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Challenges in identifying paediatric cancer predisposition syndromes: international SCOPE survey and SIOPE expert consensus recommendations

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Cancer Predisposition Syndromes (CPS) are heritable genetic conditions associated with an increased risk of developing various cancers throughout life. While early identification and tumour surveillance can improve outcomes, CPS are often underdiagnosed in clinical practice. To evaluate clinicians' perspectives and identify barriers to CPS identification and care across Europe, we conducted the SCOPE study: a three-part cross-sectional survey of paediatric haematology/oncology professionals, followed by a modified Delphi consensus process with members of the SIOP Europe Host Genome Working Group. A total of 185 paediatric oncologists from 22 countries participated in the survey. More than 40% of participants reported low or uncertain confidence across different CPS-related tasks, particularly in counselling families (64.3%) and interpreting germline genetic findings (57.3%). Access to clinical geneticists and dedicated CPS clinics were predictors of higher confidence for some domains, while individual experience and institutional patient volume had limited influence. Regular use of universal CPS screening tools was low (42.3%), with most clinicians relying on personal judgement rather than structured criteria. The most cited barriers were lack of screening guidelines (57%) and difficulties in interpreting results (35.1%). Regular training and workshops, availability of genetic counsellors or educators for patient support, and patient-friendly education material were most cited as areas of improvement. The Delphi process led to three recommendations: (1) improve clinician training and communication strategies, (2) integrate CPS screening into standard treatment plans, and (3) develop accessible, patient-centred educational materials. These recommendations highlight opportunities to enhance CPS care through structured support, interdisciplinary collaboration, and systematic screening approaches.

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BACKGROUND

Cancer predisposition syndromes (CPS) are heritable genetic conditions that increase an individual's lifetime risk of developing cancer. Recent studies report that 8–18% of childhood cancer patients have an underlying CPS [1–5]. Timely recognition of a CPS has various clinical implications which can include treatment modifications for the primary malignancy. For example, excess toxicity to certain chemotherapy agents and radiotherapy can occur in CPS such as Li-Fraumeni syndrome, Bloom Syndrome, or Nijmegen Breakage syndrome, while resistance to some chemotherapy agents such as temozolomide occurs in patients with mismatch repair deficiency, who may instead respond to newer agents such as immune checkpoint inhibitors [6–9]. Children with a CPS additionally have an elevated risk of developing second primary neoplasms (SPN). Among childhood cancer survivors, SPN represent the second leading cause of mortality, after the primary malignancy [10–12]. A study conducted in Switzerland calculated the cumulative incidence of any SPN from diagnosis to 20 years

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after first primary cancer diagnosis in patients with CPS to be 23% versus 2.7% in those without [13]. Studies in 5-year childhood cancer survivors found that genetic predisposition and cancer treatments were the major contributors to SPN [14, 15]. Together, these studies emphasise the clinical importance of timely detection of CPS. Importantly, identifying a CPS can also inform surveillance protocols and lead to cascade genetic testing of other at-risk family members, benefiting the broader family by facilitating timely neoplasm detection and improving management.

Despite these implications, CPS are underdiagnosed in clinical practice, where testing is based on clinical suspicion, compared to research cohorts that apply systematic germline genetic testing to all childhood cancer patients [13, 16]. Referral for genetic testing often relies on the treating oncologist's discretion and is typically guided by clinical features suggestive of a CPS, such as specific tumour types, congenital anomalies, presence of developmental delay, haematological, endocrine, distinct skin findings, or a suggestive family history [17]. Access to germline sequencing is limited in some institutions, reimbursement issues may hinder genetic work-up, and subjective case-by-case referral practices can result in missed or delayed CPS diagnoses.

To support healthcare providers, various screening tools have been developed for identifying at-risk patients and improving CPS detection. These include syndrome-specific tools, cancer-specific tools, and tools that can be applied to all children with cancer, regardless of tumour type or clinical features. Examples include the Jongmans' criteria, an easy-to-use, single-page selection sheet (modified later by Ripperger et al.), and the MIPOGG tool, an app-based algorithm that utilises both universal and tumour-specific criteria to give a binary recommendation for or against genetic referral [17–19].

While some efforts have assessed CPS screening and management practices at the institutional level, including the LEGEND-COST survey, there are limited data capturing individual clinician perspectives across Europe [20]. To address this gap and to better understand the current landscape of CPS management practices in Europe, we initiated, with the support of the European Society for Paediatric Oncology (SIOPE), the Survey on Cancer Predisposition Identification and Observation Practices in Europe (SCOPE). Here, we report the results from the three-part survey of paediatric oncology professionals, along with consensus recommendations derived from a modified Delphi approach to improve paediatric CPS care in Europe.

METHODS

Study design and population

We developed a cross-sectional, three-part online survey, which was distributed to paediatric oncologists and haematologists across Europe. National collaborators from 27 countries were identified and tasked with disseminating the survey to members of their respective professional societies. We used REDCap (Vanderbilt University, Nashville, TN, USA, version 15.01.11) to distribute the survey and collect the information. Upon signing in to the survey and providing their email, participants received up to five reminders to complete the survey. The ethics committee of the canton of Bern waived an ethics evaluation (Req-2022-00455).

Outcomes

The primary outcome was clinician confidence across four CPS-related domains, assessed using a 5-point Likert scale where 1 indicated very low confidence; 2, low confidence; 3, uncertainty, 4, high confidence; and 5, very high confidence. Participants rated their confidence in: (i) deciding which childhood cancer patients need genetic work-up ("How confident do you feel with evaluating childhood cancer patients for their risk of having an underlying genetic cause for their cancer diagnosis – which patient needs genetic work-up?); (ii) utilising somatic genetic data ("How confident do you feel with interpreting the results of somatic (tumour) genetic findings in childhood cancer patients?"); (iii) utilising germline genetic findings ("How confident do you feel with interpreting the results of germline genetic findings in childhood cancer patients?"); (iv) counselling families on CPS ("How confident do you feel with family counselling in the event of identification of a cancer predisposition syndrome in a childhood cancer patient?").

