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Rare in prevalence, but common in needs: Supportive needs of parents caring for a child with rare disease

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The representation and coordination of programs in early intervention for children with rare diseases indicate that support for parents may be lacking as well. To gain knowledge about the needs of parents caring for a child with a rare disease, this research aimed to identify the supportive needs of affected families and their interrelations and to investigate the relationship between different aspects of needs with the place of residence and the age of the parents.

The research was conducted on a sample of 43 parents (41 mothers, 2 fathers) of preschool children (median 3 years, mean age 3 years, SD = 1.3) with rare diseases. Socio-demographic characteristics were evaluated, and the Parental Need Scale for Rare Diseases (PNS-RD) was used to assess parental needs.

The results indicate that parents reported medium to high levels of psychosocial needs (PNS-DR mean Total Score = 42.8, SD = 7.3). The parents living in rural areas expressed a greater level of supportive needs, which was particularly relevant to the emotional burden. No difference in any aspects of needs was found among parents of different age groups. Emotional issues and parents' need for support in understanding their child's illness were positively related (r = 0.48, p = 0.013).

Our study indicates that the supportive needs of parents of children with rare diseases are characterized by significant emotional effort and an urgent demand for effective social support networks. The obtained insights provide guidelines for a better understanding of parental needs through a comprehensive framework that takes into account emotional, informational, and systemic factors that will lead to improved outcomes for both parents and children. Furthermore, our results highlight the importance of improving family-centered care approaches.

Keywords: PARENTS; RARE DISEASES; NEEDS ASSESSMENT; QUALITY FO LIFE

INTRODUCTION

Rare diseases are a prominent public health problem in countries across Europe. Available data indicate that 6 – 10 % of the total population suffers from a rare disease (1). Although there is no universally accepted definition of rare diseases, within the current political and legislative context, rare diseases are closely linked to a definition based on their prevalence (2). Prevalence is the

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most appropriate indicator, as it allows for the measurement of the number of people with a disease at a given point in time with the possibility of international comparison of data (2, 3).

The course of the disease is chronic and progressive, resulting in permanent disability. In the process of diagnosing rare diseases, the challenges are the diversity of their symptoms. Symptoms and combinations of different symptoms, as well as the worsening of the disease, can also occur at different ages. Despite significant scientific and medical advances that have improved diagnostics and prognosis for children with rare diseases, concerns remain regarding the time required for diagnosis, the level of care provided to children and their parents, and their participation in research (4). The quality of life of those affected is undoubtedly impaired, especially due to the lack or complete loss of independence.

In order to improve a child's health, achieve social and cognitive development and help overcome the many obstacles that children with rare diseases and their families face in everyday life, effective early support is needed, and this process is successful when action is taken for the child's wellbeing at the right time, in the family itself. By strengthening strategic planning at the local level, a solid foundation is laid for creating a support system and connecting various services intended for children and their families to respond to their needs.

The impetus for this research is the needs for rehabilitation practice, which indicate insufficient representation and coordination of programs in early developmental support for children with rare diseases, as well as insufficient education and support for families in the earliest period of a child's development.

The literature consistently emphasizes the importance of utilizing validated measurement tools to identify the specific supportive needs of parents raising children with rare diseases. One such tool is the Parental Needs Scale for Rare Diseases (PNS-RD), which effectively outlines diverse areas of need, including the desire for practical information, emotional support, and assistance navigating healthcare systems (5). By applying these structured assessments, professionals can better tailor interventions and resources to match the specific needs of families, thus optimizing support delivery.

While we can assume that parents caring for a child with a rare disease share common needs regardless of disease or country of residence, we cannot generalize current findings in the literature, while access to healthcare and early intervention services can differ.

The present study examines the supportive needs of parents caring for a child with a rare disease, their interrelations, and different aspects of needs in relation to the place of residence and the age of parents.

