RARE DISEASES AND THE QUALITY OF LIFE OF CAREGIVERS

Jan Michalík
Palacky University Olomouc,
Czech Republic

Abstract

The quality of life of persons caring for a family member with rare disease is discussed in the article and the results of the quantitative research (N = 298) in the Czech Republic are presented. The idea of the research is to determine basal psycho-social markers defining the quality of life of persons caring for a family member with rare disease and describe phenomena characterizing the conception of understanding the term quality of life by representatives of the target group (and compare this conception with how the term is understood by the intact population). Long-term care for a child with a serious, incurable disease means a significant change of life situation including not only personal growth, but also social, partner, health and economic associations. In the present research it has been focused on the existence of states and changes that were evaluated by caregivers themselves.

Key words: *quality of life, children with rare diseases***,** *socio-psychological stability, expectations.*

Introduction

The presented results are a part of an extensive research of the quality of life of persons caring for a family member with rare disease that was conducted in the Czech Republic in years 2009-2011 as a part of a research supported by the Czech Grant Agency (The quality of life of persons caring for a family member with severe disability 406/09/0177).

The subject of the research were areas comprising the model of the quality of life of people who over a long period of time care for a family member who suffers from some of the so called rare diseases. Researchers set as a project goal to bring new and not yet detected data about the situation of these persons in the indicated area. They worked on the premise that the long-term care for persons with various presented diseases usually means significant disadvantage in the series of monitored characteristics (values, possibilities, opportunities, facts) in comparison with persons who are not burdened with the long-term care for such a diseased family member.

The target research group (long-term caregivers who care for a person suffering from some of the rare diseases) has stood away from the attention of specialized workplaces so far (when sporadic and partial researches concerned primarily persons with disability themselves – here the diseased). The presented issues closely correspond to the research boom of the last decade (in the CR and the world) focused on the research of the quality of life (for details see the summarizing publication focused on research and research results in this matter (Mareš, 2006). Research subject-matters of these researches are focused on the area of intact population and partially also population with health disability (generally the quality of life is connected with health, see researchers' generally accepted term HRQL - *health-related quality of life*). None of domestic or foreign researches has yet followed the quality of life of caregivers regarding the evaluation of their opinion and attitude spectrum to questions that comprise the immanent part of their everyday life connected with care.

If the generally accepted fact says that the care of close persons is beneficial for the health of the diseased, then in case of diseases in summary known as rare diseases this factor is absolutely unquestionable (Ješina, Magner, Poupětová, et al., 2011). It is a group of various diseases that are characterized by a relatively rare presence in population. Nevertheless, at present a few hundreds of these diseases with various prevalence are registered. Rare diseases usually occur shortly after birth when 4-5% of new-borns and infants are affected (for example some inborn developmental defects, spinal atrophy, hereditary metabolism disorders, cystic fibrosis etc.), but they can become apparent later during childhood (for example, Rett syndrome, osteogenesis imperfecta), adolescence (for example neurofibromatosis) or adulthood (e.g. Huntington disease or some forms of thyroid cancer). About 80% of rare diseases have genetic causes, nonetheless, the etiology of the disease remains unknown for most patients. In case of wrong and late diagnoses, mainly in diseased patients for whom there exists the possibility of treatment, it comes to irreversible health damage. This leads to a significant psychological trauma of not only patients, but also their families, including distrust to health-care system (Michalík, Zeman, et al., 2010). The characteristic feature of the care for a family member (usually a child) with such disease is high demands on the care that is only hardly comparable with other social events of this type. On the other hand, there also exist big differences among rare diseases regarding the seriousness of the impact on the quality of life of the diseased – and thus usually also a caregiver.

The essence and aim of the project was to *determine basal psycho-social markers defining the quality of life of persons caring for a family member with rare disease and describe phenomena characterizing the conception of understanding the term quality of life by representatives of the target group* (and compare this conception with how the term is understood by the intact population). Consequently, to discriminate different characteristics of the conception of the quality of life of persons of the target group and to prepare on their basis general methodology applicable in other researches when finding out markers connected with understanding and perception of the content of the term *quality of life* of the target group. Key changes in value and psycho-social dispositions of probands (caregivers) were determined, monitored and analysed on the basis of such realized quantitative research.

