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Addressing unmet needs in pregnancy and family planning of people living with rare and low-prevalence diseases: results of the "ERN transversal working group on pregnancy and family planning" survey

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Abstract

Background Pregnancy and family planning for individuals living with rare and low prevalence diseases present unique medical, psychological, and logistical challenges. The European Reference Networks (ERNs) were established to address healthcare disparities and enhance patient care for rare diseases across Europe. The ERN Transversal Working Group on Pregnancy and Family Planning was created to identify common unmet needs and to develop targeted actions to improve healthcare delivery. As part of this initiative, a survey was conducted to gather insights from patients, caregivers, and family members about their experiences and challenges related to pregnancy and family planning.

Methods The survey was co-designed by healthcare professionals and patient representatives from 20 ERNs. It covered various domains, including fertility preservation, pre-conceptional counselling, psychological support in the

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pre-conceptional counselling, pre-implantation diagnosis, prenatal diagnosis, family planning, pregnancy monitoring, post-pregnancy monitoring, lactation and newborn management. The survey, available in multiple languages, was distributed via online platforms between February and July 2022. Quantitative responses were analysed descriptively, while qualitative data from open-ended questions were processed using word frequency analysis.

Results A total of 769 responses were collected, with 574 from patients and 155 from caregivers. The majority of respondents were female (90%) and aged 31–40 years, primarily from Germany, France, Spain, and Italy. The most pressing concerns identified included lack of access to accurate medical information, limited psychological support, inadequate pre-conceptional counselling, and challenges related to pregnancy monitoring and postpartum care. The need for multidisciplinary healthcare teams and improved education on reproductive health was emphasised. Word frequency analysis highlighted key concerns, with "inform," "support," and "risk" being the most recurrent terms.

Conclusions This study underscores the widespread need for comprehensive, patient-centred approaches in pregnancy and family planning for individuals with rare and low prevalence diseases. Improved access to specialised healthcare teams, psychological support, and clear, standardised medical information is essential. These findings advocate for the development of harmonised European policies and multidisciplinary strategies to enhance reproductive healthcare.

Plain english Language summary People living with rare diseases often face extra challenges when it comes to pregnancy and family planning. To better understand these issues, a group of experts and patient representatives from 20 European Reference Networks (ERNs) worked together to conduct a survey. A European survey of 769 people, mostly women aged 31 to 40 from Germany, France, Spain, and Italy, revealed significant gaps in care. The most common concerns shared by patients included difficulty finding clear medical information, not enough psychological support, gaps in counselling, and challenges with checking on health during and after pregnancy. Many felt that better teamwork among different healthcare providers and improved education on reproductive health would make a big difference. In summary, this study underscores the urgent need for better reproductive healthcare for people with rare diseases.

Keywords Rare diseases, Pregnancy, Reproductive health, Unmet needs, Psychological support, Patient-centered care european reference networks (ERN)

Background

A rare disease is defined as a condition that affects a small percentage of the population, and the specific threshold can vary by region. For example, in the European Union, a disease is considered rare when it affects no more than 1 in 2,000 people [1].

In response to the complexities surrounding rare and low-prevalence diseases, the European Commission established the European Reference Networks (ERNs). The ERNs are virtual networks of centres of expertise across the European Union, bringing together experts and patient representatives in rare or complex conditions. The 24 ERNs have been working since 2017 across various areas including clinical practice guidelines, education, registries, patients' care pathways, and other important topics [2–5].

The period spanning from preconception to postpartum is inherently challenging, but this complexity is markedly amplified in the context of rare diseases. For people living with these conditions and their caregivers, this journey often involves a labyrinth of medical, emotional and logistical challenges [6, 7].

Starting from these premises, a dedicated ERN Transversal Working Group (WG) on Pregnancy and Family Planning was created by the Coordination Team of ERN ReCONNET to promote collaboration among all the rare disease areas covered in the 24 ERNs and develop common actions [8]. The WG is composed of different Health Care Professionals and patient representatives belonging to more than 20 ERNs with the main mission to join efforts across the different disease areas and countries in order to develop common actions towards the improvement of the management of family planning in rare and low prevalence diseases in the EU.

