

Questionnaire survey on healthcare providers' awareness of PGT-M and their perceptions of the preference of individuals diagnosed with cancer predisposition syndromes: A single-center study in Japan

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Abstract

Background

Many cancer predisposition syndromes (CPSs) are inherited in an autosomal dominant manner, meaning pathogenic variants in causative genes have a 50% likelihood of being transmitted to offspring. Hence, a CPS diagnosis during reproductive age may impact family planning. Preimplantation genetic testing for monogenic disorders (PGT-M) and prenatal diagnosis (PND) are reproductive options that may help prevent the transmission of these variants. However, PGT-M awareness among healthcare providers (HCPs) in oncology settings in Japan remains unclear. This single-institution study surveyed HCPs' awareness of PGT-M and their perceptions of patients' preferences regarding these reproductive options.

Methods

HCPs at the Cancer Institute Hospital of the Japanese Foundation for Cancer Research were surveyed using an anonymous self-administered online questionnaire.

Results

Among 499 respondents, 7% (36/499) reported being "aware" of PGT-M, 14% (71/499) had "heard of it," and 79% (392/499) were "not aware." Over 90% of respondents thought that some individuals with CPS would be interested in or seek information about PGT-M or PND.

Conclusion

In this study, approximately 20% of HCPs in an oncology facility were aware of PGT-M. Greater awareness among oncology HCPs and support systems that enable individuals with CPSs to make informed reproductive decisions is required.

INTRODUCTION

Preimplantation genetic testing for monogenic disorders (PGT-M) for cancer predisposition syndromes (CPSs) has been actively discussed in Japan; however, domestic guidelines on what information to provide, and when, remain lacking. Hereditary breast and ovarian cancer (HBOC), a representative CPS caused by pathogenic BRCA1 or BRCA2 variants [1], can affect partner relationships and childbearing decisions [2–4]. Reproductive options for avoiding transmission include PGT-M and prenatal genetic diagnosis (PND), and several countries recommended informing individuals or couples diagnosed with a CPS about these options [5, 6]. The Human Fertilisation and Embryology Authority (HFEA) lists over 2,000 disorders approved for PGT-M [7].

Preimplantation genetic diagnosis was first performed in 1990 for couples at risk of X-linked diseases [8],

initially restricted to severe childhood-onset conditions. In 1999, however, PGT-M targeting the *TP53* gene—one of the CPS genes—resulted in the birth of a child without pathogenic *TP53* variant [9].

Subsequently, the European Society of Human Reproduction and Embryology (ESHRE) in 2003, and the HFEA in 2006 approved PGT-M for adulthood-onset hereditary cancers with incomplete penetrance [10, 11], and in 2018 the Practice Committee of the American Society for Reproductive Medicine (ASRM) and the Society for Assisted Reproductive Technology endorsed PGT-M for hereditary cancers, emphasizing reproductive freedom [12]. ESHRE reported approximately 1,400 PGT-M procedures in 2018, about 5% for HBOC [13]. Unlike PND, PGT-M is performed before implantation, avoiding the difficult decision of whether to continue an affected pregnancy.

In Japan, PGT-M for severe childhood-onset hereditary disease was approved in 2004. In 2022, promoted by an application from a patient with hereditary retinoblastoma, the Japan Society of Obstetrics and Gynecology (JSOG) revised its "Clinical opinion on Preimplantation Genetic Testing for Monogenic Disorders (PGT-M)" [14]. Since then, discussion of PGT-M for CPSs has increased, though HCPs' awareness of it remains unclear.

We previously reported the initial findings from a single cancer center survey of Japanese oncology HCPs regarding their awareness of PGT-M and their perceptions of patients' preference toward it [15]. Here, we analyzed additional data from the same survey to further examine challenges associated with PGT-M for CPSs in Japan.