METHOD

Study design

The study took place from November 2021 to March 2022. Participants were parents of children with rare diseases, who were up to seven years old and included in the early developmental program at the Centre for Rehabilitation Zagreb. Participants were sent an invitation letter from the Centre for Rehabilitation Zagreb to take part in the study. Forty-two participants enrolled in the study.

Ethical approval, number 738-01-11- 01-25-2 , for the study was obtained from the ethics committee of the Centre for Rehabilitation Zagreb.

Participants

Inclusion criteria were parents of children with a rare disease who were up to seven years of age and who were able to speak and understand Croatian. Participants provided written informed consent to take part in this study.

Procedure

To measure the overall support needs of parents caring for a child with a rare disease, we have used the validated Parental Needs Scale for Rare Diseases (PNS-DR) translated into Croatian language (7).

The scale includes four domains on the perceived level of Understanding the disease, Working with health professionals, Emotional issues, and Financial needs. Next to the domain score, the scale provides the overall needs score.

Data analysis

Statistical analysis was performed using the licensed program STATISTICA6.1 (StatSoft Inc., Tulsa, Oklahoma, USA). Descriptive statistics were computed for sociodemographic data. We used mean and standard deviation for continuous data and frequencies. Testing the difference between the total scores on support, satisfaction, emotional problems, and financial needs in relation to the age of the parents and place of residence was performed using the t-test for independent samples.

The relationship between the total scores on support, satisfaction, emotional problems, and financial needs was tested using correlation and regression analysis. Statistical testing was performed at a statistical significance level of 95 % (*p*<0.05).

RESULTS

Demographic characteristics

Forty-three parents (two fathers) participated in the study with a mean age of 38.1 (SD = 12.5). The mean age of the children was 3.2 years (SD = 1.3).

Most of the respondents, 95.2 %, had only one child with a rare disease, of which progressive neuromuscular diseases were the most common (31 %). In a few cases, Rett syndrome (4.8%) and DiGeorge syndrome (7.1 %) were present, while other rare diseases were isolated cases.

Most parents (85.7 %) were not suspected or diagnosed as carriers of the disease. Three parents were diagnosed as carriers of the disease, while three parents were suspected of being carriers.

The largest number of parents at the time of the study were between 25 and 44 years of age and most of them were married (88.1 %) and lived in the city area (66.7 %) (Table 1).

Table 1. Sociodemographic characteristics of parents and children

		n	%
Parent	Mother	41	95.3
Parents' age	18 -34	21	48.8
	35 - 54	22	51.1
Child's age	0 - 3	16	37.2
	3 - 5	15	34.8
	5 - 7	12	27.9
Place of residence	Urban	29	67.4
	Rural	14	32.5
Marital Status	Married	38	88.3
	Divorced	5	11.7

The overall level of supportive needs

On average, parents reported medium to high levels of need across all items of the parental needs scale (Total Score mean = 41.7; Table 2). The lowest levels of need were reported regarding the domain of understanding the disease. Parents reported medium levels of satisfaction regarding the scale related to working with healthcare professionals. The greater need for support was evident in the domain of emotional burden. A group comparison of the overall level of supportive needs related to the place of residence revealed a greater need for support in parents living in rural areas (*p*<0.05; Table 2). A comparison of parents between younger and older age groups did not reveal any significant group differences.

The level of parental need to understand the disease

The mean parental need to understand the disease was 9.7. Parents in rural areas (mean = 10.9) need almost the same level of support in understanding the disease as parents in the city (mean = 9.1). (Table 2). No difference was also found among parents in different age groups.

Level of parent satisfaction with cooperation with health professionals

The mean parent satisfaction with cooperation with health professionals was 10.1. The independent samples t-test shows that the level of satisfaction related to cooperation with healthcare professionals did not differ significantly among parents in different age groups or based on place of residence (*p*>0.05) (Table 2).