The first action of the WG was to identify the common unmet needs and current challenges that people living with rare and low-prevalence diseases faced before, during and after pregnancy independent of the type of the disease. To that, the WG members agreed to develop two distinct surveys. The first survey targeted Health Care Professionals and was aimed at gathering professional insights [9], and the second was aimed at collecting the perspectives of patients, caregivers, and family members living with a rare and complex condition to directly understand their personal experiences and challenges in this context.

This paper aims to present the results of the survey of patients, caregivers and family members, combining classical quantitative and qualitative methods, to identify their unmet needs related to pregnancy and family planning.

Methods

This was a cross-sectional, observational study based on an online survey, developed and disseminated by the ERN Transversal Working Group on Pregnancy and Family Planning. The study targeted adult individuals (≥ 18 years) living with rare and low-prevalence diseases, as well as their caregivers and family members. Inclusion criteria included: [1] voluntary participation; [2] being a patient, caregiver, or family member with experience related to pregnancy and/or family planning in the context of a rare disease; and [3] ability to complete the survey independently in one of the provided European languages. There were no formal exclusion criteria beyond the age under 18 and inability to complete the questionnaire. Recruitment was carried out online through social media, ERN communication channels, and mailing lists, ensuring a broad representation across the EU. Although no formal sample size calculation was performed, the target was to obtain at least 500 responses to allow for meaningful descriptive analysis. Ultimately, 769 complete responses were collected.

Health Care Professionals and patient representatives from 20 European Reference Networks (ERNs) have taken part in the ERN Transversal Working Group on Pregnancy and Family Planning: ERN on bone disorder (ERN BOND), ERN on craniofacial anomalies and ear, nose and throat disorders (ERN CRANIO), ERN on endocrine conditions (Endo-ERN), ERN on epilepsies (EpiCARE), ERN on inherited and congenital anomalies (ERNICA), ERN on respiratory diseases (ERN LUNG), ERN on skin disorders (ERN Skin), ERN on adult cancers (EURACAN), ERN on urogenital diseases and conditions (ERN Eurogen), ERN on neuromuscular diseases(ERN EURO-NMD), ERN on eye diseases (ERN EYE), ERN on genetic tumour risk syndromes (ERN GENTURIS), ERN on diseases of the heart (ERN GUARD-Heart), ERN on congenital malformations and rare intellectual disability (ERN ITHACA), ERN on paediatric cancer (ERN PaedCan), ERN on hepatological diseases (ERN RARE-LIVER), ERN on connective tissue and musculoskeletal diseases (ERN ReCONNET), ERN on immunodeficiency, autoinflammatory and autoimmune diseases (ERN RITA), ERN on Transplantation in Children (ERN TRANSPLANT-CHILD) and ERN on Rare Multisystemic Vascular Diseases (VASCERN).

The survey was co-designed by a group of both clinicians and patients' representatives from different ERNs, in order to ensure the relevance and the transversality of the survey. This process was coordinated by representatives from the ERN ReCONNET Coordination Team. The co-design group has agreed first on the transversal domains to be covered in the survey. These included: fertility preservation, pre-conceptional counselling, psychological support in the pre-conceptional counselling (i.e. counselling before a planned pregnancy), pre-implantation diagnosis and prenatal diagnosis, family planning counselling (e.g. contraception or planning the time of pregnancy also related to the disease activity), pregnancy monitoring, post-pregnancy monitoring, lactation counselling, and newborn management counselling. A preliminary draft of the survey was then discussed by the co-design group and a new, improved versions of the survey were circulated within the co-design group in order to implement their comments. The final version of the survey was then approved, uploaded to the EU-Survey platform [10] and tested by the co-design group.

