The Impact of the Lack of Supportive Care Needs of Parents Caring for a Child with a Rare Disease

The Effects on Parents and the Strategies Proposed by

the Netherlands, Belgium, and Germany

Bachelor Thesis

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Abstract

Parents have often unmet needs while caring for a child with a disease. While the supportive care needs of parents in the field of oncology have been researched, the field of rare diseases lacks scientific knowledge about the supportive care needs of parents. The aim of this thesis is firstly, to research the lack of supportive care needs of parents caring for a child with a rare disease and secondly, to examine the strategies towards supportive care needs, proposed in the rare disease national action plans drafted by Belgium, Germany, and the Netherlands. A narrative literature review was conducted with 18 articles and for the strategies, the national action plans were used. The results of the literature review indicated that Emotional, Psychological, and Physical Needs were the outcome of Informational, Practical, and Social Needs. The amount of interactions between certain needs have been further researched. These preliminary results were the base of a modified framework that could help health care practitioners (HCPs) in the guidance of parents through the difficult process with their child. Moreover, the framework can help parents to overcome the unmet needs, which are eventually also beneficial for the child. The strategies proposed by Belgium, Germany, and the Netherlands show the general lack of information on rare diseases and the supportive care needs. However, the national action plans provide a sufficient foundation to support new technologies and researches. Yet, the focus of most researches is understandably concentrated on the patient and their rare diseases, though caregivers and parents caring for a child should not be neglected and further research on supportive care needs will help the burden of caregivers and parents be more acknowledged.

1. Introduction

Around 6000-8000 rare diseases have been identified in the world and around 350 million people are affected with a rare disease, nonetheless, little attention to research and medical care is given, because a single rare disease will only affect a couple thousands or ten thousands of people globally. To what is considered a rare disease, multiple definitions exist. Yet in Europe, a disease is considered “rare” or “orphan” if it is affecting less than 1 per 2000 individuals (Eurodis, 2007). Rare diseases are mostly genetic, however, rare diseases can also be infectious diseases. Most diseases develop during childhood due to its genetic origin; nonetheless, rare diseases can also occur at a much later stage of life ("Orphanet: About rare diseases", 2017).

In the European Union (EU) it is estimated that between 27 - 36 million people are affected with a rare disease (EUCERD, 2014). Since the combined burden of all rare diseases is considerable, the focus of research has shifted predominantly towards the medical and care needs of rare diseases a decade ago. However, only since 2007, within the Seventh Framework Programmes for research, around €620 million was established to fund 120 research projects on rare diseases which focus lays on Europe-wide studies of natural history, pathophysiology and the development of preventive, diagnostic and therapeutic interventions for rare diseases, while rare diseases are known to humans since decades ago ("Rare diseases", n.d.; European Commission, 2013). In 2014 the Horizon 2020 took over the funding of the Seventh Framework Programmes and since then limited but increasing orphan drugs – drugs for rare diseases – are reaching patients for treatment. Unfortunately, the majority of rare diseases have still no effective treatment.

Additionally, due to the nature of being rare, hence not common, a frequent problem is that rare diseases are mostly difficult to diagnose (Denis, Mergaert, Fostier, Cleemput & Simoens, 2010). Furthermore, the pathway to a diagnosis for most rare diseases is long and hard, because a physician is more likely to search for common diseases instead of a rare disease. This will lead to situations in which parents of young rare disease patients and the rare disease patients live in capricious and indecisive times while waiting on the diagnosis, which lays a psychological burden on those patients and relatives (Pelentsov, Fielder & Esterman, 2016). Moreover, if the disease can be diagnosed, many conditions have a significant impact on the life of patients and their relatives; patients could be impaired, handicapped (more) through time, or even worse, the

disease might also be even life-threatening. Henceforth, well-provided (psychological) help should be provided to address the supportive care needs, because patients and parents will go through a process of grieving (Pelentsov, Fielder & Esterman, 2016).

Also, other needs should be properly addressed to fulfill all needs in parents and patients because next to Psychological Needs, also social, emotional, practical, informational, and Physical Needs have a significant impact on parents of rare diseases patients (Pelentsov, Fielder & Esterman, 2015). Pelentsov et al. revealed 15 supportive care needs that are divided into three areas: (1) Feeling boxed-in outside the box, (2) Practicalities of care, and (3) Relationships. Within these three areas, parents feel most vulnerable and require support in order to maintain a certain mental health status.

Research into rare diseases is predominately focussed on biomedical research. Only a few researched the mental health aspect and even less concentrates on the supportive care for parents of rare disease patients; instead, most of the researchers delved into problems that are now occurring, such as the lack of orphan drugs and treatment, and the urgent request of more research, instead of giving solutions, advice, or recommendations to decrease the psychological burden of parents with rare disease patients (Barlow & Ellard, 2006; Picci et al., 2013; Rare Disease UK, 2016; Zurynski, Frith, Leonard & Elliott, 2008). Nonetheless, some researchers look into parents that have a child with a (common) chronic disease such as cancer (Hoekstra-Weebers et al., 2001; Kerr et al., 2004; Kerr et al., 2007; Martinson et al., 1999; Martinson et al., 1997; Santacroce, 2002; Sawyer et al., 2000; Shields et al., 1995). While there are a lot of similarities between cancer and rare diseases, there are also differences. Most common cancers have many tests to diagnose cancer and most cancers are curable when the cancer is found in an early stage, while this is the contrary for rare diseases.

As is mentioned above, research into rare diseases is predominately focused on biomedical research. Due to this biomedical focus, children with rare diseases and their relatives have received little attention in scientific and medical communities on behalf of their mental health, while 350 million people – children and adults – in the world have a rare disease. Since the coverage is so limited, patients and their family do not obtain the supportive needs it should receive, which represents in certain – not yet researched – mental health effects.

The aim of this thesis is to understand the mental health effects and the strategies used to fulfill the supportive care needs by the Netherlands, Belgium, and Germany. Therefore, the main research questions are as follows: (1) What are the effects of the lack of supportive care needs of parents caring for a child with a rare disease; and (2) what are existing strategies in the Netherlands, Belgium, and Germany to fulfill the supportive needs of parents?

2. Theoretical Framework

The Parent Supportive Care Needs Framework (P-SCNF), developed by Pelentsov et al. (2015) (Figure 2, page 4), is a framework developed as an answer to the Supportive Care Needs Framework (SCNF) of the Cancer Care Ontario Foundation in Canada. The SCNF framework (figure 1) helps a health care professional (HCP) to guarantee that the supportive care needs of cancer patients are being met within all areas of care and along the disease experience (Fitch, 2008). The SCNF consists of seven domains that are each influenced by external factors such as, age, gender, SES, family, culture, social support, education, and personality. The seven domains include: practical, spiritual, social, psychological, informational, emotional and physical needs. However, Kerr et al. (2004; 2007) have evaluated the SCNF and criticized it, because Kerr et al. had troubles to classify the supportive care needs of parents. Nonetheless, the SCNF is the most widely accepted framework – not only in the oncology – for supportive care needs.

Figure 1: The Supportive Care Needs Framework (Fitch, 2008)

Pelentsov et al. (2015) determined that a new framework (page 4) should be developed for parents that are caring for children with rare diseases, since some disease experiences as described (i.e. recurrent disease) in the SCNF are not applicable for most rare diseases and

because many supportive care needs in the SCNF fell into more than one domain. Also, according to Pelentsov et al. (2015),

> *"The SCNF does not seem to describe or imply any suggestion of causality. Yet, it is apparent that some needs are likely to cause others. For example, some Practical Needs of parents (e.g. financial needs) are likely to lead on to Emotional Needs, which if not resolved, can lead to psychological distress."* (p. 489).

Consequently, Pelentsov et al. suggested a conceptual framework for parents caring for rare disease patients. This framework will be used for this thesis (figure 2).

The P-SCNF consist of six domains: Emotional needs, Social needs, Informational needs, Psychological needs, Physical needs, and Practical needs. Each containing multiple items or

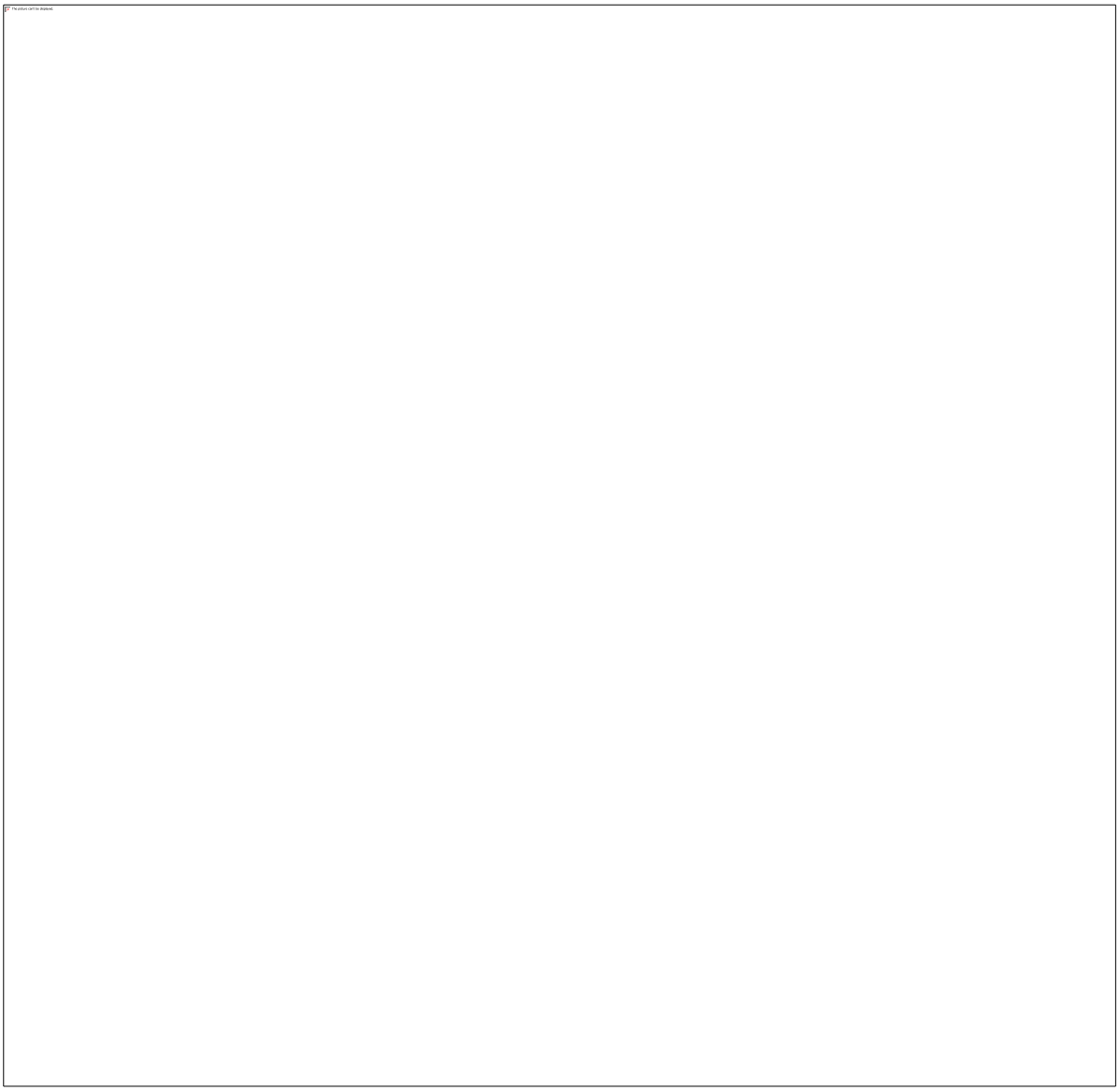


Figure 2: The Parent Supportive Care Needs Framework (Pelentsov et al., 2015)