Parental self-assessment of emotional issues

The mean parent self-assessment of emotional issues was 12.4. The parents in rural areas expressed a higher level of emotional burdens (mean

Table 2. Overall parental needs in families with different places of residence and the age of parents

	Total (n=42)	**Families living in urban areas (n=28)**	**Families living in rural areas (n=14)**	***p***
	M (SD)	M (SD)	M (SD)	
Understanding the disease [A]	9.7 (4.5)	9.1 (4.5)	10.9 (4.5)	0.229
Working with healthcare professionals [A]	10.1 (3.8)	9.8 (3.1)	10.3 (4.1)	0.689
Emotional issues [B]	12.4 (2.9)	10.5 (2.9)	12.9 (2.7)	**0.031**
Financial needs [B]	9.3 (3.6)	8.7 (3.6)	9.6 (3.5)	0.472
Total score [C]	42.8 (7.3)	37.7 (5.2)	43.2 (5.7)	**0.015**

[A] Possible score 4–20; [B] possible score 3–15; [C] possible score 14–70; in all cases, higher scores indicate higher need/less satisfaction.

Table 3. The interrelationship between different aspects of parental needs

	r
Understanding the disease – Working with healthcare professionals	0.243 (p=0.786)
Understanding the disease – Emotional issues	**0.482 (p=0.013)**
Understanding the disease – Financial needs	0.294 (p=0.553)
Working with healthcare professionals – Emotional issues	0.316 (p=0.169)
Working with healthcare professionals – Financial needs	0.170 (p=0.660)
Emotional issues – Financial needs	0.381 (p=0.071)

= 12.9) than parents living in urban areas (mean = 10.5) (Table 2). No difference was found between parents of younger and older age groups.

The level of financial needs of parents

The median financial needs of parents was 9.3 and there was no difference between parents of younger and older age groups and between parents living in the city (mean = 8.7) and rural (mean = 9.6) areas (Table 2).

The interrelations between different aspects of parental needs

Correlation and regression analysis showed that there is a significant positive association between emotional problems and parents' need for support in understanding their child's illness (p<0.05, Table 3), i.e. the greater the emotional problems parents have, the more support they need for their child to learn about the illness and explain their child's illness to other people in their immediate environment (Figure 1). No significant correlations were found among other aspects of parental needs (Table 3).

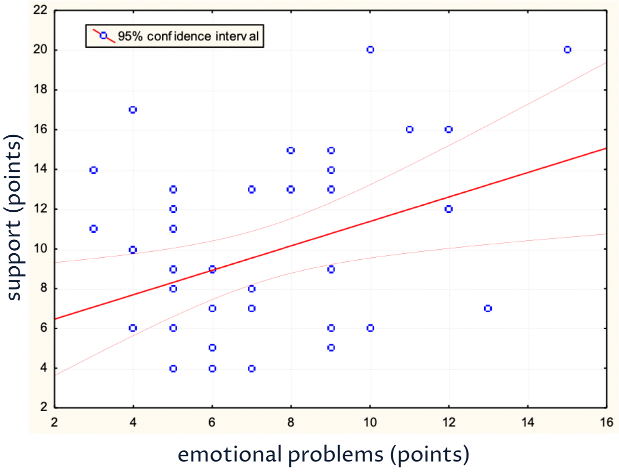

Figure 1. The relationship between emotional problems and parents' need for support in understanding their child's illness

DISCUSSION

The present study examined the supportive needs of parents caring for children with various types of rare diseases. The study revealed a consistent need for additional support, mainly in the fields of emotional and income-/disease subtype-specific burden.