The survey was composed of 53 closed and 25 open-ended questions (Supplementary Material, Appendix A, Fig. 1). For each domain, the following aspects were explored: the relevance attributed to the topic, the need

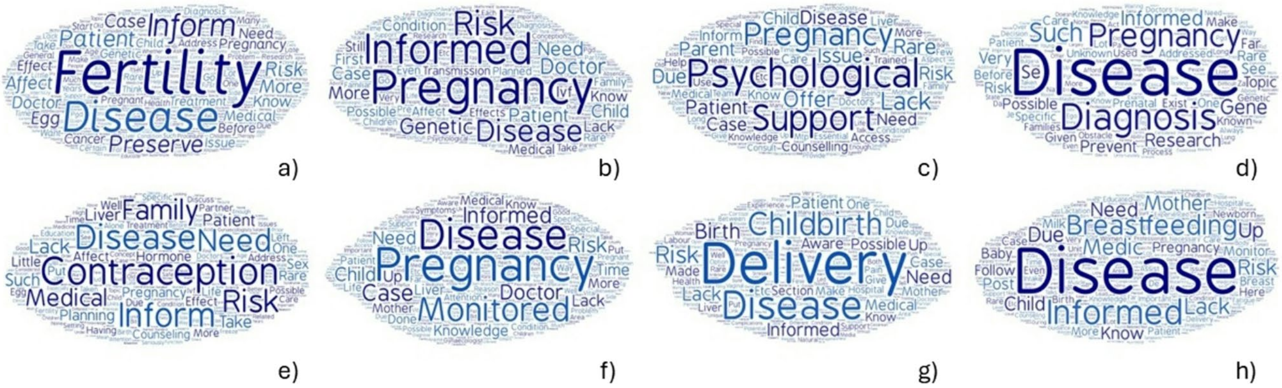

Fig. 1 Word clouds with the most frequently mentioned terms in response to unmet needs across various topics: **a** fertility preservation **b** pre-conceptional counselling **c** psychological support in the pre-conceptional counselling **d** pre-implantation diagnosis and prenatal diagnosis **e** family planning **f** pregnancy monitoring **g** pregnancy delivery **h** post pregnancy monitoring, lactation counselling and newborn management counselling. The size of each term in the cloud corresponds to its frequency in the survey responses

for education in that area, potential educational activities for patients and unmet needs. Regarding the assessment of unmet needs, the survey included a question specifically focused on delivery. Additionally, to capture a more comprehensive perspective of the postpartum period, post-pregnancy monitoring, lactation counselling, and newborn management counselling were combined into a single question. Finally, to gain a broader overview, the respondents were invited to share general comments about their overall pregnancy experience. The questionnaire was initially developed in English and subsequently translated into Bulgarian, Dutch, French, German, Italian, Polish, Portuguese, and Spanish to ensure wider accessibility.

The survey, in each of its translated languages, was made available through the EuSurvey Platform [10] and was promoted by various channels, including social media and email campaigns. The questionnaires were officially launched on 28th February 2022 and remained accessible to participants until 31st July 2022. Since the survey was completely anonymous and no personal information was collected, approval of the Institutional Board Review was not needed, and patients consented to participate in the study by replying to the survey.

After the data collection phase was concluded, the WG met virtually to discuss the results of a preliminary analysis and to agree on the most important findings. The open-ended questions about unmet needs were sent to volunteers for each language to translate the answers and identify the main issues. Finally, the closed-ended questions were analysed with a descriptive analysis and the open-ended questions with a qualitative analysis approach. In particular, the latter were analysed using the online software Wordart, which identified the most frequent words. The results for each domain were then represented in dedicated word clouds and tables of absolute frequency. In the survey results, for each topic that explored unmet needs, the most significant answer was reported, providing clear insights into the primary concerns and challenges.

Results

A total of 769 responses were collected, of which 574 were from patients, 155 were from caregivers, and 40 were from other people, mainly healthcare professionals (Supplementary Material, Appendix B, Fig. 1). These latter were excluded from the analysis, as the survey was not directly targeted towards them. The questionnaires were collected from nearly all the ERNs as detailed in Table 1. Among them, ERN ReCONNET was the most represented, accounting for 15.45%, followed by ERN RITA with 14.27%. Thirty-six respondents did not specify their associated ERN.

