EMRaDi
EUREGIO MEUSE-RHINE
— RARE DISEASES

REPORT OF WORK PACKAGE 1

WP1.1 REPORT ON THE DEMANDS AND NEEDS OF PATIENTS

Lead partner of Work Package	**Maastricht University**
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AUTHORS' LIST

Name/Surname	Institution name	Contact e-mail
Leading Author(s)		
Rok Hrzic	Maastricht University	r.hrzic@maastrichtuniversity.nl
Co-Author(s)		
Timo Clemens	Maastricht University	timo.clemens@maastrichtuniversity.nl
Peter Schröder-Bäck	Maastricht University	peter.schroder@maastrichtuniversity.nl
Helmut Brand	Maastricht University	helmut.brand@maastrichtuniversity.nl

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The Interreg V-A Euregio Meuse-Rhine (EMR) programme invests almost EUR 100 million in the development of the Interreg-region until 2020. This area stretches out from Leuven in the west to the borders of Cologne in the east, and runs from Eindhoven in the north all the way down to the border of Luxemburg. Over 5.5 million people live in this cross-border region, where the best of three countries merges into a truly European culture.

With the investment of EU funds in Interreg projects, the European Union directly invests in the economic development, innovation, territorial development and social inclusion and education of this region.

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PROJECT DESCRIPTION

"EMRaDi" stands for **Euregio Meuse-Rhine Rare Diseases**.

This project involves a **cross-border cooperation** between health insurers, university hospitals, patient associations and a university in the Euregio Meuse-Rhine. It is part of the European Union INTERREG V-A Euregio Meuse-Rhine programme.

Thanks to their long experience in cross-border healthcare, the project partners have decided to join forces in the specific field of rare diseases. This EMRaDi project is innovative in the sense that it is a patient-oriented and cross-sectoral project. The consortium of partners includes the major health players who support rare disease patients and their relatives in their day-to-day rare disease patient pathway.

Through **the project activities**, the EMRaDi project aims to:

- increase the transparency of needs and availability of services in the field of rare diseases in the Euregio Meuse-Rhine (EMR);
- develop EMR models of *rare disease patient pathways* in order to draw up patient-oriented recommendations in synergy with national and European developments, and;
- improve the network of health care providers, health insurance providers and patient organisations, and raise (public) awareness of rare diseases.

The general long-term aim is to **improve the quality of life of these patients**.

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LEGAL ISSUES

The methodology utilised in this report does not pose a threat to individual health or privacy, therefore a formal bioethical review was not deemed necessary under Dutch law (WMO).

EXECUTIVE SUMMARY

Rare diseases are an important source of disease burden for health systems in the European Union and worldwide. Despite representing disparate pathological processes that require highly individualised approaches to diagnosis, treatment and support, there are important common features of patients' experience with rare diseases, especially in contrast to more common diseases. A key question is whether there are common patient needs across the spectrum of rare diseases, and whether main domains of such needs could be identified. To accomplish this task, a systematic literature search and thematic analysis were performed as part of the Euregio Meuse-Rhine Rare Diseases project. The literature search identified 37 records of relevance, including studies, commentaries, and patient organisation reports. The thematic analysis uncovered three key domains of rare disease patients' needs: (1) diagnosis, (2) information and understanding, (3) medical care and psychosocial support. To our knowledge, this study is the first attempt at a rigorous synthesis of the literature on the needs of rare disease patients as a whole, and provides an empirical framework for future research into patient satisfaction and development of health and social services aimed at supporting rare disease patients.

1 INTRODUCTION

In the European Union, rare diseases are defined as illnesses with a prevalence of 5 in 10,000 or lower, which includes approximately 5-8,000 different diseases that affect 6-8% of the population [1]. The Euregio Meuse-Rhine Rare Diseases (EMRaDi) project was initiated in 2016 and includes partners representing key stakeholders (hospitals, insurance companies, patient organisations, and research institutions) from the cross-border region (EMR), which includes territories in Belgium, Germany, and the Netherlands. The project aims to improve our understanding of the interactions between rare disease patients and health services and explore the impact of cross-border care in this context, with the ultimate goal of improving the provision of care patients need in terms of access, quality, and efficiency [2]. Work package 1 of EMRaDi focuses on understanding the needs of rare disease patients on the one hand, and their actual healthcare utilisation on the other.