Research illustrates that the burden on parents is markedly high due to the ongoing medical needs of their children, compounded by issues of accessibility and coordination in healthcare services. Firstly, our results indicate that the emotional and psychological toll on parents is notable. The emotional and psychological challenges faced by parents are often exacerbated by a lack of timely and accurate information regarding their child's condition. Research highlights that many parents initially rely on informal sources, such as the internet, to gather knowledge about their child's rare disease, often feeling the need to become "experts" about their situation to effectively advocate for their child's care (8, 9). This proactive approach, while empowering, can lead to frustration when healthcare professionals are not familiar with the nuances of rare disorders or provide insufficient guidance (9, 10). This dynamic underscores the necessity for enhanced communication and support from healthcare providers, which can significantly alleviate parental stress and improve mental health outcomes (11, 12). In particular, the variability in the presentation and progression of rare diseases complicates healthcare management and adds to parental anxiety about the future. Coping with this uncertainty is a pervasive theme in the literature, with parents often employing both proactive and avoidant coping strategies (13, 14). These strategies include seeking social support networks, such as peer groups, which serve as vital resources for emotional support and shared experiences. Studies indicate that such networks can foster resilience and mitigate feelings of isolation (15, 16). Moreover, the emotional burden is not merely an individual concern; it can reverberate through the family system. Parents may find that their relationships with partners and siblings become strained under the pressure of caregiving demands. Specific stressors, such as managing daily care routines and attending numerous medical appointments, often lead to conflict and a sense of co-dependence among family members (11, 17, 18). Indeed, the interplay of stressors can culminate in profound disruptions in family functioning, suggesting that support services must consider the family as a unit rather than only focusing on the affected child (18, 19).

In addressing the support needs of these parents, social networks and peer support have shown to be particularly beneficial. This aligns with findings that stress the importance of collective advocacy and mutual support among families facing similar challenges (15, 20). Parents often rely on informal networks to fulfill their needs for emotional and practical support, as formal systems may fail to offer satisfactory solutions (17, 21, 22). Moreover, the lack of comprehensive informational resources further exacerbates feelings of isolation and helplessness, indicating a significant gap in needed support structures (6, 7).

Finally, it is critical to recognize the systemic barriers that parents face in accessing health and social support services. Research points to the detrimental effects of fragmented care models and insufficient resources tailored for families dealing with rare diseases (9, 23). For effective support, health systems must adapt to the intricacies of rare disease care by ensuring families receive not only the medical resources they need but also consistent psychological and emotional assistance throughout their healthcare journey (24, 25, 26). This approach is essential in mitigating the overall burden on parents, allowing for a focus on holistic family health and well-being. Educational interventions that enhance parents' understanding of their child's rare disease are imperative. These programs can empower parents with the skills necessary to navigate healthcare complexities and actively engage in decision-making processes surrounding treatment and care (10, 11). Furthermore, psychoeducational initiatives can aid parents in improving their emotional coping skills and provide practical strategies for managing daily care challenges, thereby enhancing overall family well-being (6, 16).

This study has some limitations. The first stems from patient selection, with mothers accounting for almost all participants. As only two fathers responded to the questionnaire, fathers' perspectives on issues related to parental supportive care needs remained underrepresented. The second arises from a relatively small sample size that presents parents from urban and rural areas gravitating to the capital of Croatia. Therefore, further research is required to validate these findings in other regions of the country.

CONCLUSION

The landscape of support needs for parents of children with rare diseases is characterized by significant emotional strain, a quest for knowledge, and an urgent demand for effective social support networks. Understanding their needs through a comprehensive framework that considers emotional, informational, and systemic factors will lead to improved outcomes for both parents and children. The integration of tailored interventions, improved communication with healthcare providers, and the fostering of peer support mechanisms would represent a significant advancement in addressing the unique challenges faced by these families.