Study methodology

Researchers chose quantitative methodology based on the choice of a highly representative sample of probands, questionnaire research and statistical processing of data with classification of the first and the second level and determining statistical importance of the collected data (chi-square– Pearson's coefficient).

Research in the group of persons caring for a child with rare disease

The research population was chosen (beyond originally planned project content) from the basic population of persons caring for a person with a rare disease on the whole territory of the CR. The questionnaire was distributed predominantly to persons who care for the mentioned person. Cooperation with specialized doctors was applied and these representatives of patients' organizations uniting such care-givers allowed addressing of respondents with preservation of full anonymity and the necessary level of protection of (delicate) data of respondents and possibly persons they care for.

In total 593 questionnaires were distributed and 298 returned for data procession (i.e. 50,2%). In total, the opinions were obtained from 113 persons caring for a child with cystic fibrosis, 47 persons caring for a child with Achondroplasia, 59 persons caring for a child with some of the metabolic diseases (and other 33 persons caring for a child with mucopolysacharidosis) and finally 46 persons caring for a child with spinal muscular atrophy. This research was done in autumn 2010.

Research with persons from control group

Measured attitude results (opinions, evaluations, data information) in the researched population of care-givers laid the foundations for conducting a similar comparative research in the group of persons who do not care for a family member with a severe disability. Whereas a high level of territorial representativeness was reached with caregivers' group, the choice method of respondents of the control group was different regarding the way of arranging and logically the researched sample far less represents opinions of the basic population. Still, the author believes that results obtained from the control population (see below) are sufficient for conducting a meaningful comparison. Participants were chosen randomly from students of the combined form of study programme at Palacky University in Olomouc. Characteristics of the basic control population sample in general features (see below) corresponded to caregivers' group regarding gender of respondents (women prevailed). Distribution of the measuring tool took place in September and October 2011.

Basic requirement on research participants was formulated this way: person who cares for a child in household (family) but at the same time does not care for any family member who has a serious disability. The choice of participants was random, app. 510 students, both males and females, were addressed, however, childless persons did not take part in the filling out. Thus, in total 305 questionnaires from control population respondents were evaluated. It is observed that the control group persons are also "caregivers", but they care for a child without disability, i.e. they are not persons who do not have any experience with the care for another person.

Table 1. Sample characteristics

CARING PERSONS	CONTROL GROUP
LENGTH OF CARE FOR FAMILY MEMBER	
up to 5 years: 108 persons (36%), up to 15 years: 124 persons (42%), up to 25 years: 40 persons (13%), over 25 years: 12 persons (4%). Did not respond: 14 persons (5%).	more than 10 years: 62% less than 10 years: 30% Did not respond: 8 %
THE MOST CARING PERSON IN FAMILY	
Women – 227 (76%). Men – 0 Take turns – 67 persons (22%). Did not respond – 4 persons (2%).	Women – 137 (45%). Men – 4 (2%). Take turns – 111 persons (36%). Did not respond – 53 persons (17%).