PATIENTS AND METHODS

Respondent recruitment

Participants were healthcare providers at the Cancer Institute Hospital of the Japanese Foundation for Cancer Research (JFCR) and its affiliated institutes. Survey invitations were emailed to all JFCR staff (approximately 2,800), though only healthcare providers (estimated at approximately 2,200) were eligible to respond. Clinical specialty data were not collected to preserve anonymity. Respondents included clinical healthcare professionals and patients care support personnel (e.g., physicians, nurses, allied health professionals, and clinical assistants).

Questionnaire survey method

The survey used a self-designed online questionnaire. Respondents were informed that it addressed cancer and reproductive health and provided consent before participating. Responses were collected anonymously from November 25 to December 31, 2022 (approximately 5 weeks). The questionnaire

comprised 27 items, including open-ended and branching questions, and took approximately **10–15 minutes** to complete.

Statistical analysis

Between-groups differences were analyzed using the chi-square test. Adjusted standardized residuals were calculated to identify the specific cells contributing to significant differences, and Cramér's V was used to assess effect sizes. Post hoc pairwise comparisons were conducted using Fisher's exact test, with p-values adjusted by the Benjamini-Hochberg method (Benjamini & Hochberg, 1995). For two related questions, agreement between respondents was assessed using linear weighted Cohen's kappa coefficient, followed by comparison of the response distributions using the Wilcoxon signed-rank test. Statistical significance was set at $p < .05$. Missing responses for individual items were excluded from the statistical analysis on an item-by-item basis. All analyses were conducted using js-STAR XR+ release 2.5.0j for macOS and R for macOS Cocoa GUI (Urbanek et al., 2025).

RESULTS

Participant characteristics

A total of 509 individuals accessed the survey; of these, 499 provided consent and were included in the survey [15]. Most participants completed the survey in full, though missing data were observed for some items.

Of those 499 respondents, 117 (24%) were male, 381 (76%) were female, and 1 (0.2%) identified as neither (Table 1). By profession, 264 (53%) were nurses, 108 (21%) were physicians/dentists, and 127 (25%) other medical staff (included genetic counselors). Ages ranged from 20s to 70s, and oncology experience ranged from less than 1 year to ≥ 20 years.

Awareness of PGT-M and PND

For PGT-M, 7.2% (36/499) were "aware," 14.2% (71/499) had "heard of it," and 79% (392/499) were "not aware." For PND, the corresponding proportions were 76% (380/499), 20% (99/499) and 4% (20/499) (Figure 1a). The Wilcoxon signed-rank test showed significantly higher awareness of PND than PGT-M ($z = 18.04, p < .001$; cross-tabulation in Supplementary table S1).

Differences in awareness by respondent characteristics are shown in Supplementary Tables S2-S5. Significant differences were observed for PGT-M by age and parental status, and for PND by age. Multiple comparisons (Benjamini-Hochberg adjusted) showed significant differences between physicians/dentists and nurses, and between physicians/dentists and other healthcare providers for both PGT-M and PND; no significant difference was found between nurses and other healthcare providers (Supplementary Table S5).

Among 107 respondents (21.4%) aware of PGT-M ("aware" or "have heard of it," Figure 2a), 23% (25/107) knew the difference between PGT-M and PND. Among the 57 respondents with such knowledge, 21% (12/57) did not know that PGT-M requires in vitro fertilization, and 46% (26/57) correctly identified that PGT-M in Japan is primarily indicated for serious childhood-onset genetic disorders.

HCPs' perceptions of preferences for PGT-M and PND among individuals with CPSs

Respondents received a brief explanation of PGT-M and PND before answering. Overall, 56% (277/499) believed some individuals with CPSs wish to undergo PGT-M, and 38% (190/499) believed some would seek information about it; corresponding rates for PND were 65% (323/499) and 30% (Figure 1b). Cohen's kappa indicated high agreement between perceptions of PGT-M and PND ($\kappa = 0.68$, 95% CI: 0.61–0.76); however, the Wilcoxon signed-rank test revealed a significant difference ($z = 4.11$, $N = 90$, $p < .001$), with ratings of patients' preferences for PND ($M = 3.59$, $SD = 0.6$) significantly higher than for PGT-M ($M = 3.49$, $SD = 0.62$) (Table S6a, S6b).

