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Parents' expectations regarding case management for rare diseases in Switzerland: mixed-method findings from an online survey

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Summary

AIMS OF THE STUDY: This pilot study aims to enhance understanding by examining parents' specific views on the requirements, content and objectives of case management and advanced care coordination for children with rare diseases during childhood. The findings of this study are expected to offer valuable insights and recommendations for existing and future initiatives in clinical practice and research, with the goal of improving the comprehensive, child-centred and family-orientated approach to case management.

METHODS: This pilot study is part of an ongoing prospective study (SPACE), involving parents and families from various networks in Switzerland. Participants were parents recruited from the Children with Rare Diseases (KMSK) network consisting of families with children with rare diseases. The survey questionnaire covered demographic information; expectations and perceived need for case management; assessment of their quality of life and their child's suffering; and evaluation of interprofessional and interdisciplinary communication. Qualitative data from free-response answers were analysed using Mayring's content analysis and descriptive statistics were used to analyse quantitative data from Likert-scale questions.

RESULTS: The study included 108 respondent families from among the 775 in the KMSK, a 14% response rate. The age of their children ranged from 0.4 to 24 years (mean: 8) and their level of suffering in the past six months varied, with 31.5% indicating intense or very intense suffering. In terms of case management, 15.8% of families reported access while 32.4% expressed a need but did not have access to it. The study identified three categories of parental expectations regarding case management, emphasising the importance of interprofessional collaboration, effective communication and comprehensive support.

CONCLUSIONS: The findings shed light on the high need for case management support with a current undersupply in Switzerland and an association with reduced parental quality of life, highlighting the necessity for diverse support and assistance to effectively manage the challenges faced by families with children with rare diseases.

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Introduction

A disease is defined as rare when it affects fewer than 1 in 2000 people. There are over 6000 rare diseases [1], cumulatively affecting 30 million people in Europe and 400 million people worldwide [2]. The prevalence of rare diseases is rising, a phenomenon attributed to several factors: an increasing incidence due to enhanced periconceptional care and techniques; advancements in diagnostic techniques; as well as innovative and more effective treatments that increase survival rates and the number of people living with rare diseases over time [3, 4]. In general, infants, children and adolescents have unique characteristics regarding development, dependency on caregivers, differential epidemiology, demographic pattern and financial issues, which health services and researchers need to incorporate into an inclusive, child-centred and family-orientated definition of medical necessity [5]. Rare diseases however pose even more specific challenges to patients, families and healthcare systems. Most rare diseases are chronic, genetic, involve multiple body systems and therefore need various medical specialties; moreover only a few conditions have an effective treatment [6]. As a result, families are often confronted with a "diagnostic odyssey". Unsurprisingly more than half of families indicate that their primary care provider does not sufficiently understand the condition [6, 7]. Traditional healthcare models often fall short in addressing the complex needs of these patients, resulting in fragmented care, suboptimal outcomes and increased healthcare costs [8].

Parents play a central role in their child's healthcare journey, acting as advocates, decision-makers and primary caregivers [9]. They possess unique insights into their child's condition and are vital for coordinating and ensuring continuity of care. However, effectively managing these complex situations requires the consolidation, coordination, prioritisation, communication and support of a vast amount of information, needs and resources within the child's complex system [8]. These tasks are time-consuming and resource-intensive, placing additional strain on parents and their support system, especially in the face of an already difficult and complex situation. The collaboration between parents and healthcare professionals, with a strong emphasis on the quality, not just quantity, of life, is crucial. In a study on transition, Bigby et al. underscore