Because a comprehensive cataloguing of needs of patients with rare diseases could aid future research by identifying critical domains of need and related gaps in the current provision of services, to support the stakeholders in the EMR in providing more value to rare disease patients, and to support subsequent activities in the EMRaDi project, a thematic analysis of literature has been undertaken. The aim was to identify key dimensions of health-related needs that are as particular to rare disease patients as possible, with the purpose of providing an empirical framework to aid future evaluation and development of health services for rare diseases in the region and elsewhere.

2 METHODS

2.1 PAPER SELECTION

An electronic search was performed in the CINAHL (EBSCO), PsychINFO (EBSCO), EMBASE (OVID) and MEDLINE (OVID) databases using the following keyword strategy:

1. rare diseases (and its synonyms in Mesh)
2. patient (as the stakeholder of interest)
3. need* OR experience* OR challenge* OR burden*
4. #1 AND (#2 ADJ6 #3)

The value of the adjacency operator was determined empirically, with a view of including documents that mention the second and third search terms in the same sentence.

Search results were limited to the English language, while no temporal limitation was used. The database search was performed on 23 August 2017.

The titles and abstracts of the identified papers were screened using the following key inclusion criterion: any study, report, or other document that includes a description of rare disease patient needs from the perspective of the patients themselves. Studies and other documents that exclusively

focused on a description of the rare disease itself, its epidemiology, pathophysiology, clinical manifestations, treatment options, costs and economic aspects of treatment were excluded. Also excluded were documents that focused solely on health policy or care-organizational issues related to rare diseases without reporting on how they relate to the explicit needs of rare disease patients. The full text of records of interest was reviewed and a final selection made using the same criteria.

2.2 DATA ANALYSIS

Due to the identified literature sources encompassing a very heterogeneous set of research questions and methods, the data analysis followed the procedure of thematic analysis laid out by Thomas and Harden [3]. After identifying the articles of interest, their full text was coded, looking for statements related to expressed patient needs, negative experiences possibly amenable to health system interventions, and similar topics. The codes were generated inductively, evolving as more of the literature was processed, leading to the second step of an identification of wider areas of need or experiences expressed by the patients (descriptive themes) [Figure 2]. In the last step, these wider areas were interpreted as belonging to domains of rare disease patients' needs (analytical themes). These domains are listed and discussed in more detail in the Results section.

3 RESULTS

The search identified 198 records, with a further 54 potentially interesting records identified through a hand-search of references. The relatively high number of references sourced by hand-searching was the result of the literature not consistently using the word "patient" when it referred to specific categories of patients (e.g. mothers, families, etc.), not consistently using the phrase "rare disease" when detailing information on specific rare diseases, and due to the substantial amount of grey literature that exists in the field. Finally, 37 documents were included in the final selection and are used to craft the thematic analysis [*Figure 1*].

3.1 CHARACTERISTICS OF INCLUDED STUDIES

All but one document was published after the year 2000, with more than half published since 2010. A diverse geographic representation was achieved, with about half of the included records stemming from individual European countries or representing the European Union as a whole, and the rest originating from the United States of America, Australia or elsewhere. Not surprisingly, the methods most often employed were qualitative in nature, either interviews with rare disease patients or their carers, focus groups, or narrative or content analyses. Surveys, however, were utilized in almost all studies that involved wider geographic regions. Lastly, a wide range of diseases was included, with a little over half the records studying more than one rare disease. For a more detailed representation of included records, see Table 1.