Table 1 ERN of the respondents. Note: in some cases, more than one ERN was selected in the same answer

Please select to which ERN your centre belongs to	*N*	%
ERN ReCONNET	144	15.45%
ERN RITA	133	14.27%
ERN RARE-LIVER	89	9.55%
ERNICA	69	7.40%
ERN GUARD-Heart	53	5.69%
Endo-ERN	52	5.58%
ERN LUNG	42	4.51%
ERN BOND	37	3.97%
I don't know	36	3.86%
MetabERN	32	3.43%
ERN EURO-NMD	31	3.33%
ERN-Skin	31	3.33%
ERN eUROGEN	25	2.68%
ERN-RND	23	2.47%
ERN CRANIO	20	2.15%
ERN ITHACA	18	1.93%
ERN VASCERN	18	1.93%
ERN-EYE	18	1.93%
ERN ERKNet	17	1.82%
ERN EURACAN	16	1.72%
ERN GENTURIS	11	1.18%
ERN PaedCan	7	0.75%
ERN EpiCARE	6	0.64%
ERN TRANSPLANT-CHILD	4	0.43%

With respect to the characteristics of the respondents, the majority were patients (75%), mainly female (90%) and most commonly aged between 31 and 40 years. The participants predominantly originated from 17 countries, with the highest representation from Germany (27%), France (23%), Spain (16%), and Italy (11%). Furthermore, approximately 13% of the respondents came from other countries that were not specifically listed in the survey. The characteristics of the respondents are detailed in Table 2.

In a broad overview, the survey results revealed that "Pre-conceptional counselling" and "Pregnancy monitoring" were the topics most frequently rated as "Very important/Important" by respondents, with 95.7% and 95.3%, respectively. Conversely, the domains of "Pregnancy Monitoring" and "Post pregnancy monitoring" were the only topics that respondents marked as "Not Important", even if with a small percentage (1.0% for both). Overall, the data indicate a consistently high level of importance attributed to all domains explored. Even topics often under-addressed in clinical settings, such as psychological support (91.0%) and lactation counselling (92.0%), were strongly valued by participants. A complete overview of the respondents' perceptions of the importance of the different topics of the survey is

Table 2 Sociodemographic characteristics of the respondents

	Patients (%) *n* = 574	Caregivers (%) *n* = 155	Patients and Caregivers (%) *n* = 729
N	74.6	20.2	94.8
Gender			
Male	8.4	15.5	9.9
Female	91.5	84.5	90.0
Other	0.2	0.0	0.1
Age			
18–20	0.9	0.6	0.8
21–30	16.6	7.1	14.5
31–40	38.2	27.7	35.9
41–50	23.2	37.4	26.2
51–60	13.6	18.7	14.7
61–70	5.6	7.1	5.9
71–80	2.1	1.3	1.9
Countries			
Belgium	3.7	6.5	4.3
Bulgaria	0.7	3.2	1.2
Croatia	0.0	1.3	0.3
Denmark	0.4	0.0	0.3
Finland	0.4	0.0	0.3
France	26.5	9.7	22.9
Germany	26.6	28.4	27.0
Greece	0.5	0.0	0.4
Ireland	1.1	1.3	1.1
Italy	8.8	17.4	10.7
Latvia	0.0	0.6	0.1
Lithuania	0.5	0.6	0.6
Malta	0.2	0.0	0.1
Poland	0.2	0.6	0.3
Portugal	0.9	1.3	1.0
Slovenia	0.2	0.0	0.1
Spain	16.6	15.5	16.3
Other country	12.9	13.5	13.0

summarized in Table 3. This table outlines the distribution of responses categorizing each domain as "Very important/Important", "Moderately Important/Slightly Important" and "Not Important".

The unmet needs are visually represented through word clouds (Fig. 1a-h) in which the size of each term corresponds to its frequency in the responses. For a more detailed quantitative analysis, the frequency of these terms is tabulated in the Supplementary Material, Appendix B, Tables 1, 2, 3, 4, 5, 6, 7 and 8. Upon analysing all the responses, the most recurrent words that emerged were "inform," "need" "risk" "support" and "psychological".