causalities which clarifies the domain. Kerr et al. (2007) pointed out that no study tried to explore all the domains of supportive care needs, which is a big disadvantage. Moreover, the P-SCNF is a preliminary version and has also not been researched yet (Pelentsov et al., 2015). Also, causalities have not been investigated and the P-SCNF does not entail correctly the effects of the lack or abundance of supportive care needs or to which extent the supportive care should be given. However, the disadvantages do not weigh against the advantage of having a framework dedicated to the supportive care needs of parents caring for a child with a rare disease.

To see to which extent the supportive care should be given to the parents, a secondary framework will be used. This framework is a different model of Margaret Fitch (2008) (figure 3). This diagram shows the provision of service based on the proportion of patients requiring the assistance.

The framework consists of a upside-down pyramid with different levels of care provision. The first level of care provision is required for 100% of the patients/parents and 20% will be satisfied with only this level of care. 80% will flow to the second level of care provision and need also additional information and education, and approximately 30% will be satisfied with this level of

Figure 3: Service provision based on proportion of patients requiring assistance (Fitch, 2008)

care. Therefore, 50% need also the third level of care provision, which entails specialized or expert professional intervention for symptom management or psychosocial distress. This level fulfills the need of 35-40% of patients/parents and this results in 10-15% requiring also the last level of care provision, which entails intensive and on-going complex interventions.

Despite the fact that this diagram is originally made for cancer patients, the model can serve as an additional model to the P-SCNF and it can be used to answer both research questions, because the strategies used by the Netherlands, Belgium, and Germany can be compared with the amount of service/care per level.

3. Methods

This chapter will discuss the methodology used for this thesis. Firstly, the methodology of the narrative literature review for the first research question will be presented and secondly, the methodology of the second research question will be presented. This thesis used a narrative literature review to answer the first research question. The Maastricht Library search engine and Google have been used as Metasearch engine to search for relevant literature. PubMed, ResearchGate, PsychInfo and Google Scholar have been ultimately used to access the literature since these databases have a complete selection of articles in the field of mental health and

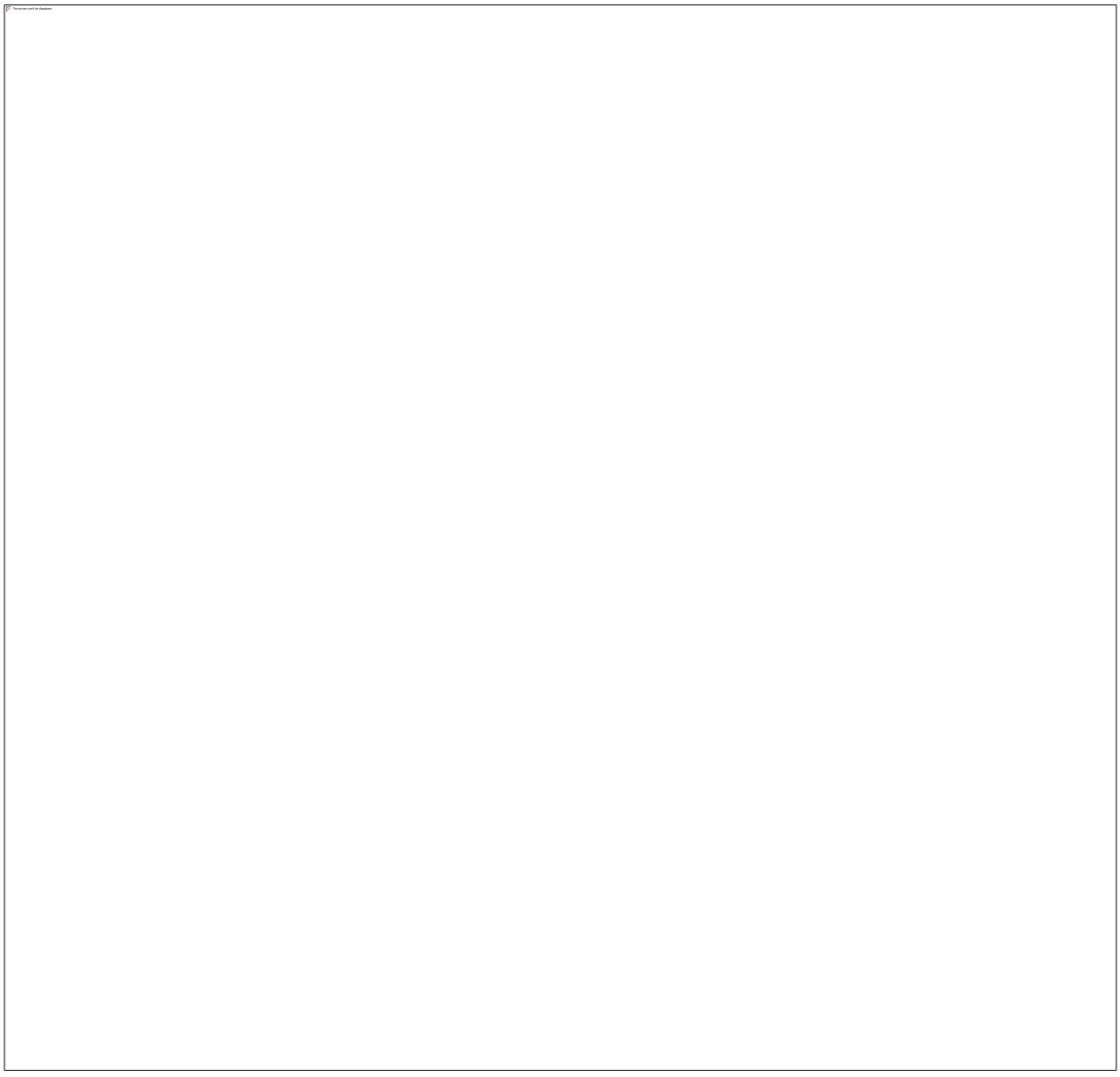


Figure 4: Flow diagram of the selection process for the literature review

medical illnesses. Also, the references of the articles that were found by PubMed, ResearchGate, PsychInfo and Google Scholar have been scanned for other relevant literature that was not found by the above search strategy.

Initially, the keywords that have been used in the search engine primary target specific articles of supportive care needs for parents caring for young rare disease patients, thus a narrow search strategy. Eventually, due to the lack of sufficient literature about parents caring for rare disease patients, the amount of usable research did deplete. Therefore, synonyms of the keywords of the narrow search strategy were added to zoom out the search strategy and, after determining the significance of the newly found articles, the literature of (more common) chronic diseases, cancer, and infectious diseases has been used. However, due to the broad search, irrelevant articles were also found. With the help of Boolean terms a total of 79 articles were found and after a screening on the title and abstract, language, and date of publication (<5 years) 46 articles were left. After reading all 46 articles, studies that were literature reviews or not including any supportive care needs were removed from the list. Also, articles that did cover parents/caregivers caring for children were removed. In the end, 14 articles remained, but 4 articles were added based on the literature list of the 46 articles. Hence, ending with 18 articles that were included in the literature review.

For the second research question, a hand search of websites has been used to determine the strategy used by the Netherlands, Belgium, and Germany. Google was used to find relevant reports and documents submitted by the government or government organizations. Eventually, four reports were found, one each for Belgium and Germany, and two for the Netherlands.

4. Results

This chapter will be split into three sections. The first (4.1) and second section (4.2) will cover the findings of the first research question: “What are the effects of the lack of supportive care needs of parents caring for a child with a rare disease?” and in the third section (4.3) the findings of the second research question will be presented: “What are existing strategies in the Netherlands, Belgium, and Germany to fulfil the supportive needs of parents?”. For the first section, firstly, an overview will be given on the findings of the literature review. Secondly, the findings of the literature review will be explained per domain and per item. For the second section, the interactions between the different domains are presented.

4.1 Literature Review

The literature review consisted of 18 studies each covering multiple supportive care needs of carers. In total there were 10 quantitative studies and 8 qualitative studies. Out of the 18 studies, 8 studies covered carers of cancer patients, 3 studies covered carers of chronic disease patients, and 7 studies covered carers of rare disease patients. The characteristics of the studies can be found in table 1 (page 10). Table 2 (page 17) consists of the items of the P-SCNF domains. A score was given if an item was identified in a study, otherwise, the field was left empty and this was repeated for every study. The percentages in the last column present the proportion of the particular item in comparison with the number of studies with that domain.

Within the explanation of the findings of the different P-SCNF-domains, a preference was made in the following order: rare diseases, chronic diseases, and as for last cancer, in order to emphasize the main focus of this thesis: rare diseases.

4.1.1 Social Needs

The domain Social Needs have been identified in 16 studies. Items that are listed under the domain Social Needs are isolation & loneliness, work/life balance, the relationship between HCPs and parents, social support from their family and friends, and social expectations of their friends and family.