the importance of a trusting relationship between the case manager and parents, often the result of many months of dedicated effort [10]. With an increasing focus on the quality of life (QoL), paediatric palliative or advanced care teams have become important pillars in coordinating and supporting complex care systems, bringing together a diverse team of physicians, nurses, social workers, psychologists, therapists and other allied healthcare professionals. The ideal approach is one that is child-centred, family-orientated, comprehensive, holistic and financially sustainable. However, the reality is often different, as families frequently face challenges such as uncoordinated appointments, ineffective communication, limited resources, inconsistent knowledge and the overwhelming burden of being the main coordinators of care [11, 12]. Addressing these issues presents numerous challenges and requires multiple potential solutions. Concepts and terms like case management, care coordination, advanced care, comprehensive and well-managed care, continuity of care, care integration, team-based care, patient-centred medical home, chronic care model, accountable care organisations, standards of care, national networks of expertise and one-stop-shop services have been described as potentially effective and efficient ways for fostering collaboration and communication among stakeholders and supporting them [2, 13, 14]. While these terms are used inconsistently within the healthcare continuum, their shared goal is to improve patient outcomes and care efficiency. In this context, we will refer to the overarching concept of supporting coordination and care in rare diseases as case management (CM). The etymology of "case management" derives from the Latin words *casus* meaning "event" or "situation" and *manu* and *agere* meaning "handle". Together, "case management" historically suggests guiding or overseeing specific events or situations. The concept of case management originated in early 20th-century social work, evolving as a response to the growing intricacies of social service systems following the industrial revolution. During the 1970s, it was introduced in medicine as a response to the need for coordination and continuity of care amid the complexity of the healthcare system, guiding patients through the system, ensuring that they received appropriate care and helping to control costs [15]. Over time, as healthcare systems and patient needs evolved, so too did the role and scope of case management, expanding into various specialties and adapting to new models of care. Today, it is recognised as an essential discipline in healthcare and defined as a "collaborative process of assessment, planning, facilitation, care coordination, evaluation and advocacy for options and services to meet an individual's and family's comprehensive health needs through communication and available resources to promote patient safety, quality of care, and cost effective outcomes" [16].

These standards emphasise the expertise and value of case managers in empowering individuals to access quality healthcare, advocating for patients' needs and serving as financial stewards. Despite the increasing prevalence of chronic complex and rare conditions in childhood and the proposed need for case management and associated concepts, there has been little research focused on parents' experiences of navigating the healthcare system. Notably, very few studies have been conducted on case management and care coordination from the perspectives of parents, who are a primary target group for case management in paediatric care. This pilot study aims to enhance our understanding by examining parents' specific views on the requirements, content and objectives of case management for them and their children with rare diseases. The findings of this study are expected to offer valuable insights and recommendations to existing and future initiatives in clinical practice and research, with the goal of enhancing a comprehensive, child-centred and family-orientated approach to case management.

Population and methods

This pilot study is part of an ongoing prospective study called the Swiss Pediatric Advanced Care and Ethics (SPACE) study, which began in 2023 and involves a cohort of parents, families and healthcare professionals from various networks and teams in Switzerland. Participants for the current pilot study were parents recruited from a well-established network known as the Children with Rare Diseases (KMSK, *Kinder mit seltenen Krankheiten*) family network [17, which consists of 775 families, children affected by diverse complex chronic conditions, including rare diseases, plus their parents. KMSK is a unique Swiss non-profit organisation which since 2014 has been connecting affected families mainly in the German-speaking part of Switzerland from the moment of diagnosis, and provides advisory as well as occasional financial support. Membership of KMSK is free and members have no obligations.

Invitations to participate in the survey were sent to parents via email, providing them with detailed information about the study's purpose, procedures and confidentiality measures. The questionnaire was in German (see appendix). Prior to taking part in the survey, interested parents were required to provide informed consent. Out of the total of 775 potential participant families, 108 individual questionnaires were completed (14% response rate). If both parents participated, they were counted as one participant family. We did not receive multiple questionnaires from any family. To safeguard participants' anonymity, each questionnaire was assigned a code for identification purposes, a measure that rendered specific follow-up or analysis of non-responders impossible. The questionnaire requested a diagnosis and a description of symptoms from all 108 parents having a child with a rare and complex chronic condition. Due to the rarity of these conditions and the challenges of anonymisation, specific diagnoses are not reported. After a first review of the presented results showed a rich and saturated spectrum of answers and considering the presumed high psychological and time burden on the parents, we limited our follow-up to only one email reminder sent to everyone. The online survey was set up using Tivian Unipark, EFS Fall 2022 (Tivian XI GmbH, Cologne, Germany).

Measurements

The research team developed a survey questionnaire with two main parts consisting of multiple sections for collecting relevant data. The first part covered various areas such as demographic information, parents' expectations of a case manager, assessment of child's suffering and parents' QoL, evaluation of interprofessional and interdisci-

plinary communication and perceived need for case management. Parents were given the opportunity to indicate their expectations of a case manager, with the option to provide their top three priorities or express their thoughts in a few sentences. Additionally, participants were asked to rate their own and their child's QoL and suffering over the past 6 months, their evaluation of interprofessional and interdisciplinary communication ("satisfaction Likert scale") and their perceived need for case management using a 5-point Likert scale. The second part contained in-depth open questions on various topics in the context of rare disease, which will be published separately.

