Journal of Pediatric Nursing (2015) xx, xxx–xxx

ELSEVIER

The Supportive Care Needs of Parents With a Child With a Rare Disease: A Qualitative Descriptive Study

Lemuel J. Pelentsov BHSc, GDip, BN [a,*], Andrea L. Fielder PhD, BSc [b], Adrian J. Esterman PhD, MSc, BSc, DLSHTM, PACE [c]

[a] *School of Nursing and Midwifery, Division of Health Sciences, University of South Australia, City East Campus, Adelaide, SA, Australia*

[b] *Sansom Institute for Health Research and School of Nursing and Midwifery, Division of Health Sciences, University of South Australia, City East Campus, Adelaide, SA, Australia*

[c] *Sansom Institute of Health Service Research and School of Nursing and Midwifery, Division of Health Sciences, University of SA, Centre for Chronic Disease Prevention, James Cook University, Cairns, QLD, Australia*

Received 17 September 2015; revised 27 October 2015; accepted 28 October 2015

Key words:
Rare diseases;
Parents;
Children;
Supportive care needs;
Experiences

There are few studies that exist which focus specifically on parents with a child with a rare disease. The purpose of this study was to better understand the lived experiences and supportive care needs (SCN) of parents caring for a child across a spectrum of rare diseases. A qualitative descriptive approach was used to guide the research, and four semi-structured focus group interviews were conducted with 23 parents (17 mothers and 6 fathers). Participants described 'feeling boxed-in outside the box' due to a number of limitations unique to their child's disease, daily practical challenges in providing care and the various relational impacts of caring for a child with a rare disease were discussed. The results from this study help to give clearer direction for health professionals on where to focus future efforts in better meeting the supportive care needs of parents and their child with a rare disease.

RARE DISEASES ARE a large and diverse group of life-threatening or chronically-debilitating illnesses, the majority of which originate in early childhood, are genetically-based, and have no cure, existing pathways of care or appropriate medical interventions (Aymé & Schmidtke, 2007; de Vrueh, 2014; Eurordis, E. O. f. R. D., 2005). The definition of rare diseases varies globally. The United States of America (USA) defines rare diseases as diseases that affect fewer than 200,000 persons, Europe (EU) less than 1 in 2000, and Australia less than 1 in 10,000 (Zurynski, Frith, Leonard, & Elliott, 2008), though many individuals suffer from diseases far less frequent. Approximately 8000 rare diseases exist, many of which have no formal title and are difficult to diagnose (Bellgard et al., 2013; Denis, Mergaert, Fostier, Cleemput, & Simoens, 2010). Although individually uncommon, collectively rare diseases are considered a global public health concern, with approximately 6–10% of the world's population affected (Schieppati, Henter, Daina, & Aperia, 2008). Repeatedly, rare diseases are overlooked by medical, scientific and political communities as they are considered a diffuse minority. Yet, increasing epidemiological and scientific data exist to show that rare diseases are an important public and social issue that poses significant challenges to communities (Schieppati et al., 2008). Despite this, few studies exist describing the impact of rare diseases on the lives of individuals, their family, health professionals and healthcare services (Zurynski et al., 2008). This study, focuses on the supportive care needs of parents caring for a child with a rare disease.

Parents caring for a child with a rare disease face significant multidimensional daily challenges. Their

* Corresponding author: Lemuel J. Pelentsov, BHSc, GDip, BN, Lecturer.
E-mail addresses: lemuel.pelentsov@unisa.edu.au, andrea.fielder@unisa.edu.au, adrian.esterman@unisa.edu.au.

http://dx.doi.org/10.1016/j.pedn.2015.10.022
0882-5963/

commitment to caring for their child will span many years and involve a lifetime of sacrifice (Barlow & Ellard, 2006; Nuutila & Salanterä, 2006). Providing appropriate care for their child results in changes to work patterns, income and domestic responsibilities. Parents often require additional specialist health literacy, care giving skills and resources beyond normal requirements to provide effective parenting (Kirk & Glendinning, 2004; Sartore, Lagioia, & Mildon, 2013). Although parents of children with chronic health problems face similar issues, parents of children with rare diseases face additional problems as diagnosis can be delayed or undetermined, support groups (if in existence) are geographically scattered and healthcare skills and resources limited (Jaffe, Zurynski, Beville, & Elliott, 2010).