Continued Table 1

RESPONDENTS' GENDER	
197 women (66%), 96 men (32%), 5 persons (2%) did not indicate gender.	259 women (85%), 39 men (13%), 7 persons (2%) did not indicate gender.
RESPONDENTS' AGE	
up to 30 years: 35 persons (12%), up to 50 years: 224 persons (75%), up to 70 years: 35 persons (12%). Did not respond: 4 persons (1%).	up to 30 years: 55 persons (18%), up to 50 years: 231 respondents (76%), up to 70 years:11 respondents (4%). Did not respond: 8 persons (2%).
RESPONDENTS' EDUCATION	
Primary education: 36 persons (12%) Secondary education: 173 persons (58) Higher education: 23 persons (8%) University education: 63 persons (21%) Did not respond: 3 persons (1%).	Primary education: 4 persons (1%) Secondary education: 152 persons (50%) Higher education: 30 persons (10%) University education: 114 persons (37%) Did not respond: 5 persons (2%).
EMPLOYMENT	
Full-time: 144 persons (48%), Part-time: 37 persons (12%) Without employment: 113 persons (38%) Did not respond: 4 persons (2%).	Full-time: 276 persons (90%) Part-time: 8 persons (3%) Without employment: 18 persons (6%) Did not respond: 3 persons (1%).
FAMILY STATUS	
Single: 14 persons (5%) Married: 203 persons (68%) Divorced: 33 persons (11%) Widowed: 21 persons (7%) Did not respond: 27 persons (9%)	Single: 27 persons (9%) Married: 247 persons (81%) Divorced: 23 persons (8%) Did not respond: 8 respondents (2%).

In both cases the measuring was delivered by a multi-level anonymous questionnaire comprising both its own content items and constant items, functional psychological items, control items composed of mostly structured (closed) and partially non-structured (open) form. Next, rating scales and standardized SEIQoL methodology were applied (note SEIQoL questionnaire evaluation is done in a separate study). The collected data were statistically processed with the first level classification (49 basic contingency tables) with graphic support.

Analysis of the research results

In the next part a brief summary of results in individual monitored areas has been presented. To simplify the table part the results in the item "did not respond" are not stated.

Arrival and existence of stressful factors of caregivers

In this part the answers of respondents monitoring the arrival and existence of stressful factors that are connected with (in particular) the longterm care were monitored.

Table 2. Arrival and existence of stressful factors affecting socio-psychological stability

Monitored factor/frequency	Very often - often		Rarely		Not at all	
	Caregiver	Control	Caregiver	Control	Caregiver	Control
Loss of feeling "I have things under control"	**44 %**	30 %	48 %	61 %	8 %	8 %
Arrival of depression and hopelessness	**38 %**	16 %	38 %	45 %	24 %	39 %
Feeling of exhaustion	**65 %**	54 %	28 %	41 %	7 %	5 %
Loss of the ability to enjoy free time	**54 %**	33 %	30 %	41 %	16 %	26 %
Loss of the ability to rejoice	**34 %**	18 %	42 %	45 %	24 %	36 %
Feeling of losing personal prospects	**41 %**	17 %	36 %	47 %	23 %	36 %

On the basis of analysis (Table 2) of the answers of the respondents in the part of the questionnaire that monitors opinions on the existence and development of negative effects or feelings as tiredness, the loss of the ability to rejoice, arrival of massive depression and hopelessness or the loss of personal life prospects the following can be stated:

In all monitored items the respondents from the caregivers' group showed statistically significantly more frequent arrival and perception of "negative" feelings and conditions in comparison with the respondents from the control group. Striking differences in "the loss of the ability to rejoice" and the feeling of "the loss of personal prospects" can be observed.

It is interesting that in case of "the feeling of exhaustion" statistically significant difference is also observed although in value "very often and often" 54% persons from the control group indicated this item in comparison with 65% of care-givers. It is obvious that this item reflects "imperfections" of the focus of quantitatively oriented researches on measuring "subjective" categories of perceiving and experiencing of respondents. It is quite doubtless that care-givers experience (objectively) incomparable situations requiring enormous psychological and physical performance – in comparison with persons from the control group. However, persons from the control group also experience difficult times – they prepare for their employment in the combined study course, i.e. they work, study – and at the same time they care for a (healthy) child. Also these persons logically experience feelings of tiredness. That is why – objectively – the measured results are significant. The care for a family member with a rare disease leads to a massive arrival of hopelessness and depression of the respondents, they experience the feeling of exhaustion or the loss of the ability to enjoy free time. The control population persons come the closest to caregivers in the feeling of exhaustion. The results of "predictive" items, i.e. the loss of the ability to rejoice and particularly the loss of personal prospects of caregivers are seen as alarming for the future psycho-somatic setting of respondents caring for a family member with a rare disease.