Due to the exploratory nature of this pilot study, we did not provide a specific definition of case management to parents. This approach was taken to capture a wide range of aspects they deemed relevant.

Content and statistical analysis

The qualitative data were obtained from the free-response answers, which were analysed using Mayring's content analysis [18, 19] via the following steps:

1. The respondents' free-response answers, including keywords or complete sentences, were transcribed into an Excel file.
2. Content categories (codes) and overarching coding families and corresponding definitions per code were developed inductively, and their descriptions were documented in a code manual. The code manual included definitions and examples.
3. The first author then assigned the respondents' answers to the appropriate content categories, i.e. codes and coding families based on the code manual.
4. Frequencies of responses were calculated.

Mayring's content analysis provides a systematic and replicable approach for interpreting patterns and themes within semi-quantitative questionnaire data, ensuring both qualitative depth and quantitative reliability.

Descriptive statistics were used to analyse the quantitative data obtained from the responses to the Likert-scale questions. Pearson's chi-squared test was applied to determine the associations between variables. P values lower than 0.05 were considered statistically significant. Statistical analysis was conducted using IBM SPSS Statistics.

The study was reviewed and approved by Ethics Review (CEBES), the institutional review board of the Institute of Biomedical Ethics and History of Medicine at the University of Zurich (CEBES #2023-02).

Overall, the survey methodology employed in this study aimed to gather data on the expectations of parents of children with rare and complex chronic conditions regarding case management. The semi-quantitative approach allowed for both qualitative insights through free-response questions and quantitative analysis through Likert-scale responses, providing a comprehensive understanding of the parents' perspectives.

Results

108 of the 775 families participated after two mailings within 4 weeks (14% response rate). Table 1 presents the characteristics of the study sample. The mother only responded for the majority, 88.9%, of respondent families, the father only for 6.5% and both parents for 4.6%. If both parents participated, they were counted as one family/participant. We did not receive multiple questionnaires from any family. The mean age of the children in the study was 8 years (median: 7 years) with a standard deviation of 4.8 years and an age range of 0.4–24 years. In terms of the child's level of suffering in the past six months, 45.4% of parents reported that their child experienced no or little suffering, 23.1% indicated that their child experienced moderate suffering, and 31.5% reported that their child endured intense or very intense suffering. Regarding the parents' QoL, 45.3% reported their QoL as good or very good, 35.2% indicated that it was moderate and 19.5% reported their QoL as bad or very bad.

Table 2 presents the current support received by the families (n = 108) for various aspects of care. With regard to the outpatient nursing service (called *Kinderspitex* in Switzerland), 25% of families reported receiving the service and needing it; 4.6% reported receiving it but did not consider it necessary; only 1.9% of families expressed a need for this service but were not receiving it; the majority, 66.7%, reported not receiving nor needing outpatient nursing.

Regarding personal care assistance (*Assistenzperson*), 19.4% of families reported receiving and needing this assistance; 8.3% expressed a need for personal care assistance but were not receiving it; the majority, 67.6%, reported not receiving nor needing such assistance.

The number of different outpatient therapies (like physical therapy, occupational therapy, etc.) received by the families had a mean of 3.88 per patient with a standard devia-

Table 1:
Study sample characteristics (n = 108).

		n (%)
Role, n (%)	Mother	96 (88.9%)
	Father	7 (6.5%)
	Both	5 (4.6%)
Age of child, in years	Mean (SD)	8 (4.8)
	Range	0.4–24
Child's level of suffering in the past six months, n (%)	No or little	49 (45.4%)
	Moderate	25 (23.1%)
	Intense or very intense	34 (31.5%)
Parent's quality of life, n (%)	Good or very good	49 (45.3%)
	Moderate	38 (35.2%)
	Bad or very bad	21 (19.5%)

tion of 1.1, indicating that almost all families have multiple professions to coordinate in addition to a medical specialist.

Regarding case management, 15.8% of families had experienced or benefited from case management in some capacity. On the other hand, 32.4% of families reported not receiving case management, but they expressed a need for such support. And 26.9% reported not receiving nor needing case management.