Awareness of CPSs and clinical experience with patients diagnosed with CPSs and their families

Of 499 respondents, 464 (93.0%) reported being "very familiar" or "somewhat familiar" with CPS (Figure 2b). Among those, 30% (138/464) had clinical experiences with either patients with CPS or their family members; of those, half (70/138) had treated patients whose family planning was directly affected by a CPS diagnosis, and 18.1% (25/138) had treated patients who forwent childbearing specifically because of a CPS diagnosis rather than cancer itself.

Stratifying PGT-M awareness by CPSs familiarity (Supplementary table S7a), 36.4% (12/33) of those "very familiar" with CPSs were unaware of PGT-M, rising to 80.5% (347/431) among those "somewhat familiar" and to 94.1% (32/34) among those who had never heard of CPSs ($\chi^2 (4) = 64.546$, $p < .001$, Cramér's $V = .255$). Post hoc comparisons showed significant differences between "very familiar" and "somewhat familiar" ($p < .001$) and between "very familiar" and "never heard of it" ($p < .001$), but not between "somewhat familiar" and "never heard of it" ($p = .192$) (Supplementary table S7b).

Attitude toward PGT-M for CPSs

Respondents were asked about their attitude toward PGT-M for childhood-onset and adult-onset CPS, both as HCPs and as individuals (Figure 3); for both onset types, respondents were more likely to endorse PGT-M from a personal perspective than as HCPs. Linearly weighted Cohen's kappa indicated consistency between these perspectives within individuals (childhood-onset: $\kappa = 0.69$, 95% CI: 0.63–0.76; adulthood-onset: $\kappa = 0.75$, 95% CI: 0.70–0.81) (Supplementary table S8 a and b). The Wilcoxon signed-rank test showed no significant difference between perspectives for childhood-onset CPS ($p = .2077$), but a significant difference was found for adulthood-onset CPS ($p < .05$) (Supplementary table S8c).

DISCUSSION

This survey was conducted at the Cancer Institute Hospital of JFCR, which has provided care for individuals with CPSs for over 20 years through multidisciplinary collaboration. Notably, it coincided with the JSOG's 2022 revision of its clinical opinion on PGT-M [14], a time when PGT-M practice in Japan was expected to reach a turning point.

Only 21% of respondents were aware of PGT-M (16 physicians/dentists, 9 nurses, 11 other providers), compared with 96% for PND (100% among physicians/dentists), indicating a substantial awareness gap between the two tests among HCPs at this oncology facility. Awareness also varied by age, with opposite trends: PGT-M awareness was higher among respondents in their 60s and lower in their 20s ($p = .0069$, Cramér's $V = .179$), whereas PND awareness was higher in their 30s and lower in their 60s ($p = .0265$, Cramér's $V = .159$). Furthermore, over 90% of HCPs believed some individuals with CPS wish to undergo, or seek information about, PGT-M as well as PND, although the reported percentage was higher for PND than for PGT-M.

Indeed, a CPS diagnosis at reproductive age has been reported to influence reproductive choices [2–4], and several HCPs in our survey similarly reported having encountered individuals whose reproductive choices were influenced by a CPS diagnosis. Reflecting this, some international guidelines recommend providing information on reproductive options to individuals diagnosed with CPS at reproductive age, enabling informed decision-making. Furthermore, some have argued that PGT-M, by avoiding the difficult decision of whether to continue an affected pregnancy required by PND, may be an ethically preferable option in certain cases, although this view is not universally held [12].

Despite this potential advantage of PGT-M, our survey found that a higher proportion of HCPs believed individuals diagnosed with CPS would want PND than would want PGT-M.