Secondary outcomes included clinicians' self-reported knowledge and the use of CPS screening tools, perceived barriers to CPS care, and the availability and adequacy of institutional resources, including educational tools.

Survey content and explanatory variables

The survey was structured into three parts: (1) demographics, CPS screening approaches, and confidence of healthcare providers as described above; (2) screening tool use, institutional and clinician-level barriers to screening implementation; (3) genetic work-up timing, methods used for testing, and perceptions of educational needs.

Explanatory variables included sex, country of practice, years of experience post-specialisation, number of institutional primary cancer diagnoses, access to a CPS clinic, access to a clinical geneticist, and availability of protected research time. Institutional patient volume was self-reported and dichotomised for the analysis as <100 or ≥100 new paediatric cancer diagnoses per year. Countries were assigned a priori to geographical regions following regional mapping and were as followed: Northern, Eastern, Southern, Western Europe, and Western Asia (Cyprus, Israel, Turkey). Additional domains assessed included the integration of CPS screening into clinical workflows (e.g., frequency, staff involvement, and use of electronic health records), decision-making processes for recommending genetic evaluation, timing and modality of genetic testing, handling and reassessment of genetic variants, patient adherence to follow-up, and clinician-reported educational needs (Supplementary Material S1).

Participants were presented with a matrix listing both universal and CPS-specific tools. For each tool, they were able to select one of the following options: "I don't know this tool"; "I know this tool but don't use it"; "I very rarely use it (< 10% of patients)"; "I sometimes use it (10-50% of patients)"; "I regularly use it (> 50% of patients)"; "I use it in almost all patients (100% or close to)". For analysis, we grouped those who responded either "I don't know this tool", "I know this tool but don't use it," and "I rarely use it" together as low use. "Sometimes use" was categorised as moderate use, and "regular use/ almost always use" were categorised as high use. Usage proportions were calculated separately for each tool due to missing matrix responses. To summarise overall use across all universal screening tools, participants were categorised into low, moderate, and high use categories based on their highest reported usage of any universal screening tool.

Statistical analysis

Descriptive statistics summarised demographics, resource access, confidence, and tool usage. We performed a complete case analysis for all data entered into statistical analysis. Chi-squared tests were used to assess associations between demographic variables and completion of each survey part.

We constructed univariable and multivariable logistic regression models to assess predictors of clinician confidence across four domains. For the primary analysis, confidence was dichotomised by summarising scores of 4–5 on a 5-point Likert scale as "high confidence" and comparing them to scores of 1–3 as "low or unsure confidence". For each domain, all predictors with a univariable p value < 0.1 were included in the corresponding multivariable models along with four a priori predictors: sex, ≥10 years of experience in paediatric oncology, access to a clinical geneticist, and access to a dedicated CPS clinic. As a secondary analysis, the same modelling strategy was applied using linear regression, treating the original Likert scores [1–5] as continuous outcomes.

All statistical analyses were conducted in R Statistical Software (v4.1.0, R Core Team 2021).

Delphi rounds and consensus statement development

Preliminary results of the SCOPE study were presented at the annual SIOPE Host Genome Working Group meeting held in Bern, Switzerland, in January 2025. Members of the working group, including paediatric oncologists, haematologists, and clinical geneticists with expertise in CPS, were given the opportunity to express interest in participating in modified Delphi rounds [21]. Each round had a defined focus: clinician confidence and tool use [1], guideline dissemination and resources needed for clinicians, patients, and families [2], and consensus statement creation [3]. In each

Table 1. Demographic characteristics of SCOPE survey participants across all three parts.

Characteristic	Part 1 ($n = 185$)	Part 2 ($n = 114$)	Part 3 ($n = 85$)	p value*
Sex				
Female (%)	61.1	61.1	57.6	0.73
≥10 years of experience (%)	57.3	57.9	56.5	0.98
Protected research time (%)	71.9	69.3	71.8	0.88
Regions of workplace				0.89
Northern Europe (%)	27.6	21.9	24.7	
Eastern Europe (%)	9.2	8.8	10.6	
Southern Europe (%)	35.7	41.2	34.1	
Western Europe (%)	24.3	24.6	29.4	
Western Asia (%)	3.2	3.5	1.2	
Institution with ≥ 100 cases/year (%)	28.1	29.8	29.4	0.94
Access to a paediatric geneticist (%)	80.0	77.2	82.4	0.66
Access to a dedicated CPS clinic (%)	50.3	50.0	47.1	0.88

CPS Cancer Predisposition Syndrome, *n* number.
*p values calculated using Chi2 test.

round, participants engaged in structured voting polls combined with open discussion to interpret the survey results and refine statements. Feedback from the first two rounds informed the development of the consensus statement, which was finalised and voted on in round three. Consensus was predefined as at least 70% agreement between participants.

RESULTS

Demographics

A total of 185 clinicians completed Part 1 of the SCOPE survey, with 114 completing Part 2, and 85 completing Part 3 (Table 1). The majority of respondents were female (61.1%) and experienced (57.3% had ≥10 years of practice following completion of their specialist degree in paediatric haematology/oncology). Most reported having protected research time (71.9%) and working in small- to medium-volume centres, with only 28.1% working at institutions caring for more than 100 new childhood cancer cases per year. Geographically, participants represented all regions of Europe. Comparisons across survey parts showed no significant differences in sex, experience, protected research time, region, centre patient volume, or CPS clinic access (all $p > 0.05$), indicating similar demographics across survey parts.