To-date, few studies have investigated the supportive care needs of parents with a child with a rare disease. Those that have, typically are qualitative studies that focused on a specific disease, involved small sample sizes or are limited to a particular country or culture (O'Connor & Hemmings, 2014). Parental support needs most frequently discussed in the literature relate to: parents feeling socially isolated and needing more support (Aytch, Hammond, & White, 2001; Coffey, 2006; Duffy, 2011; Pelentsov, O'Shaughnessy, Laws, & Esterman, 2014; Speraw, 2006), need for general medical information regarding child's disease (Fisher, 2001; Graungaard & Skov, 2006; Hendriks, De Moor, Oud, & Franken, 2000; Kerr, Harrison, Medves, & Tranmer, 2004; Kerr, Harrison, Medves, Tranmer, & Fitch, 2007), financial concerns and healthcare costs (Gallo, Hadley, Angst, Knafl, & Smith, 2008; Goble, 2004; Nahalla & FitzGerald, 2003; Neil-Urban & Jones, 2002; Zierhut & Bartels, 2012), and the physical and emotional burden of caring for a child with a rare disease (Glenn, 2015; Hummelinck & Pollock, 2006; McGrath, 2001; Strehle & Middlemiss, 2007; van Scheppingen, Lettinga, Duipmans, Maathuis, & Jonkman, 2008; Yiu & Twinn, 2001). Until recently, studies which have attempted to identify the supportive care needs of parents in a comprehensive fashion, have been limited to childhood cancer (Kerr et al., 2004; Kerr et al., 2007). Although the past three decades has seen significant gains in research being conducted into rare diseases, mainly clinical research (Batshaw, Groft, & Krischer, 2014), studies specifically exploring parental supportive care needs remain scarce. A recently published review by the current authors sought to gather and appraise all available published literature investigating the supportive care needs of parents with a child with a rare disease (Pelentsov, Laws, & Esterman, 2015) and was the first to provide a detailed synopsis of parental supportive care needs in rare diseases irrespective of disease. In this literature review, it was found that although the affected children had very diverse health problems and needs, the supportive care needs of the parents were in fact quite similar. Given the paucity of research, the current study sought to answer the following research question: what are the supportive care needs of parents caring for a child with a rare disease, and are they similar over all types of rare diseases? It is envisaged that this information will lead to more appropriate individualised supportive care for parents, and improve the way health providers identify needs, tailor support and plan and implement services for the rare disease community. The qualitative study we report here will contribute to addressing this gap.

Method

Aim

The aim of the current study was to better understand the experiences and supportive care needs of parents caring for a child with a rare disease.

Study Design

A qualitative descriptive approach was used for this study as described by Sandelowski (2000), because we desired a straight description of the phenomena. Qualitative descriptive studies are considered less interpretive than other forms of qualitative research (e.g., phenomenology) and produces findings that are close to the data, and of low inference (Sandelowski, 2010). We do however, note that many of the analytic techniques recommended by Sandelowski, such as the use of codes and thematic analysis, overlap with other philosophical approaches such as grounded theory and phenomenology. In this study, we ensured that analysis and interpretation of findings remained data-near and are representative of the views and experiences of the participants. Focus groups were chosen over individual interviews to provide participants with an opportunity to use the responses of others to engage and stimulate deeper discussion.

Sampling and Recruitment

Participants were included if they were a mother or father of a child (aged 18 years or younger) diagnosed with a rare disease or whose child was suspected by health professionals or the parents of having a rare disease but not formally diagnosed. The latter inclusion criterion was implemented as many parents have an affected child with no formal diagnosis. A mixture of purposeful, convenience and snowball sampling techniques was used to recruit sufficient numbers, given the difficulty in recruiting populations associated with rare diseases (Griggs et al., 2009; O'Connor & Hemmings, 2014), whilst also representing a wide range of variation between participants. Whilst participants may share similar experiences and needs, they may also have distinct differences (e.g. level of support) depending on the type and severity of their child's rare disease. Because of the difficulty of recruitment, a pragmatic approach was taken to include any parents available to participate in the focus groups, including parents of children with both diagnosed and undiagnosed conditions. Parents of children with cancer were also included since all childhood cancers are considered rare diseases (Kotecha, Kees, Cole, & Gottardo, 2015) and also included parents of children with cystic fibrosis (1:3000) (Fajac et al., 2013). Although the latter is not formerly