One possible explanation is a difference in awareness: respondents appeared less familiar with PGT-M than with PND. Although brief explanations were provided before the survey questions, HCPs' greater pre-existing familiarity with PND, relative to PGT-M, likely shaped their responses. This is considered to have resulted in slightly more "likely exist" responses for PND than for PGT-M.

Several factors may explain why awareness of PND was higher than that of PGT-M. First, the two tests have been considered in differing clinical contexts.

Before JSOG revised its clinical opinion on PGT-M in 2022, PGT-M indications in Japan were limited to severe childhood-onset genetic conditions. The facility surveyed specializes in cancer care, and most cancers, including many CPSs, develop in adulthood. Unlike the hereditary conditions traditionally indicated for PGT-M, many CPSs are characterized by incomplete penetrance and have generally been considered actionable. Consequently, the conditions targeted by PGT-M were largely outside the respondents' field of expertise. Although discussions regarding PGT-M in the context of CPS have been taking place even before the 2022 revision of the clinical opinion, it cannot be ruled out that these discussions were limited to affected individuals and their families, as well as a subset of HCPs.

Second, in contrast, PND is a reproductive test that anyone—including HCPs themselves— might consider, making it a highly relevant issue for many prospective parents.

Third, the introduction of non-invasive prenatal cell-free-DNA screening in Japan in the 2010s sparked widespread public and professional debate, and the JSOG's clinical opinion on PND further emphasized non-directive counseling to support informed decision-making [16].

Fourth, PND is included in genetics education for medical and nursing students, whereas PGT-M is not yet a mandatory topic [17, 18], giving HCPs more exposure to PND information.

Studies from other countries support this awareness-implementation gap. Among 145 obstetricians, internists, and pediatricians managing patients with genetic disorders, 88% agreed that at-risk parents should be informed about PGT-M, yet only 23% had made referrals, with 63% citing unfamiliarity as a barrier [19].

Similar findings have been reported for nurses: among 201 surveyed, 78% had never heard of PGD (now commonly referred to as PGT-M), and among the 18% (37) who had, 59% described their knowledge as limited; overall, 89% of respondents cited lack of confidence in their understanding of PGD as a barrier to discussing it with patients [20].

Since the 2022 revision of JSOG's clinical opinion, the practice of PGT-M for CPSs in Japan has been evolving. According to JSOG data as of September 2026, the number of PGT-M applications and approvals has been increasing, with applications submitted for conditions such as retinoblastoma, rhabdoid tumor predisposition syndrome, Li-Fraumeni syndrome (LFS), familial adenomatous polyposis, and von Hippel-Lindau disease. Approved cases have been reported for LFS and retinoblastoma [21]. The number of PGT-M applications for CPS in Japan is expected to increase further, potentially including adulthood-onset conditions. Given this clinical demand, raising HCPs' awareness of PGT-M is increasingly important, as it could facilitate appropriate response to patients' inquiries and smoother referrals to specialized genetic counseling.

One way to raise awareness of PGT-M is to include it explicitly in cancer treatment guidelines. This could help standardize knowledge and promote collaboration between oncology and reproductive medicine. In Japan, the Fertility Preservation Clinical Practice Guidelines 2024 and Diagnostic and Treatment Guidelines for Hereditary Breast and Ovarian Cancer 2024 now mention PGT-M in the context of fertility preservation. International clinical guidelines and position statements recommend that patients be informed about reproductive options, including PGT-M and PND. However, Cabana et al. reported that clinical practice guidelines alone have limitations in increasing HCP awareness [22].

Fertility preservation offers a comparable precedent: although guidelines have long recommended informing eligible patients [23, 24], HCP awareness was initially low even among oncologists [25], and guidelines alone proved insufficient—multidisciplinary collaboration and referral to reproductive specialists were additionally needed. Similar strategies may help raise PGT-M awareness in Japan.

