Protocol

Assessing Psychosocial and Supportive Care Needs of Families With Children and Adolescents Diagnosed With Cancer Predisposition Syndromes: Protocol for the KiTDS-Care Mixed Methods Study

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Abstract

Background: Cancer predisposition syndromes (CPSs) are identified in approximately 10% of pediatric patients with cancer, and the number of children known to be affected by CPSs is steadily increasing. Families of children and adolescents with a CPS face substantial psychosocial challenges, including uncertainty regarding genetic risk, communication, long-term surveillance requirements, and concerns about future health outcomes. Despite these challenges, little is known about the psychosocial burden, supportive care needs, and current care situation of affected families in Germany.

Objective: The KiTDS-Care (Children with Tumor Predisposition Syndrome and their Families–Analysis of the current Care Situation and Support Needs, and Derivation of Care Recommendations) study aims to (1) comprehensively assess the current psychosocial care landscape, psychosocial burden, and supportive care needs of families with children and adolescents diagnosed with a CPS in Germany and (2) develop recommendations for improving psychosocial and family-centered care based on the study findings.

Methods: KiTDS-Care is a mixed methods study. First, a systematic review will synthesize existing evidence on the psychosocial experiences and support needs of families affected by a CPS. Second, a nationwide cross-sectional survey will be conducted among parents and adolescents or children aged ≥7 years to assess depression, anxiety, quality of life, supportive care needs, and health care use using standardized questionnaires. Finally, qualitative semistructured interviews with families and health care professionals will explore psychosocial experiences, barriers to care, and perceived gaps in existing support services in greater depth. Quantitative data will be analyzed using descriptive and inferential statistical methods, while qualitative data will be analyzed using structured content analysis. On the basis of the integrated findings, recommendations for psychosocial care will be developed and refined with input from families and health care professionals.

Results: The study is funded by German Cancer Aid for 36 months from October 2024. Data collection for the systematic review began in January 2025. Recruitment for the cross-sectional survey and qualitative interviews started in May 2025 and will continue until April 2027. A sample of 90 families is planned. As of January 2026, participant recruitment is ongoing, and 52 participants from 24 families have been enrolled. Data analysis is expected to be completed in July 2027, and publication of the primary study results is anticipated in October 2028.

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Conclusions: The KiTDS-Care study will provide comprehensive insights into the psychosocial situation and supportive care needs of families affected by a CPS in childhood and adolescence. The findings are expected to identify gaps in current care structures and inform the development of targeted, family-centered psychosocial support services for affected families in Germany.

Trial Registration: German Clinical Trials Register DRKS00035594; http://drks.de/search/de/trial/DRKS00035594

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KEYWORDS

cancer predisposition syndromes; pediatric cancer; psychosocial care needs; family-centered care; psychosocial stressors; psychosocial burden

Introduction

According to current scientific evidence, approximately 10% of children and adolescents with cancer have a cancer predisposition syndrome (CPS) [1-3]. About 30% of CPS cases are caused by de novo genetic variants, with rates reaching up to 50% in certain syndromes such as neurofibromatosis type 1 [4-7]. In Germany, 2000 to 2500 children and adolescents are newly diagnosed with cancer each year, corresponding to approximately 200 to 250 CPS diagnoses annually [8]. In 60 to 75 of these cases, no other family member with a known CPS is identified. With increasing awareness and advances in diagnostics, the number of identified CPS cases is expected to rise, leading to a growing need for genetic counseling and psychosocial support [3,9]. Identifying a CPS has far-reaching consequences for children and their families. Treatment strategies are adapted to reduce acute and long-term toxicities, and preventive screening programs are initiated to enable early detection of secondary or additional malignancies. Where appropriate, individualized therapies are used to balance treatment efficacy and side effects. Genetic counseling also supports families in making informed decisions regarding family planning and preventive options [3,9-12].