Clinician confidence

Many clinicians reported having low or unsure confidence in key domains of CPS management (Fig. 1). More than half indicated low or unsure confidence in most clinician confidence areas. Low or unsure confidence was highest for counselling families with known CPS (64.3%), utilising germline genetic information (57.3%), and utilising somatic genetic information (50.3%). More than 40% of participants reported low or unsure confidence in identifying patients for genetic workup (41.7%). Median Likert confidence scores were 4 (IQR 1) for referral to genetic work-up, 3 (IQR 1) for utilising somatic and germline genetic findings, and 3 (IQR 2) for counselling families on CPSs. Among clinicians with access to a dedicated CPS clinic, confidence remained low: 63.8% in counselling families, 55.2% in utilising germline findings, 47.4% in utilising somatic findings, and 31.9% in identifying patients for genetic work-up.

In multivariable logistic models, access to a clinical geneticist and access to a dedicated CPS clinic were associated with higher clinician confidence in some tested domains (Table 2 & Supplementary Material Tables S2 & S3). Access to a clinical geneticist was associated with significantly higher self-reported confidence among clinicians in identifying patients for genetic work-up (OR: 2.99, 95% CI: 1.35–6.89) and interpreting somatic results (OR: 2.53, 95% CI: 1.17–5.78). Similarly, access to a CPS clinic increased odds ratios for higher confidence in identifying patients at increased risk for CPS and suitable for genetic work-up (OR: 2.78, 95% CI: 1.42–5.53). Clinician level characteristics such as sex, years of experience, or working in a high-volume centre were not consistently associated with confidence. Workplace region had no significant association in most logistic models. However, clinicians based in Southern Europe had borderline lower odds of "high confidence" in counselling families compared to those in Western Europe (OR: 0.45, 95% CI: 0.20–1.02, $p = 0.058$) and this association reached significance in the corresponding linear model ($\beta = -0.49$, 95% CI: −0.93 to −0.05, $p = 0.031$).

Approaches to CPS evaluation

Clinical decision-making regarding genetic evaluation was heterogeneous when asked about the most common scenarios (with multiple answers per respondent possible). Roughly half of respondents (50.8%) reported relying primarily on their own clinical experience to determine whether a child requires further evaluation for a possible CPS, while 41.1% reported using a structured tool such as a checklist or algorithm. 29.7% indicated that they follow treatment protocols in which germline testing or referral is embedded. Only a minority (12.4%) indicated that all patients were routinely referred to a clinical genetics professional. Universal germline testing on all or nearly all patients (≥ 98% of patients) was reported by 8.1% of participants.

When asked how often participants personally communicated germline genetic test results to patients and families, 41.1% of clinicians reported doing so in all or nearly all cases, 15.7% in about half of cases, and 43.2% rarely or never. When clinicians did not disclose results themselves, respondents reported that genetic counsellors most often assumed this role: in all or nearly all cases for 28.6% of respondents, in about half of cases for 15.7%, and rarely or never for 55.7%

Most clinicians reported limited use and knowledge of universal screening tools (Fig. 2A). Even among better known tools (e.g., MIPOGG), reported usage varied widely with a majority of respondents reporting low use (57.5%). When respondents were grouped by their highest reported use of any universal screening tool, 42.3% remained in the low use category and 14.4% in the moderate use category (Fig. 2B). When knowledge and usage were examined separately, a large proportion of clinicians either