Gietel-Habets et al. noted that, although genetics professionals bear the primary responsibility for providing PGT-M information, other specialists can serve as important facilitators, helping patients take the initial step toward genetic counseling [26]. As cancer care increasingly relies on multidisciplinary and interprofessional collaboration, leveraging these established networks may be an effective way to raise organizational awareness of PGT-M.

Because few facilities in Japan offer PGT-M, collaboration with internal and external specialists is essential for seamless patient consultation and referral. Fertility preservation networks may be particularly well suited to this role: some cancer survivors with preserved fertility may seek PGT-M in the future, and the multidisciplinary relationships already built through these programs could similarly help raise HCP awareness of PGT-M. Notably, some respondents in our survey indicated that participating in the survey itself was a valuable learning experience, suggesting that even small-scale initiatives can serve as catalysts for raising awareness among HCPs.

Ethical considerations are central to discussions of PGT-M. As the ASRM Ethics Committee has noted, reconciling conflicting perspectives can be challenging; however, reproductive autonomy must remain a priority [27]. HCPs must thus provide neutral, balanced information that supports informed decision-making and respects each couple's values and beliefs.

Limitations

This survey has several limitations. First, it reflects the perspectives of HCPs at a single cancer facility and does not capture the views of HCPs elsewhere or individuals diagnosed with CPS themselves; the generalizability of the findings to oncology HCPs in Japan is therefore limited. Second, to preserve

anonymity, respondents' specific medical specialties were not recorded, precluding analysis of associations between specialty and responses. Third, selection bias is possible, as HCPs with a particular interest in reproductive medicine or cancer predisposition syndromes may have been more likely to respond. Fourth, the self-developed questionnaire had not been sufficiently validated for reliability and validity.

Finally, because the survey was conducted in late 2022, awareness and clinical practices may have since changed.

Conclusion

This survey assessed the awareness of PGT-M among HCPs at an oncology facility and their perceptions of patient demand. The findings indicate that awareness remains limited, although HCPs recognize potential demand for PGT-M among patients. To advance educational initiatives and establish clinical support systems, multifaceted discussions among HCPs are essential.

Declarations

Data availability

The datasets generated during the current study are available from the corresponding author upon reasonable request due to ethical restrictions.

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Author contribution

H.A.: Conceptualization, methodology, investigation, data curation, formal analysis, and writing—original draft. A.U.: Conceptualization, supervision. All authors reviewed and approved the final manuscript.

Ethical declaration

This study was approved by the Ethics Committee of the Cancer Institute Hospital of the Japanese Foundation for Cancer Research (Approval No. 2022-GB-063) and was conducted in accordance with the Declaration of Helsinki. The submission of the questionnaire constituted informed consent.

Conflict of Interest

A. U., K. K., and K. I. received research grants from Pfizer Inc. for unrelated projects. The other authors declare no competing interest. This study received no support from Pfizer Inc.

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Table

Table 1. Participant characteristics (n = 499)

