

RESEARCH

Being a Mother of a Child with Prader-Willi Syndrome: Experiences of Accessing and Using Formal Support in Croatia

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The aim of this qualitative study is to obtain insight into the experiences of mothers of children with PWS with the formal support system in Croatia. The study was conducted in 2013 with five mothers of children with PWS. Thematic analysis was used as the analytic method. The results highlight four major themes related to mothers' experience of accessing and using formal support: professionals' lack of knowledge about PWS results in living without support; professionals' lack of empathy; professionals' commitment to improve the well-being of families; and getting support through membership in the Association of Persons with PWS Croatia. The social support in Croatia is defined by the lack of knowledge and lack of empathy of professionals, which results in disrespect for the human rights and dignity of children with PWS. Raising the awareness about PWS is crucial for improving the quality of life of families living with PWS.

Keywords: Mothers of children with PWS; Social services; Health care system; Living with rare disease

Introduction

Rare diseases are mostly inherited and life threatening (Knight and Senior 2006; Grut and Kvam 2013; European Commission 2015), with prevalence that is five out of every 10,000 persons (Orpha.net 2012). One of them is Prader-Willi Syndrome (PWS) with an estimated prevalence of one out of every 10,000–30,000 persons (Yearwood et al. 2011; Cassidy et al. 2012; Orpha.net 2014). It is found in all races and both genders (Dudley et al. 2008; Yearwood et al. 2011). The major characteristics of PWS are developmental and cognitive delays (mild to moderate intellectual disability), dysmorphic features, hypogonadism, short stature, and a characteristic facial appearance (Cassidy 1997; McCune and Driscoll 2005; Cassidy and Driscoll 2009; Cassidy et al. 2012; Miller 2012; Paschos, Bass and Strydom 2014). Clinical features include infantile lethargy and hypotonia (weak muscle tone) alongside poor suckling and feeding difficulties in early infancy (Cassidy and Driscoll 2009, Cassidy et al. 2012), as well as hyperphagia during early childhood (Key and Dykes 2008; Cassidy et al. 2012; Miller, Lynn, Shuster and Driscoll 2013; Dimitropoulos, Ho and Feldman 2013). Dysregulation of appetite-satiety patterns may cause obesity (McCune and Driscoll 2005, Dudley et al. 2008; Cassidy and Driscoll 2009; Yearwood et al. 2011; Miller 2012) which can be potentially life-threatening (Mazaheri et al. 2013). Typical behavioural phenotype includes temper tantrums, stubbornness, manipulative, compulsive and oppositional behaviours, aggression, self-injurious behaviour, and sleep abnormalities (Cassidy and Driscoll 2009; Wulffaert, Scholte and Van Berckelaer-Onnes 2010; Cassidy et al. 2012). These problems result in lower quality of life (Caliandro et al. 2007; Driscoll et al. 2014).

Most of the studies on PWS focus on its medical aspects. Different studies on social aspects of PWS can provide a better understanding of life quality of families living with PWS. They can also be used to provide more adequate formal social support or to obtain insight into daily routines of families living with PWS. For instance, parents of children with PWS place great efforts in dietary control, management of behavioural problems or keeping children engaged in an activity (Allen 2011; Dimitropoulos et al. 2013; Pignatti et al. 2013). That is why social support is important for them. There are two aspects of social support: emotional (listening, counselling, and providing encouragement) and informational (knowledge, information, and advice) (Young 2006; Guralnick et al. 2008; Tsai and Wang 2009). Family members, friends, neighbours, and colleagues provide informal support (Leutar, Ogresta and Milić Babić 2008), while formal support involves healthcare professionals, social workers, teachers, professional organisations, and support groups (Tsai and Wang 2009). Formal support is crucial for the well-being of families of children with disabilities (Leutar et al. 2008; Orgassa 2008) because it determines their quality of life (Pansini

2011). Although medical and non-medical professionals should support parents of children with PWS (Caliandro et al. 2007), different studies highlight that professionals made up only a small proportion (8%) of the support system (Hodapp et al. 1997). Parenting a child with PWS often results in pessimism (Hodapp, Dykens and Masino 1997).

