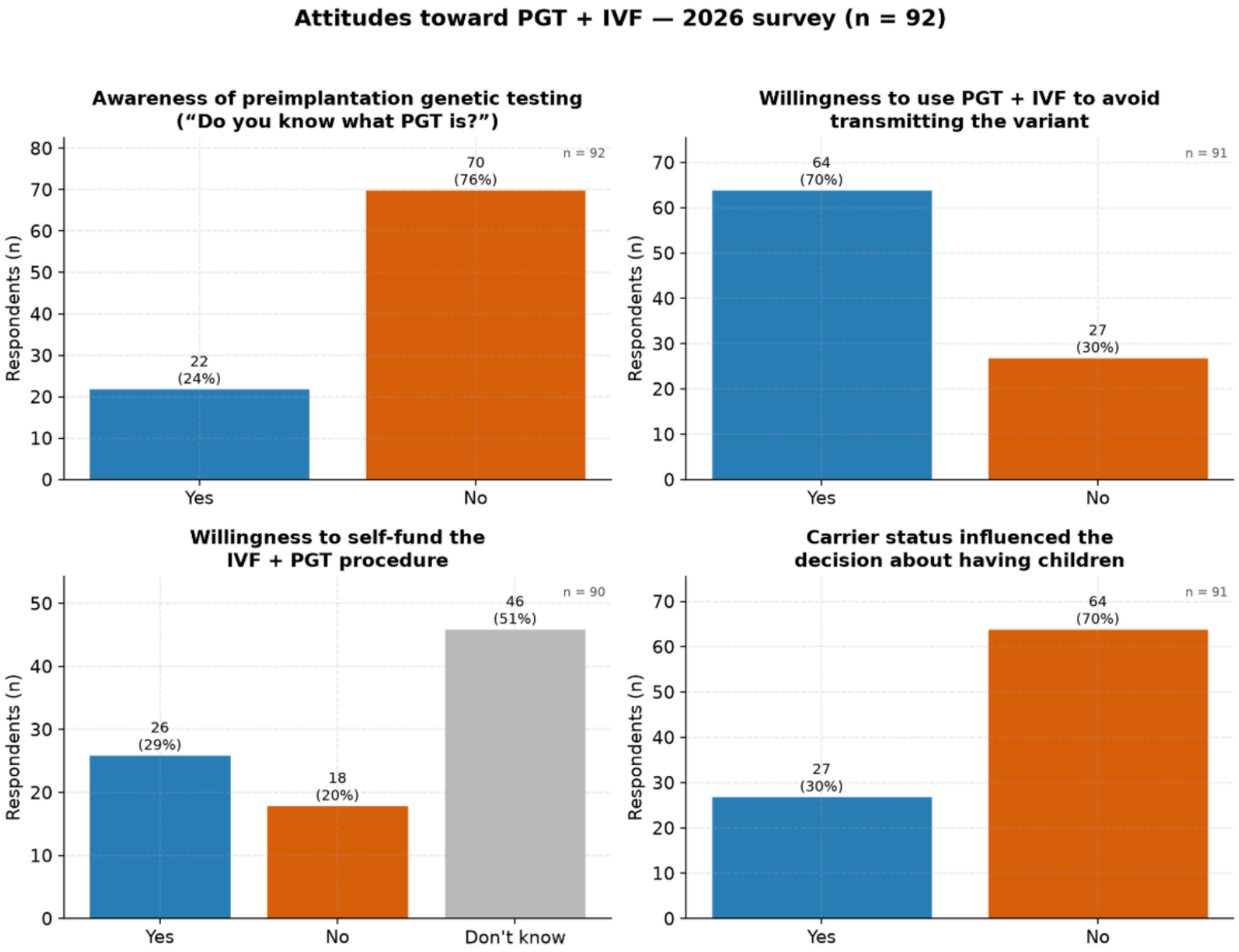


Figure 5

Core attitudes toward PGT-M + IVF (2026, n = 92).