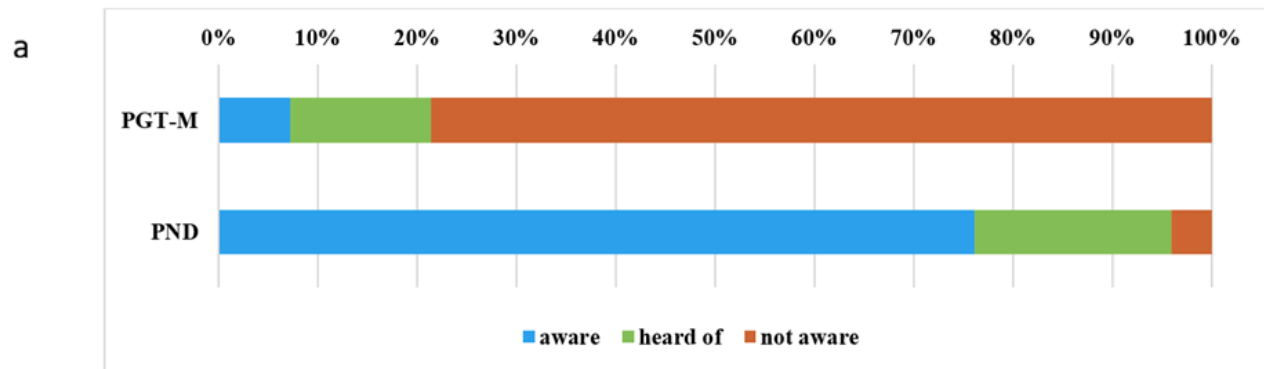
Characteristic(p-value)	n = 499 (%)
Gender ^a (p<.001)	
Male	117(23.4%)
Female	381(76.4%)
Neither	1(0.2%)
Professional variables ^a (p<.001)	
Physician, Dentist	107(21.4%)
Nurse	264(52.9%)
Clinical laboratory technologist	35(7%)
Nursing assistant	19(3.8%)
Pharmacist	18(3.6%)
Clinical Research Coordinator	6(1.2%)
Medical social worker	5(1.0%)
Certified Genetic Counselor	5(1.0%)
Other	40(8.0%)
Presence of children ^b (p<.005)	
Yes	215(43.1%)
No	284(56.9%)
Age ^a (p<.001)	
Up to 29 years old	84(16.8%)
30–39	158(31.7%)
40–49	147(29.5%)
50-59	77(15.4%)
60–69	30(6.0%)
Over 70 years old	3(0.6%)
Years of clinical experience of oncology ^a (p<.001)	
Up to 1	33(6.6%)
3	57(11.4%)

5	56(11.2%)
10	117(23.4%)
15	91(18.2%)
20	56(11.2%)
> 20	89(17.8%)

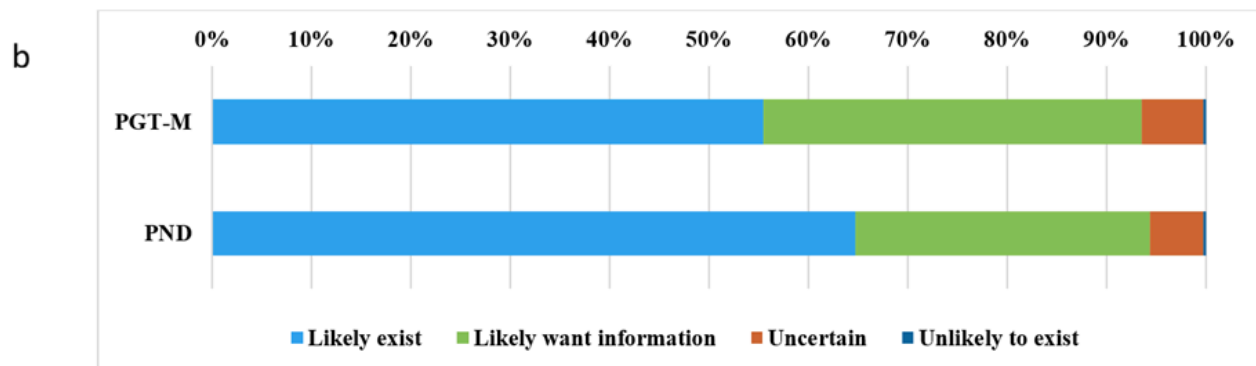
Note: ^a: Analyzed using the Chi-square test. ^b: Analyzed using Fisher's exact test ($\alpha = 0.05$, two-tailed). p -values indicate statistical significance

($p < .05$)

Figures



Note. PND = prenatal diagnosis; PGT-M = preimplantation genetic testing for monogenic disorders.



Note. PND = prenatal diagnosis; PGT-M = preimplantation genetic testing for monogenic disorders.