It is very often the case that people with rare diseases are not recognised as potential users of the social welfare system in Croatia. Some of the most important proposals and guidelines from the First National Conference on Rare Diseases in Croatia (2010) are to improve access to health care and social services, to protect patients' social rights and the right to multidisciplinary approach to care (Rodwell and Aymé 2014). According to McGravey and Hart (2008), families living with a rare disease generally use generic disability services. Although there are three Referral Centres for rare diseases in Croatia, there is no national registry of rare diseases (Rodwell and Aymé 2014). This is the result of a lack of a classificatory process in health care, while a lack of specific social care services results in inadequate social care (Ministry of Health 2015). For example, in 2014, there were 3,279 parent caregivers, 579 children were in an early intervention programme, and an integration programme involved 368 children with developmental disabilities in Croatia (Ministry of Social Policy and Youth 2015). It is not known if some of them are children with PWS. Mrcić and Nola (2008) state that in Croatia most physicians do not meet with a single patient who has a rare disease during their entire career, which results in the lack of special treatment and low awareness about rare diseases. The invisibility of families living with PWS in Croatian society is often the result of professionals' lack of knowledge and leads to limited support.

Methods

A qualitative research approach is particularly useful for fields in which little is known about phenomena under investigation (Loberg 2009). This approach can provide insights into participants' perspectives about the phenomenon and highlights what is important for participants (Berkwits and Inui 1998; Chambers 2007). There is a lack of knowledge about mothering a child with PWS and support, which is provided by the formal support system. In Croatia, the formal support system includes salaried professionals working in healthcare, social care, education, religious organisations, NGOs, etc.

The aim of this qualitative study was to obtain insight into the experiences of mothers of children with PWS with the formal support system in Croatia. The research question was 'What are mothers' experiences of accessing and using formal support services?' Their experiences can provide better understanding of social aspects of PWS.

This study was conducted in 2013 with five mothers of children with PWS. Two of them live in the capital city, Zagreb, and the other three live in rural and urban locations in the eastern part of Croatia, namely Osijek-Baranja County. Participants' age ranged from 28 to 62, while the mean age was 44. Two of them are single mothers, and three are mothers of an only child. Four of them graduated from high school, and one graduated from college. Their children are 6, 10, 11, 13 and 35 years old.

The sample was purposive. The lead author contacted the president of the Croatian Alliance for Rare Diseases and the president of the Association of People with PWS to obtain contacts of potential participants. According to the Ethics Committee of the Faculty of Law in Zagreb, ethical approval in social work research must be obtained when participants are children or people under guardianship and the topic is very sensitive. This study did not need ethical approval because it did not include sensitive personal data, the topic was not very sensitive, and the participants were not vulnerable. Regardless, this study was conducted in accordance with ethical principles. The lead author conducted all interviews in participants' homes. Complete information on the research (voluntary participation, confidentiality, data anonymization and protection, the right to withdraw from research at any time) was provided to the participants before each face-to-face interview. After being informed they gave verbal informed consent. The interviews were digitally recorded and lasted approximately 60 minutes. To achieve confidentiality and data anonymization, the lead author transcribed all of the interviews.

Data analysis

The analytic method in this study is thematic analysis. It is used for identifying, analysing and reporting themes within data (Braun and Clarke 2006; Fereday and Muir-Cochrane 2006; Vaismoradi, Turunen and Bondas 2013). The phases of thematic analysis are the following: familiarisation with data, generation of initial codes, searching and reviewing themes, defining and naming themes, and producing the paper (Braun and Clarke 2006). First, the material was transcribed. Each author read and reread the transcribed material to familiarise themselves with the data. During the process of reading, the authors coded interesting features of the data from each individual transcript. The aim was to define as many potential codes as possible. The codes were sorted into potential themes. These themes were reviewed, defined and named. The thematic map was constructed to define relations between them. The most descriptive quotes were selected for illustration of the defined themes. Investigator triangulation was made to ensure the trustworthiness of the results (Leech and Onwuegbuzie 2007).