Table 1: Study characteristics by Authors, Sample, Method, Findings/Conclusions, and Domains of P-SCNF

Authors	***Sample***	***Method***	***Findings/Conclusions***	***Domains of P-SCNF***
Armoogum, Richardson & Armes, 2012	98 informal caregivers (ICG) caring for adult bone marrow transplant patients	A cross-sectional survey to describe the needs of ICG and patients, and their relationships between unmet needs and psychological morbidity.	High unmet need and psychological morbidity among informal caregivers of adult bone marrow transplant patients.	Emotional, Informational, Physical, Practical, Psychological & Social
Bourke-Taylor, Pallant, Law & Howie, 2013	152 mothers caring for a school-aged child with a developmental disability	Cross-sectional survey mail-out survey with follow-up phone call to find relationships between sleep disruptions, health and care responsibilities.	Mothers that experience the highest sleep disruptions reported poorer mental health and capacity to participate in health-promoting activities.	Emotional, Physical, Practical, Psychological & Social
Collins et al., 2013	23 current and bereaved carers of adults with Primary Malignant Glioma (PMG)	In-depth interviews to explore the concerns of patients with PMG, to understand the care needs during palliative care, and to obtain the experiences of bereaved family members.	Carers reported that they were surprised by the enormity of caring and the complexity of living with the unspoken future. Also, they reported that there was a lack of care coordination, lack of continuity in care, lack of individualized information, lack of preparation by the health system to assist carers, and lack of emotional support. Moreover, carers have still emotional problems, because of the loneliness of caring and the ongoing suffering for bereaved carers.	Emotional, Informational, Physical, Practical, Psychological & Social

Table 1: Study characteristics by Authors, Sample, Method, Findings/Conclusions, and Domains of P-SCNF (continued)

Authors	***Sample***	***Method***	***Findings/Conclusions***	***Domains of P-SCNF***
Girgis et al., 2012	443 caregivers of cancer survivors	A comprehensive survey measuring the primary outcome and psychological variables.	Prevalence of unmet needs significantly decreased over time. However, over a third of caregivers still reported unmet needs, such as caregivers' well-being and relationships. Increased interference in activities due to caregiving, anxiety, depression, avoidant and active coping, and out-of-pocket expenses was associated with reporting of more unmet needs and unmet psychosocial services.	Emotional, Informational, Practical, Psychological & Social
Heckel et al., 2015	150 dyads of patients newly diagnosed with cancer and their carers, aged 18 and older	Carers completed the Supportive Care Needs Survey-Partners & Caregivers (SCNS-P&C45) and both carers and patients completed the Centre of Epidemiologic-Depression Scale (CES-D).	Most common unmet needs were in the domains of information and health care service needs. Also, 36% of patients were at risk of clinical depression. Also, a moderate positive relationship was observed between unmet needs and carer depression.	Emotional, Informational, Physical, Practical & Social
Heese et al., 2013	605 carers of glioma patients in at the end of their life	Carers answered a 15-item questionnaire focussing on medical, logistic, financial, and mental health support given in the last 4 weeks of the patient.	The medical support was given a 7.2 (10-point scale), nursing service an 8.1, but mental health support a 5.5. In 22.9% of the cases, no mental health support was given for the caregivers.	Emotional, Practical, Psychological & Social

Table 1: Study characteristics by Authors, Sample, Method, Findings/Conclusions, and Domains of P-SCNF (continued)

Authors	***Sample***	***Method***	***Findings/Conclusions***	***Domains of P-SCNF***
Javalkar et al., 2017	150 mothers and children with a chronic condition	Cross-sectional with a web-based survey with 22 items about the caregiver burden.	Patients with lower child self-efficacy, frequent primary care provider, and emergency room visits, and the amount of medicines and injections were predictors of increased caregiver burden. Programs and interventions to improve patients' health-related self-efficacy alleviated the caregiver burden.	Emotional, Practical & Psychological
Kasparian et al., 2014	15 patients and 8 carers of patients with Von Hippel-Lindau disease	Semi-structured interview to explore patients' and carers' psychological difficulties.	Diverse range of experiences were reported, among: sustained uncertainty about future tumour development, frustration regarding the need for lifelong medical screening, strained family relationships, difficulties communication with others about VHL, perceived social isolation and limited career opportunities, financial and care-giving burdens, complex decisions in relation to childbearing, and difficulties accessing expert medical and psychosocial care.	Emotional, Informational, Physical & Psychological
McDowell, Duffy & Parkes, 2015	123 families with a young person (4-27 years) with severe cerebral palsy	Standardised background proforma and validated questionnaires on provided care in the UK.	Over 70% of the families needed more information on their child's disability. Too often parents have no family-centred care, while this lowers significantly health-related quality of life.	Emotional, Informational, Practical & Social

Table 1: Study characteristics by Authors, Sample, Method, Findings/Conclusions, and Domains of P-SCNF (continued)

Authors	***Sample***	***Method***	***Findings/Conclusions***	***Domains of P-SCNF***
Monterosso & Kristjanson, 2008	24 parents of children who died from cancer	Semi-structured interview to obtain feedback from parents of children who died from cancer about their understanding of palliative care and supportive care received during their child's illness.	Parents live in chronic uncertainty and apprehension throughout their child's cancer illness trajectory. Parents were in shock and disbelief; had to adapt and suspend their own life; and had to accept that the child had also influence on the decision making. Moreover, when transferred to palliative care, parents felt being cut-off from the hospital. Also, the parents would have liked to have more quality time with their child during palliative care. More open and honest communication by the health professionals could have prevented Informational Needs, Emotional Needs, and Social Needs.	Emotional, Physical, Practical, Psychological & Social
Muskat et al., 2015	42 participants consisting of youth with autism spectrum disorder, their parents and health-care providers	Semi-structured interview to understand individuals' experiences of health and illness grounded within the context of health care.	Parents had problems with communicating with the healthcare professionals and the degree of flexibility of healthcare providers.	Emotional & Social
Pelentsov, Fielder & Esterman, 2016	23 parents with a child with a rare disease	Qualitative descriptive approach with focus groups.	Parents had troubles with social isolation while caring for their child with a rare disease. Also, parents needed to feel that somebody was taken responsibility for their child's care. Additionally, the parent had problems with their own relationship and had the need to speak with other parents with similar diseases, because other parents could not relate to their problems.	Emotional, Informational, Physical, Practical, Psychological & Social

Table 1: Study characteristics by Authors, Sample, Method, Findings/Conclusions, and Domains of P-SCNF (continued)

Authors	***Sample***	***Method***	***Findings/Conclusions***	***Domains of P-SCNF***
Pelentsov, Fielder, Laws & Esterman, 2016	301 parents with a child with a rare disease	An online survey consisting of 45 questions about the perceived level of satisfaction with receiving care, experiences and needs of providing daily care, the impacts of disease on relationships, the emotional and psychological burdens of disease, and parents overall satisfaction with the support received.	54% was dissatisfied with the knowledge and awareness of the disease of health professionals. 45% had financial problems while caring and 58% had problems with socials isolation. Most emotions that were reported in the week of the survey were: anxiety and fear, anger and frustration, and uncertainty.	Emotional, Informational, Physical, Practical, Psychological & Social
Somanadhan & Larkin, 2016	8 parents caring for a child with Mucopolysaccharidoses (MPS)	Three in-depth interviews with each parent over a 17-month period to understand and interpret parents' experience of living and caring for a child with MPS.	Caring for a child with a rare disease, and in this particular MPS, a chronic, progressive and degenerative disease, has a large impact on the family's life. Parents had many uncertainties in the beginning of the disease, but these became less when the disease developed. Yet, parents described that they had uncertainties before the diagnosis, were devastated, but relieved during the diagnosis, had constant guilt towards their child, because the child would not be able to have a normal life. Parent felt that they were in 'No man's land' and also described that the 'future is unknown', because no cure is available.	Emotional, Physical, Psychological & Social

Table 1: Study characteristics by Authors, Sample, Method, Findings/Conclusions, and Domains of P-SCNF (continued)

Authors	***Sample***	***Method***	***Findings/Conclusions***	***Domains of P-SCNF***
Tenniglo et al., 2017	11 patients with childhood cancer and 18 parents with a child with cancer	3 focus groups with an open discussion to investigate which supportive care topics patients and parents regard as most important and the preferred role they which to fulfil in decision making.	Patients and parents were satisfied with the provided care and appreciated that appointments were clustered. Also, patients and parents preferred to be actively involved in decision-making in the provided care. However, they had troubles with psychosocial care and felt that they had a lot to cope in the first weeks and regretted the absence of a periodic assessment regarding they had Psychological Needs.	Emotional, Informational, Psychological & Social
van Scheppingen, Lettinga, Duipmans, Maathuis & Jonkman, 2008	11 families with a child with Epidermolysis bullosa (EB)	Semi-structured interviews to discover the problems that parents experience with a child with EB and the differences in problems between mildly and severely affected children.	Parents had multiple problems divided into three main themes: the burden of EB on their child's life, (2) the care burden on the parents, and (3) the impact of EB on care providers. Children with severely EB had a bigger impact on the life of parents.	Emotional, Informational, Physical, Practical, Psychological & Social
Yoshida et al., 2014	61 bereaved parents of patients with paediatric cancer in Japan	A multi-centre questionnaire survey to explore the distressing experiences of parents of patients with intractable paediatric cancer in Japan and to explore support they regarded as necessary.	Most distressing experiences were that parents realised that their child was going to die, parents seeing their child suffering, and to plan the funeral. More than 80% of the parents found it essential or very necessary to visit the child every day, to discuss the treatment options, and to have up-to-date information about the treatment.	Emotional, Informational, Physical, Practical & Psychological

Table 1: Study characteristics by Authors, Sample, Method, Findings/Conclusions, and Domains of P-SCNF (continued)

Authors	***Sample***	***Method***	***Findings/Conclusions***	***Domains of P-SCNF***
Zurynski et al., 2017	462 parents caring for children with a rare disease in Australia	An online survey send to families with a child aged <19 years to research the experiences of diagnosis and perceived consequences of diagnostic delays.	When diagnoses are delayed, families, parents and their child, had frustration, stress, and anxiety. 8% of the respondents had waited more than 3 years for a diagnosis. Most delayed diagnoses were devoted due to the lack of knowledge among health professionals. Most parents were satisfied with the diagnosis. However, 16% were dissatisfied, because of inadequate information about the disease, lack of sensitivity among health professionals when giving the diagnosis, and lack of psychological support.	Emotional, Informational, Physical, Practical & Psychological