Figure 1

(a) Comparison of awareness levels between preimplantation genetic testing for monogenic disorders (PGT-M) and prenatal diagnosis (PND) (*n* = 499).

(b)Healthcare providers' perceptions of preference for regarding PGT-M and prenatal genetic diagnosis among individuals diagnosed with CPSs and Their families in Japan. (*n* = 499).

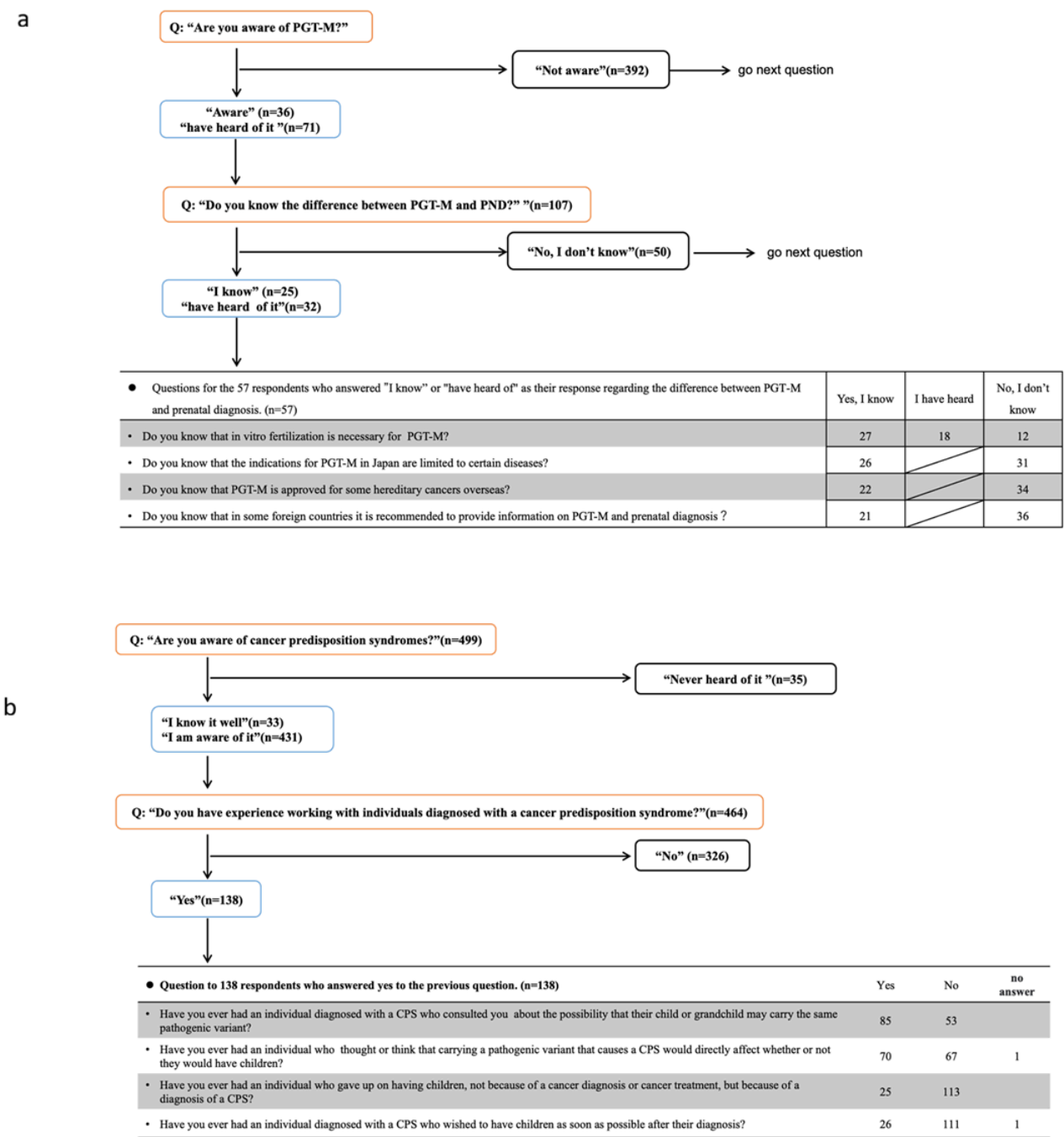


Figure 2

a. Flow of questions regarding PGT-M and answers.