Changes of chosen social facts during the care

Next, extensive area mapping the perception of the given state, status of a caregiver and his/her family within selected social situations or states were monitored, also the existence of possible resilient factors counteracting stressful factors – as they were recorded above.

Table 3. Changes observed by the respondents in selected areas during the period of last 5/10 years

Monitored factor/frequency	Yes or more likely yes		Rarely		More likely no or no	
	Care-giver	Control	Care-giver	Control	Care-giver	Control
Increased isolation from other people	51 %	27 %	19 %	18 %	30 %	56 %
Deterioration of health	36 %	31 %	32 %	26 %	32 %	43 %
Decline in family economic situation	53 %	32 %	22 %	18 %	25 %	50 %
Deterioration of relationship among family members	15 %	41 %	22 %	21 %	63 %	15 %
Change in family social life	49 %	23 %	21 %	29 %	30 %	48 %
Feeling of enrichment from the care/ situation	36 %	71 %	28 %	13 %	36 %	16 %
Feeling of pride of what I achieved	53 %	79 %	29 %	11 %	18 %	10 %

The monitored items in this part were respondents' opinions on possible changes that could have occurred during the period of caring for a diseased child regarding the economic situation and social life of caregivers and their families (Table 3). Generally it can be said that the overall majority of care-givers has noticed during the course of care (during more than 10 years) a significant decline in economic situation. Care for a seriously diseased child means not only physical and mental strain, but it also brings difficulties for economic securing of family. Possible notions of the part of the public about "allowance paradise" are false and do not reflect the real state of the issue.

It is necessary to take into account that many of rare diseases are infaust already in childhood and adolescence. This fact could have also manifested in perception of the economic level of family life. Related influence that manifests in evaluation of the economic level of family is increased economic demand (aids, care cosmetics, special diet, additional charge for health care) of long-term care during the time when the deterioration of patient's health occurs.

Since 2012 in the CR significant changes also have been observed in the social support for families caring for a family member with a serious health disability (Otevřený dopis, 2012) and consequently also for most families in the present research. Particularly speaking about single mothers (mother and diseased child or his/her sibling) it is likely to expect another deterioration of the described situation. It is confirmed by the results from the study of mothers caring for children with retarded psychological development that showed unequivocally a higher risk of emerging of socio-economic problems (Emerson, Hatton, Llewellyn, 2007).

Another item monitored within the study was the "social life of family", i.e. changes in family contacts with the outer environment. It usually includes meeting with friends, participation in culture and social events etc. Almost 50% of respondents confirmed that social contacts of their families were limited (however, it is necessary to mention that within the group of care-givers who care for example for a child with mucopolysacharidosis this option was indicated by full 90% of care-givers!) The feeling of social exclusion and social isolation is common in caring families.

Further, it is necessary to pay attention to the item monitoring "deterioration of relationships among family members". Here quite a surprising result is observed. Only 15% of care-givers confirm deterioration of relationships among family members in the group of care-givers in comparison with 41% of persons from the control group! Negative influence of home preparation for university studying on the quality of relationships among family members can be inferred. However, this item is considered as supplementary or controlling to items presented further in this part.

It is necessary to interpret the results from the items monitoring “feeling of enrichment from care/situation” and “feeling of pride of what the respondent has achieved” individually. Just the first of the items is sometimes presented as an “immanent” part of “faith” of care-givers whether a serious disease or health disability is concerned. It is becoming apparent that care-givers are “very realistic” when judging “benefits” of such a situation. Facing the strenuousness of such care the results of the present research overshadow these general characteristics (or rather prejudices). Care-givers are good at evaluating “benefits” and “expenses” of care… The feeling of significant enrichment was observed in persons from the control group who “have and care for a child” – a healthy one… Similarly the feeling of pride of people from the control group (almost 80%) on what they have achieved was observed. This can be related to the results of their university studying.

It is necessary to emphasize that all mentioned concerns more women who are dominant care-givers.