Pre-pregnancy

> *"I see an urgent need for action in the timely information of patients".*

Fertility preservation emerged as a significant concern and was deemed "very important/important" by 87.3% of respondents, with a slightly greater emphasis among patients (88.6%) than among caregivers (82.4%). The predominant issues identified include the lack of information from experts regarding reduced fertility due to medication (such as chemotherapy) or the disease itself, fertility preservation options not being covered by health insurance in some countries and the absence of adequate alternatives such as surrogacy. These concerns are echoed in the recurrent use of the words "Inform" and "Preserve".

> *"My gynaecologist is not familiar with the condition and does not see any complications".*

Pre-conceptional counselling, defined as counselling before pregnancy, was similarly regarded as very important/important by both patients and caregivers (95.7%), with a smaller portion (4.3%) considering it moderately/slightly important. The most common problems described were the limited awareness of pregnancy risks among specialists as well as the lack of information on the relationship between disease/pregnancy and hereditary risks. These aspects are emphasized by terms such as "Informed", "Risk" and "Genetic".

> *"Psychological help is unheard of".*

Psychological support in the context of pre-conceptional counselling was seen as crucial by 91% of participants. The challenges highlighted include the need for reassurance by specialists, insufficient emotional support in multidisciplinary teams and a lack of systematic organization in providing this support. These needs are represented by the words "Support" and "Lack".

> *"It is a real obstacle course to have access to this type of care and the waiting period is very very long".*

Pre-implantation and prenatal diagnosis were considered "very important/important" by 94% of the respondents. The principal unmet need in this domain was the unavailability and long waiting times for the diagnostic tests, a frustration captured by the frequent mention of the word "Diagnosis".

> *"Lack of standard of care for contraception under immunosuppressive medication. Patients are left alone".*

Family planning, encompassing contraception and timing pregnancy in relation to disease activity, was rated as "very important/important" by 94.5% of the respondents.

Table 3 Rate of relevance of the explored topics

Rate of relevance (% of respondents)

Topic	**Very important/Important**			**Moderately Important/Slightly Important**			**Not Important**		
	Patients	Caregivers	Patients and Caregivers	Patients	Caregivers	Patients and Caregivers	Patients	Caregivers	Patients and Caregivers
Fertility Preservation	88.6	82.4	87.3	11.4	17.6	12.7	0.0	0.0	0.0
Pre-conceptional counselling	95.7	95.7	95.7	4.3	4.3	4.3	0.0	0.0	0.0
Psychological support in the pre-conceptional counselling	90.7	92.0	91.0	9.3	8.0	9.0	0.0	0.0	0.0
Pre-implantation diagnosis and Prenatal diagnosis	93.3	95.9	94.0	6.7	4.1	6.0	0.0	0.0	0.0
Family Planning counselling	94.3	95.5	94.5	5.7	4.5	5.5	0,0	0,0	0,0
Pregnancy Monitoring	96.4	90.8	95.3	2.7	7.7	3.7	0.9	1.4	1.0
Post pregnancy monitoring	93.6	92.4	93.3	5.4	6.8	5.7	1.0	0.8	1,0
Lactation counselling	91.9	92.5	92.0	8.1	7.5	8.0	0.0	0.0	0.0
Newborn management counselling	92.5	97.5	93.8	7.5	2.5	6.2	0.0	0.0	0.0

The most significant necessity was related to finding suitable contraception methods that are compatible with the disease, as indicated by the recurring terms "Contraception" and "Disease".

Pregnancy

> *"Good and frequent cooperation between different doctors is important".*

Pregnancy monitoring, involving checkups and surveillance, was viewed as "very important/important" by 92.3% of the survey participants. Notably, this topic was more significant for patients than for caregivers, (96.4% versus 90.8%, respectively). Common issues identified from the open-ended question revolved around a significant lack of information and a strong need for comprehensive follow-up care involving a multidisciplinary team. This team should ideally include trained specialists, gynaecologists, obstetricians, midwives and professionals providing psychological support. Key terms such as "Informed" and "Monitored" were frequently used, highlighting these concerns.