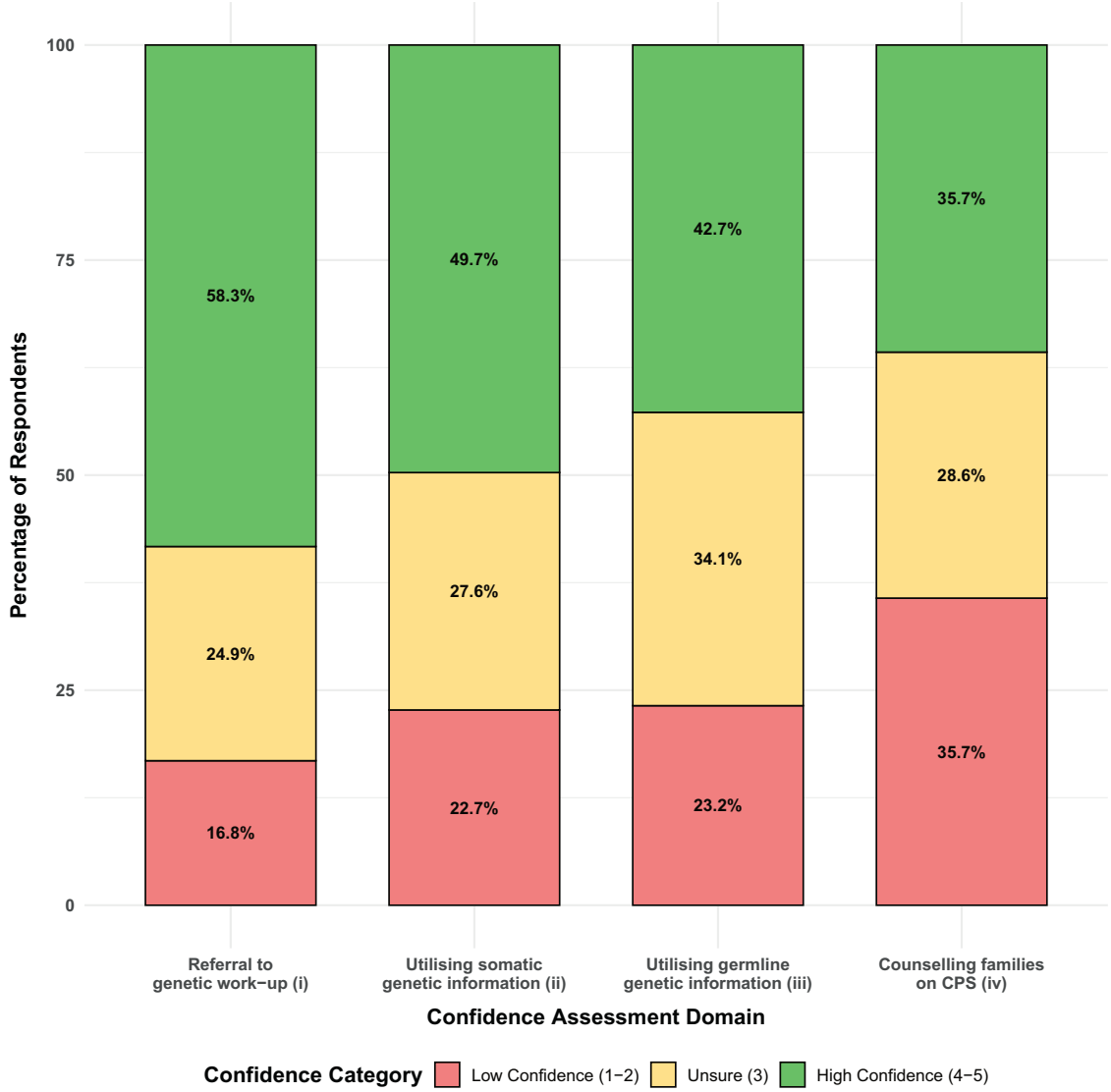

Fig. 1 Distribution of clinician confidence across four cancer predisposition syndrome (CPS) management domains ($n = 185$ respondents for each domain): (i) referral to genetic work-up, (ii) use of somatic genetic information, (iii) use of germline genetic information, and (iv) counselling families on CPS. Confidence was assessed on a 5-point Likert scale and grouped into low (1–2), unsure (3), and high (4–5).

did not know any universal tool (30.7%) or knew at least one tool but did not use it (13.2%).

Barriers

Clinicians reported various barriers to screening for CPS. The most common barrier was "Lack of Guidelines" (57%, Fig. 3). When asked which resources would best support their ability to educate families about surveillance and CPS, 80% of clinicians indicated a need for regular training/workshops. Over 70% also reported needing more dedicated personnel for patient support, such as genetic counsellors or educators, and about two-thirds citing needing more patient-friendly education material (Fig. 4).

Delphi process

Participation included 8 individuals in Round 1, 9 in Round 2, and 7 in Round 3 (6 voting members). All participants were paediatric oncologists, haematologists, or clinical genetics specialists with expertise in cancer predisposition syndromes. In Round one, experts identified and discussed key reasons for low clinician confidence in CPS counselling, most notably uncertainty in interpreting germline genetic results, lack of training, absence of patient-friendly information, and limited access to genetic counselors. Barriers to CPS screening tool use were primarily attributed to insufficient awareness or training. Strategies to improve screening practices included embedding CPS screening into paediatric oncology treatment protocols, incorporating CPS content into formal training curricula, and integrating CPS tools into electronic medical records. Participants also emphasised the importance of endorsement of screening guidelines by major oncology societies and the development of mandatory training programs.

In Round two, participants discussed the perceived lack of standardised CPS screening guidelines, information and educational needs of families affected by CPSs. Participants emphasised that although some screening guidelines exist, many oncologists are unaware of them or are unsure of which guidelines to follow. Additional challenges included limited dissemination of guidelines, organisational barriers such as coordination with families for follow-up, and difficulties maintaining follow-up. Regarding family educational resources, 71% of participants agreed that development of patient-friendly materials was the most urgent need.

Table 2. Multivariable logistic regression assessing predictors of clinician confidence across four CPS-related domains.

	Genetic work-up (i)		Utilising somatic genetic information (ii)		Utilising germline genetic information (iii)		Counselling Families (iv)	
Predictor	**OR (95%-CI)**	***p***	**OR (95%-CI)**	***p***	**OR (95%-CI)**	***p***	**OR (95%-CI)**	***p***
Sex (female)	1.42 (0.72–2.83)	0.31	1.57 (0.84–2.95)	0.16	1.33 (0.71–2.49)	0.38	0.79 (0.4–1.51)	0.47
≥10 years of experience	1.51 (0.78–2.94)	0.22	1.11 (0.6–2.05)	0.74	0.67 (0.36–1.24)	0.2	1.17 (0.61–2.25)	0.63
Access to a paediatric geneticist	2.99 (1.35–6.89)	0.0080*	2.53 (1.17–5.78)	0.022*	1.85 (0.85–4.24)	0.13	1.6 (0.71–3.84)	0.27
Access to a dedicated CPS clinic	2.78 (1.42–5.53)	0.0031*	1.35 (0.72–2.56)	0.35	1.37 (0.73–2.61)	0.33	0.88 (0.45–1.72)	0.71

CI confidence interval, *CPS* Cancer Predisposition Syndrome, *OR* odds ratio.

In Round three and following minor wording revisions, all recommendations achieved 100% "strong agreement" (Table 3). The recommendations cover (1) training on CPS risk assessment, interpretation of germline results, and communication with families; (2) inclusion of clinical screening for CPS during initial cancer evaluation; and (3) development of general, accessible educational materials.