REFERENCES

1. Feltmate K., Janiszewski PM, Gingerich S, Cloutier M. Delayed access to treatments for rare diseases: who's to blame? Respirology. 2015;20(3):361–9. doi:10.1111/resp.12498.
2. Richter T, Nestler-Parr S, Babela R, et al. Rare Disease Terminology and Definitions-A Systematic Global Review: Report of the ISPOR Rare Disease Special Interest Group. Value Health. 2015;18(6):906–14.
3. Hytiris M, Johnston D, Mullen S, Smyth A. Experience of health care at a reference centre as reported by patients and parents of children with rare conditions. Orphanet J Rare Dis. 2021;16(1).
4. Khair K, Pelentsov L. Assessing the supportive care needs of parents with a child with a bleeding disorder using the parental needs scale for rare diseases (PNS-RD): a single-centre pilot study. Haemophilia. 2019;25(5):831–7. doi:10.1111/hae.13826.
5. Pelentsov L, Fielder A, Laws T, Esterman A. Development of the parental needs scale for rare diseases: a tool for measuring the supportive care needs of parents caring for a child with a rare disease. J Multidiscip Healthc. 2016;9:425–33. doi:10.2147/jmdh.s113898.
6. Pelentsov L, Fielder A, Laws T, Esterman A. The supportive care needs of parents with a child with a rare disease: results of an online survey. BMC Fam Pract. 2016;17(1). doi:10.1186/s12875-016-0488-x.
7. Gómez-Zúñiga B, Pulido-Moyano R, Fernández M, García-Oliva A, Armayones M. The experience of parents of children with rare diseases when communicating with healthcare professionals: towards an integrative theory of trust. Orphanet J Rare Dis. 2019;14. doi:10.1186/s13023-019-1134-1.
8. Smits R, Vissers E, Pas R, Roebbers N, Feitz W, Rooij I, Verhaak, C. Common needs in uncommon conditions: a qualitative study to explore the need for care in pediatric patients with rare diseases. Orphanet J Rare Dis. 2022;17(1). doi:10.1186/s13023-022-02305-w.
9. Witt S, Schuett K, Wiegand-Grefe S, Boettcher J, Quitmann J. Living with a rare disease - experiences and needs in pediatric patients and their parents. Orphanet J Rare Dis. 2023;18(1). doi:10.1186/s13023-023-02837-9.
10. Anderson M, Elliott E, Zurynski Y. Australian families living with rare disease: experiences of diagnosis, health services use and needs for psychosocial support. Orphanet J Rare Dis. 2013;8(1). doi:10.1186/1750-1172-8-22.
11. Lim Y. Impact of raising children with rare diseases on parental quality of life and family functioning. Int J Rare Disord. 2023;6(1). doi:10.23937/2643-4571/1710053.
12. Schiller J, Towne M, Epstein R, Thornton J, Suslovitch V. How parents of children with ataxia-telangiectasia use dynamic coping to navigate cyclical uncertainty. J Genet Couns. 2023;33(2):301–13. doi:10.1002/jgc4.1727.
13. Picci R, Oliva F, Trivelli, F, et al. Emotional burden and coping strategies of parents of children with rare diseases. J Child Fam Stud. 2013;24(2):514–22. doi:10.1007/s10826-013-9864-5.
14. 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;28(1):80–90. doi:10.1007/s10897-018-0294-9.
15. Currie G, Szabo J. Social isolation and exclusion: the parents' experience of caring for children with rare neurodevelopmental disorders. Int J Qual Stud Health Well-being. 2020;15(1). doi:10.1080/17482631.2020.1725362.
16. Cardinali P, Migliorini L, Rania N. The caregiving experiences of fathers and mothers of children with rare diseases in italy: challenges and social support perceptions. Front Psychol. 2019;10. doi:10.3389/fpsyg.2019.01780.
17. Ammann-Schnell L, Groeschel S, Kehrer C, Frölich S, Krägeloh-Mann I. The impact of severe rare chronic neurological disease in childhood on the quality of life of families – a study on mld and pch2. Orphanet J Rare Dis. 2021;16(1). doi:10.1186/s13023-021-01828-y.
18. Belzer L, Wright S, Goodwin E, Singh M, Carter B. Psychosocial considerations for the child with rare disease: a review with recommendations and calls to action. Children. 2022;9(7):933. doi:10.3390/children9070933.
19. Titgemeyer S, Schaaf C. Facebook support groups for pediatric rare diseases: cross-sectional study to investigate opportunities, limitations, and privacy concerns. JMIR Pediatr Parent. 2022;5(1):e31411. doi:10.2196/31411.
20. Zanello G, Chan C, Pearce D. Recommendations from the irdirc working group on methodologies to assess the impact of diagnoses and therapies on rare disease patients. Orphanet J Rare Dis. 2022;17(1). doi:10.1186/s13023-022-02337-2.
21. Boettcher J, Denecke J, Barkmann C, Wiegand-Grefe S. Quality of life and mental health in mothers and fathers caring for children and adolescents with rare diseases requiring long-term mechanical ventilation. Int J Environ Res Public Health. 2020;17(23):8975. doi:10.3390/ijerph17238975.
22. Currie G, Szabo J. It is like a jungle gym, and everything is under constructio: the parent's perspective of caring for a child with a rare disease. Child Care Health Dev. 2018;45(1):96–103. doi:10.1111/cch.12628.
23. Zurynski Y, Deverell M, Dalkeith T, et al. Australian children living with rare diseases: experiences of diagnosis