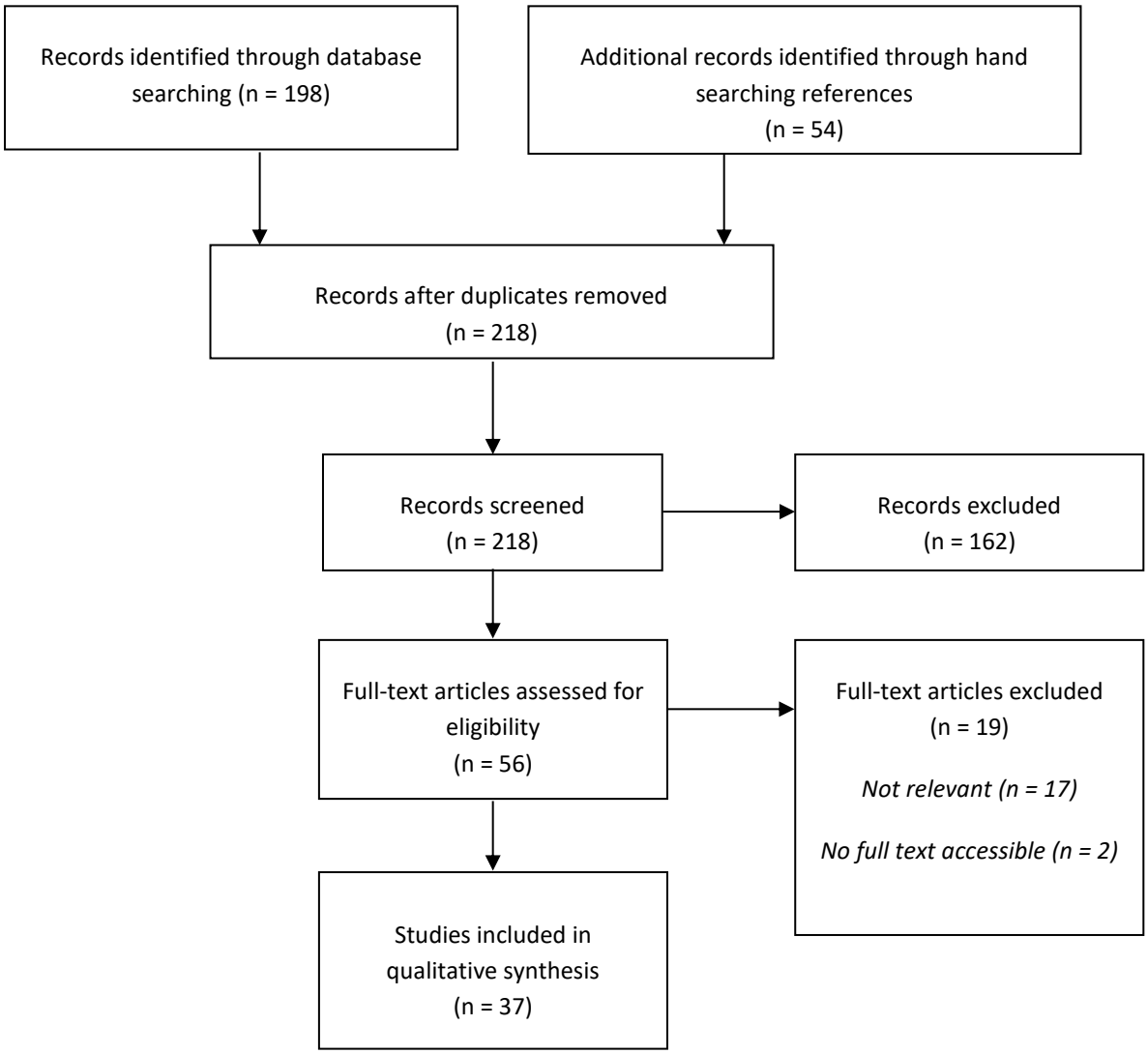

Figure 1 PRISMA diagram of the flow of identified records

3.2 RARE DISEASE PATIENT NEEDS

Figure 2 summarizes the process of experiences specific to rare disease patients generating specific needs, their characteristics and key stakeholders that could be targeted by a service. The needs have been categorized into three dimensions: (1) diagnosis, (2) information and understanding, (3) care and support, with the last consisting of physical, psychological, and social aspects.

3.2.1 DIAGNOSIS

The most salient feature of rare diseases according to the literature surveyed seems to be the challenging period in a patient's life between the onset of symptoms and their medical explanation in the form of a diagnosis (if that ever indeed occurs). Patients are exposed to a host of difficulties during this period, including a minimization of their health issues or their denial [4-7], intense psychological distress including feelings confusion, fear and frustration [5, 7, 8], and frequent failures at diagnosis or misdiagnoses [5-7, 9-12]. This *"odyssey",* which often takes several years and numerous trips to various medical specialists [5, 6, 9-19], also leads to a delay in treatment [6, 14, 20, 21], with often permanent damaging consequences for the patient's wellbeing [6, 9, 11, 20, 21].