Table 2: Summary of items found

Type of need		*n*[1]	*%*
Social needs	Work/Life balance	9	56,3
(16 papers)	Health care practitioners partnerships	7	43,8
	Social expectations	7	43,8
	Isolation & loneliness	9	56,3
	Family & friends support	6	37,5
	Partner & siblings relationship	5	31,3
Informational needs	Information easy to access & is relevant	7	53,8
(13 papers)	Early & definitive diagnosis	4	30,8
	Resources for family & friends	6	46,2
	Information on child's illness	10	76,9
	Child's future health needs	9	69,2
	Available services	8	61,5
Emotional needs	Anger & Frustration	6	33,3
(18 papers)	Fear	7	38,9
	Uncertainty & Worry	10	55,6
	Shock & Denial	2	11,1
	Powerlessness	8	44,4
	Guilt & Blame	5	27,8
	Stress & Anxiety	12	66,7
	Emotions related to:		
	Social Needs	10	55,6
	Informational Needs	13	72,2
	Practical Needs	17	94,4
	Physical Needs	8	44,4
	Psychological Needs	13	72,2
Practical needs	Childcare & Other carers	5	35,7
(14 papers)	Finances	7	50,0
	Accessing services & support	9	64,3
	Respite & Leisure	3	21,4
	Transport	8	57,1
	Work & Employment	5	35,7
	Home modifications	2	14,3
Physical needs	Health problems associated with parent also having the disease	2	16,7
(12 papers)	Fatigue & Exhaustion	10	83,3
	Poor sleep	7	58,3
	Loss of appetite	1	8,3
	Weight loss	0	0,0
	Headaches	0	0,0
	Frequent illness	2	16,7
Psychological needs	Self-worth	4	28,6
(14 papers)	Stress	11	78,6
	Depression & Anxiety	7	50,0
	Coping	7	50,0

[1] Sometimes multiple items per domain were found in a study, therefore the amount of items per domain can be higher than the amount of studies with a particular domain.

In the context of supportive care needs for parents caring for children, the most reported domain of Social Needs is **isolation & loneliness** (56,3%, table 2 page 17). Apparently, companionship is highly appreciated among carers of patients. In a qualitative study of Somanadhan and Larkin (2016) the researchers spoke with parents who cared for a child with Mucopolysaccharidosis, a rare, life-limiting condition, which has no cure. Parents felt that they were in 'no man's land' while waiting for a diagnosis. Another example of isolation reported in the study of Pelentsov, Fielder and Esterman (2016) was that a parent reported that she was isolated, because of her child's disease to avoid infection. She was unable to have face-to-face contact with other parents of the same disease because there was a risk of cross-infection. Accordingly, parents have reported in a survey by Pelentsov, Fielder, Laws & Esterman (2016) that 58% have lost friends, following the birth of their child with a rare disease. Mostly because the caring for the child took too much time (Collins et al., 2014; Pelentsov, Fielder, Laws et al., 2016).

Additionally, Collins et al. (2014) found that bereaved parents missed the opportunity to talk to someone, while the child was dying. They had to keep an 'uphold face' to protect the child from outside judgements, but on the other hand, they experienced fear, uncertainty, and grief, which they want to share with friends and family. The parents even talk about a 'pre-caring life', because the demands of caring for their child was extending their capabilities (Collins et al., 2014).

Parents had also troubles finding the golden mean in the **work/life balance** since caring for a child with a rare disease costs a lot of time (Pelentsov, Fielder & Esterman, 2016). The time spends in work and caring for the child resulted in almost having no time left for social interaction with friends, family, or other parents. Also, it is difficult to find an employer that understands the situation of the parents. This leads to more stress and anxiety because some parents are dependent on their work's income. Correspondingly, parents voiced in the study of Pelentsov, Fielder and Esterman that being at work was a social distraction from the situation at home. However, some rare diseases require continuous care, which prohibits some parents from working and therefore creates a limitation in Social Needs.

In seven studies the importance of a good **relationship between HCPs and parents** is accentuated (Collins et al., 2013; Kasparian et al., 2014; Monterosso & Kristjanson, 2008; Muskat et al., 2015; Pelentsov, Fielder & Esterman, 2016; Pelentsov, Fielder, Laws & Esterman,

2016; Somanadhan & Larkin, 2016). Often parents feel frustrated that the HCPs have not enough knowledge about rare diseases and feel powerless that the HCPs do not want to learn about the disease to help the child and the parents. Therefore, parents believe they cannot act as a health care team that takes care of the child (Kasparian et al. 2015; Pelentsov et al., 2016b; Somanadhan & Larkin, 2016). Parents that had a good relationship with the HCPs and had the feeling that the HCP was interested and sensitive to the situation, felt they had someone to fall back to, which relieved stress and uncertainties (Kasparian et al. 2015). However, differences exist in the relationships and the support that is given to the parents by HCPs between occupational support groups (Heese et al., 2013). Nurses were given an 8 out of 10 on the caregivers' perception of the overall support given, physicians a 7 out of 10, but psychologists a 5 out of 10. Additionally, parents in a study of Muskat et al. (2015) stressed the importance of HCPs not speaking to the parents about the child in his room or in the vicinity of the child assuming the child would not understand it is about him. Parents often know better how to explain delicate information to their child and, according to Muskat, parents should always be given the choice and the opportunity to be the person that explains treatments, symptoms, or other sensitive information to their child.

Pelentsov, Fielder, Laws et al. (2016) reported that friends and family of parents caring for a child with a rare disease had difficulties to relate to the problems the parents were experiencing and had problems understanding the seriousness of some diseases, whereas parents reported in the study of Pelentsov, Fielder, Laws et al. as well that they would like to receive **social support from their family and friends**. In a study of Heese et al. (2013), the researchers found that parents perceived the support of nonprofessional sources as the most valuable support. However, 23% of the questioned caregivers reported that they received no support from their family members during the final stage of caring for their family member, whereas this stage was perceived as the severest.

Moreover, parents have the urge to attend social gatherings but have troubles to be present at such meetings, because their child might require special assistance or care (Pelentsov, Fielder & Esterman, 2016). Parents reported in the study of Pelentsov, Fielder and Esterman (2016) that the **social expectations of their friends and family** are too high while caring for their child, because the parents are expected to be at social gatherings. However, often parents report that

they feel exhausted after a day of caring for their child (Armoogum, Richardson & Armes, 2012; Bourke-Taylor, Pallant, Law & Howie, 2013; Girgis et al., 2012; Kasparian et al., 2014; Pelentsov, Fielder & Esterman, 2016; Pelentsov, Fielder, Laws & Esterman, 2016). Girgis et al. (2012) found that parents had the understanding that friends and family knew what is happening with the parents and the child in the first 6 months after the diagnosis, but that after 12 months parents have difficulties with friends and family, because they do not understand or acknowledge the impact of caring for a child with a disease.

Generally speaking, parents are dealing with a double-edged sword. On the one hand parents wish to receive support from friends and family, but on the other hand, parents believe that the social expectations of friends and family are too high and parents have not enough time to find the golden mean in work/life balance.

4.1.2 Informational Needs

In total 13 papers had mentions of Informational Needs. In some papers parents mentioned that they had to wait a long time until an HCP finds a diagnosis, which has significant impacts on them, the parents, but also on their child and family, including parents, feeling concerned, frustrated and having stress while waiting (Pelentsov, Fielder & Esterman, 2016; Zurynski et al., 2017). Moreover, Zurynski et al. also found that there were serious consequences while waiting on the diagnosis, such as delays in getting early intervention programmes and treatments, worsening symptoms and disease progression, and wrong medications been given to the patients. Correspondingly, parents blamed the lack of knowledge among HCP as the main perceived reason for delayed diagnoses and the lack of a diagnosis.

Zurynski et al. (2017) reported that receiving an **early and definitive diagnosis** is life-changing and can relieve the burden on parents. Yet, some parents of the study mentioned that they were dissatisfied with the diagnosis, because it lacked information and, according to the parents, the HCP had no sensitivity towards the parents. As said above, receiving a diagnosis can relieve the burden of parents, but this depends on how susceptible the parents are in receiving such information. Parents are in a state of grieve and HCPs should be aware of the fact, that some diagnoses can upset families.

Apart from diagnosis, parents have many questions. How old will my child become? Does my home need modifications? Is there a treatment? In the study of Pelentsov, Field and Esterman (2016) it is clear that when a rare disease is found, most parents know nothing about it. Parents have a lot to digest when a disease is diagnosed and they want to receive as much **information about the child's disease** as possible (Collins et al., 2013; Girgis et al., 2012; Heckel et al., 2015; Kasparian et al., 2014; McDowell, Duffy & Parkes, 2015; Pelentsov, Fielder & Esterman, 2016; Pelentsov, Fielder, Laws et al., 2016; Tenniglo et al., 2017; Yoshida et al., 2014; Zurynski et al., 2017). Yet, a rare disease is mostly connected to scarce information. Moreover, after the diagnosis parents were disappointed with the information given by HCPs (Zurynski et al., 2017) and searching online is for parents on various occasions a wonderful tool, but also a dangerous tool, because when information is found it can be frustrating and scary, because parents might learn how the disease might develop over time (Somanadhan & Larkin, 2016). Another aspect is that parents want to know as much as possible because they feel the HCPs will give more information and include the parents more in decisions if the parents show that they are knowledgeable and skilled in their child's disease (Pelentsov, Fielder & Esterman, 2016).

Unfortunately, some diseases are so rare, that there is no **information that is easy to access and relevant**. In the study of McDowell, Duffy and Parkes (2015) it is clear that parents lack information about the simplistic areas of the disease, such as information about the therapy the child is in, or what the disease caused. Also, 81% of 123 participants were dissatisfied with the amount of information that is easy to access (i.e. booklets or videos). Parents often need this kind of **information to give to family and friends**, who are not interested in reading long articles or have to scrape information out of the internet. In the study of Pelentsov, Fielder, Laws et al. (2016) more than 1/3 of the 237 parents of children with a rare disease felt they need to be educated by the HCPs about how to explain the disease to other parents, family, and friends, because parents will experience uncertainties if they cannot explain fully their child's disease.

Parents have also many questions about their **child's future health needs**. Especially parents of children with progressive diseases or disease which develops over many years are craving to know what kind of health necessities the child might need, which can be relieving for the parents to know. Yet, too much information can be harmful to the parents, causing anxiety, worry and stress (Pelentsov, Fielder & Esterman, 2016). However, in the study of Pelentsov, Fielder and

Esterman parents communicated towards the researchers that information about their child's future health needs can strengthen them emotionally, but also psychologically to the upcoming situation, and prepare them to make necessary life-adjustments.