To date, no data from Germany are available on the needs and preferences of families with a child or adolescent with cancer regarding CPS screening, genetic testing decisions, or coping with a CPS diagnosis. A hereditary tumor predisposition can represent a lifelong burden for children and their parents. Therefore, investigating the psychosocial consequences of screening, genetic testing, and living with a CPS is essential to support families throughout the process and its long-term implications (eg, intensified surveillance) [13].

International studies have examined the psychosocial situation of children and adolescents with a CPS and their parents, showing that the burden differs depending on cancer status and family context [13-17]. Families face particular emotional challenges during transition phases, and parents of children with cancer may experience CPS testing as an additional stressor due to time pressure and the diagnostic context [16]. Communicating genetic results is especially sensitive for adolescents, who report that a CPS diagnosis can affect their self-image [16-19]. A recent review by Patenaude and Wakefield [13] highlights research gaps in the area of the familial psychosocial situation and its effects. Identifying stressors and support needs is essential to prevent or reduce psychological burden in affected children, adolescents, and their parents, underscoring the need to address CPS-related challenges in addition to cancer care. CPS-related stressors include regular surveillance examinations, ongoing cancer risk, concerns about affected family members, and challenges related to life and family planning. In addition, care is complicated by a lack of specialized expertise, the need for lifelong monitoring, and difficulties during transition to adult health care.

Studies on specific CPSs (eg, Li-Fraumeni syndrome, retinoblastoma, and familial adenomatous polyposis) indicate that affected children do not necessarily have an increased risk of mental health disorders but face CPS-specific stressors, such as social isolation, fear for family members, and reduced quality of life [13]. Current care is predominantly medically focused, emphasizing frequent preventive and follow-up examinations. Preventive screening recommendations exist for only a subset of >100 CPSs, and these recommendations are largely expert-based, with demonstrated survival benefits limited to Li-Fraumeni syndrome and constitutional mismatch repair deficiency [15,18]. These intensive surveillance programs are viewed with ambivalence by patients and parents: on one hand, they are experienced as reassurance; on the other hand, adolescents and parents experience anxiety related to examinations and reminders of cancer risk. Nevertheless, the benefits of tumor prevention are perceived as outweighing the challenges [18]. There is evidence suggesting that psychosocial burden depends on whether and which other family members are affected [20]. Evidence from adult populations with genetic tumor predispositions demonstrates the need for targeted psychosocial support to improve coping with hereditary cancer risk [16,18].

It remains unclear whether international findings on psychosocial burden and support needs can be transferred to Germany, given differences in health care structures and genetic counseling practices. Unlike many countries with established genetic counselor roles, genetic counseling in Germany is typically limited to the testing context and is further constrained by a shortage of qualified professionals [21]. Importantly, psychosocial support for families affected by a CPS is inherently multidisciplinary and is provided across different health care professions, including clinical geneticists, oncologists (in the context of cancer diagnosis and treatment), nurses, psychologists, psycho-oncologists, and, where available, genetic counselors [22]. Although each of these professional groups contributes to addressing different aspects of psychosocial care,

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coordinated and structured support pathways remain limited in routine clinical practice [23].

Although genetic counseling plays an important role in the communication of genetic risk information, no specific, structured psychosocial intervention concept for families affected by a CPS currently exists in Germany. Identifying family-specific needs and burdens across disciplines is therefore essential to inform the development of integrated, patient- and family-centered care approaches.

Although existing research has largely concentrated on the decision-making process regarding genetic testing and the initial communication of a CPS diagnosis [23], considerably less is known about the long-term psychosocial burden associated with living with a CPS, including repeated surveillance procedures, ongoing uncertainty, and the cumulative impact on family functioning. Accordingly, this study will deliberately target families who are in the postdiagnostic phase and engaged in long-term follow-up care. The quantitative survey will be conducted to capture current psychosocial outcomes and support needs during routine care and surveillance, providing a cross-sectional snapshot of lived experiences beyond the diagnostic decision-making period. The qualitative interviews will be conducted during the same overall study period and will explore in greater depth how families experience long-term management of CPSs, including coping with ongoing risk awareness, medical surveillance, and communication within the family system. By focusing on this phase, the study will address a key gap in the literature regarding the sustained psychosocial impact of CPSs beyond the initial diagnostic and testing context.