Results

The results concerning accessing and using formal social support are presented in chronological order based on participants' need for formal support services: participants' experiences of accessing and using support provided by obstetricians on a maternity ward; support provided from physicians, social workers and teachers when PWS was diagnosed; and lastly support from mothers who also have children with PWS.

Professionals' lack of knowledge about PWS results in living without support

The theme of professionals' lack of knowledge about PWS is based on participants' experiences of accessing and using support provided by health and social care professionals during their children's infancy and early childhood. The participants' first experiences with the formal support system demonstrate obstetricians' lack of knowledge about PWS:

At the maternity ward I asked to speak to a paediatrician and I told him that something was wrong with my child, but he said that everything was normal – it had just been a difficult birth. They told me I was talking nonsense, and my child was simply suffering the consequences of labour. I saw that she could not suckle. (4)

I had a caesarean so I had not seen her for three days. She was in the intensive care ward, and I was in the maternity ward. I could not go to see her because I was on an IV – I could not go as long as I was on it – and then, after three days, I stood in front of a nurse and asked if I could see my child, even though no physician had said a thing to me. My husband came and told me our daughter was in intensive care and they did not know what was wrong with her. The obstetricians told me that he thought she would be able to suckle the following day and that was it. That is literally all the information I got. In fact, during those 14 days in hospital, we did not get any quality information, but – her heart stopped beating, she had bacteria, all in all... nothing. I saw she was different from other children, but no one said anything. No information, no support from obstetricians. (3)

Diagnosing PWS undeniably requires a certain period of time. However, the characteristics of the newborn described above should have been clear indicators for the obstetrician signalling deviations in development. The inability to suckle and hypotonia are indicative signs of possible PWS. In addition to their lack of education, the obstetricians failed to perceive the relevant characteristics of the newborn and reassured mothers, stating that they exaggerated the infants' conditions. According to experiences of two participants, the diagnostic procedure concerning PWS has not been changed in two decades. One of them also stated that she did not have any support from the health care system over the entire period of the child's first 8 years of life. Physicians who are knowledgeable about PWS work in the capital city exclusively, so their support depends on mothers' financial and organisational opportunities:

No one here in my town was able to recognise it. When we arrived in the capital city, the physician said it was PWS the moment she opened the door. (2)

Of the 20 specialists I asked for information, I found one who knew about PWS and made an effort to provide me with some relevant information. She was a geneticist and explained the developmental process to me, what to pay attention to. (1)

Social workers showed a lack of knowledge:

Support – oh, how hard I find it to say that word. For no one ever wanted to say anything to me about all the rights I thought my son had until he was 10. He is beautiful, he looks so nice, and that was what I was told about him. They did not know what PWS was – one of them last year said she saw a documentary on PWS on TV and that she understood the condition then. Financial support, nil. I gave up on it totally when he was five. (5)

Croatian social workers do not recognise PWS as a developmental disability and consider it as condition that can result in disability because they are not educated about rare diseases. On this account, children with rare diseases often cannot be provided with adequate social care. Two participants stated that they were not able to initiate the process of obtaining the status of parental caregiver, resulting in their children being unable to exercise the rights and access services accorded to children with disabilities because they look 'healthy'. Inadequate support resulted in parents' activism in gathering information and sharing knowledge. The participants began to educate and raise physicians' awareness about PWS:

There was not one text in Croatian to be found. I translated some foreign literature. Later, I used to buy books and distribute them to physicians. I lacked help from physicians. Today they have made a little progress. They know something about PWS. (1)

Whenever the physician thought I had some experience that could be useful to others, he sent people to see me. I educated people in some way. (4)

It is not questionable that support between parents of children with PWS is very important, but participants' experience indicates that they were left to their own devices. They were presented with the responsibility for supporting and educating other parents of children with PWS.