Importance of case management and association with other factors

A large majority of participants, 83.3% (n = 90), rated case management as important or very important. No participant described case management as not important. 41.7% (n = 45) saw a pressing need for improvement in their own current situation regarding case management, while only 17 (15.8%) of families have or had access to some form of case management (table 1).

Pearson's chi-squared test showed a significant association (p = 0.002) between the child's level of suffering and the parent's QoL and a significant association (p <0.001) between parents' evaluation of interdisciplinary communication and coordination and the need for case management. We found no significant association between the child's level of suffering or the parent's QoL and their need for and appraisal of case management. Similarly, we did not find any association between support with outpatient nursing or supportive assistance and case management.

Parents' expectations of case management

"Families have been experts for years, diagnosis is their life, and it's not just a diagnosis. [Case management] certainly requires a lot of sensitivity and empathy" (mother).

The primary objective of the survey, conducted among parents of children with complex chronic rare conditions, was to gain insight into the specific areas where the support and assistance of a case manager could be beneficial in effectively managing their child's condition. 87 respondent families (80.6%) had clear expectations of case management and described one or more expectations, with a total of 160 expectations described in short sentences. Data analysis yielded three distinct coding families that encompassed the various functions of case management, as presented in table 3. Coding families were named "Coordination and collaboration", "Counselling and interpretation" and "Support and empowerment". General and specific quotes from parental answers are given below.

Coordination and collaboration

"I constantly have to inquire everywhere: when is the next appointment, why haven't we received it, has the doctor's report been sent, what happens next..."

"[We] would have wished for a roundtable discussion with the future involved disciplines and possibly a social worker who could explain what is important now, what will be important in the future, and what needs to be considered. Contacts to support groups or even, if possible, contact with those affected in Switzerland/region would be valuable. It would also be nice if it's possible to have another phone call about 1–2 weeks after receiving the diagnosis to address any remaining questions."

Under the "Coordination and collaboration" category, parents emphasised the significance of interprofessional collaboration to facilitate efficient communication and information exchange among healthcare professionals, as well as coordination with schools. Managing financial aspects effectively was another crucial concern, and parents sought support in handling funding-related matters, including assistance with specific needs applications, managing the payroll of assistants and monitoring overall costs. Knowledge management emerged as a vital aspect of case management, involving the maintenance of patient records, the collection and archiving of pertinent information and the dissemination of care plans as a central point of contact within an inter- and transprofessional network. Additionally, parents valued timely coordination of appointments, efficient time management and guidance for managing symptoms when specialist care was not readily available.

Table 2:
Current support of families (n = 108).

		n (%)
Outpatient nursing, n (%)	Receiving + need	27 (25%)
	Receiving + do not need	5 (4.6%)
	Not receiving + need	2 (1.9%)
	Not receiving + do not need	72 (66.7%)
	Missing answers	2 (1.9%)
Personal care assistance, n (%)	Receiving + need	21 (19.4%)
	Receiving + do not need	3 (2.8%)
	Not receiving + need	9 (8.3%)
	Not receiving + do not need	73 (67.6%)
	Missing answers	2 (1.9%)
Number of different outpatient treatments	Mean (SD)	3.88 (1.1)
	Range	0–"4 or more"
Case management, n (%)	Receiving + likely need	17 (15.8%)
	Not receiving + need	35 (32.4%)
	Not receiving + do not need	29 (26.9%)
	Not receiving + unclear whether need	26 (24.1%)
	Missing	1 (0.9%)

Counselling and interpretation

"Many times, we also feel overwhelmed in the moment and may not even be able to ask the right questions. Or we lack the courage or clarity. Sometimes, the good questions only come to mind on the way home, but do we still ask them then?"

Within the "Counselling and interpretation" category, parents expressed the need for a trusted "translator" to serve as a bridge between doctors and themselves for effective communication, representing parental opinions and accompanying them during discussions. They also sought comprehensive counselling that involved second opinions and exploration of alternative disciplines when necessary. Access to assistive technology, and to up-to-date research and therapy options and support for problem-solving were identified as critical components within this category.

Support and empowerment

"At the time of diagnosis, help was lacking everywhere! No support: neither psychological, financial, emotional, nor professional... Nothing! Then you have no energy to gather information, exchange experiences, and search everywhere..."