Summarizing feeling of happiness and expectations of the respondents to the future

In the final part of the research the questions that allowed respondents to express summarizing evaluations of their situation and expectations to the future were modelled. Most caregivers feel the change in the perception of life values, their own personal growth and gained experience connected in many cases with the feeling of pride and self-confidence (Table 4). However, considerable number of the respondents rejects in connection with the care for a diseased child to talk about any “benefits”. In the area of “losses” many respondents feel total disillusion and talk about the loss of “the sense of life, the loss of everything”. It was also focused on experiencing the “feeling of happiness” in actual time.

Pregnancy usually brings a great feeling of happiness and so the more devastating is the news that the long expected child has health disability. At this moment the feeling of happiness is replaced with the feeling of hopelessness, fear and despair. The feeling of happiness returns individually to every caregiver in a different way and different time. It depends on how quickly an individual is able to cope with this fact and how strong he/she is. Someone cannot manage it at all – more exactly – in some cases it is obviously not possible to induce such feeling permanently or fully.

Person (child) suffering from any of rare diseases will have certain problems during his/her whole life and handicap. Caretaker realizes this fact very well and that is why due to concerns about life and happiness of their child with disability they themselves are not able to enjoy the feeling of happiness from life. Nevertheless 1% of respondents indicated that they feel completely unhappy. Dominant feeling for caregivers is “reserved happiness”. It was indicated by full 42,3% of the caregivers.

It is necessary to mention that for example in the group of persons who care for a child with Achondroplasia (short stature) full 83% feel very happy – on the contrary to the group who care for a child with mucopolysacharidosis where nobody chose this option!

Table 4. Feeling of happiness of the respondents at present

At present I feel	Caregiver		Control	
	a. f.	**r. f.**	**a. f.**	**r. f.**
Absolutely happy	23	7 %	68	22 %
Very happy	64	21 %	169	52 %
A little happy	133	45 %	37	13 %
Nor this nor that	26	9 %	7	3 %
Rather unhappy	33	11 %	11	5 %
I am unhappy	15	6 %	4	2 %
Absolutely unhappy	4	1 %	2	1 %
Total	298		298	

Another summarizing item focused on the prediction of the future development (expectation) in relation to measured elements of socio-psychological stability of caregivers (Table 5). Again a 7-point scale was evaluated where value 1 denoted the possibility “I fear the future” and value 7 the possibility “I absolutely look forward to it”.

Table 5. Expectations of respondents to the future

Feeling of happiness of respondents at present	Caregiver		Control	
	a. f.	**r. f.**	**a. f.**	**r. f.**
I absolutely look forward to the future	23	8 %	35	15 %
I look forward to it moderately	60	22 %	156	48 %
I look forward to it little	39	13 %	64	23 %
Nor this nor that	20	7 %	6	1 %
I rather do not look forward to it	35	7 %	16	7 %
I do not look forward to it	21	7 %	11	4 %
I fear the future	100	36%	10	2 %

Thus, it was a combination of actual psychological temper and the expected prognosis of the future development – usually connected with the disease and in a certain way also expected demands on the future care for a diseased child. It must be remembered that a large part of diseased children suffer from a progressive disease that is in many cases (conclusively) infaust. It is possible to state that this item has clearly and simply confirmed the difference between both groups of the respondents that was obvious in previous items. Regarding socio-psychological perspective, the arrival of resignation and negative states it was caring parents who always indicated worse results by a certain amount.

Full 36% of respondents from the group of caregivers chose the option “I fear the future” in the item focused on the future expectations. In contrast to this only “mere” 2% of respondents from the control group chose this option. If the summarizing survey adding both positive and negative expectations to the future is chosen, then 43% of the respondents from the group of caregivers and 86% from the control group look forward to the future… In this item significant differences inside the group of caregivers were also observed. Again the influence of demand of the care on attitudes and judgements of caregivers was evident – see the following Table 6.