Professionals' lack of empathy

Participants' experiences about professionals' lack of empathy refer to support provided by physicians or teachers when PWS was diagnosed. They described how physicians exhibited a lack of empathy, and dehumanising tendencies, toward their children when they delivered the diagnosis:

I was told 'He has something. It's called PWS. It is a syndrome and we do not know anything about it'. The feeling was such that for two days I wanted to throw myself out the window. If there had been no bars on the window, who knows what I could have done. You know what you have to cope with – you have no instructions, no ray of hope. They told me to leave him in an institution because nothing would come out of it – it would just ruin my life. (1)

All participants stated that they received no support when receiving diagnosis although it was, emotionally and socially, an extremely sensitive moment associated with parental acceptance of the child's condition:

The physician is the one who first suspected PWS and she asked me very coldly why I was crying, since my child would die in about 6 months anyway, and then she turned and walked out the door, and I remained raving. (5)

In Croatian society physicians very often take an authoritarian approach, one based on their social status. They do not provide patients with support and abandon them to deal with the diagnosis by themselves. The participants' description of their emotional reaction to the disclosure of the diagnosis indicates a fatalistic reaction to a PWS diagnosis. These experiences come from participants whose children are the same age.

Their experience with the educational system indicates a perceived undesirability of the child attending a mainstream school, no investment in the educational process, and resistance:

They only wanted to move him out of school as soon as possible. If you cannot change a teacher, change the environment. In another school, we also experienced resistance. Support, no. If a teacher does not have a single positive word about him, it means she does not accept him as a person. I find it very difficult for him. She told him he could not learn how to write, and then I paid for a teacher who taught him. She would not give him a song to learn or let him perform at a school event. Terrifying things, those are too painful for me. (5)

Professionals' commitment to improving the well-being of families

In certain instances inadequate support became satisfactory after a proper diagnosis of PWS had been made:

Never mind the maternity hospital department. Now one physician is trying to improve her quality of life, regularly checks up on her, takes care of her, and gives her hormones. I trust her because she treats my daughter as if she were her own child. (5)

Physicians' commitment is marked by meeting families' needs, monitoring the child's condition and the fact that they possess special knowledge of PWS. The support from social workers is described as a potential source of financial aid:

Speaking of finances, I cannot complain – we get disability benefits. I get disability benefits from Social Welfare and that is it – since we are given no other rights. (3)

The Croatian social welfare system grants children with developmental disabilities access to realising different rights or different services only if the disability is classified as severe or profound. Rare diseases are not even listed as causes of serious or severe disabilities in regulations that establish social rights. Therefore, social workers working in centres for social welfare are recognised as sources of emotional support through empowerment and provision of information about rights:

Social workers have been of great help from the beginning. These were the people who were very sympathetic. They listened to me, and the physicians – they do not listen. They have always encouraged me and suggested a way of coping. They always fought by my side on the issue of rights. (1)

Another participant noted that she was informed about the right to work part-time, which enabled her to be much more dedicated to her child. It should be noted that all of the participants stated that they received financial and emotional support primarily from the members of their immediate family, such as parents, sisters, brothers or sisters-in-law. They described the involvement of their own parents as total commitment or the maximum subordination of their own life. In addition, two participants stated that teachers were also engaged by controlling the diet of their children since it is one of the main challenges in caring for children with PWS:

He gets full support from the teacher – she's my friend, his second mother. She looked into the syndrome thoroughly, all of the children in the class are educated about it, there is no food in the classroom, no one talks about food and no one eats in front of him. When he eats an apple for snack, only his teacher keeps him company. She knows how to avoid his psychological outbursts. (1)

Obtaining support through membership in the Association of Persons with PWS Croatia

The support gained by means of membership in the Association of Persons with PWS Croatia represents a kind of a bridge from informal to formal support. It represented support from other mothers of children with PWS. The membership enabled realisation of growth hormone treatment for two participants. Through providing emotional and informational support, which they call moral support, the Association also provides them with safety:

We call each other and it makes it all easier for me because I did not know anyone before to share my experience with when all of this happened. It is a lot easier when you can share it with someone, to know that I'm not alone or the only one with this problem. I get moral support from the Association. (2)

This association does not really function as it should. We just talk to each other, help each other –it is more a kind of moral support. There is nothing else we could do. (5)

The last statement shows that sometimes moral, emotional and informational support is not enough. It indicates that some mothers of children with PWS may have greater expectations from the association, not only to receive support from mothers who have similar experiences.