The "Support and empowerment" category underscored the importance of providing comprehensive support to enhance the wellbeing of both the child and the entire family, including siblings. Parents valued assistance in resolving communication issues or conflicts, via mediators, as well as support with daily tasks and activities. Additionally, they highlighted the need for help in navigating hospital procedures, advocating for their child's best interests and addressing educational matters and job placements as essential areas for empowerment.

Overall, table 3 shows the need for multifaceted support and assistance in various areas to effectively manage the unique challenges they face.

Discussion

Our pilot study aimed to explore the specific needs and preferences of parents of children with rare diseases in relation to case management, with a particular focus on case management. The findings shed light on the high need for case management support with a current undersupply in Switzerland and the specific challenges and aims that are deemed crucial by parents in supporting their families dealing with rare diseases. Most parents had certain expectations of case management and expressed a broad array of needs for existing or future services. To our knowledge, this is the first study on the need and use of case management and the largest sample of parents in the context of rare disease, case management and/or paediatric palliative care services. The challenges and aims of case management were categorised into three main areas: "coordination and collaboration", "counselling and interpretation" and "support and empowerment". In general, parents considered case management to be highly important. However, the study found that only 15.8% of parents had access to some form of case management, while a larger proportion, 32.4%, expressed a current need for case management but did not have access to it (in contrast to the 1.9% who needed outpatient nursing but lacked access to it), which can be seen as underprovision of the studied population.

Similar to previous studies, knowledge management played a central role in maintaining patient records and disseminating care plans. Timely and efficient coordination of appointments and guidance on symptom management were also mentioned as areas where case managers were involved. Considering that 54.4% of respondents reported their child to be experiencing some level of suffering and the relatively low frequency of 9 mentions of symptom management (table 3), it could be inferred that a majority of parents may view a case manager rather as a supplementary member of the medical team, potentially facilitating the communication and management of unassessed symptoms. In agreement with existing literature, financial management support was identified as essential. This support helps in handling funding-related matters efficiently, saves the family time and resources, and enhances their QoL [8, 9].

Another previously less-discussed topic emerged in the realm of "counselling and interpretation", revealing the necessity for case managers to serve as trusted "translators" who facilitate effective communication between doctors and parents. Additionally, parents expressed the need for thorough counselling, encompassing second opinions and exploration of alternative approaches when necessary. Vital components within this category included access to assistive technology, up-to-date information, comprehensive electronic patient records and overarching problem-solving support.

Several issues overlap with existing professional roles (e.g. assistive technology with occupational therapists, financial management with social workers). In paediatric palliative care, for example, collaboration with these professionals is integral to delivering holistic services, working in conjunction with the case manager. This collaboration streamlines the case manager's role, which becomes akin to controlling symptoms, focusing on identifying needs, precisely directing enquiries to the appropriate team members and continuously assessing the positive impact of these actions. This approach enhances efficient care coordination while optimising the collective efforts of the healthcare team.

In the "support and empowerment" category, the significance of holistic support for both the child and the entire family, including siblings, was underscored. In this regard, case managers were recognised as mediators in resolving interpersonal conflicts, as well as helping in daily tasks and activities. Furthermore, they were deemed essential for helping to navigate the hospital, advocating for the child's best interests and addressing educational and job placement matters.

Combining our results with previous publications, future guidelines for rare disease should (a) prioritise diversity and unintended consequences for equity, (b) raise awareness about disparities, (c) support effective collaboration, (d) provide training for all stakeholders on the specific needs of children and families with rare disease, (e) implement a child-centred and family-orientated approach, (f) collect and analyse more data (e.g. a minimum dataset including psychosocial aspects), (g) support and guide necessary policy changes for rare diseases, (h) build strong

community support networks and (i) promote transparent communication to mitigate inherent inequities in rare disease care and research [6, 8, 11, 20–23].

Limitations of our study include the relatively small number of parents with older children, which may have led to an underrepresentation of transition-related needs. Given that the groups requiring outpatient nursing and personal care assistance – but not receiving it – are the smallest, at 1.9% and 8.3%, respectively, it is possible that these groups are underrepresented in the study. They may not have had the capacity or energy to participate.