A French study found that the patients did not mind the delay as much, as long as they were taken seriously, treated with consideration, and the physicians were transparent about their limitations [18]. Other studies concurred regarding the importance of an adequate and respectful approach to disclosing the diagnosis [13, 28].

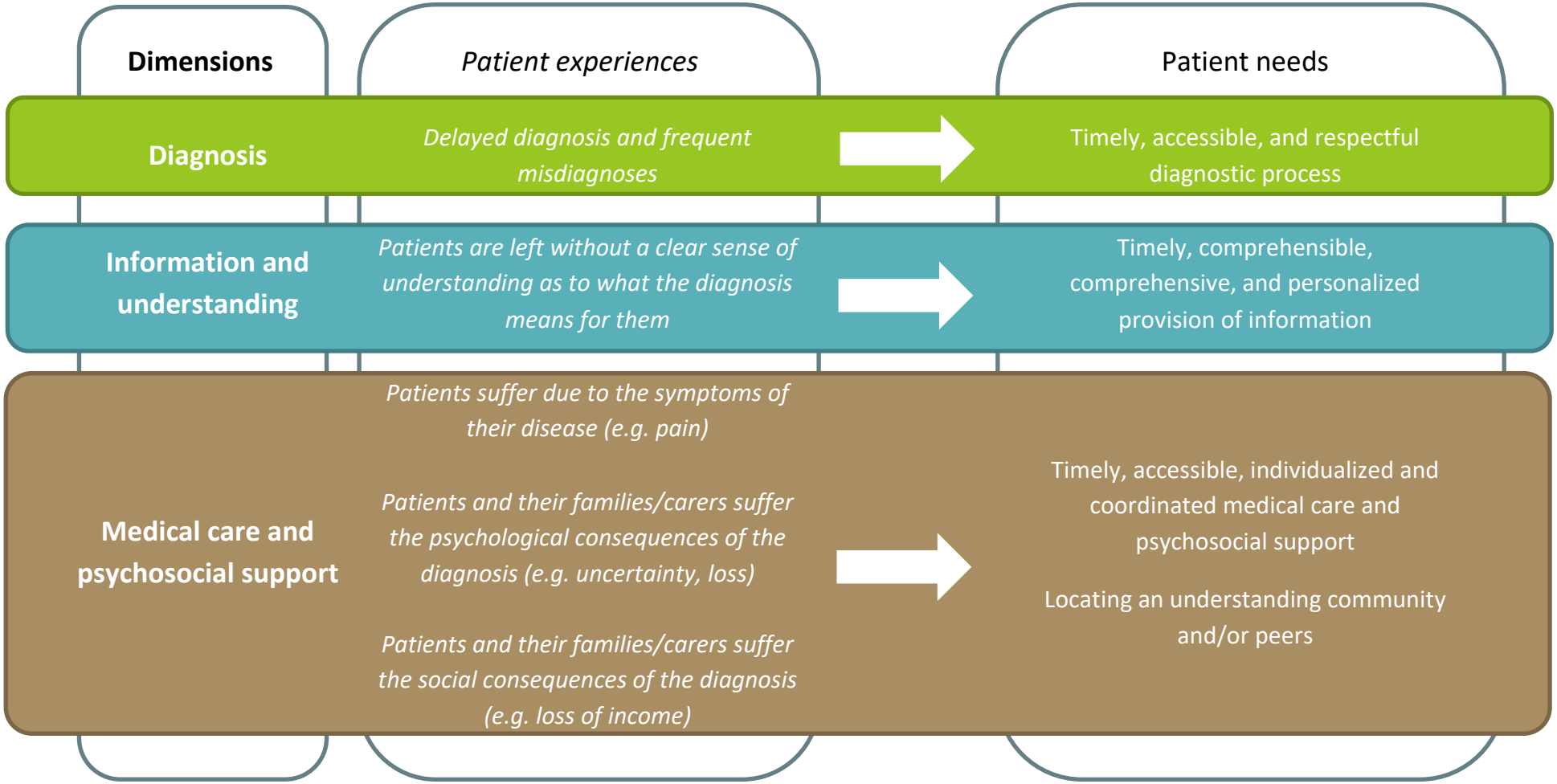

Figure 2 Dimensions of needs of rare disease patients as a process

Table 1. Characteristics of included studies

Study	Country	Methodology	Diseases included	Needs listed / themes
Christensen et al. [5]	USA	Interviews	Hermansky–Pudlak Syndrome	Diagnosis, expert patient, community
Garau [17]	UK	First person narrative	Leiomyosarcoma	Diagnosis, access to information, access to treatment
Garrino et al. [8]	Italy	Interviews	Various	Diagnosis, access to treatment, access to information, support with everyday activities, inability to work, community, independence
Hutchinson [22]	USA	Narrative review	Hairy cell leukemia	Community, psychosocial support, expert patient, access to information, access to treatment
Kesselheim et al. [6]	USA	Focus groups	Various	Diagnosis, access to treatment, access to information, expert-patient, financial support, psychosocial support
Wagland et al. [23]	Australia	Interviews	Multiple myeloma	Isolation – access to information, community
Walker [24]	Online / USA	Content analysis	Rare vascular diseases	Access to information, community, access to treatment
Caputo [25]	Italy	Content analysis	Various	Access to treatment, support with everyday activities, psychosocial support
Walker [26]	Online / USA	Content analysis	Thoracic outlet syndrome	Community, access to information
Znidar et al. [27]	EU	Survey	Gaucher disease	Access to treatment, community, access to information, psychosocial support
Anderson et al. [13]	Australia	Survey	Various	Diagnosis, psychosocial support, financial support, access to treatment
Grut & Kvam [28]	Norway	Interviews	Various	Expert patient, lack of expertise, diagnosis, access to information, access to treatment
Budych et al. [29]	Germany	Interviews	Various	Expert patient, lack of expertise, access to information
Weng et al. [14]	Taiwan	Interviews	Russel-Silver syndrome	Psychosocial support, diagnosis, lack of expertise, access to information, financial support
Gahl & Tifft [7]	UK	Commentary	Various	Diagnosis, organization of care, psychosocial support
Gunderssen [30]	Norway	Interviews	Various	Access to information, lack of expertise, diagnosis, community
Kelly & Dowling [31]	Ireland	Interviews	Multiple myeloma	Access to information, community, psychosocial support
Molassiotis et al. [32]	UK	Interviews	Multiple myeloma	Independence, access to information, psychosocial support