Similarly, **available services for the child** are hard to find (Pelentsov, Fielder, Laws et al., 2016). In the survey, 72.5% of the parents (n=240) said that they are desperate to know more about the services the child might receive in the future and 60.8% of the parents said that they are desperate to know more about the services that are presently available for the child. Identically, in the study of McDowell, Duffy and Parkes (2015) 70% of the asked families (n=123) were unsatisfied with the information given about the types of services offered by the HCPs.

Informational Needs have a tremendous impact on the lives of parents. Too little and too much information can be harmful, but receiving a definitive diagnosis remains the most critical Informational Need.

4.1.3 Practical Needs

In the following section, the Practical Needs will be illuminated covering access to services and support, financial support, transportation, leisure and respite time, work and employment, childcare and other carers, and home modifications.

In addition to the need of information for available services, parents stated in nine studies that they require assistance to **access services and support** for their child (Armoogum, Richardson & Armes, 2012; Collins et al., 2013; Heese et al., 2013; Monterosso & Kristjanson, 2008; Pelentsov, Fielder & Esterman, 2016; Pelentsov, Fielder, Laws et al., 2016; van Scheppingen, Lettinga, Duipmans, Maathuis & Jonkman, 2008; Yoshida et al., 2014; Zurynski et al., 2017). Parents often do not know which services can be provided to their child, which can be exhausting for the parents (Pelentsov, Fielder & Esterman, 2016; Pelentsov, Fielder, Laws et al., 2016). Moreover, in some cases, the process to obtain a formal diagnosis can take a long time (Zurynski et al., 2017). Parents have to repeatedly fight for their child's rights to receive specialist treatment because health insurance will not cover certain services until a diagnosis has been found (Pelentsov, Fielder & Esterman, 2016). Parents will not only feel powerless towards the current system, but also it gives the parents stress, anxiety, and frustration (Armoogum,

Richardson & Armes, 2012). Another problem is that parents often want to be in contact with other parents caring for a child with the same disease. Despite the internet, many parents do not know about the existence of any patient organisation, yet are relieved when they find others being in the same situation because they will not feel 'alone' anymore (personal communication, 28th of February 2017). Also, sharing information about possible treatments, good centres of expertise, or just a place where the parents can ask questions, have a good impact on the Emotional and Psychological Needs (Pelentsov, Fielder and Esterman, 2016). Additionally, finding **childcare** that is able to care for your child is hard to find. Parents caring for a child with Epidermolysis Bullosa (EB), a rare genetic blistering-skin disorder, claimed they could only find childcare for a couple times per week, instead of the whole week (van Scheppingen et al., 2008). They claimed that the carers could not bear the stress, albeit the most important thing to do for the carers, was to dress the child. However, the process of dressing the child is extremely painful and the carers could not endure the anxiety of torturing the child. Therefore, the parents said in the study of van Scheppingen et al. they had subsequently different carers, because one left after the other.

Another aspect of Practical Needs is the **financial support** for parents. Reaching a formal diagnosis is necessary for receiving financial support from health insurers. However, the problem exists with rare diseases that a diagnosis is sometimes hard to reach. One parent said that she had to visit more than ten HCPs to receive a diagnosis for her child and that in the whole time of travelling to all kind of different places and hospitals she never received any financial support (personal communication, 28th of February 2017). Another parent in the study of Pelentsov, Fielder and Esterman (2016) called it "feeling boxed-in outside the box", which can be translated as feeling neglected by all the systems because your child is not fitting in one of the pictures and therefore not receiving any financial support. Additionally, more than half of the participants (n=236) of the study of Pelentsov, Fielder, Laws and Esterman (2016) stated that they cannot afford special equipment or clothes when the child needs it. Also, more than half of the participants stated that they did not have enough money to pay for **home modifications**, baby-sitting, or respite care. Moreover, almost half of the parents could not afford medical care, therapy, or transportation for the child. However, most of the parents could pay for medications.

As mentioned above, **transportation** is also a practical need that parents need help with. Not only to receive a diagnosis, but also to bring your child to school, leisure activities, or hospitals for treatment or check-ups, transportation is sometimes required. The Netherlands have different centres of expertise for rare diseases across the country. While this ensures that parents and patients have the best care centred in one place, instead of different centres with mediocre knowledge, this can result in a centre of expertise in the north of the Netherlands, while living in the south. Resulting in a 3-hour drive for every check-up. In the study of van Scheppingen et al. (2008) parents mentioned that the organisational aspects of care were exhausting and time-consuming. Parents have stated in the study of Monterosso and Kristjanson (2008) that they felt relieved when the hospital arranged transportation and planned all appointments on one day because they could then focus on their child instead of arranging practical details.

Due to the daily task of caring for your child, **leisure and respite time** was brought to a minimum (van Scheppingen et al., 2008). Parents mentioned in the study of Scheppingen et al. that leisure interests of holidays or visiting relatives was difficult and hard to plan. Also, in the study of Bourke-Taylor et al. (2013) parents had no time for leisure, because parents were woken up regularly by their child. This detained parents from doing any leisure activities, because they were too exhausted. Yet, Pelentsov, Fielder, Laws and Esterman (2016) saw that leisure and respite time was needed among parents to relieve stress, anxiety and uncertainties that parents often cope with.

Work and employment is hard to combine with a child with a rare disease (van Scheppingen et al., 2008). As mentioned above, it is difficult to find specialized childcare for children with rare diseases, because it is a demanding occupation. In the demographics details of the study of Pelentsov, Fielder, Laws and Esterman (2016) it becomes clear that often one of the parent works and one parent takes care of the child, or both are part-time earners.

The practicalities of employment, arranging childcare, and the financial burden of parents show the implication of caring for a child with a rare disease. Moreover, Practical Needs are diverse and parents mostly require assistance from others – HCPs, friends, and family – to comfort them.

4.1.4 Physical Needs

In the framework of Pelentsov et al. (2015) seven different items of Physical Needs are presented: health problems associated with parent(s) also having the disease; fatigue & exhaustion; poor sleep; loss of appetite; weight loss; headaches; and frequent illnesses. Not every Physical Need is represented in the 18 studies, yet are connected to other needs as will be discussed in section Interactions Between Needs.

While the item **health problems associated with parent(s) also having the disease** has been reported in two papers, it provides a big impact for a family to have two or more persons with a rare disease to care about (Kasparian et al., 2015; Pelentsov, Fielder & Esterman, 2016). Kasparian et al. interviewed families affected by Von Hippel-Lindau disease, which is a rare autosomal dominant genetic condition. The persons affected by this disease will develop multiple tumours throughout their life and have a mean life expectancy of 59.4 years. The mean age at diagnosis of tumours is 26 years (range 4 – 68 years). Due to the dominant allele, parents will have a 75% chance of giving the disease to their children. Kasparian et al. spoke to parents who did not know about VHL before their child was born, but some parents have mentioned that they would not have had any children if they had been diagnosed beforehand. Guilt and anger were some of the reported emotions after their children were also diagnosed with VHL. However, it did increase the relationship between the family members, because it is for persons easier to talk about VHL with family members who are also carrying the VHL disease.

Out of eight papers that have reported signs of Physical Needs, **fatigue & exhaustion** has been reported the most (83.3%). Heckel et al. (2015) found that carers had high levels of stress causing other needs, among which of fatigue and exhaustion. Moreover, Heckel et al. questioned carers of newly diagnosed cancer patients and found also that other factors were causing Physical Needs for the carers – and in particular fatigue and exhaustion – including finding information about services, possible alternative treatments, outcomes of cancer, and caring for the Physical Needs of the patient. In addition, **poor sleep** resulting from stress is also common among carers.

Armoogum, Richardson and Armes (2012) found in their questionnaire, that caregivers had trouble looking after their own health when given care to a relative. 33% of the respondents indicated that they had a **loss of appetite** while giving care. Yet, in no other studies a loss of appetite was reported. Moreover, signs of **weight loss** or **headaches** were not reported in the 18

studies of the literature review. However, two studies reported that parents had **frequent illnesse**s due to the intensive care the parent have to give to their child (Bourke-Taylor, Pallant, Law & Howie, 2013; Pelentsov, Fielder, Laws et al., 2016).

Physical Needs were mostly found in combination with other needs, for example, finding information about services (Informational Needs) was a reason to be fatigued and exhausted (Heckel et al., 2015). Relationships and interactions between needs will be further discussed and examined in section 4.2 Interactions Between Needs.

4.1.5 Emotional Needs

Having a child with a rare disease can be stressful. Emotions such as stress, guilt, anger, frustration, etc. are common among parents.

Most emotions are related to Practical, Informational, and Psychological Needs (Table 2). For example, financial problems (Practical) were often reported as stressful (Armoogum, Richardson & Armes, 2012; Collins et al., 2013; Heckel et al., 2015; Heese et al., 2013; Pelentsov, Fielder & Esterman, 2016; Pelentsov, Fielder, Laws et al., 2016; van Scheppingen et al., 2008). Moreover, delayed diagnosis (Informational) gave a lot of uncertainty and frustration, because parents felt that the HCP was not putting enough effort to diagnose the child's disease (Girgis et al., 2012; Pelentsov, Fielder & Esterman, 2016; Pelentsov, Fielder, Laws et al., 2016; Zurynski et al., 2017). Pelentsov, Fielder and Esterman (2016) reported that during pre-diagnosis parents had troubles coping with the complex situation of the child because neither the parents nor the HCP had concrete solutions for the child. Moreover, parents felt insecure, because they did not know how the disease would evolve. More about emotions will be discussed in section 4.2 Interactions Between Needs.

4.1.6 Psychological Needs

Maintaining the mental health of parents is important for the functioning of the parents. Hence the significance of this domain. Psychological Needs consists of 4 items: self-worth, stress, depression & anxiety, and coping. While the domain Emotional Needs also have the item 'stress', here it includes chronic stress and chronic distress, instead of acute stress.

Depression and anxiety is a common problem among parents. Moreover, parents have a greater prevalence of depression than patients (Kasparian et al., 2014). Pelentsov, Fielder, Laws et al.

(2016) reported in their study that 41% of parents caring for a rare disease patient was being treated for a depression. Also, 10% is being treated for anxiety since the birth of their child.

Self-worth is a difficult concept to grasp. In some articles there is spoken of self-esteem or self-respect, but in terms of parents caring for child with a rare disease self-worthiness can be understood as the ability to care and the confidence the parent has (Collins et al., 2013; Girgis et al., 2012; Pelentsov, Fielder & Esterman, 2016). During the diagnosis parents' world is turned upside-down and a lot of parents have trouble **coping** with the 'truth' and are blaming themselves for the disease of their child and have reportedly less confidence (Pelentsov, Fielder & Esterman, 2016). Therefore, their self-worth is decreasing. Pelentsov, Fielder and Esterman also mentioned that low self-worth could be an indication of a depression.