Table 6. Expectations to the future-care-givers group – inner division

Disease type	**1 I fear it**	**2**	**3**	**4 neither this nor that**	**5**	**6**	**7 max. I look forward to it**
Cystic fibrosis	**33,63%**	9,73%	11,50%	6,19%	13,27%	16,81%	7,08%
Achondroplasia	10,64%	2,13%	17,02%	8,51%	17,02%	**38,3%**	6,38%
Metabolic diseases	**38,98%**	1,69%	5,08%	5,08%	11,86%	20,34%	11,86%
Spinal muscular atrophy	**34,78%**	13,04%	8,70%	8,70%	8,70%	13,04%	10,87%
Mucopolysacharidosis	**45,45%**	6,06%	21,21%	6,06%	15,15%	6,06%	0,00%

Modus values are presented. Modus values for diseases that are incurable or even infaust (mucopolysacharidosis) are in the area “I fear it”. In contrast to this caregivers of a child with achondroplasis (short stature) dominantly feel as “very happy”. This example is given for its illustrative and important significance. Even though the group of caregivers who care for a child with rare disease and the control group are dominantly compared – it is appropriate to show inner diversity of the first population group. Again it confirms the influence (and demand) of the care on the quality of life of members of the given population group.

Conclusion

Long-term care for a diseased child with a serious, incurable to infaust disease means a significant change of life situation including not only personal growth, but also social, partner, health and economic associations. Description of the whole issue exceeds limits of this text. However, it can be rightly stated that it is one of the most difficult life trials that a person can be exposed to these days. Published studies show that parents of children with serious psychomotor retardation are exposed to a higher risk of stress and consequently development of psychiatric difficulties (Emerson, Mcculloch, Graham, et al., 2010). At the moment the questions about experiencing (accepting) the fact of health disability by the individual himself/herself that is naturally considered with acquired diseases at certain age are put aside.

In the present research it has been focused on the existence of states and changes that were evaluated by caregivers themselves. The fact that the described situations and states are significantly influenced by the combination of the following factors – seriousness of the disease and its consequences, expected or real length of care and expected and real demand of care applies to all the acquired data. To illustrate “social” connotations of the measured values, the research results of the control group that consisted of caregivers who care for a child without health disability or disease are also stated. The present research proved obligatory connection between the fact of the care for a child with rare disease and the quality of life of caregivers – in comparison with the life of the “common” population, here represented by the control group members.

Care for patients with a serious rare disease is demanding and places extra demands on caregivers. Naturally – in the given segment of results it is obvious that it also places extra demands on the knowledge of real conditions of the quality of life of the diseased, caregivers and also towards members of helping professions – doctors, social workers, teachers, psychologists and others. Common expenses on the rare disease (health care, social protection, losses due to unemployment of caregivers) amount to tens and hundreds millions of euro. Thus, it is essential to include measuring of the impacts of this support to the quality of life of the diseased and caregivers. The results stated in this article are a contribution for starting the discussion in the Czech Republic.

References

1. Emerson, E., Hatton, C., Llewellyn, G., et al. (2006). Socio-economic position, household composition, health status and indicators of the well-being of mothers of children with and without intellectual disabilities. *Intellect Disabil Res*, *50 (12),* 62-73.
2. Emerson, E., Mcculloch, A., Graham, H., et al. (2010). Socioeconomic circumstances and risk of psychiatric disorders among parents of children with early cognitive delay. *Intellect Dev Disabil*, *115(1),* 30-42.
3. Ješina, P., Magner, M., Poupětová, H., et al. (2011). Mukopolysacharidóza I - klinické projevy u 24 dětí z České republiky a Slovenska. *Česko-Slovenská Pediatrie*, *66 (4)*, 215-225.
4. Mareš, J. (2006). *Kvalita života u dětí a dospívajících I*. Brno: MSD.
5. Michalík, J., Zeman, J. (2010). *Mukopolysacharidóza*. Olomouc, Společnost pro MPS.
6. Otevřený dopis: Důstojnost rodinám se zdravotně postiženými dětmi (2012). In *Informace Národní rady osob se zdravotním postižením č, 14*. Internet access: <http://www.nrzp.cz/aktualne/informace-predsedy-nrzp-cr/742-informace-c-14-2012-otevreny-dopis-.html>.