> *"Humanize and naturalize pregnancy and childbirth".*

Focusing on delivery, the prominent issue identified was planning the appropriate delivery (vaginal delivery, caesarean section, induced labour, etc.) to minimize the risks associated with rare diseases. This perspective was underlined by the frequent use of the words "Childbirth" and "Risk".

After pregnancy

> *"Education and postpartum counselling, including breastfeeding and infant care, are crucial regardless of the patient's medical condition".*

Post-pregnancy monitoring, which refers to surveillance of the days and weeks after delivery, was rated as highly important by 93.3% of respondents and not important by only 1%. Lactation counselling was seen as "very important/important" by 92% of the participants. Newborn management counselling, which focuses on monitoring and handling the newborn, was regarded as "very important/important" by 93.8% of the respondents, with caregivers paying slightly more attention (97.5%) than patients did (92.5%). The most stressed elements were the need for information on managing the post-pregnancy period and guidance on breastfeeding, especially regarding potential contraindications related to medications. The prevalence of the words "Informed" and "Breastfeeding" underscored these challenges.

Further comments and educational activities

> *"If sometimes the mother receives some concern, the father is totally forgotten! To raise a child you must be two parents and in case of a disability, you must be strong together".*

Table 4 Preferred educational activities of the respondents in all the domains. The results are outlined in a range format and show the spectrum of minimum to maximum preferences expressed across various topics

Favourite kind of educational activities	Patients (%)	Caregivers (%)	Patients and caregivers (%)
Leaflets/brochure and educational materials	19.97–24.40%	18.92–23.35%	19.78–23.96%
Specific face to face education sessions	18.89–30.29%	18.23–29.61%	18.75–30.1%
Online educational material	12.88–15.05%	15.72–18.48%	13.48–15.67%
Link to useful website	19.04–21.86%	15.12–17.87%	18.60–20.82%
Contact to patient organisation(s) or patient forum	13.86–20.16%	16.52–25.78%	14.61–21.36%
Other	1.50–2.77%	0.90–2.11%	1.39–2.57%

The general comments rested on the essential requirements for both informational and emotional care throughout all stages of pregnancy. This support should extend beyond the expectant mother, encompassing the co-parent, the entire family, and caregivers as well. This recurrent need is expressed by the terms "Informed," "Support" and "Psychological" (Supplementary Material, Appendix B, Fig. 2; Table 9).

Regarding the preferred methods of educational activities, patients and caregivers expressed a preference for face-to-face education sessions, (18.75–30.1%) and, secondarily, for educational materials such as leaflets (Table 4). As an alternative, online platforms, including websites and online educational material, along with connections to patient organizations, were suggested as effective secondary means for providing education and information.

Discussion

This study aimed to explore the experiences of pregnancy and other aspects of reproductive life among patients with rare and low prevalence diseases and their caregivers and family members, uncovering the unmet needs of the community.

The respondents represented over 17 countries across Europe, with the highest participation from Germany, France, Spain, and Italy. Additionally, the majority of respondents were within the age range of 30 to 50 years, representing the demographic characteristics most likely to be actively considered or experiencing pregnancy.

The ERN questionnaire for patients and caregivers revealed several key issues, particularly those concerning the psychological sphere, the need for information and health care supported by a multidisciplinary team.

Key findings and interpretation

A notable finding was the substantial requirement for psychological support, highlighting the emotional and mental health challenges accompanying pregnancy in these conditions. This underscores the fact that for these patients, pregnancy transcends physical aspects, encompassing significant psychological dimensions [11].

The lack of access to information was another major concern. This gap in knowledge reveals the necessity for more accessible, comprehensive information about the implications of rare diseases on pregnancy and vice versa. Similar gaps in the provision of preconception care have been documented in both general and rare disease populations, suggesting a broader systemic issue in reproductive health education [12–15]. To address insufficient awareness, primary methods should be include face-to-face education sessions and the distribution of informational leaflets offering direct interaction and tangible resources. Second, leveraging online sources and establishing contact with patient organizations provide additional, accessible channels for information and support. Interestingly, the survey revealed that patients, despite being predominantly in the 30–50 age range, expressed a clear preference for receiving information directly from Health Care Professionals and via paper leaflets rather than from online sources.