The primary outcomes of The KiTDS-Care (Children with Tumor Predisposition Syndrome and their Families–Analysis of the current Care Situation and Support Needs, and Derivation of Care Recommendations) study are psychosocial functioning and health-related quality of life (HRQoL) in children and adolescents with a CPS, as well as in their parents and siblings. Psychosocial functioning is operationalized as emotional and behavioral problems (children) as well as depression and anxiety (parents), assessed using validated standardized instruments (SDQ [Strengths and Difficulties Questionnaire], Patient Health Questionnaire–9 [PHQ-9], and Generalized Anxiety Disorder–7 [GAD-7]), while HRQoL is assessed using established HRQoL measures (Kinder Lebensqualität Fragebogen [KINDL; Children's Quality of Life Questionnaire] and Ulm Quality of Life Inventory for Cancer Patients [ULQIE]).

Secondary outcomes include perceived supportive care needs, psychosocial stressors related to genetic risk, family communication, and long-term medical surveillance, as well as the use and perceived adequacy of existing psychosocial and supportive care services. In addition, barriers to and facilitators of accessing psychosocial support will be explored from the perspectives of affected families and health care professionals as qualitative outcomes.

An exploratory objective of the study is to develop evidence-based recommendations for improving psychosocial and family-centered care for CPS-affected families, based on the integrated findings from the quantitative and qualitative study components.

Methods

Study Design

The KiTDS-Care study will use a convergent mixed methods design with an embedded qualitative component and an integrated systematic review. Quantitative and qualitative data will be collected within the same overall study period and analyzed independently before being integrated at the interpretation stage. Given the limited existing evidence on psychosocial outcomes and care needs in families affected by a CPS, the qualitative interviews will serve 2 complementary purposes: first, to provide in-depth expert and experiential insights from affected families and health care professionals; and second, to ensure that potentially relevant psychosocial domains are not overlooked in the purely quantitative assessment. In this context, the qualitative component will enhance the comprehensiveness of the study by identifying context-specific issues, care gaps, and emerging themes that may not be captured by standardized survey instruments. The systematic review will further complement these primary data sources by providing an evidence-based conceptual framework. The findings of the systematic review will inform the interpretation of the empirical results and help contextualize the study findings within the broader research landscape.

Study Population

The study population consists of pediatric patients diagnosed with a CPS and their parents. Only the child or adolescent is required to have a confirmed CPS diagnosis. The study includes (1) parents of patients aged ≤18 years with a confirmed CPS diagnosis and (2) patients aged >7 years with a confirmed CPS diagnosis. Inclusion and exclusion criteria are presented in Textbox 1.

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Textbox 1. Inclusion and exclusion criteria for participating families.

Inclusion criteria

- Confirmed diagnosis of a cancer predisposition syndrome in the child or adolescent patient
- Patient aged ≤18 years
- Sufficient German-language skills among participating family members
- Written informed consent from parents or legal guardians and assent from children or adolescents, where appropriate

Exclusion criteria

- Excessive physical or psychological burden of the patient or parents, as determined by clinical or therapeutic judgment
- Refusal to participate in the study by the legal guardian and/or patient, where applicable
- Children or adolescents who have not been fully informed about their diagnosis

Health care professionals will be eligible for participation in the qualitative interviews if they meet the following criteria: (1) be currently employed in a clinical or psychosocial care setting involved in the diagnosis, treatment, follow-up, or psychosocial care of patients with a CPS; (2) belong to one of the following professional groups: clinical geneticists, oncologists, pediatric oncologists, nurses, psychologists, psycho-oncologists, or other health care professionals involved in genetic counseling or psychosocial support; (3) have direct or indirect experience with the care of patients or families affected by a CPS; and (4) have sufficient proficiency in German to participate in a qualitative interview.