DISCUSSION

This is the first study to our knowledge examining how paediatric haematology/oncology clinicians across Europe individually identify and manage CPS, highlighting significant variability in confidence, screening tool use, and resource availability. Unlike previous institutional surveys, this study captures individual clinician perspectives. Our survey found that many paediatric oncologists/haematologists report low or unsure confidence in evaluating patients for genetic work-up, utilising somatic and germline findings, and counselling families on CPS. Confidence was lowest in areas where communication and interpretation are most complex; specifically, 64.3% of clinicians reported low or unsure confidence in counselling families with a confirmed CPS, and over half reported low or unsure confidence in applying somatic or germline findings to patient care. This may reflect variability across disease groups as somatic findings are more routinely applied in some cancers (e.g., leukaemia) than in others. A majority of respondents reported feeling confident in identifying which patients should be referred for genetic evaluation to identify those with a CPS. However, a substantial proportion of clinicians in our survey (41.7%) still reported low or unsure confidence in this domain.

Interestingly, this difference in confidence was not explained by seniority or institutional patient volume but instead was associated with access to CPS-specific resources, particularly clinical geneticists and a dedicated CPS clinic. This suggests that improving access to CPS specialists may have a greater impact on confidence than individual experience alone. We assume that this reflects multidisciplinary teams and structured case reviews in these institutions. Here, the access to clinical geneticists increased also the likelihood of feeling confident with utilising somatic genetic information while the presence of a dedicated CPS clinic did not influence this. However, confidence levels across the domains remained low or unsure for a substantial proportion even among clinicians with access to dedicated CPS clinics: 64% of respondents reported low or unsure confidence in counselling families (66% in those without access to a CPS clinic), and about half lacked confidence in applying germline (55%) and somatic (47%) genetic findings (compared to 60% and 54% without a CPS clinic, respectively), suggesting that access to specialists alone may not be sufficient. The only domain that was strongly affected by the access to a CPS clinic was evaluating which patients would benefit from genetic work-up (low or unsure confidence in 32% with access to a CPS clinic versus 57% in those without, $p = 0.001$, Supplementary Material Fig. S4). Higher confidence in interpreting somatic results among clinicians with access to clinical geneticists may reflect multidisciplinary case review where somatic and germline results are discussed together. Presence of a dedicated CPS clinic alone was not associated with the same increase in confidence and might be due to its role in care of already identified CPS patients and not workup of patients. A U.S.-based survey of primary care providers showed similar findings, where many respondents similarly expressed uncertainty about when to refer patients to genetics services in various clinical scenarios including developmental delay, early cardiovascular risk factors, and personal or family history of cancer [22].

Although most respondents in our study had more than ten years of experience, confidence was particularly low in counselling families with confirmed CPS. This aligns with another key finding:

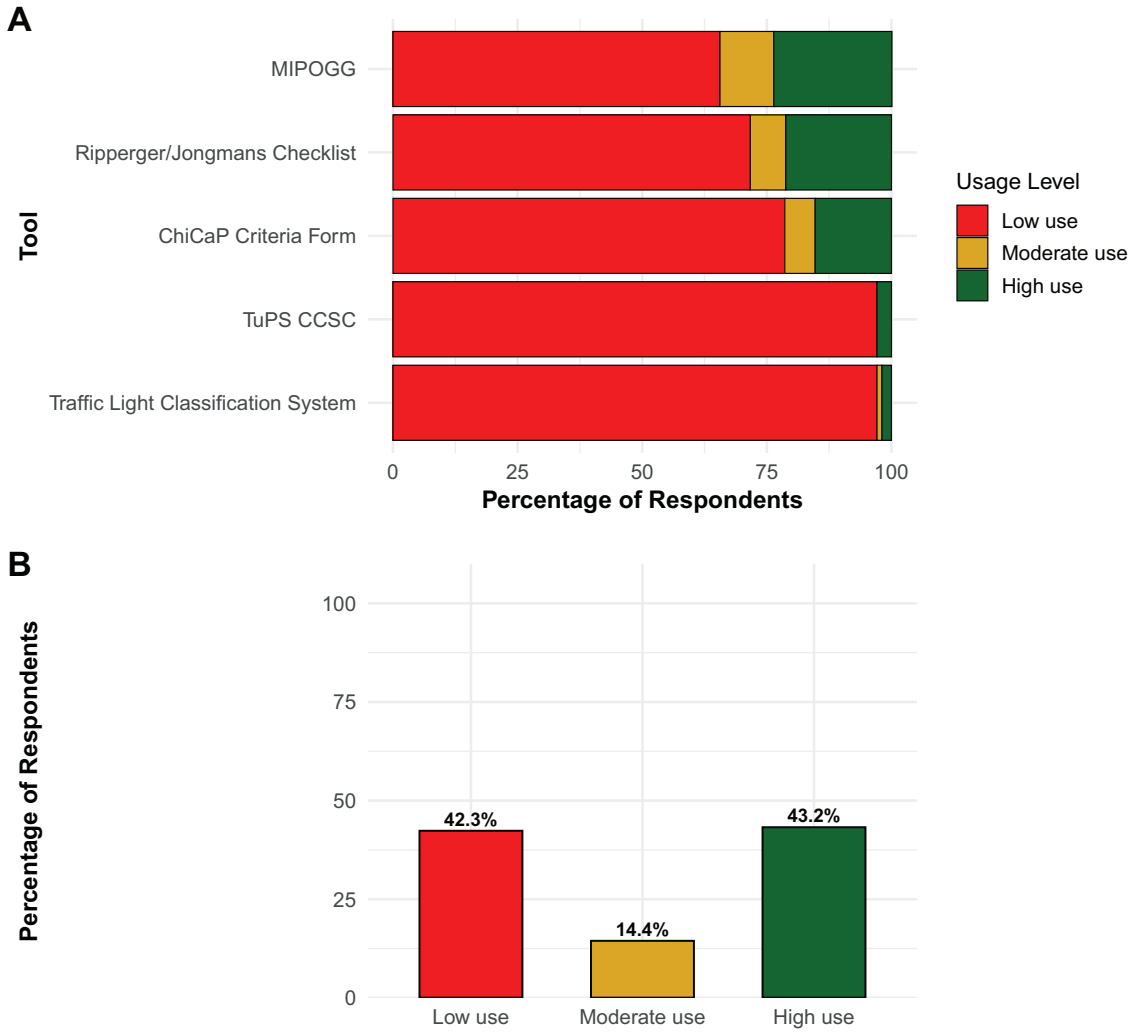

Fig. 2 Usage of universal cancer predisposition syndrome (CPS) screening tools among 114 respondents of the SCOPE survey. **A** Individual-level responses on self-reported use and knowledge of five universal CPS screening tools, categorised as low, moderate, or high use. **B** Grouped distribution of respondents based on their highest reported usage level of any universal CPS screening tool. Legend: MIPOGG McGill Interactive Paediatric OncoGenetic Guidelines, ChiCaP Childhood Cancer Predisposition Criteria Form, TuPS CCSC Tumour Predisposition Childhood Cancer Syndrome Checklist.

80% of respondents expressed a belief that regular training and workshops are a necessary resource to support patient education. Apart from a possible lack in clinical knowledge, this points towards a gap in ability to communicate complex genetic information in a way that is accessible, age-appropriate and supportive. Neither years of experience nor access to a CPS clinic positively affected this domain.

We observed that the availability of CPS screening tools did not translate into widespread use. Although validated resources such as the Jongmans/ Ripperger checklist or the MIPOGG tool exist, only 42.3% of respondents in our survey reported regular use of any universal screening tool. This may in part be explained by the fact that over half of respondents reported primarily relying on their own clinical experience to decide which patients require further genetic evaluation. The LEGEND-COST survey, which assessed CPS practices at the institutional level, found that although many institutions were familiar with screening tools, they were applied in just 24% of centres [20], which would suggest an increased uptake in our more recent survey. While clinical expertise is valuable, it may lead to inconsistency in identifying at-risk patients, especially in cases where CPS signs are subtle. Furthermore, as recommendations on which patients may benefit from genetic evaluation may change over time, such updates would likely be reflected in regularly updated tools but might be missed by clinicians who do not use them. These findings suggest that while screening tools are known to many clinicians, routine implementation remains a key barrier to their effective use. Other contributing factors may include lack of time, limited integration into hospital workflows, or unclear endorsement at the institutional level. As less than 10% of respondents reported performing genetic testing for all or most patients, the limited uptake of screening tools cannot be explained by universal testing practices. Taken together, these findings underscore the need for structured implementation strategies and organisational support to ensure successful integration of screening tools as a standard of care.

Various barriers in screening for CPS in paediatric patients were identified: Lack of CPS screening guidelines, difficulty interpreting results, financial issues, and limited access to genetic testing. For the latter, we cannot elaborate on which genetic testing might be available in the different institutions, but likely, different testing approaches might also play a role here (single gene, panel versus whole-genome sequencing). The most reported barrier to CPS screening in our study was “lack of CPS screening guidelines”.