Psychological support and access to information are extremely important, not only for patients but also for their families and caregivers. It is vital to recognize the emotional and informational requirements of this wider support network, as they play a fundamental role in the care and overall well-being of patients affected by rare diseases.

The study revealed issues with the unavailability or inaccessibility of essential diagnostic tests and medical counselling, particularly in critical areas such as fertility preservation, pre-implantation, prenatal diagnosis, and contraception. Difficulties in accessing contraception can result in poor contraceptive coverage, increasing the risk of unplanned pregnancies, which can have significant health implications for both the mother and fetus. These challenges highlight notable gaps in healthcare provision for female patients with rare diseases [16]. Although our survey focused on adults, the importance of early reproductive counselling during adolescence should not be overlooked. Pediatricians can play a key role in engaging directly with young people over 15, alongside parental advocacy, to support informed decision-making and smoother transition to adult care [17].

Relevance for healthcare policy and practice

A trained multidisciplinary team, including specialists in rare diseases, gynecologists, obstetricians, midwives and mental health professionals, is essential to provide comprehensive care to patients with rare diseases during pregnancy. This team approach ensures that all the aspects of the patient's health, previously discussed, are addressed effectively [18–20]. Easy access to multidisciplinary teams can be ensured through primary care providers as local points of contact for referrals. Studies in various healthcare settings have highlighted similar structural gaps and call for the integration of preconception and reproductive counselling into general and primary care systems [21, 22]. A shared care model across centres, aligned with the ERN perspective, could enhance accessibility by bringing specialized care closer to patients, reducing travel burdens, and ensuring high-quality care.

The qualitative analysis provided additional information by offering insights into the patients' experiences. Owing to this approach, the personal dimension of the patients was captured, enriching the understanding of their necessities beyond standard quantitative data [7]. Key terms such as "information" "support" and "risk" were frequently mentioned underscoring unmet needs otherwise not obtainable from closed-ended questions.

Despite the diversity of rare diseases, countries, and ERNs represented in the survey, the responses revealed common issues across these groups. This finding suggests that many challenges during pregnancy are universal among patients with rare diseases, irrespective of their specific condition or geographic location. Owing to these findings, the unmet needs identified can be addressed in the rare and low-prevalence community through the ERN Transversal Working Group, which can develop common actions aimed at improving the aspects identified in the survey with dedicated activities, such as the development of educational material to be adopted across Europe. Moreover, owing to the ERN Transversal Working Group, each ERN Representative can discuss the results collected from their own specific disease areas and develop disease-specific initiatives within their respective ERN, providing an additional contribution to the improvement of family planning management in their disease area across the EU.

However, significant differences exist among European countries, particularly in terms of legislation. Previous analyses of rare disease policies confirm wide variability in care models, financing, and strategic priorities across Europe, which continues to affect access to services and patient engagement [3, 23]. For example, fertility preservation is regulated by diverse policies, resulting in disparities in access. These variations pose a challenge to developing harmonized guidelines but also serve as a call to action for lawmakers to ensure equitable access to pregnancy and fertility management across all European countries.

Strengths and limitations

The survey was distributed online via different social media platforms, as well as to different patient organisations, thanks to the support of the ERNs and of the respective patient organisations. Due to the online distribution, some bias related to selection, self-selection and response bias might have been encountered. To reduce these biases, mitigation strategies were implemented, such as diversifying the channels of online distribution, increasing accessibility with the availability of multiple language translations, and providing clear instructions and context for participation to the respondents.