Given that this was a pilot study, our questionnaire was broad yet as concise and short as possible. For instance, QoL of parents was assessed solely using a subjective 5-point Likert scale. This approach is justifiable based on previous studies [24, 25]. The current absence of formal case management services might have constrained participants' familiarity with these concepts, potentially leading to responses centred on theoretical ideals instead of actual experiences. It is important to note that the codes and definitions presented in table 3 stem from an interpretation of patterns and themes identified in brief semi-quantitative questionnaire data, as outlined by Mayring's approach [19]. Since we only employed the chi-squared test for the specific hypothesis and did not use other statistical models, we refrained from using listwise or pairwise deletion of variables, any form of imputation or inter-/extrapolation of missing values. Although our study involved a relatively high number of participants, the response rate of 14% was relatively low, and the sample was limited to one country, one language (German) and one network – as befits a pilot study. Subsequently our findings may not be generalisable to other settings or populations. The identified pressing issue of high bureaucracy when accessing necessary financial support may be specific to Switzerland, where financial assistance for families is available but subject to a complex and sometimes overwhelming process. It is also important to acknowledge that our sample may not be representative of rare diseases, given the relatively low response rate. To date, obtaining comprehensive data that cover the prevalence and complexity of rare diseases, es-

Table 3:
Parents' expectations from case management.

Categories	Codes	Definition/examples	n
Coordination and collaboration			**160**
	Interprofessional collaboration	Focuses on interprofessional communication and information exchange, including communication with schools, and the coordination of information to ensure everyone involved has the same information	40
	Financial management	Refers to administrative assistance for issues related to funding, such as standardising and expediting processes, managing extensions and applications, handling financial queries, payroll and monitoring cost coverage	36
	Knowledge management	Involves maintaining an overview of information, assisting with document archiving, managing patient records, promptly obtaining reports, acting as a central point of contact, collecting and archiving knowledge and disseminating it, accordingly, often including a care plan	29
	Coordinating appointments	Involves coordinating appointments efficiently, avoiding unnecessary appointments when possible	20
	Time management	Focuses on managing time effectively, monitoring schedules, and prioritising important tasks	10
	Symptom management	Involves interpreting and coordinating prescriptions and medication optimisation, as well as managing symptoms when a specialist is not required or available	9
	Collaborative spaces	Refers to coordinating and offering home visits and roundtable discussions to facilitate exchange among relevant parties	5
	Emergency triage	Focuses on maintaining accessibility and performing triage functions in emergency situations	4
	Assisted correspondence	Involves managing phone calls and following up emails	4
	Responsibilities	Involves clarifying roles and responsibilities to avoid duplication of efforts	3
Counselling and interpretation			**55**
	Translator	Refers to acting as a neutral "translator" and trusted person who supports and advocates for effective communication between doctors and parents. This includes accompanying parents during discussions, strengthening and representing parental opinions	18
	Second opinion	Involves counselling from a broader perspective and drawing on experience to consider alternatives, seek second opinions or involve new disciplines when necessary	14
	Assistive technology and assessment	Focuses on organising assistive technology, clarifying responsibilities and identifying beneficial resources and services	7
	Research	Assisting in the research of new and up-to-date information about the illness, as well as the latest therapy options, including monitoring their effectiveness and, if necessary, providing suggestions for improvement and therapy adjustments based on the latest advancements	8
	Problem solver	Acts as a point of contact for questions and supports problem-solving in all aspects	4
	Legal advice	Offers legal counselling and guidance	2
	Synopsis	Involves having comprehensive knowledge of the entire case and providing guidance throughout the process	2
Support and empowerment			**34**
	Family wellbeing	Providing support to enhance the family's overall wellbeing, including siblings, by consistently checking in on their situation and addressing their needs with appropriate assistance	14
	Mediator	Providing immediate assistance in resolving communication issues or conflicts by acting as a mediator	6
	Daily support	Offering support in general questions, daily tasks and activities	2
	Hospital navigator	Providing guidance and support before, during and after hospital stays	5
	Child's advocate	Takes a holistic view of the child's wellbeing and places them at the centre, advocating for their best interests	4
	Education navigator	Addresses issues and conflicts in the school setting, assists in the search for educational opportunities and helps with job placements	3

pecially within the context of complex chronic conditions, has remained beyond our reach. It is important to note that while rare conditions and complex chronic conditions intersect, they are not synonymous so it is of paramount importance to further characterise these distinct groups, gain insight into their unique needs and identify the necessary associated services. To address these limitations, we intend to carry out a prospective study to further study and implement the identified case management strategies. Additionally, we aim to determine the role and potential opportunities of existing reference centres for rare diseases, such as KOSEK (National Coordination of Rare Diseases, *Nationale Koordination Seltene Krankheiten*) and to include all parts of Switzerland.