Participation is voluntary, and informed consent will be obtained prior to the interview. Efforts will be made to ensure variation in professional background, care setting (eg, hospital, outpatient clinic), and level of experience to capture a broad range of perspectives.

Recruitment

Participants are being recruited by the patient collective within the Pediatric Oncology Network Bavaria (Kinderonkologisches Netzwerk Bayern [KIONET]). In KIONET, the pediatric oncology departments of the university hospitals in Augsburg, Erlangen, Munich (Ludwig-Maximilians-University [LMU] and Technical University of Munich), Regensburg, and Würzburg have joined forces to improve the medical care and cure rates for children and adolescents with cancer [24].

Patients with a CPS aged ≤18 years and their families who have an appointment at one of the hospitals in KIONET during the recruitment period will be informed about the study during the medical consultation and invited to participate in the survey.

Patients may present either with or without a personal history of cancer. In the KiTDS-Care study, both groups will be included where applicable, as psychosocial experiences and support needs are expected to differ between these subgroups. Specifically, individuals with a previous cancer diagnosis may experience additional burden related to treatment history, late effects, and ongoing surveillance, whereas those without a cancer history may be more strongly affected by anticipatory anxiety, uncertainty regarding future cancer risk, and decision-making related to preventive surveillance and genetic risk communication. Including both groups allows a more comprehensive understanding of the full spectrum of CPS-related psychosocial challenges. Additionally, patients will be invited to participate in the study through self-help organizations or parent associations.

Systematic Review

As a basis for the systematic analysis of the current care situation, the first step was a systematic review of the psychosocial stressors, burden, and support needs of families with a child or adolescent with a CPS.

We will use the PRISMA (Preferred Reporting Items for Systematic Reviews and Meta-Analyses) statement to ensure the transparency and scientific quality of our systematic review [25]. In line with the research question, relevant studies will be identified using electronic database searches, manual searches of reference lists, and searches for citing publications of relevant publications. Relevant studies will be selected through title and abstract screening based on predefined inclusion and exclusion criteria, which are derived from the evidence-based population, intervention, comparison, and outcome (PICO) framework [26]. Full-text screening will be conducted to determine the final inclusion of studies according to the inclusion and exclusion criteria. From the included studies, relevant data will be extracted, aggregated, and summarized through a narrative synthesis. For more details, refer to the corresponding PROSPERO registration (CRD42025618671) [27].

Quantitative Survey of Patients and Parents

Measurement instruments (Table 1 [28-38]) were selected based on their relevance to the study objectives, prior use in pediatric psycho-oncology, and psychometric quality. Whenever possible, validated German-language instruments with established reliability and validity were used to assess psychosocial outcomes and supportive care needs. Relevant sociodemographic data, as well as experiences of the diagnostic process, will be collected via self-developed items. In addition, several instruments were selected to ensure comparability with an existing historical dataset of pediatric oncology patients and their families available to the research group. This will enable exploratory comparisons between families affected by a CPS and families of children with cancer regarding psychosocial burden and supportive care needs.

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Table 1. Overview of constructs and measurement instruments in the KiTDS-Care study.

Constructs	Instruments
Parent questionnaire	
Sociodemographic and medical parameters	Self-developed items, partially based on Psycho-Oncological Basic Documentation [28]
Experience of the CPS[a] diagnosis and experiences in the care context	Self-developed items; for example, diagnosis notification (timing, person, and setting) and understanding of the disease (eg, probability of illness and consequences)
Quality of life	Ulm Quality of Life Inventory for Cancer Patients [29]
Psychological distress (anxiety, depression, and progression anxiety)	Generalized Anxiety Disorder–7 [30], Patient Health Questionnaire–9 [31], and progression anxiety scale [32-34]
Family functioning (including communication)	FAD-GF[b] [35], FAD[c] communication subscale
Social support	Oslo Social Support Scale [36]
Support needs and use	Based on Supportive Care Needs Survey–Short Form 34 [37] and self-developed items
Psychosocial stress factors	Self-developed questions regarding psychosocial stress factors during diagnosis and care
Child questionnaire (age 7-10 years)	
Quality of life	KINDL[d] [38]
Experience of the CPS diagnosis	Self-developed items; for example, diagnosis notification (timing, person, and setting) and understanding of the disease (eg, probability of illness and consequences)
Child questionnaire (age 11-18 years)	
Quality of life	KINDL [38]
Experience of the CPS diagnosis	Self-developed items; for example, diagnosis notification (timing, person, and setting) and understanding of the disease (eg, probability of illness and consequences)
Experiences in the care context	Self-developed items
Family functioning (including communication)	FAD-GF [35] and FAD communication subscale
Support needs	Self-developed items
Medical data	
Cancer disease	Diagnosis, date, treatment protocol, and treatment elements
CPS	Diagnosis (including gene or gene variant), date, screening examinations, frequency, number and age of affected individuals, inheritance, and abnormalities