Discussion

The results of this study provide an answer to the research question about mothers' experiences of accessing and using formal support services in Croatia. The first result of this study is that professionals' lack of knowledge of PWS results in living without support. According to the results of some earlier studies, the lack of knowledge on the part of obstetricians or paediatricians results in failure to achieve an early diagnosis and early multidisciplinary care, which could greatly improve the health and quality of life of new-borns with PWS (Cassidy and Driscoll 2009; Lovell and Mason 2012; Grut and Kvam 2013). Furthermore, mothers of children with rare diseases or developmental disabilities experienced a lack of knowledge as exhibited by physicians (Chaij, Han and Graziano 2014; Glenn 2015). Physicians provide them with inadequate and inaccessible information about their children's health condition or support they can obtain (Alvarez McHatton and Correa 2005; JCHR 2008; McGarvey and Hart 2008; Edwardraj et al. 2010; Glenn 2015).

The results of this research are virtually the same. Some of the participants were convinced that their children did not have health problems or difficulties, claiming that hypotonia and a poor sucking reflex were results of childbirth. Indisputably, there are many causes of hypotonia, although hypotonia and a poor sucking reflex are indicative signs of PWS (Cassidy and Driscoll 2009; Chaij et al. 2014; Paschos, Bass and Strydom 2014). The diagnosis of a rare disease is less definite (Picci et al. 2015), but 99% of PWS cases can be diagnosed with a simple molecular test, DNA methylation (Cassidy and Driscoll 2009). The participants in this study did not receive any information about growth hormone treatment which improves growth, physical phenotype and body composition in children with PWS (Cassidy and Driscoll 2009; Mazaheri et al. 2013). The results of different studies also demonstrate that mothers were told there would be no improvement in the child's condition (McGracey and Hart 2008), and suggested that an institutional setting would be the best solution for their children (Leutar 2012). All of these results highlight the extent to which physicians' lack of knowledge determines the quality of life.

The results of some earlier Croatian studies on parenting children with developmental disabilities also show physicians' lack of specific knowledge concerning disability, lack of support when the diagnosis is provided, failure to provide information or implement an early intervention programme, as well as inadequate support as exhibited by physicians and social workers (Leutar 2012; Milić Babić, Franz and Leutar 2014). The majority of parents (34%) think physicians did not provide them with enough information about their children's health (Cazin, Cindrić and Piščenec 2014). According to experiences of two participants in this study, over the last decade there appeared no positive differences and changes in accessing formal support or concerning awareness of PWS in Croatia. These results are very similar to the results of some earlier Croatian studies (Lisak 2013, 2014). Based on experiences of participants in this study, the only improvement in health care is the fact that there are only a few physicians with adequate knowledge of PWS working in the capital of Croatia. If mothers want to book an appointment with them, and they live in the county in which participants in this research reside, they have to travel over 186 miles one-way for an appointment with them. These physicians can empower mothers and act on their behalf after PWS is diagnosed (Knight and Senior 2006). Pelentsov, Laws and Esterman (2015) state that need for information regarding a child's condition and need for support from health professionals were the most common social and informational needs of parents caring for a child with a rare disease. Additionally, physicians' lack of action can be based on their lack of knowledge about PWS or it can be related to the fact that the condition is labelled "rare" (Grut and Kwam 2013).

One of the results of this study is also about social workers' lack of knowledge about PWS. In Croatia, children with disabilities can gain access to their rights in the social welfare system only if a rare disease results in a severe or profound disability. Children with PWS cannot access early intervention services because Croatian social welfare system does not recognise them as children with developmental disabilities. Early intervention is aimed at children under the age of three, exceptionally up to seven years of age (The Social Welfare Act 157/13, 152/14, 99/2015).