For future research, recommendations on healthcare structure and care pathways, it is crucial to enhance case management structures to consolidate, communicate and comprehend complex information, integrating existing but insufficiently implemented ideas of case management and care coordination. Future strategies, such as electronic patient records, need careful integration to cater to the specific needs of rare diseases, including the above-mentioned aspects for future guidelines. Adequate outcome indicators should be established to evaluate the impact of healthcare and psycho-social-spiritual interventions, considering both patient and family outcomes. Additionally, involving patients and families in decision-making processes through shared decision-making principles can lead to more empathic, supportive and co-responsible care pathways. To achieve a more comprehensive and coordinated care approach for children with rare diseases and their families, it is essential to strike a balance between economic necessity and integrated care needs. Nationwide and internationally connected integrated and financially sustainable care programmes should be fostered to address the unique needs of people with rare diseases while promoting a healthy society with shared needs. Overall, case management serves as a necessary basis for future steps in providing optimal care for this population and should be a top priority for collaborations between existing teams and disciplines.

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References

1. Nguengang Wakap S, Lambert DM, Olry A, Rodwell C, Gueydan C, Lanneau V, et al. Estimating cumulative point prevalence of rare diseases: analysis of the Orphanet database. Eur J Hum Genet. 2020 Feb;28(2):165–73. http://dx.doi.org/10.1038/s41431-019-0508-0.
2. Castro R, de Chalendar M, Vajda I, van Breukelen S, Courbier S, Hedley V, et al. Rare Diseases. In: Amelung V, Stein V, Suter E, Goodwin N, Nolte E, Balicer R, editors. Handbook Integrated Care [Internet]Cham: Springer International Publishing; 2021. pp. 763–82. [[cited 2023 Jul 17]]. http://dx.doi.org/10.1007/978-3-030-69262-9_44.
3. Auvin S, Irwin J, Abi-Aad P, Battersby A. The Problem of Rarity: Estimation of Prevalence in Rare Disease. Value Health. 2018 May;21(5):501–7. http://dx.doi.org/10.1016/j.jval.2018.03.002.
4. Fraser LK, Gibson-Smith D, Jarvis S, Norman P, Parslow RC. Estimating the current and future prevalence of life-limiting conditions in children in England. Palliat Med. 2021 Oct;35(9):1641–51. http://dx.doi.org/10.1177/0269216320975308.
5. Stille C, Turchi RM, Antonelli R, Cabana MD, Cheng TL, Laraque D, et al.; Academic Pediatric Association Task Force on Family-Centered Medical Home. The family-centered medical home: specific considerations for child health research and policy. Acad Pediatr. 2010;10(4):211–7. http://dx.doi.org/10.1016/j.acap.2010.05.002.
6. Bogart K, Hemmesch A, Barnes E, Blissenbach T, Beisang A, Engel P, et al.; Chloe Barnes Advisory Council on Rare Diseases. Healthcare access, satisfaction, and health-related quality of life among children and adults with rare diseases. Orphanet J Rare Dis. 2022 May;17(1):196. http://dx.doi.org/10.1186/s13023-022-02343-4.
7. Molster C, Urwin D, Di Pietro L, Fookes M, Petrie D, van der Laan S, et al. Survey of healthcare experiences of Australian adults living with rare diseases. Orphanet J Rare Dis. 2016 Mar;11(1):30. http://dx.doi.org/10.1186/s13023-016-0409-z.
8. Baumbusch J, Mayer S, Sloan-Yip I. Alone in a Crowd? Parents of Children with Rare Diseases' Experiences of Navigating the Healthcare System. J Genet Couns. 2018 Aug;28(1):80–90. http://dx.doi.org/10.1007/s10897-018-0294-9.