[a]CPS: cancer predisposition syndrome.

[b]FAD-GF: Family Assessment Device–General Functioning.

[c]FAD: Family Assessment Device.

[d]KINDL: Kinder Lebensqualität Fragebogen (Children's Quality of Life Questionnaire).

Medical parameters related to the CPS (eg, diagnosis, including gene variant, date of diagnosis, number and gender of affected individuals, relationship, frequency and extent of screening examinations, and abnormalities) and, if applicable, those related to the cancer (eg, diagnosis, date of diagnosis, and physical functionality) will be transmitted in pseudonymized form via the clinics and assigned to the questionnaires using a unique participant code. This will apply only to patients recruited through KIONET, while for participants recruited through other channels, the collection of medical data will be based on self-report by the parents.

Qualitative Interviews

Qualitative interviews will be conducted using a semistructured approach. We have developed 2 interview guidelines (1 for parents and 1 for health care professionals), based on theoretical considerations and prior experiences. Central aspects are addressed in the form of open questions and further explored through targeted follow-up questions.

After agreeing to participate in an interview, the involved parents and health care professionals can submit their contact details to the project team. They will then be contacted by a member of the project staff, and an appointment for the interview will be scheduled. The interviews will be conducted using a semistructured interview guideline and can take place via phone or through a videoconference system. In both guides, the different perspectives will cover not only the psychosocial stressors, burden, and support needs of the families but also areas such as coping with the diagnosis, understanding of the diagnosis, experiences in the care context (eg, diagnosis notification, decision-making processes, and course after diagnosis), use of services, and wishes for psychosocial support. In the parent interviews, the handling of siblings will be specifically addressed. The interviews with health care

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professionals aim to complement the perspectives of affected families by providing an expert view on the provision of psychosocial care within CPS services. Health care professionals are included due to their central role in different stages of the CPS care pathway, including diagnosis, genetic counseling, surveillance, and psychosocial support.

Sample Size

In the cooperating clinics, >400 patients with a diagnosed CPS are treated, and around 40 patients are diagnosed with a CPS each year. Considering the inclusion and exclusion criteria, as well as other potential reasons for nonparticipation, a sample size of 90 families for a recruitment period of 24 months can be expected.

For the interviews with families, a sample of approximately 10 to 15 parents whose child's diagnosis was made >12 months ago and 10 to 15 parents whose child's diagnosis was made less than 12 months ago will be targeted based on theoretical saturation [39].

A sample of 15 to 20 health care providers is also targeted for the qualitative interviews.

Documentation, Data Storage, Data Security, and Data Protection

Written consent is required for participation. All children or adolescents and their families who agree to participate will receive the study materials along with an assigned pseudonymization code (1 questionnaire per participating family member, detailed study information, and an information letter for an interview, along with a stamped and addressed return envelope). Each family will be assigned a unique code, which will serve as a pseudonym, ensuring that no direct conclusions can be drawn about the families. Only the clinics will maintain a list linking the pseudonyms to personal data. This list will remain within the clinics and will not be shared with the research institutes. After providing consent to participate in the study, families will receive an individual family-related access code, which will allow parents and children to fill in the online survey. The families can choose to either complete paper-based questionnaires or complete the survey via an online data collection method using the REDCap (Vanderbilt University) program. The written consent form will remain in the clinics. The medical data will be documented by hospital staff and sent, along with the corresponding pseudonymization code, to the research institutes. The access code will be linked to the pseudonymization code, ensuring that the pseudonymized medical data can be associated with the families.