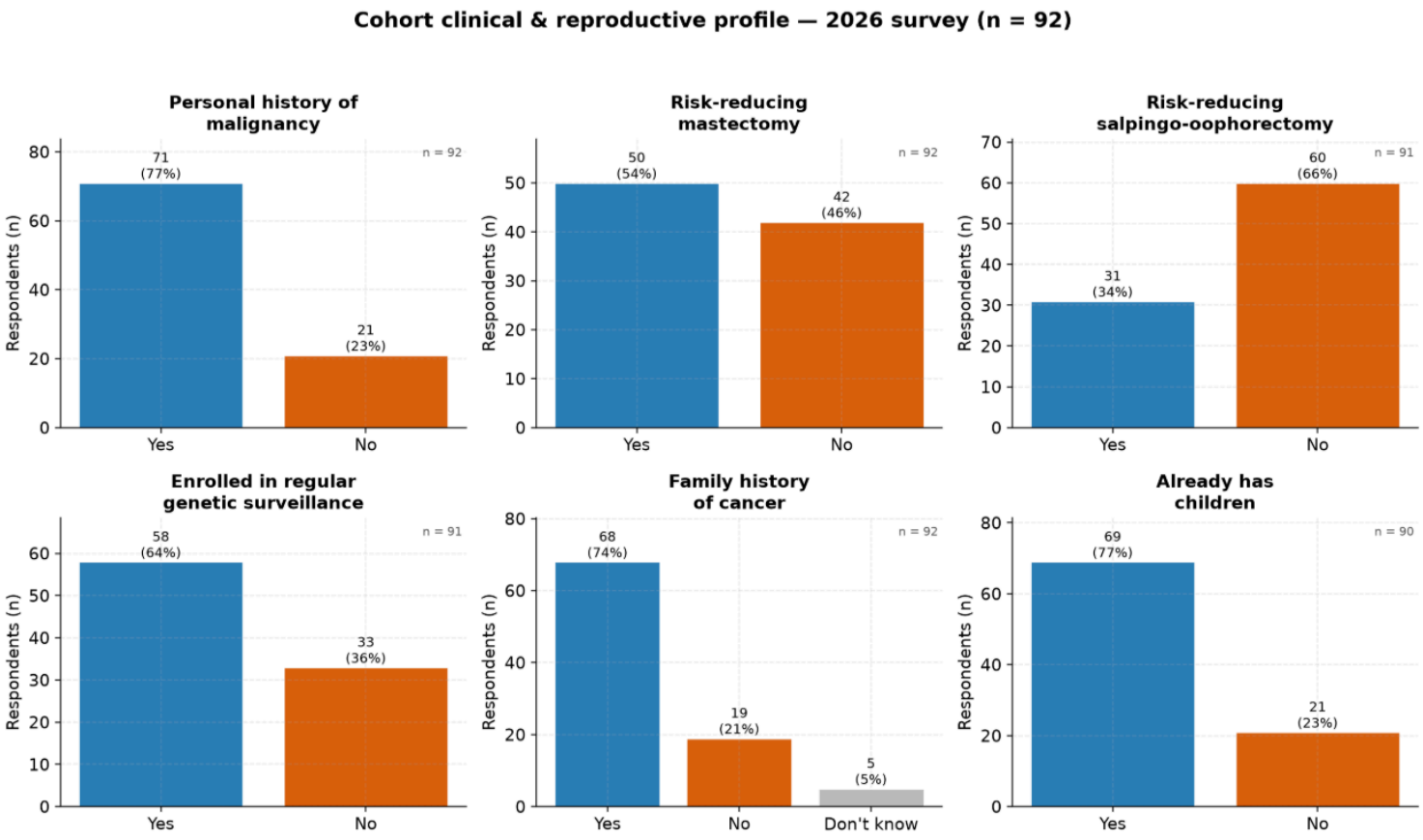


Figure 6

Clinical and reproductive profile of the cohort (2026, n = 92).