International and domestic law guarantee early intervention as one of children's rights (Convention on the Rights of Persons with Disabilities 2006; Schlumpf et al. 2006; Cassidy and Discroll 2009; Driscoll et al. 2014; The Social Welfare Act 157/13 152/14 99/2015). Moreover, the system of formal support is obliged, according to domestic and international legal regulations, to ensure that children with disabilities have access to rights and services (UN 2006; The Social Welfare Act 157/13 152/14 99/2015). Children with PWS cannot gain just mentioned rights because for the social welfare system they do not have developmental disabilities. So can we talk about prevention of disability, adequate social support or early intervention if children with PWS are not recognized as potential users? One of the problems is the lack of an official database about children with rare diseases in Croatia. When seeking information on the exact number of people with PWS in Croatia, in conversation with employees of the Croatian Institute of Public Health, the received information was that the Office does not have data on persons with PWS. The answer was that in the last 25 years not a single hospital has submitted information on the birth of a child suspected to suffer from PWS. This kind of database could improve quality of life and services form families living with PWS.

Professionals' lack of empathy is another result of this study. Different studies show that mothers of children with developmental disabilities often deal with rudeness, indolence, emotionlessness, lack of empathy, inadequate support and dehumanisation of children from social workers and physicians (Huyard 2009; Tsai and Wang 2009; Pansini 2011; Chaij et al. 2014). They felt a lack of professionals' interest in their child's condition (McGracey and Hart 2008; Tsai and Wang 2009; Pansini 2011; Chaij et al. 2014). The results from Leutar and Oršulić's study (2015) shows that parents of children with developmental disabilities estimated that centres for social welfare and schools are not important sources of support in their lives (Leutar and Oršulić 2015). The participants in the Blaži and Kolarić study (2015) stated that professionals are not emphatic and do not provide systematic information how to achieve social rights for their children. Power-related aspects of the physician-patient relationship can be one of the explanations about professionals' lack of empathy. This kind of imbalance of power between physician and mothers of children with PWS can be based on assumed knowledge and expertise of physicians, or the fact that families of children with PWS need support and help. In addition, social status in Croatian society of physicians and other experts can be one of the reason of this kind of behaviour.

Mothers of children with rare diseases expect health professionals to express their awareness of their limited knowledge, and to be interested in their worries and aspirations (Huyard 2009). They need well-equipped and highly knowledgeable professionals (McGravey and Hart 2008; Allen 2011; James 2013), and action-oriented and ability-related information about life expectancy, care and the consequences of rare disease (van den Borne et al. 1999; McGravey and Hart 2008; Cassidy and Driscoll 2009; Huyard 2009; Dimitropoulos et al. 2013; James 2013; Mazaheri et al. 2013; Chaij et al. 2014). Picci et al. (2015) conducted a study that highlights the importance of training and information-providing of physicians for parents of children with rare diseases. The results from this study show that participants did not receive, in the most cases, that kind of support during the early years of their children. Maybe the lack of knowledge and lack of empathy can also be explained by the fact that medical experts are provided with little or no information about rare diseases during their formal education in Croatia. Lack of support resulted in developing mothers' own resources and skills for achieving information and knowledge about PWS. Most parents of children with rare diseases use the internet to access information about rare diseases (McGravey and Hart 2008; Glenn 2015). So can we talk about support from helping professions if mothers of children with PWS feel they are all by themselves in many aspects of their lives?