Chronic **stress** is also a major burden for parents. Like acute stress, most of the times, chronic stress was a result of unmet supportive care needs. For example, delayed diagnosis resulted in chronic stress among parents of children with a rare disease, because parents were afraid of what the future might bring and because parents do not know what is wrong with their child (Zurynski et al., 2017). Moreover, the lack of knowledge among HCPs was also a reason that parents had higher stress levels (Pelentsov, Fielder & Esterman, 2016).

4.2 Interactions Between Needs

This section will elaborate on the interactions between needs in two parts. In the first part, a small explanation will be given on the preceding of this section. In the second part, the relations between the different needs are explained via different graphs.

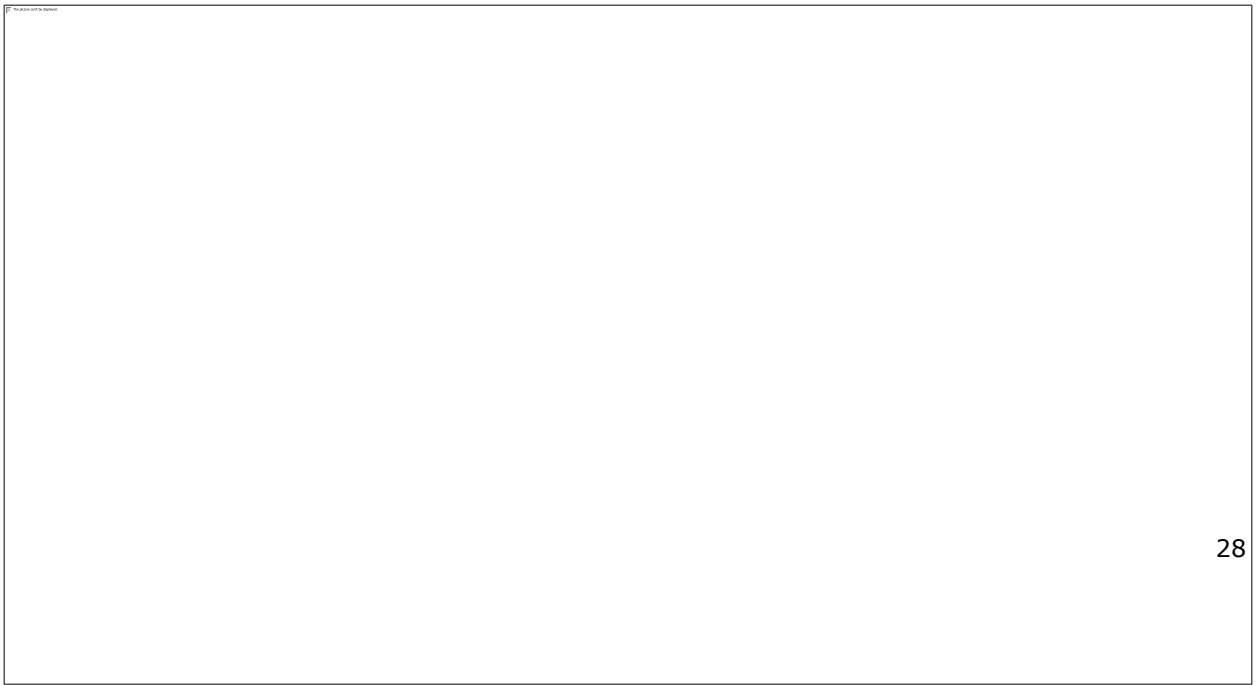
During the calculation of the P-SCNF's items in Table 2, it became clear that in every included study, signs of Emotional Needs were present (n=18). Also, the number of studies with Psychological Needs (n=14) and Physical Needs (n=15) was high. In section 4.1 emotions were repeatedly the result of the lack of supportive care needs. Therefore, causal interactions between Practical, Informational, and Social Needs with Physical, Emotional and Psychological Needs have to be explored to confirm the relationships.

Firstly, all the articles have been scanned to find the result of a certain need. For example, if a parent mentions that he got stress due to financial problems it means that there are Emotional Needs (results) from Practical Needs (cause). This has been counted for every article in the

literature review and the results are presented in graph 1 and graph 2 (page 29 and 29). Graph 1 represents the number of interactions per article between certain needs. The height of the bar is based on a calculation of the amount of interactions (which could be multiple per article) divided by the number of articles which had a single domain in it. Therefore, the height of the bar can be compared to other bars, because the main goal of this bar chart is to compare the number of interactions between different domains. To clarify with a theoretical example, if *domain A* and *domain B* have both 40 interactions. If the interactions are not divided by the number of articles, the bars would be the same. Yet, 5 out of 20 articles included *domain A* and 20 out of 20 articles included *domain B.* As a result, *domain A* has 8 interactions per articles, thus a higher bar, while *domain B* has only 2 interactions per article, thus a smaller bar. The proportions of interactions within each need are represented in graph 2 (page 29).

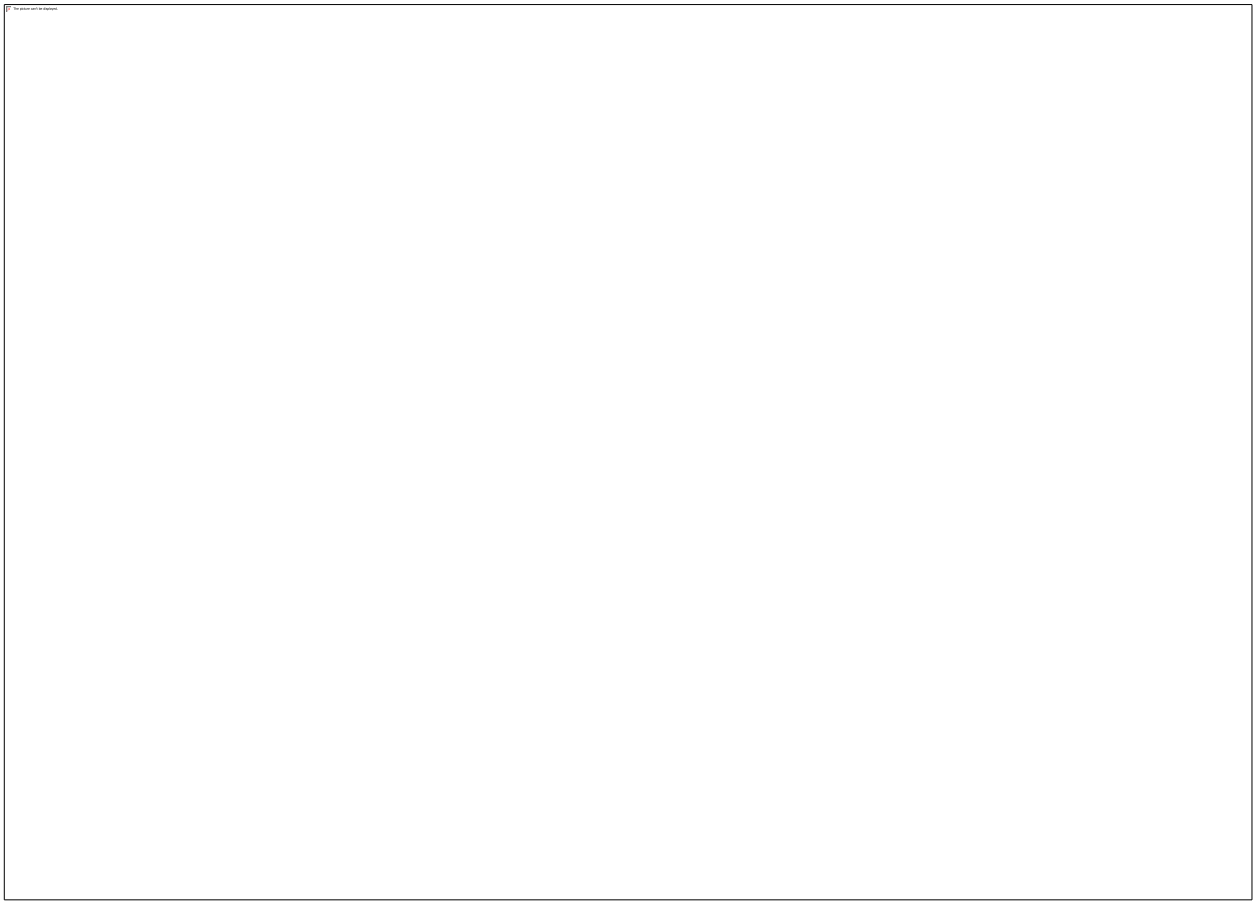
As can be seen in graph 1 there are many interactions with Social, Informational, Emotional, Physical, and Psychological Needs as a result, thus the height of the bar. On the other hand, the amount Practical Needs that are a result of other needs is marginal. This indicates that mostly Practical Needs are causes instead of results. However, there are some exceptions. In the study of Yoshida et al. (2014) parents cope with a lot of emotions and indicated they need help with the emotions, for example, by counselling a psychologist. This is a Practical Need from an Emotional Need. You could argue that the domain Practical Needs is a two-way street, thus being a cause and a result in regards to other needs. Yet, you could also argue that the concept of Practical Needs is too broad, and should be therefore split into two different needs.

Nevertheless, Practical Needs are often the cause of another need, moreover, the proportion of



Practical Needs is the highest among every other domain as can be seen in graph 2. In other words, parents will receive Social, Informational, Emotional, Physical, and Psychological Needs mostly from Practical Needs. Then again, you could argue that the concept of Practical Needs is too broad, thus the number of interactions.

Social Needs and Physical Needs have respectfully the third and fourth amount of interactions and are almost identically with each other. Surprisingly, the amount of Social Needs coming from Physical Needs and the number of Physical Needs coming from Social Needs is very low; both have one interaction. Bourke-Taylor et al. (2013) noticed that mothers with poor sleeping patterns, due to their child's disease are fatigued and exhausted. For this reason, the mothers had trouble to be socially active, while they require social interaction. Thus, Social Needs coming from Physical Needs. Visa versa, Somanadhan and Larkin (2016) found in their study that parents had trouble maintaining their work/life balance and were fatigued and exhausted by it. As shown above, it is surprising how close certain needs interact with each other.