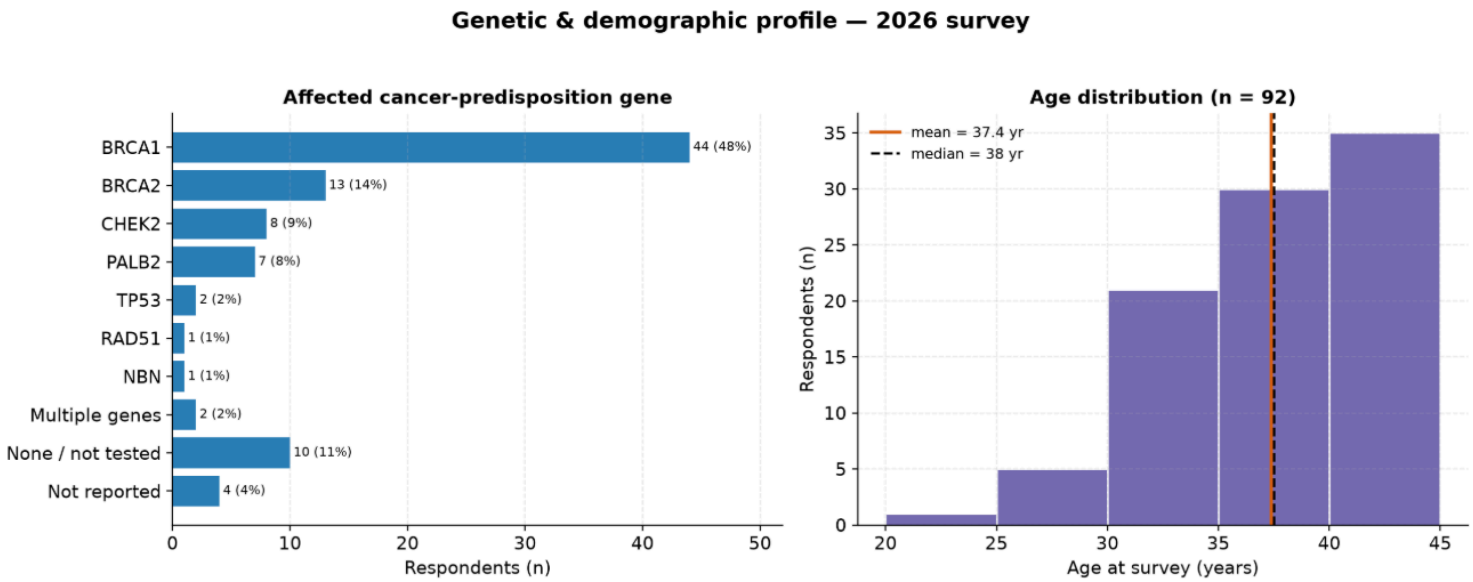


Figure 7

Affected cancer-predisposition gene and age distribution (2026, n = 92).

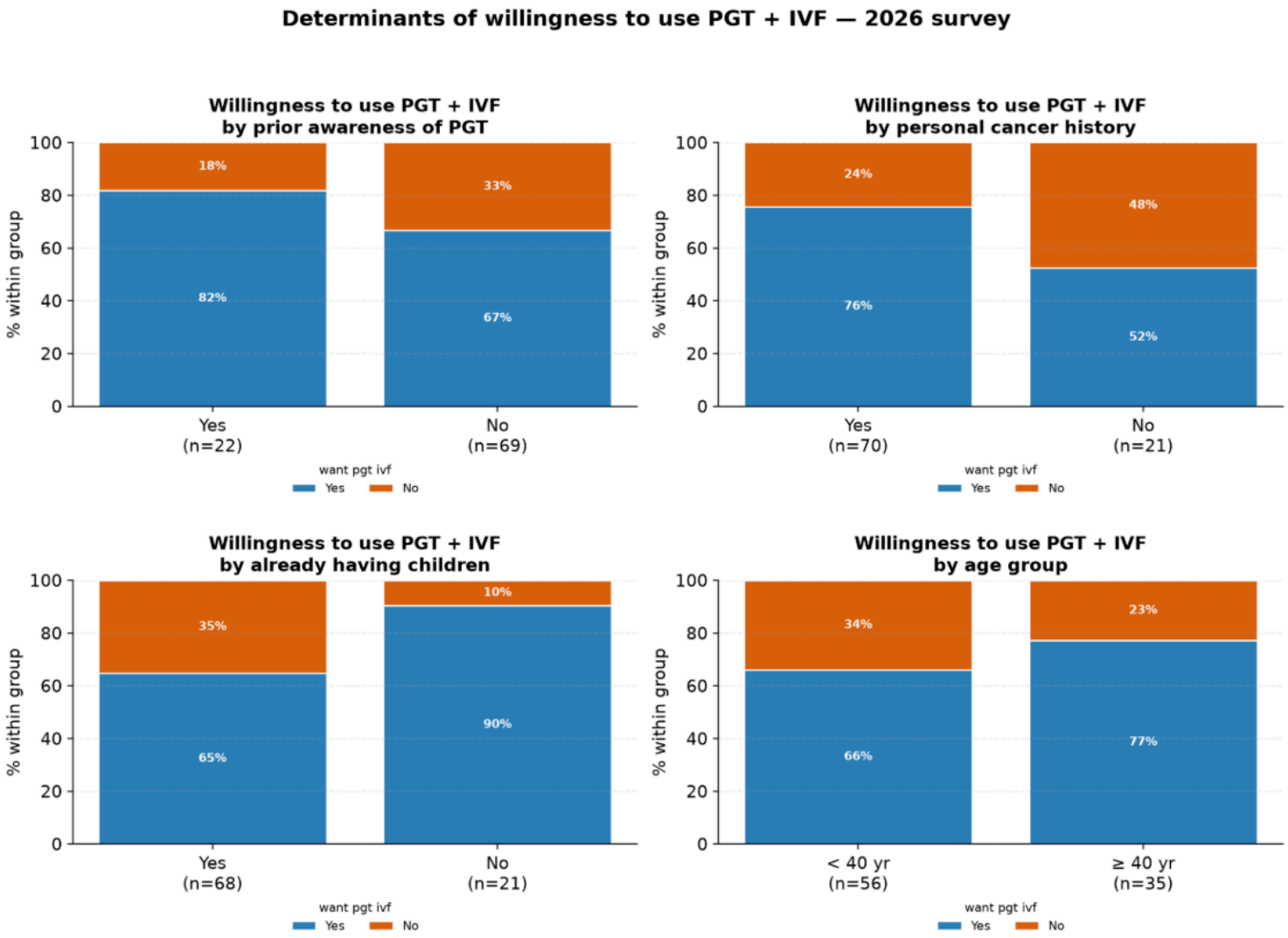


Figure 8

Willingness to use PGT-M + IVF stratified by candidate determinants (2026).

Supplementary Files

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- SupplementaryMaterial1QuestionnaireEnglish.docx