9. Ankeny EM, Wilkins J, Spain J. Mothers' Experiences of Transition Planning for Their Children with Disabilities. Teach Except Child. 2009 Jul;41(6):28–36. http://dx.doi.org/10.1177/004005990904100604.
10. Bigby C, Ozanne E, Gordon M. Facilitating Transition. J Gerontol Soc Work. 2002 Dec;37(3–4):25–43. http://dx.doi.org/10.1300/J083v37n03_04.
11. Simpson A, Bloom L, Fulop NJ, Hudson E, Leeson-Beevers K, Morris S, et al. How are patients with rare diseases and their carers in the UK impacted by the way care is coordinated? An exploratory qualitative interview study. Orphanet J Rare Dis. 2021 Feb;16(1):76. http://dx.doi.org/10.1186/s13023-020-01664-6.
12. Walton H, Simpson A, Ramsay AI, Hudson E, Hunter A, Jones J, et al. Developing a taxonomy of care coordination for people living with rare conditions: a qualitative study. Orphanet J Rare Dis. 2022 Apr;17(1):171. http://dx.doi.org/10.1186/s13023-022-02321-w.
13. Auvin S, Bissler JJ, Cottin V, Fujimoto A, Hofbauer GF, Jansen AC, et al. A step-wise approach for establishing a multidisciplinary team for the management of tuberous sclerosis complex: a Delphi consensus report. Orphanet J Rare Dis. 2019 Apr;14(1):91. http://dx.doi.org/10.1186/s13023-019-1072-y.
14. Schultz EM, McDonald KM. What is care coordination? Int J Care Coord. 2014 Jun;17(1–2):5–24. http://dx.doi.org/10.1177/2053435414540615.
15. Kersbergen AL. Case management: a rich history of coordinating care to control costs. Nurs Outlook. 1996;44(4):169–72. http://dx.doi.org/10.1016/S0029-6554(96)80037-6.
16. Fraser K, Perez R. The Evolution of Case Management and the Professional Case Manager. In: Fraser K, Perez R, Latour C, editors. CMSA's Integrated Case Management: A Manual For Case Managers by Case Managers [Internet]1st ed. Springer Publishing Company; 2018. http://dx.doi.org/10.1891/9780826169518.0001.
17. KMSK [Internet]. [cited 2023 Aug 8]. Available from: https://www.kmsk.ch/
18. Mayring P. Qualitative Inhaltsanalyse. In: Mey G, Mruck K, editors. Handbuch Qualitative Forschung in der Psychologie [Internet]Wiesbaden: VS Verlag für Sozialwissenschaften; 2010. pp. 601–13. [[cited 2023 Oct 4]]. http://dx.doi.org/10.1007/978-3-531-92052-8_42.
19. Mayring P. Qualitative Inhaltsanalyse–ein Beispiel für Mixed Methods. Mix Methods Empirischen Bild. 2012;1:27–36.
20. Anzelewicz S, Garnier H, Rangaswami A, Czauderna P. Cultural, geographical and ethical questions when looking to enroll pediatric patients in rare disease clinical trials. Expert Opin Orphan Drugs. 2017 Aug;5(8):613–21. http://dx.doi.org/10.1080/21678707.2017.1348293.
21. Halley MC, Halverson CM, Tabor HK, Goldenberg AJ. Rare Disease, Advocacy and Justice: Intersecting Disparities in Research and Clinical Care. Am J Bioeth. 2023 Jul;23(7):17–26. http://dx.doi.org/10.1080/15265161.2023.2207500.
22. Sparring V, Tschank J, Handler K, Mansoory E, Konzett-Smoliner S, Lindgren P. Social and economic impact of a case management approach for people with rare and complex conditions in Salaj. Romania. 2018 Oct;18 s2:61.
23. Tumiene B, Graessner H. Rare disease care pathways in the EU: from odysseys and labyrinths towards highways. J Community Genet. 2021 Apr;12(2):231–9. http://dx.doi.org/10.1007/s12687-021-00520-9.
24. Krzych ŁJ, Lach M, Joniec M, Cisowski M, Bochenek A. The Likert scale is a powerful tool for quality of life assessment among patients af-

ter minimally invasive coronary surgery. Kardiochir Torakochirurgia Pol. 2018 Jun;15(2):130–4. http://dx.doi.org/10.5114/kitp.2018.76480.

25. Ventegodt S, Merrick J, Andersen NJ. Measurement of quality of life II. From the philosophy of life to science. ScientificWorldJournal. 2003 Oct;3:962–71. http://dx.doi.org/10.1100/tsw.2003.76.

Appendix: questionnaire

The appendix is available for download as a separate file at https://doi.org/10.57187/s.3401.