For patients not recruited at a KIONET clinic but through self-help organizations, the medical data will be provided via a parent's report. Affected children aged ≥7 years will be surveyed, provided that their parents consent to their child's participation.

Ethical Considerations

This study is being conducted in accordance with the Declaration of Helsinki and has received ethics approval from the ethics committee of LMU Munich (24-0322) and the ethics committee of University Medicine Greifswald (BB 194/24). Written informed consent will be obtained from all parents or legal guardians prior to participation. In addition, age-appropriate assent will be obtained from children and adolescents in accordance with their developmental stage and understanding. Participants will receive a copy of the signed consent form, while the original will be stored in the clinical study records. Participation is voluntary, and withdrawal is possible at any time without any consequences for medical care or other services.

Given the sensitive nature of genetic risk information and potential psychological distress, special attention will be given to participant safety and emotional well-being during data collection. Children and adolescents will only be invited to participate after they have been fully informed about their CPS diagnosis. Furthermore, the qualitative interviews with families will be conducted by researchers who are also trained as psychological psychotherapists. These researchers will also serve as contact persons for participants throughout the study period and will be available for support in case of emotional distress or questions related to study participation. Participants may skip questions or discontinue participation at any time without providing justification. If clinically relevant psychological distress is identified, participants will be provided with information on appropriate psychosocial support services and referral pathways. In urgent cases, established clinical care pathways will be activated.

All data will be pseudonymized and handled in accordance with applicable data protection regulations. Genetic and family-related information will be treated as highly sensitive data and stored securely with restricted access. Only authorized study personnel will have access to identifiable information.

Statistical Analysis

All statistical analyses will be performed using appropriate statistical software (eg, R version 4.5.3 [Foundation for Statistical Computing] or SPSS version 29 [IBM Corp]). Descriptive statistics will be used to summarize sociodemographic characteristics and study outcomes. Continuous variables will be reported as means and SDs or medians and IQRs, depending on distribution, and categorical variables as frequencies and percentages.

For inferential analyses, group comparisons (eg, between patients with a CPS with and without cancer history) will be conducted using *t* tests or Mann-Whitney *U* tests for continuous variables and chi-square tests for categorical variables. Associations between psychosocial outcomes (eg, anxiety, depression, quality of life, and supportive care needs) and potential predictors will be examined using multivariable linear or logistic regression models, depending on the distribution and nature of the outcome variables. Normative comparisons will be conducted for selected validated instruments (eg, GAD-7, PHQ-9, and KINDL) using age- and gender-adjusted reference values and 1-sample *t* tests. To account for the nonindependence of observations within families (parent-child dyads and sibling clusters), mixed-effects models with random intercepts for family units will be applied, provided that the available sample size allows sufficiently powered analyses. Otherwise, analyses will be considered exploratory. Relevant covariates will include

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age, sex, diagnosis status, time since diagnosis, and socioeconomic indicators, as well as clinical variables where available.

Missing data will be assessed for patterns and mechanisms of missingness. If data are assumed to be missing at random, multiple imputation using chained equations will be performed. Sensitivity analyses will be conducted to compare imputed and complete-case results. For exploratory analyses involving multiple outcomes, false discovery rate (FDR) correction will be applied. All tests will be 2-sided, with $P<.05$ considered statistically significant.