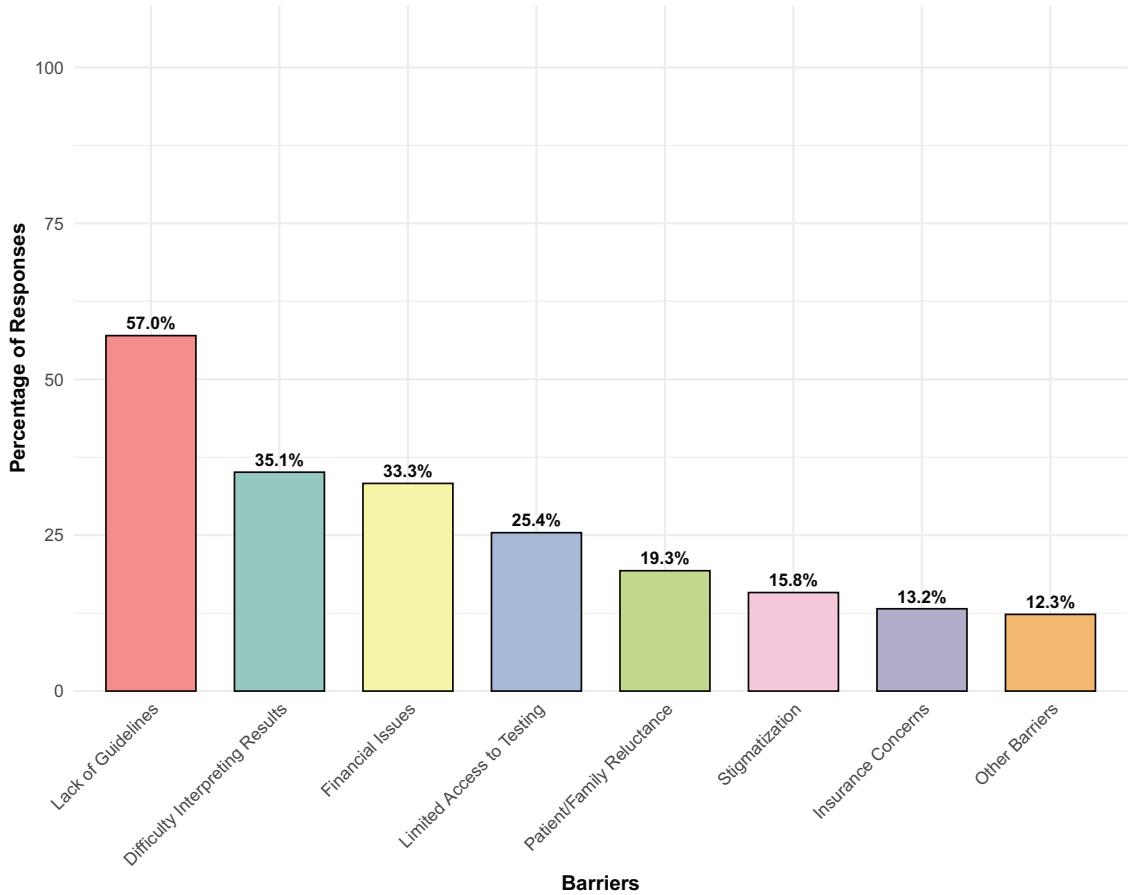

Fig. 3 Reported barriers to screening for cancer predisposition syndromes (CPS) among 114 respondents of the SCOPE survey.

More than half of the respondents (57%) identified this as a key obstacle, despite the existence of multiple published guidelines and referral criteria [17, 23–25]. This discrepancy highlights a dissemination failure, where existing resources are not reaching clinicians and may not be integrated into existing clinical workflows. This mismatch of a perceived lack of availability was also observed in a US-based survey of members of the Children's Oncology Group (COG). Among their 181 respondents, only 34.8% reported consistently using CPS referral guidelines for suspicion of a CPS. The most common reason for not using them was the lack of available or known guidelines (34.4%), and a further 7.8% of participants were unaware that any guidelines existed [26]. These studies highlight that the problem is not in the absence of guidelines, but rather poor visibility, dissemination, and implementation support.

To address these gaps, we conducted a Delphi Consensus process with members of the SIOPE Host Genome Working Group. Across three rounds of structured reviews, experts reached full agreement on three core priorities: improving clinician training, integrating CPS screening into standard practices and protocols, and developing tailored educational materials for patients and families. Notably, all three recommendations reached 100% "strong agreement" in the final round, underscoring the urgency and shared vision for improving CPS-related care. The Delphi process added expert interpretation and recommendations of clinical changes to the survey findings by translating broad patterns into actionable guidance. While we expected training needs and improvement of screening protocols to be identified as areas of need, the group's emphasis on patient-centred materials reflects a growing awareness of the communication challenges in this field. Supporting this, qualitative studies of adolescent perspectives during genetic counselling have shown a strong preference for visual and concise materials [27]. Adolescents found infographics and videos more engaging than long information letters, suggesting that well-designed, age-appropriate materials can improve communication and empower families in complex CPS decisions. These insights reinforce the value of designing targeted educational materials that address the specific needs and desires of patients alongside their healthcare providers.

A major strength of this study is its pan-European scope and integration of both survey and expert consensus data. We assessed CPS screening and care practices at an individual level, which might differ from what institutions report. The additional consensus finding adds actionable recommendations to the survey results which are based on the input of experts from various clinical and regional backgrounds. However, several limitations should be acknowledged. Given that our survey was distributed through national collaborators, we were unable to calculate formal response rates and could not control which paediatric oncologists received the survey invitation. As a result, participation may have been limited to individuals within specific institutions or professional networks. To mitigate this, we promoted the survey through conferences and society websites. There is also a possibility of self-selection bias, where clinicians with a stronger interest in CPS may have been more likely to respond, suggesting confidence and tool use may be even lower in the broader paediatric haematology/oncology community. We also recognise that, while the pan-European scope is a strength, it may simultaneously be a limitation given