Responses were unevenly distributed, with an over-representation of some ERNs; this may reflect differences in care quality, awareness, or access to information across networks. Additionally, the diversity of diseases and healthcare systems may have masked more specific needs. Despite these limitations, the large, multilingual sample from 20 ERNs ensured broad representation allowing the study to capture common challenges experienced across the rare disease community. The mixed-methods approach and co-design with patient groups enhanced the relevance and depth of findings. The over-representation itself may signal where awareness and engagement are higher-offering a model to strengthen participation in less represented networks.

Conclusions

In conclusion, this study suggests that family planning for people living with rare and low-prevalence diseases should be a journey where their needs for psychological support, comprehensive information, access to care and a multidisciplinary team are adequately met towards a patient-centred approach. As the first transversal study spanning various rare and complex diseases, the common insights gained could be pivotal in developing a unified model for managing pregnancy and family planning in patients living with rare and low prevalence complex diseases, These insights may serve to inform the development of targeted policies and educational programs, contributing to the standardization and improvement of care pathways across Europe.

In this context, ERNs can act as catalysts for implementing structured support models, sharing best practices, and promoting equitable access.

Abbreviations

ERN	European Reference Network
ERN ReCONNET	ERN on Rare and Complex Connective Tissue and Musculoskeletal Diseases
ERN BOND	ERN on Bone Disorders
ERN CRANIO	ERN on Craniofacial Anomalies and Ear, Nose and Throat Disorders
Endo-ERN	ERN on Endocrine Conditions
EpiCARE	ERN on Epilepsies
ERNICA	ERN on Inherited and Congenital Anomalies
ERN LUNG	ERN on Respiratory Diseases
ERN Skin	ERN on Skin Disorders
EURACAN	ERN on Adult Cancers
ERN eUROGEN	ERN on Urogenital Diseases and Conditions
ERN EURO-NMD	ERN on Neuromuscular Diseases
ERN EYE	ERN on Eye Diseases
ERN GENTURIS	ERN on Genetic Tumour Risk Syndromes
ERN GUARD-Heart	ERN on Diseases of the Heart
ERN ITHACA	ERN on Congenital Malformations and Rare Intellectual Disability
ERN PaedCan	ERN on Paediatric Cancer
ERN RARE-LIVER	ERN on Hepatological Diseases
ERN RITA	ERN on Immunodeficiency, Autoinflammatory and Autoimmune Diseases
ERN TRANSPLANT-CHILD	ERN on Transplantation in Children
VASCERN	ERN on Rare Multisystemic Vascular Diseases
WG	Working Group
HCP	Health Care Professional
EU	European Union

Supplementary Information

The online version contains supplementary material available at https://doi.org/10.1186/s12978-025-02136-5.

Supplementary Material 1

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Authors' contributions

GF and DM contributed to the conception and design of the study, to the acquisition and interpretation of data, and were responsible for writing the manuscript. SA, AB, RB, IB, MCB, PB, MLB, EC, JEF, MF, VG, VI, ELA, ARL, SL, CN, MO, MOn, WP, BP, CR, CRJ, DS, AT, CVDV, AV, DD, JDB, CDDS, ADu, CF, GL, BL, EK, MJP, JR, IRC, RR, HS, HJVdP, TY participated in the execution of the study and data collection. CT and DZ were responsible for the analysis of the data and contributed to the drafting of the manuscript. RT contributed to all aspects of the study design. MM contributed to the conception and design of the study and to the acquisition of data. In addition, MM was also responsible for the interpretation of data and contributed to the drafting of the manuscript. All authors were responsible for drafting the article and all the authors have read and approved the final manuscript.

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Data availability

DISCLAIMER The content of this publication represents the views of the authors only and it is their sole responsibility; it cannot be considered to reflect the views of the EC and/or the Consumers, Health, Agriculture and Food Executive Agency (CHAFEA) or any other body of the European Union. The EC and CHAFEA do not accept any responsibility for use that may be made of the information it contains.

Declarations

Ethics approval and consent to participate

An ethics committee approval was not needed since the survey was completely anonymous and completed by clinicians, and patients were not recruited. The participant's consent to the study was obtained by replying to the survey.

Consent for publication

Not applicable.

Competing interests

The authors declare that they have no competing interests.

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