Qualitative Data Analysis

The qualitative data will be analyzed using qualitative content analysis based on the approaches described by Mayring and Fenzl [40] and Mayring [41]. The interviews will be digitally audio recorded and transcribed verbatim. The categorization process for the analysis of the qualitative interviews will follow a mixed approach. Specifically, it will combine a deductive approach, which will be guided by the predefined research questions, and an inductive approach, which will be informed by the data. The deductive approach will involve applying existing theoretical concepts or frameworks to categorize the data in relation to the research questions. Meanwhile, the inductive approach will allow new categories or themes to engage directly from the material, without preconceived notions. This dual approach will ensure a comprehensive and nuanced analysis, capturing both theory-driven insights and data-driven findings. A coding guideline based on this approach with anchor examples will be created and applied to a portion of the data material separately by 2 project members to increase validity and reliability. In cases of unclear category assignments and coding, these will be discussed, and the coding guideline will subsequently be adapted and optimized. The final analysis is carried out using the definitive coding guideline. The analysis will be performed using MAXQDA2022 (VERBI Software).

Development of Recommendations

Evidence-informed recommendations for psychosocial and family-centered care in treating CPSs based on the integrated findings from the systematic review, quantitative survey, and qualitative interviews will be developed. The recommendations will focus on identifying critical psychosocial burden situations, supportive care needs, and structural gaps in current care pathways across different stages of the CPS trajectory, including diagnosis communication, long-term surveillance, and transition processes.

Particular emphasis will be placed on recommendations that support health care professionals in addressing the specific psychosocial needs of affected families and adapting communication and support strategies to different developmental stages and family situations. The multidisciplinary perspectives of affected families and health care professionals will be incorporated to ensure clinical relevance and practical applicability.

The recommendations will be subjected to a 2-step validation process involving affected parents and health care professionals (with a target of 2-3 participants per group): (1) a joint meeting to present the findings and discuss the proposed recommendations, followed by (2) a written consultation phase to solicit detailed feedback on the recommendations. The feedback obtained through this iterative process will be used to refine and finalize the recommendations, which will then be disseminated to health care providers and patient advocacy groups.

Results

The KiTDS-Care study was funded in October 2024 and is currently in the recruitment phase following ethics approval. The interviews with health care professionals were conducted from May 2025 to October 2025. Recruitment of families started in July 2025 and will continue until April 2027 across participating clinical sites and patient organizations. The KiTDS-Care study aims to recruit a total of 90 families affected by a CPS in Germany. This sample size was determined based on feasibility considerations, including the expected number of eligible families within participating clinical sites and patient advocacy networks. As of January 2026, participant recruitment is ongoing, and 52 participants from 24 families have been enrolled. The target sample is considered sufficient to allow descriptive analyses of psychosocial burden and support needs, subgroup comparisons where feasible, and the integration of quantitative and qualitative findings within the mixed methods design. Data collection for the systematic review began in January 2025. Initial study results are anticipated to be available in October 2027.

Discussion

Anticipated Findings

We anticipate that families affected by a CPS in Germany will experience a substantial and multidimensional psychosocial burden. The nature and extent of this burden may differ between families with and without a history of pediatric cancer, reflecting challenges related to cancer diagnosis and treatment as well as the ongoing uncertainty, surveillance demands, and perceived future cancer risk associated with CPSs. In particular, we expect elevated psychological symptoms and reduced HRQoL in affected children and adolescents as well as their family members, alongside significant supportive care needs. These challenges are likely to be particularly pronounced in relation to ongoing uncertainty regarding cancer risk, repeated surveillance procedures, genetic testing decisions, and communication within the family system.

Although systematic evidence from the German health care context is limited, existing international studies similarly report increased psychosocial problems in CPS-affected families [17-19]. However, differences in health care organization, particularly the integration and accessibility of genetic counseling and long-term psychosocial support, limit the transferability of these findings to Germany. Our study therefore addresses an important gap by generating context-specific evidence within a health care system where structured psychosocial and genetic support pathways are still developing.