4.3 National Action Plans

This section will elaborate on the existing national action plans of Belgium, Germany and The Netherlands. Every country in the EU is obliged to have a national plan for rare diseases to improve the prevention, diagnosis, treatment, and revalidation of patients with a rare disease (European Council, 2009). However, every country was free to a certain degree how these national plans were constructed. Therefore, there are some differences due to the different health systems each country has. The countries will be compared based on the conceptual Service Provision framework of Fitch (2008). This framework (page 5) consists of four items in an upside pyramid. The thought about the upside pyramid was that 20% of 100% of the people requiring supportive care, were satisfied with an on-going basis of the provision of relevant information, basic emotional support, good communication and astute symptom management, thus broad service provision. Others (approx. 30%) also might need narrower support, such as additional information or education, but also an encouragement to seek help and engage in peer support groups. Between 30-40% also need more specialised or expert professional intervention for symptom management and psychological distress. The last item is required for approx. 10-15%. This includes intensive on-going complex interventions.

The different national plans will be scanned for items that contribute to supportive care needs of patients and, indirect, caregivers of patients with a rare disease. In the end, each country will be set side by side based on the four items and a comparison will be given.

4.3.1 Belgium

The Belgium Plan for Rare Diseases (Belgisch plan voor Zeldzame Ziekten) consists of 4 domains with in total 20 items (Belgisch Plan voor Zeldzame Ziekten, 2017). The focus points of these domains were: (1) improved access to diagnostics and information for the patient; (2) optimisation of care; (3) knowledge management and information; and (4) governance and sustainability of the plan. Each item in the national plan is in the plan being discussed on their aims, motivation, budget, legal basis, and description.

The first domain of the national plan is important for the supportive care needs of patients and caregivers because it is focused on centres of expertise. Belgium has eight centres, each with their own speciality. These centres help patients and parents caring for children with a rare disease with the diagnosis and treatment of their disease. When a patient gets referred to a

centre of expertise, a care coordinator is assigned to the patient and the patient gets a multidisciplinary team based on the disease. The care coordinator is the main contact for patients and caregivers and all the information will go via the care coordinator. Additionally, due to the approval and financing of genetic counselling by the Belgium government, centres of expertise are allowed to perform genetic tests. As a result that the time to reach a diagnosis is reduced and more specific diagnoses can be delivered to the patient with an advantage that the patient can start earlier with specialised treatment. Also, genetic counselling provides information for the family concerning their heredity and their family planning. Another aspect of the national plan is that every centre should provide a call centre for 24 hours per day. This ensures that caregivers and patients have a safety-net if there is an urgent question.

A big advantage for HCPs is the Multidiscipline Patient Dossier (MPD). This dossier consists of important information about the patient and as the name suggests, every HCP involved with the caring of the patient have access. Currently, patients have no access to the MPD, but the Belgium government is planning to grant access to the patient, non-medical caregivers and relatives of the patient in the foreseeable future.

The Belgium government is also working on a project: “Unmet Medical Need”. This project aims to make treatments and orphan drugs easier and faster available for patients with a rare disease. Moreover, the project assists patients with the funding of the orphan drugs, since these are mostly non-deductible and expensive. A workgroup connected with the project will evaluate the needs of patients with a rare disease and will give recommendations to insurers, the government, and organisations of HCPs.

One of the final parts of the Belgium Plan for Rare Diseases consists of a national register for rare diseases. This register can serve as a source of information for patients, patient organisations, caregivers, HCPs, researchers, and the government. The register will be filled with epidemiologic data, longitudinal data for patients, and an overview of the different organisations involved. Moreover, medical trials can locate patients with a rare disease easier.

4.3.2 Germany

The “Nationales Aktionsbündnis für Menschen mit Seltenen Erkrankungen” (National Plan of Action for People with Rare Diseases) has two main objectives in their report (NAMSE, 2013). Firstly, the National plan should prepare policy suggestions and secondly, the National plan should propose actions. To fulfil these objectives, the plan consists of 4 domains: (1) care/centres/networks; (2) research; (3) diagnostics; and (4) information management. Each domain consists of 3-4 items which contain all the relevant action plan standards.

Similarly to Belgium, Germany has centres of expertise where different areas of care are combined to give better care to the patient. Yet, Germany differentiates three types of care centres; type A, B or C. Type C are cooperating centres which offer patients outpatient care for a specific rare disease or specific rare disease group. These centres deliver care for patients with a definite diagnosis or a clear suspected diagnosis. Type B centres are focused on a specific rare disease or disease groups and deliver outpatient as well as inpatient care. Patients that are referred to this type of centres have also a definite diagnosis, but this centre offers interdisciplinary and multi-professional care. Type A centres also offer interdisciplinary and multi-professional care but have the expertise for a large group of diseases. Patients with a suspected rare disease and without a clear diagnosis are referred to here. The benefit for the patient is that these three types of centres are interlinked with each other and provide a solid information basis for rare diseases. Also, patients can easily be referred to another type of centre.

Delays in diagnosis can be troublesome for parents (Zurynski et al., 2017). Germany acknowledges this in their national plan and noticed that most delays come from the primary care. Mostly, HCPs in the primary care are confronted with a series of symptoms throughout the body. Therefore, diagnostic software technologies are implemented in primary care to propose a series of diagnostic tests that can be carried out in one of the types A centres. This ensures faster diagnosis and faster referral to the appropriate centre of expertise.

Germany’s National Plan also acknowledge the need for disease-specific information, such as diagnostic and therapeutic possibilities, and legal and social questions. However, information is scarce and is scattered around the internet, instead of concentrated at a single place. Therefore,

Germany has made a platform where all information is gathered and to ensure quality, all information should meet specific standards.

In conclusion, Germany has a more systematic approach to handle rare diseases. The different types of expertise centres can indeed decrease the diagnosis delays. However, parents mentioned in a study of Scheppingen et al. (2008) that travelling to different facilities was exhausting and preferred to stay in a single hospital if possible. The diagnostic software technologies can help with this, though these technological advantages have yet to be implemented.

4.3.3 The Netherlands

On the 28th of February 2017, an answer to the National Plan Zeldzame Ziekten (National Plan Rare Diseases) was presented called "The Last Advice" (Slotadvies Afstemmingsoverleg Zeldzame Ziekten) (ZonMw, 2017). The National Plan Zeldzame Ziekten consisted of six domains and in addition to the domains of Belgium and Germany, the Netherlands focused more on patient organisations, creating awareness for rare diseases, and education (ZonMw, 2013). Here the findings of the last report are presented.

"Patient organisations should be represented more in the Netherlands" was one of the main messages in the national plan. To achieve this, patient organisations are clustered in the VSOP (Vereniging Samenwerkende Ouder- en Patiëntenorganisaties), because all the patient organisations together have greater empowerment than being 'lone wolves'. Consequently, the VSOP represents the different patient organisations in the politics, caregivers, care institutions, insurers, and in the National Plan Rare Diseases.

One of the problems the VSOP notices is the little awareness of rare diseases. The public has to become conscious about the enormity of rare diseases. Moreover, HCPs and parents are not fully aware of the scope of rare diseases and new technologies to diagnose rare diseases. This causes delays in diagnosis, treatment, but also in research.

The national plan also mentioned the lack of education among HCPs. Medical schools and in-service training do not cover rare diseases and the signalling of rare diseases. Consultation clinics and general practitioners are mostly not thinking about rare diseases when different

symptoms are present, which can result in delays in diagnoses. Also, it is unknown where the expertise is gathered for a group of diseases.

In The Ultimate Advice some bottlenecks were presented. For example, some centres of expertise have no care coordinator or central contact person. Also, multidisciplinary treatment is limited in the Netherlands, because of obstacles in care institutions and legislations. Moreover, no guidelines are available for the costs of shared care. Most importantly, supportive care is undervalued, because there is little information about the effects of the lack of supportive care needs. Additionally, supportive care is only given when informal caregivers show signs of Psychological Needs, instead of giving preventive supportive care.

4.3.4 Comparison

As we take the conceptual service provision framework we can see that all countries struggle with the basic information provision, while 100% of the patients require this type of provision. First of all, all three countries have shown increased awareness towards rare diseases, but a couple of common problems exist. Firstly, HCPs are not educated enough in signalling rare diseases. People with symptoms, that could indicate a (potential) rare disease, are not referred to specialists fast enough. This is because of the amount of awareness among the first line, the first HCP of contact, is not high enough, which cause delays in receiving a clear diagnosis. Secondly, the Netherlands has no care coordinator, while Germany and Belgium showed improved results with the implementation of the care coordinator. Parents indicated in the study of van Scheppingen et al. (2008) that parents prefer to have contact with one HCP instead of many HCPs. Additionally, having a single HCP which parents can contact, is conducive to the relationship. Furthermore, parents tend to be more honest with a trusted HCP (Pelentsov, Fielder & Esterman, 2016). Finally, patient organisations should be more involved in decision making. The VSOP in the Netherlands has actively contributed to the national action plan, while the Belgium organisation (RadiOrg) and the German organisation (ACHSE) have not. In the light of patient empowerment, it would be convenient for patients to implement their problems into the national plan.

Also, patients and parents have indicated that information about their disease is scarce. According to the framework, 80% require additional information and education about their condition. Yet, this problem of scarce information is a vicious circle: the amount of research is

scarce, because of the rarity of the disease and the disease is rare, because of the low awareness and the low awareness is because of the lack of research and the rarity of the disease. If countries promote research about rare diseases, this Informational Needs should not be a problem and the vicious circle will be broken.

Parents caring for children with a rare disease tend to create Psychological and Emotional Needs which have to be dealt with. Unfortunately, only the national plan of the Netherlands mention psychological care for the patient, while parents often have a higher prevalence of depressions than the patient (Kasparian et al., 2014). Moreover, according to the framework in total 50% require interventions for symptom management or psychosocial distress and in total 10-15% need even more intensive interventions.

All things considered, it can be said that the national plans of Belgium, Germany and the Netherlands are far from perfect. Information is lacking and the supportive care needs are not addressed properly in the national plans. A set of recommendations will be given in chapter

Discussion.

5. Discussion

For this thesis the research questions were: (1) What are the effects of the lack of supportive care needs of parents caring for a child with a rare disease; and (2) what are existing strategies in the Netherlands, Belgium, and Germany to fulfil the supportive needs of parents? These questions were answered with the help of two frameworks, a narrative literature review, and the national action plans of the three countries.

Despite the fact that the Parents Supportive Care Needs Framework (P-SCNF) is relatively new and has not been used elsewhere, the original Supportive Care Needs Framework (SCNF) of Fitch (2008) has been used in a variety of studies (Kerr et al, 2004; Kerr et al., 2007; Pelentsov, Laws & Esterman, 2015). The overall findings of both frameworks are that individuals that are affected by a disease, directly or indirectly, should be provided with supportive care. However, relationships between the certain domains of the SCNF has not been studied yet.