and perceived consequences of diagnostic delays. Orphanet J Rare Dis. 2017;12(1). doi:10.1186/s13023-017-0622-4.

24. Zurynski Y, Gonzalez A, Deverell M, et al. Rare disease: a national survey of paediatricians' experiences and needs. BMJ Paediatr Open. 2017;1(1):e000172. doi: 10.1136/bmjpo-2017-000172.
25. Ruiz Y, Gerk A, Stegmann J. Mental health impact on primary and secondary prader–willi syndrome caregivers. Child Care Health Dev. 2023;50(1). doi:10.1111/cch.13162.
26. Nguengang Wakap SM, Lamber D, Olry A, et al. Estimating cumulative point prevalence of rare diseases: analysis of the Orphanet database. Eur J Hum Genet. 2019;28:165–73.

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SAŽETAK

Rijetke u prevalenciji, ali česte u potrebama: Potrebe za podrškom roditelja djece s rijetkim bolestima

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Zastupljenost i koordinacija programa rane intervencije za djecu s rijetkim bolestima ukazuje na mogući izostanak podrške roditeljima. Kako bi se steklo znanje o potrebama roditelja koji brinu o djetetu s rijetkom bolešću, cilj ovog istraživanja bio je identificirati roditeljske potrebe za podrškom te istražiti odnos između različitih aspekata potreba s mjestom stanovanja i dobi roditelja.

Istraživanje je provedeno na uzorku od 43 roditelja (41 majka, 2 oca) djece predškolske dobi (medijan 3 godine, srednja dob 3 godine, SD = 1.3) s rijetkim bolestima. Procijenjene su sociodemografske karakteristike, a za procjenu roditeljskih potreba korištena je Ljestvica roditeljskih potreba za rijetke bolesti (PNS-RD).

Rezultati ukazuju na srednju do visoku razinu psihosocijalnih potreba roditelja (medijan PNS-RD = 42,8, SD = 7,3). Roditelji koji žive u ruralnim područjima izrazili su veću razinu potreba za podrškom, što se posebno odnosi na emocionalno opterećenje. Nisu pronađene razlike ni u jednom aspektu potreba među roditeljima različitih dobnih skupina. Emocionalni problemi i potreba roditelja za podrškom u razumijevanju djetetove bolesti bili su pozitivno povezani (r = 0.48, p = 0.013).

Provedeno istraživanje pokazuje kako su potrebe za podrškom roditelja djece s rijetkim bolestima okarakterizirane značajnim emocionalnim naporom i potrebom za učinkovitim mrežama socijalne podrške. Dobiveni rezultati daju smjernice za bolje razumijevanje roditeljskih potreba kroz sveobuhvatan okvir koji uzima u obzir emocionalne, informacijske i sistemske čimbenike koji će dovesti do poboljšanih ishoda i za roditelje i za dijete. Nadalje rezultati naglašavaju važnost poboljšanja pristupa skrbi usmjerenih na obitelj.

Ključne riječi: RODITELJI; RIJETKE BOLESTI; PROCJENA POTREBA; KVALITETA ŽIVOTA