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Strengths and Limitations

A key strength of this study is its convergent mixed methods design, integrating quantitative survey data, qualitative interviews, and a systematic evidence synthesis. This approach enables a comprehensive understanding of CPS-related psychosocial burden by combining prevalence estimates, subjective experiences, and existing international evidence. The inclusion of multiple family members and health care professionals further allows a multiperspective analysis of care needs and psychosocial stressors. However, several limitations should be considered. Children and adolescents are not included in the qualitative interviews. Given the broad age range of the target population, the heterogeneity in developmental stages, and the exploratory nature of the qualitative component, we initially focused qualitative data collection on parents and health care professionals. Recruitment via clinical networks and patient organizations may introduce selection bias, potentially underrepresenting families with limited access to specialized care or those with greater psychosocial burden. In addition, the cross-sectional design of the quantitative component limits causal inference and does not allow the assessment of changes over time.

Implications

The findings are expected to contribute to the identification of critical situations associated with increased psychosocial burden and supportive care needs in families affected by a CPS. This may include a more detailed understanding of context-specific stressors and phases of heightened vulnerability across the care trajectory, such as genetic result disclosure, initiation of surveillance, and transition to adult care. Furthermore, the results may help identify families at increased risk for psychosocial problems and highlight modifiable health care–related factors that contribute to burden.

By integrating quantitative and qualitative evidence with existing literature, this study will provide a robust empirical foundation for the development of CPS-specific psychosocial interventions. Future work based on the identified burden patterns, needs, and qualitative insights into family experiences could translate these findings into a structured, syndrome-specific psychosocial intervention program tailored to the unique challenges of CPS-affected families. Such an intervention could complement existing medical surveillance programs and strengthen integrated, family-centered care pathways.

In addition, the evidence generated may inform national clinical recommendations and contribute to the integration of psychosocial care into future CPS guidelines, ensuring that the specific needs of affected families are systematically considered and that health care professionals are supported in tailoring communication and psychosocial care strategies to different patient and family groups.

Acknowledgments

The authors declare the use of generative AI (GenAI) in the research and writing process. According to the Generative Artificial Intelligence Delegation Taxonomy (2025), the following tasks were delegated to GAI tools under full human supervision: adapting and adjusting emotional tone and translation. The GenAI tool used was DeepL Write (DeepL SE). Responsibility for the final manuscript lies entirely with the authors. GAI tools are not listed as authors and do not bear responsibility for the final outcomes.

Data Availability

Due to the sensitivity of the data and the rarity of cancer predisposition syndrome in Germany, individual deidentified participant data, the data dictionary, statistical code, and other study materials will be made available only upon reasonable request to the corresponding author.

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Authors' Contributions

The study was conceptualized by MK, LI, and CB. MK is responsible for the overall conduct and coordination of the study and serves as the principal investigator. JB and VP drafted the manuscript. All authors have reviewed and provided input to the manuscript and approved the final manuscript. The study is conducted within the German Center for Child and Adolescent Health, with partner sites in Hamburg (LI) and Greifswald/Rostock (JB and CB)

Conflicts of Interest

None declared.

Multimedia Appendix 1

SPIRIT Checklist.
[PDF File (Adobe PDF File), 249 KB-Multimedia Appendix 1]

Multimedia Appendix 2

PRISMA checklist.

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[PDF File (Adobe PDF File), 83 KB-Multimedia Appendix 2]

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Abbreviations

CPS: cancer predisposition syndrome

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FDR: false discovery rate
GAD-7: Generalized Anxiety Disorder–7
HRQoL: health-related quality of life
KINDL: Kinder Lebensqualität Fragebogen
KIONET: Kinderonkologisches Netzwerk Bayern
KiTDS-Care: Children With Tumor Predisposition Syndrome and Their Families–Analysis of the Current Care Situation and Support Needs, and Derivation of Care Recommendations
LMU: Ludwig-Maximilians-University
PHQ-9: Patient Health Questionnaire–9
PICO: population, intervention, comparison, and outcome
PRISMA: Preferred Reporting Items for Systematic Reviews and Meta-Analyses
SDQ: Strengths and Difficulties Questionnaire
ULQIE: Ulm Quality of Life Inventory for Cancer Patients

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