During the literature review, it became clear that parents will receive Emotional, Physical and Psychological Needs from the lack of Social, Informational, and Practical Needs. Stress and depression were one of the most found items in the literature review and also a significant amount of parents have reported that they received treatment or medicines for depressions or anxiety problems (Javalkar et al., 2017; Kasparian et al., 2014; Monterosso & Kristjanson, 2008). According to the P-SCNF and E. Pelentsov's explanation, Physical, Emotional and Psychological Needs are independent and are interrelated with the three other needs. Yet, I believe, due to the findings in Interactions Between Needs, that Physical, Emotional and Psychological Needs are not only part of Supportive Care Needs, but also the consequence of not receiving supportive care and the solution to the first research question. In other words, when a parent has an Informational Need, such as the need to receive information about their child's future health requirements, the parent will also perceive Emotional Needs, for example, stress or fear. Conversely, parents will not get stress or fear if they do not have Informational Needs in the first place. Therefore, to get Physical, Emotional and Psychological Needs there must be a trigger, an initiation from another need. Given these points, the supportive care needs are a one-way street and not interrelated as Pelentsov suggests. Moreover, to my knowledge, the data in graph 1 and 2 are the first of its kind. Based on these findings I suggest a modification of the Parent Supportive Care Needs Framework (page 38).

Instead of the previous version of Pelentsov's framework (page 4), not all needs are interrelated to each other. Instead, there are two groups of three needs: the group of Social, Practical, and Informational Needs, the "triggers"; and the group of Physical, Psychological, and Emotional Needs, the "outcome". I have chosen for two groups because each need is not separately a result of an outcome. For example, financial problems can lead to poor sleep and powerlessness; or, not receiving an early and definitive diagnosis in combination with isolation and loneliness can lead to stress. Thus, one or multiple triggers can result in one or multiple outcomes. Additionally, the reason that Practical and Psychological Needs are in the middle is due to the strong assumption that Practical Needs is the most important trigger, and Psychological Needs is the most impactful outcome.

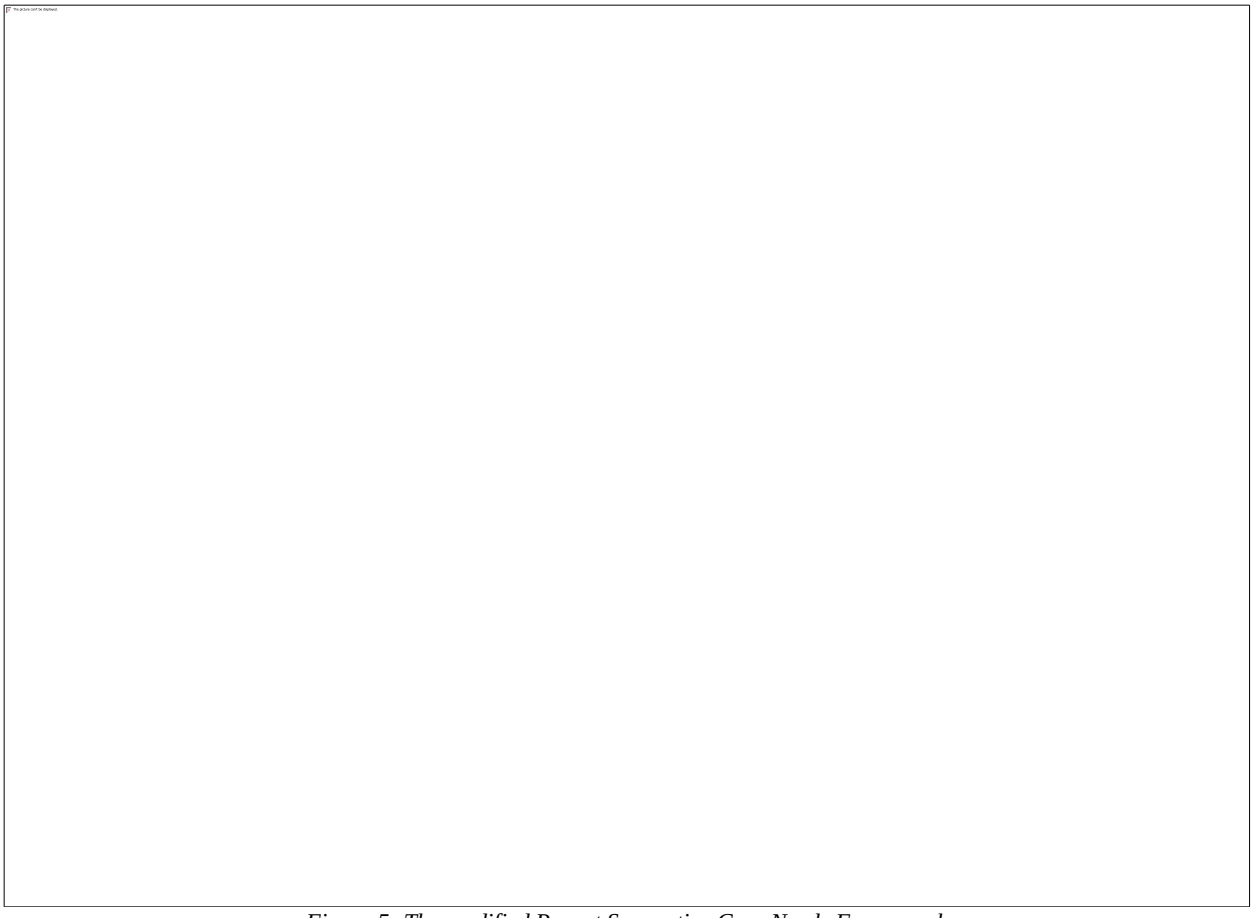


Figure 5: The modified Parent Supportive Care Needs Framework

It must be said that this modified framework is conceptual. Not only is this framework not significantly been tested, also, the framework is based on findings of a narrative literature review. Therefore, the framework must be further researched to find strong correlations between

the triggers and the outcomes. However, the framework can be used to understand the supportive care needs of parents better. Moreover, the findings are in line with the results of a report of Eurordis (EURORDIS, 2017). This report is based on a large quantitative survey with over 5,000 respondents. One of the main findings was that more than half of the participants preferred to receive support services and that there are many unmet needs in this area. The framework can serve as a guide or a tool to find the unmet needs, or triggers, that lead to Emotional, Psychological, and/or Physical Needs. Furthermore, this framework can also be used for other types of diseases, which are more common, such as chronic diseases, because the principle of a trigger and an outcome remains the same. However, the influence of certain domains will be different. For example, the influence of Informational Needs will be less, compared to rare diseases, since the rarity of a disease is mostly an indication of the amount of information available.

As been mentioned in section Comparison the national action plans are far from perfect for parents caring for a child with a rare disease. Yet, the action plans only cover the bigger picture of rare diseases and does not cover any special supportive care plans, while it can be assumed that supportive care is provided in the centres of expertise. A more in-depth research would probably acknowledge this. Furthermore, the action plans of Belgium and Germany were dated from 2013, while the Netherlands already made a reflection on their national plan (ZonMw, 2017). However, on the 6th of July 2017, a definitive set of recommendations were presented to the government of the Netherlands (VSOP, 2017). These recommendations were composed on Rare Disease Day (28th of February 2017) in the Netherlands and consist of nine focus points with several sub-items. One of these items (4.3) mentions the need for research to the psychosocial aspects of patients and their relatives. Thus, the government is now aware of the lack of the knowledge on supportive care and hopefully more research on this subject will be encouraged.

5.1 Recommendations

The amount of awareness of patients with a rare disease is increasing. The amount of research about rare diseases is expanding and correspondingly, the amount of information is growing. However, the supportive care needs of parents – and caregivers – are neglected in research and national action plans. To improve the supportive care given to parents, not only more research

has to be conducted, also parents should be involved with national action plans. Moreover, parents often receive supportive care too late, which can result in Physical, Psychological, and Emotional Needs. Therefore, a psychological assessment should be performed on parents to identify supportive care needs the moment a child is referred to a centre of expertise. A certain amount of supportive care needs can be avoided if HCPs became more aware of rare diseases and their burden in general. The lack of knowledge was one of the foremost reasons of delayed diagnoses.

5.2 Limitations

A major limitation of this study could be that also other diseases were included in the narrative literature review. However, a number of articles that included rare diseases and the burden of the parents were scarce. Yet, for future studies more chronic diseases and rare disease articles will be available, and should be used to see the interactions between the triggers and the outcomes of supportive care needs. Also, the counting of all the items was by one person and was subjective to inclusion bias. Moreover, the severity of a single item was not measured, thus, one article with a severe supportive care need weigh the same as an article with a mild supportive care need. Furthermore, an additional article would have slightly changed the graphs, because there were only 18 articles in the narrative literature review. On the other hand, not only were the number of articles on rare diseases scarce, also the time limit and ensuring the quality of the articles did contribute to the number of articles. As said before, the P-SCNF has not been used in other studies, which could be another limitation. Yet, the original SCNF has been proven to be a solid framework.

5.3 Suggestions for Future Research

As said above, the framework should be improved on the significance of the interactions. While this interaction between the triggers and the outcome was unintendedly found, further research could provide more and new information about supportive care needs of parents. Also, further implications of the lack of supportive care needs should be investigated.

Also, a more comprehensive approach to the strategies on rare diseases would provide in-depth information about the supportive care given in centres of expertise and other facilities. Further research on the approaches of supportive care should include interviews with patients and care

coordinators because this would provide inside information about supportive care since the national action plans are only the tip of the iceberg.

5.4 Conclusion

This thesis made a start on the unmet needs of parents caring for a child with a rare disease. Emotional, Psychological, and Physical Needs are seemingly the result of the lack of Informational, Practical, and Social Needs. These preliminary results were the base of a modified framework that could help HCPs in the guidance of parents through the difficult process with their child. Moreover, the framework can help parents to overcome the unmet needs which is eventually also beneficial for the child. The strategies proposed by Belgium, Germany, and the Netherlands show the general lack of knowledge of rare diseases and supportive care needs. However, the national action plans provide a sufficient foundation to support new technologies and researches. Yet, the focus of most researches is understandably concentrated on the patient and their rare diseases, though caregivers and parents caring for a child should not be neglected and further research on supportive care needs will help the burden of caregivers and parents be more acknowledged.

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