Molassiotis et al. [33]	UK	Survey	Multiple myeloma	Financial support, psychosocial support, access to information,
Berglund et al. [4]	Sweden	Survey/narrative	Ehlers–Danlos syndrome	Respect/dignity, expert patient, lack of expertise
Jaffe et al. [15]	Australia	Commentary	Various	Diagnosis, lack of expertise, access to treatment, psychosocial support
Kole & Faurisson [9]	EU	Survey	Various	Lack of expertise, diagnosis, access to treatment, access to information, psychosocial support, financial support
Limb et al. [10]	UK	Survey	Various	Access to research, access to information, diagnosis, lack of expertise, access to treatment, psychosocial support, organization of care, financial support
Packman et al. [34]	USA	Interviews	Gaucher disease	Financial support, psychosocial support, diagnosis, access to information, lack of expertise, access to treatment, coordination of care
Henderson et al. [35]	USA	Interviews	Niemann-Pick disease, type B	Psychosocial support, community, access to information, access to treatment
Huyard [18]	France	Interviews	Various	Diagnosis, respect/dignity, access to information, access to treatment, community
Kole & Faurisson [11]	EU	Survey	Various	Diagnosis, access to treatment, lack of expertise, psychosocial support, expert-patient, financial support, respect/dignity, access to information, coordination of care, access to research
Schieppati et al. [16]	EU	Commentary	Various	Diagnosis, financial support, access to care, coordination of care, social support
Witham et al. [36]	UK	Interviews	Pseudomyxoma peritonei	Lack of expertise, diagnosis, access to care, access to information, expert-patient, coordination of care, psychosocial support, independence, community
Zurynski et al. [20]	Australia	Literature review	Various	Access to care, community, psychosocial support, diagnosis, financial support, lack of expertise
Coulson et al. [37]	UK/Internet	Content analysis	Huntington's disease	Access to information, community, psychosocial support
Griffiths et al. [38]	UK	Interviews	Rare cancers	Psychosocial support, access to care, coordination of care
McAllister et al. [39]	UK	Focus groups	Various	Diagnosis, coordination of care, access to care, lack of expertise
Dyke & Leonard [21]	Australia	Survey	Rett syndrome	Financial support, diagnosis, respect/dignity, access to information, lack of expertise, coordination of care, psychosocial support
Petersen [40]	UK	Interviews	Various	Diagnosis, independence, access to information, expert-patient, community
EURORDIS [12]	EU	Survey	Various	Diagnosis, access to information, access to care, psychosocial and financial support