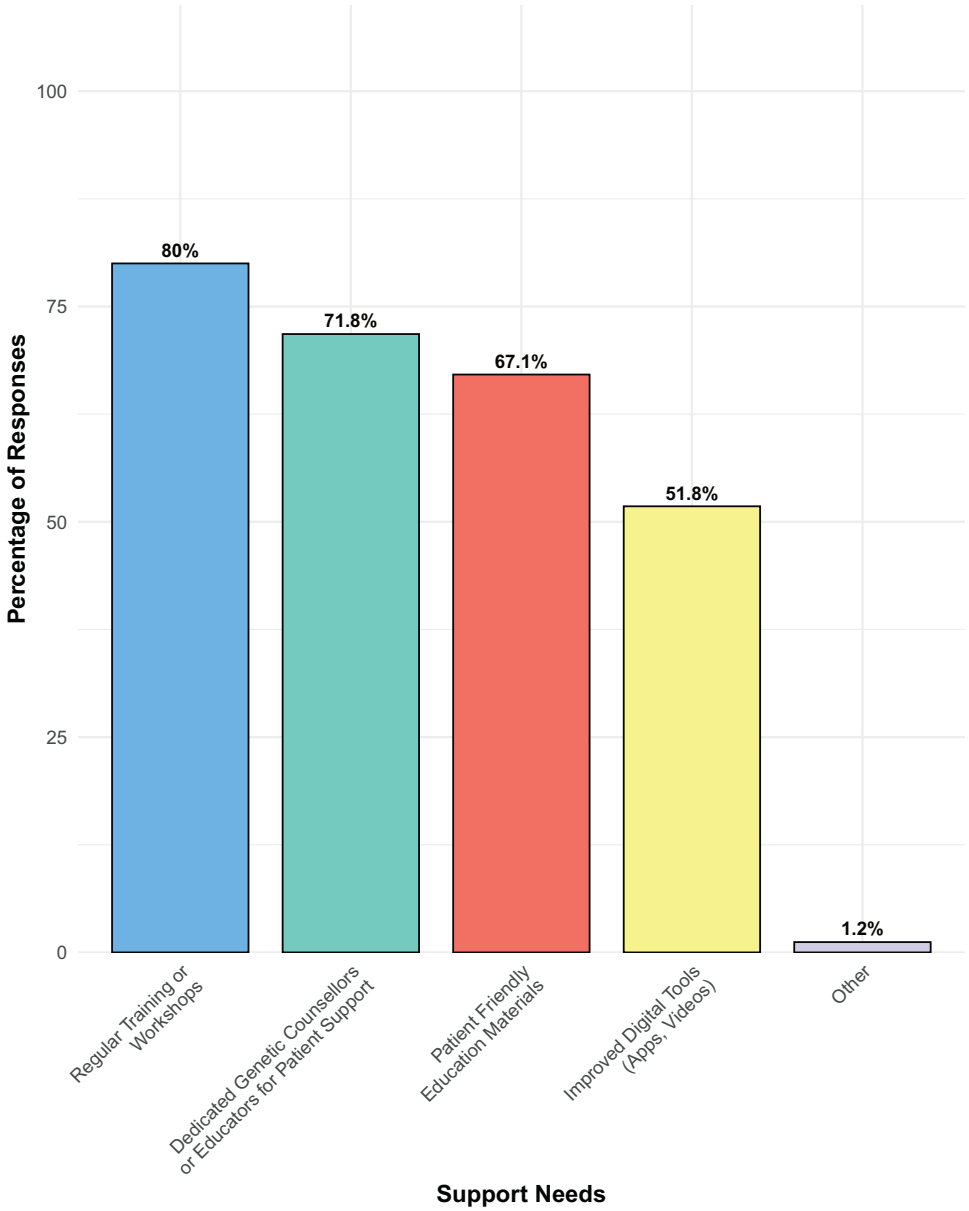

Fig. 4 Reported needs for enhancing family/patient education and surveillance related to Cancer Predisposition Syndromes (CPS) by 85 respondents of the SCOPE survey.

the substantial differences in healthcare resources, infrastructures, and policies across countries and institutions. Finally, we observed a drop-off in participation across the survey parts. Still, the broad geographical distribution and consistent demographics across parts helps mitigate concerns about bias. To analyse our data, we opted for grouping confidence levels "1: very low" and "2: low" together with "3: unsure" to be able to compare it to the levels "4: high" and "5: very high" based on the assumption that a goal for clinical care is to have specialists feel confident at basic evaluations of patients. To indicate "3: unsure" confidence in specific domains could also reflect the wish of not assessing one's own confidence level rather than a true lack of confidence, which cannot be further clarified with the data obtained in our study.

Our findings have important implications for clinical practice. Despite increasing awareness of CPS and the growing availability of screening tools, widespread implementation of standardised CPS risk assessment remains low. In our survey, less than half of clinicians reported regular use of any universal CPS screening tool. Embedding CPS screening into diagnostic and treatment protocols, as emphasised in recommendation 2, may be critical for improving uptake. National societies and institutions have a key role to play in endorsing this process. Without formal integration and accountability, screening practices will likely remain variable, and children at increased risk of harbouring a CPS may continue to be missed.

In conclusion, our study shows that paediatric oncologists and haematologists across Europe face persistent challenges in the identification and management of CPS. Confidence is low even among experienced providers, and screening tools remain underutilised. The recommendations developed through our Delphi process reflect not only the needs identified by our survey, but also an expert consensus on how to move forward. Future work should focus on implementing and evaluating structured CPS screening pathways, expanding clinician training, and developing accessible educational resources to empower patients and families across diverse healthcare settings.

Table 3. Consensus statements from the SCOPE Delphi process.

Recommendation	Statement
Recommendation 1:	We recommend that paediatric oncology providers receive training on CPS risk assessment, interpretation of germline results, and communication with families. Training should include screening tools and counselling templates.
Recommendation 2:	Clinical screening for CPS should be included in the initial cancer evaluation and treatment plan. Where possible, it should be integrated into protocols and endorsed by national societies.
Recommendation 3:	We recommend the development of general, accessible educational materials tailored to age, literacy, and language. These should be available in print/digital formats and complement counselling by clinical staff.

DATA AVAILABILITY

The underlying survey data analysed in this study are stored in a secure REDCap database and are not publicly available due to privacy and confidentiality considerations. Researchers interested in accessing the anonymised dataset may contact the corresponding author with a reasonable request. The full survey instrument is included in Supplementary File 1.

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AUTHOR CONTRIBUTIONS

JCP: Conceptualisation, Methodology, Investigation, Data preparation and curation, Formal analysis, Visualization, Project administration, Writing – all stages. MJ, FB, LL, MD, OM, CK: Conceptualisation, Methodology, Investigation, Writing – review & editing; SSK, RP, NC, ES, GC, RF: Data Analysis and Interpretation and Writing – review & editing. JR, CZ: Conceptualisation, Supervision, Writing – review & editing. NW: Conceptualisation, Methodology, Investigation, Data preparation and curation, Project administration, Funding acquisition, Supervision, Writing – all stages.

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COMPETING INTERESTS

NW reports a relationship with Swedish Orphan Biovitrum AB that includes advisory board membership, consulting, and travel reimbursement, a relationship with NovoNordisk that includes advisory board and travel reimbursement, and a relationship with Novartis that includes advisory board membership. None of these relationships are in association with the current study. LL reports an advisory board role in AMGEN, JAZZ PHARMA, CLINIGEN. JR became an employee of Novartis Pharma Basel after the setup and start of this research project.

ETHICAL APPROVAL

The ethics committee of the canton of Bern, Switzerland, waived an ethics evaluation (Req-2022-00455). All participants consented to the project prior to conducting the surveys.

ADDITIONAL INFORMATION

Supplementary information The online version contains supplementary material available at https://doi.org/10.1038/s41431-026-02040-x.

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