This study also demonstrates that a different kind of support was more adequate after an accurate provision of diagnosis. Professionals' commitment to improving the well-being of families is also one of the results of this study. The participants' experiences show that there are physicians, social workers and teachers who expend great effort to provide adequate support for them. This result is similar to the results of some earlier studies (Hoddap et al. 1997; Leutar 2012; Milić Babić et al. 2014) concerning a small number of physicians and social workers who are committed to improving the well-being of families and providing emotional and informational support to mothers of children with developmental disabilities. Results obtained by McGravey and Heart (2008) demonstrate that social workers informed mothers about rare diseases, supported them and intervened on their behalf with service providers. Formal support was provided by teachers in regular and special schools (Tsai and Wang 2009; Chaij et al. 2014), while this study shows that it was determined by a close mother-teacher relationship and the teacher's intrinsic motivation to learn about PWS and how to control dietary habits. Health of child with PWS depends on a restricted diet and strict food routines (Davies et al. 2012). Support which is described in this study resulted in improved quality of pupil's life, better inclusion in the classroom and improved class awareness of PWS. Teachers in Croatia are not provided with knowledge of rare diseases. Their education about children with developmental disabilities is basic. One of the problems is that lack of coordination between social and medical services results in inadequate assistance for mothers and prevents them from participating in economic and social activities in their communities (WHO and World Bank 2011). Lewis, Skirton and Jones (2010) show that communication between mothers and medical professionals was positive although there was a lack of communication between professionals themselves.

The results of this study also demonstrate that participants receive support through membership of the Association of Persons with PWS Croatia. The NGO sector plays a great part in supporting people with disabilities in Croatia.

This study also points out the importance of emotional and informational support, which participants received on account of their membership in an NGO. It represents a kind of a bridge from formal to informal support because it is a parental association. Mothers of children with PWS and other developmental disabilities appreciate meeting mothers of other children living with the same disabilities (van den Borne et al. 1999; Huyard 2009; Pansini 2011; Glenn 2015). It provides them with an opportunity to learn about coping strategies (van den Borne et al. 1999), and to network with other families facing the same challenges (Chaij et al. 2014). For example, mothers who participated in Glenn's study (2015) used online community venues to communicate, to seek information and to gain emotional support. Peer-to-peer support for mothers of children with PWS is very important because mothers of children with rare diseases experience stress related to a lack of expertise, social isolation and emotional demands (Dellve et al. 2006). Some earlier studies (Hoddap et al. 1997; WHO and World Bank 2011; Milić Babić 2012a, b; Lisak 2014) also demonstrate that the most significant support mothers' gain is from their family members and friends. The formal support system has only a partial role in the lives of families coping with PWS, and such is the case in the lives of people and families coping with any kind of disability. Considering this, some of the measures in the Croatian National Plan for Rare Diseases 2015–2020 (Ministry of Health 2015) are to provide education for professionals and communities concerning the issue of rare diseases, to increase knowledge of the epidemiology of rare diseases, to produce a registry of rare diseases, to increase the work quality of referral centres for rare diseases, etc.

Conclusion

This study is the first of its kind conducted in Croatia, and one of the few studies on social aspects of PWS – specifically mothers' experiences with formal support. Some of the results reproduce those of earlier studies on mothering children with developmental disabilities. The experiences show that formal support for mothers of children with PWS is defined by a lack of knowledge and awareness of PWS, which results in professionals' lack of empathy, and their perception of children with PWS as ones who cannot gain certain social rights. The results show that mothers had to use their own strategies and skills to obtain support. Although some parts of the results indicates that support was better and more adequate when PWS was finally diagnosed, it should be pointed out that mothers can receive professional support only by travelling to the capital city.

As mentioned earlier, these results are very similar to results of different qualitative and quantitative studies on experiences of mothers whose children have a rare disease or a developmental disability. The conclusion is that mothering a child with any kind of a rare disease is a very similar experience, regardless of their nationality or local place of residence where the family lives. Living with a rare disease is extremely challenging because people with rare diseases are often invisible. They are not seen as potential users of the formal support system. The results indicate that children with PWS and their families cannot gain many rights guaranteed by national and international law. Their human rights and dignity are often disregarded as well. For this reason, raising the awareness of professionals is crucial for improving the quality of life of families living with PWS. The professionals' awareness of PWS can result in community awareness, which leads to improved formal support and quality of life of families living with PWS. The results show that families living with PWS are on occasion faced with discriminatory behaviour from professionals working in the formal support system, whereby this behaviour is based on attitudes and beliefs about disability that are widespread in society.

Competing Interests

The authors have no competing interests to declare.

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