Groft [19]	USA	Commentary	Various	Diagnosis, financial support, access to care, access to information, psychosocial support, access to research

Abbreviations used: EU, European Union, UK; United Kingdom of Great Britain and Northern Ireland; USA, United States of America.

3.2.2 INFORMATION AND UNDERSTANDING

A second key need of rare disease patients was the need for understanding the diagnosis they received. In numerous studies, patients expressed an expectation that the disclosure of their diagnosis will be accompanied with a timely, thorough, and accessible explanation of what this diagnosis means for their health, their lives, and with clear information on where to turn to for treatment and other support [6, 10, 12, 17-21, 35, 36, 40]. The studies also report that this need for information is most commonly an unmet need, as the physicians either lack the expertise to provide a complete explanation [6, 7, 9, 11, 14, 20, 28, 36], which in turn was believed to be driven by a general lack of understanding of these diseases [9, 11, 19, 20, 36].

Patients therefore often turn to alternative sources of information to educate themselves. such as the internet or fellow patients in patient organisations. Especially in the case of the former, there is a rich body of research on online groups of rare disease patients and the themes most commonly discussed. Notably, these studies concurred that unlike online groups of more common diseases, rare disease groups focus more on information exchange relative to emotional support [24-26, 30, 37]. Similarly, a key function of patient organisations was identified to be information dissemination [27].

This self-education process often leads to the patients developing a substantial degree of expertise related to their disease, often leading to them knowing more than most healthcare professionals they meet. This phenomenon of an expert patient leads to further challenges to the patient, as this role reversal is uncommon (and uncomfortable) for both patients and healthcare professionals [28, 29], and places an additional burden on the patient, where they must now educate and constantly advocate for themselves with little support [5, 6, 11, 40]. It was noted by the patients that in the absence of their expertise, they believed they would not have reached the diagnosis or accessed the appropriate treatment their disease required [5, 10, 11, 40].

3.2.3 MEDICAL CARE AND PSYCHOSOCIAL SUPPORT

3.2.3.1 PHYSICAL NEEDS AND ACCESS TO SAFE MEDICAL CARE

In terms of accessing care and support for their physical needs, patients and their carers face numerous obstacles in accessing good quality care for their rare disease. First, the patients' lack of information on the treatment has been described in the section above and is not discussed further here. Second, there is a lack of existence of a recognised safe treatment approach. While this is a seemingly straight forward and unfortunately common obstacle to effective rare disease care [9], the literature allows one a more nuanced view of this obstacle. The relative lack of scientific attention to the study of rare diseases and the development of pharmaceuticals for their treatment is commonly thought to be a key cause of this obstacle and is often pointed out as political and organisational challenges [9-11, 15, 16, 19, 20, 27]. These, however, are considered outside the scope of this review and will not be discussed further. Perhaps surprisingly, patients in one study were not particularly concerned with the lack of a cure, but rather demanded that society and healthcare professionals make an earnest effort to improve their lives [18].

Another scenario described is the availability of experimental treatments or off-label drug use. The availability of the former was often in the context of a clinical trial, where access was highly controlled and limited by the patients' awareness of the study itself, as well as plagued with safety concerns and the issue of access after the trial concludes [6, 10]. Off-label drug use was associated with its own concerns, mainly the lack of knowledge by the attending physician and financial concerns of patients, related to their insurance coverage (or lack thereof) [6, 9-11, 19, 27].

The high cost of treatment was associated in the literature with concerns of insurance coverage and availability of medicines, especially in health systems with lower resource availability [6, 9, 11, 19, 27]. Even in health systems usually considered as providing universal health coverage, rare disease patients experienced substantial out of pocket expenses related to their care that placed them in financial jeopardy [9-11, 13, 20, 21, 27].