b. Flow of questions and responses regarding awareness and clinical experience of cancer predisposition syndrome

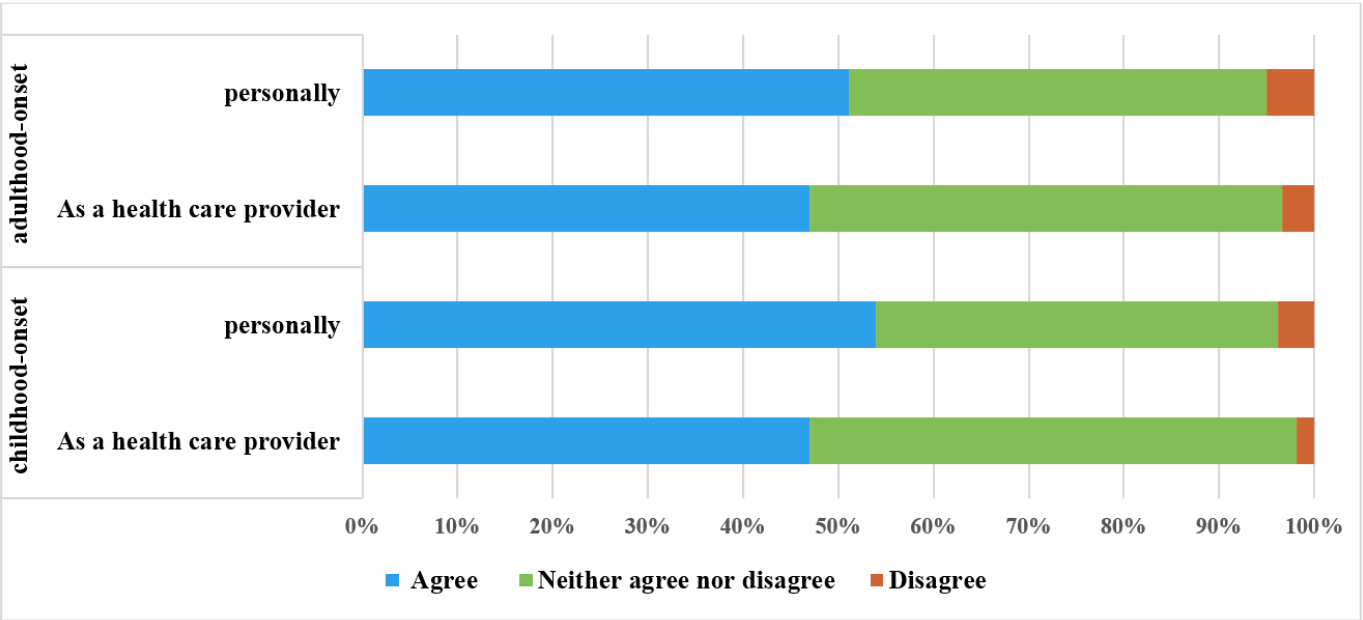


Figure 3

Comparison of Attitudes toward PGT-M for Childhood- and Adult-Onset CPS: between HCPs and Individuals.

Figure 3 shows how HCPs responded regarding PGT-M for childhood-onset CPS and adulthood-onset CPS, answering both from their professional standpoint (as HCPs) and from a personal standpoint. Response options were "Agree," "Neither agree nor disagree," and "Disagree."(n=499)

Supplementary Files

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