3.2.3.2 PSYCHOLOGICAL AND SOCIAL SUPPORT NEEDS

While the physical needs of rare disease patients might seem obvious and represent a key need, the literature identifies substantial unmet needs for psychological and social support in their pursuit of the highest attainable independence and sense of normalcy [5, 8-11, 13, 14, 23, 25, 30-36, 38, 39]. This is especially true at key stages of the disease, such as in the moment of diagnosis [10, 13, 21], reaching adulthood and being transferred to adult services [10], among others. Currently, due to a paucity of professional support in meeting psychosocial needs of rare disease patients, most of the supportive burden is carried by the patients' families and carers, who in turn respond by developing supportive needs of their own [5, 6, 8, 13, 14, 17, 21, 30-33].

In terms of psychological needs, a key fixture in the literature is the profound sense of isolation that patients often feel, since their experiences are unique and they rarely meet someone with the same diagnosis [17, 23, 32, 35]. Patients report that they battle this feeling by developing a strong sense of community, either within their family and circle of friends, within the team of healthcare professionals that treat them, or within a patient group (on- or off-line) [5, 10, 11, 15, 20, 22, 26, 27, 35, 37]. Belonging to a community seems to play a key role in fulfilling a number of other practical and emotional needs, ranging from informational needs (discussed above), to support in accessing healthcare [27] and feeling understood [26].

In terms of social functioning, a key experience of patients and their carers is their diminished ability to work or having to arrange special work conditions, which often results in loss of income [8-11, 34]. The loss of income is then combined by an increase in medical expenses [6, 9-11, 14, 20, 21] (discussed before) or an inability to acquire adequate insurance [16, 33], resulting in a need for financial assistance, which even when available and accessed is often insufficient [13, 19-21].

3.2.3.3 CARE COORDINATION

Complex needs of rare disease patients require a number of specialist services in order to be met, often in the context of multidisciplinary care. This presents a further financial and psychosocial challenge for patients, who need to keep track of the care and support they need, find ways of accessing it, ensure that all the specialists involved are aware of relevant information and developments, and deal with travelling (often abroad) to access care [9-13, 17, 19-21, 23, 27, 36].

4 DISCUSSION AND CONCLUSIONS

In this report, a systematic literature search and thematic analysis of the needs of patients with rare diseases was performed. The themes described in the 37 records finally selected and examined could be clustered into three domains of need: (1) diagnosis, (2) information and understanding, and (3) medical care and psychosocial support.

To the best of our knowledge, this is the first attempt at a rigorous drafting of a comprehensive framework of the needs of all rare disease patients, regardless of the underlying illness. Studies uncovered during the literature search focused on a specific setting [6, 8, 10, 13, 18, 20] or disease [21, 32-34], a narrowly defined outcome (e.g. experience [29] or quality of life [25]), did not utilise a rigorous approach to the question [15, 17, 19] or utilised an unknown method in determining the dimensions of need [9, 11, 12], or a combination of the above.

While collating and synthesising the studies overcomes some of the above listed issues, limitations of the current report need a fair appraisal as well. Firstly, the search and coding were performed by one researcher, possibly introducing some personal bias into the decision-making process. However, the report aims to provide a transparent account of how the decisions were made and summarises the key themes identified in the record included here, allowing for an evaluation and reproduction of the results described here. Secondly, the literature search was limited to the English language, potentially missing some publications that could be interesting for the review. Thirdly, while some grey literature was identified by virtue of it being cited by included studies, a direct search of grey literature was not performed, which again could have led to missing publications. However, the records that were included here *did* provide a convergent narrative of rare disease patient needs, so it is unlikely that the conclusions of this paper would have been significantly different, were the aforementioned potentially missing publications included. Lastly, no formal definition of a patient's need was used in coding and later stages of the thematic analysis, primarily due to a lack of a clear and unifying definition [41], but also to prevent a reductionist understanding of need (e.g. need for a specific service).

In conclusion, this report provides an empirical framework that outlines the key domains of patient needs that could be considered as particular to rare disease patients. The EMRaDi project will utilise this framework to aid its future work with rare disease patients to determine patient experience and satisfaction with existing services in the EMR region, and to recommend possible avenues for their improvement. We invite other researchers and health and social professionals to improve upon this first attempt with the final aim of understanding how best to serve people with rare diseases today and in the future.

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