The impact of Huntington disease on family carers – a literature overview

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Summary
Caring for a person with chronic disease often rests on the patient’s family. Nevertheless, most studies on the needs, quality of life and caregiver burden focus on different types of dementia, including Alzheimer’s and Parkinson’s disease, stroke, cancer or mental illnesses. Less attention has been paid to informal caregivers of patients with Huntington’s disease (HD). Meanwhile, psychosocial implications of HD are much more serious and wide-ranging. Thus, this paper aims to review the literature on the costs of caring for a person with HD (pHD). The review identifies the aspects of caregiving that are the most burdensome to family caregivers: negative experience with health and social care services, dissatisfaction with caregiving role, concern over children, loss of meaningful relationship with pHD, family breakdown and loss of social contacts and decrease in health. It also describes strategies of coping with HD. While the analysis points to the many similarities between taking care of HD patients and those suffering from other neurodegenerative disorders, it also emphasizes the factors associated with HD that are not present in other diseases: early onset, severity of symptoms, prolonged trajectory, its genetic, i.e. hereditary character and social ignorance of HD. It concludes that while in medical setting the HD patient is the one who draws professional attention, it is often the unaffected family caregiver who needs the most attention, support and help.

Key words: Huntington’s disease, family caregivers, needs

Introduction
One of the most important changes in the health care system is that due to increased life expectancy and the shift from acute to chronic diseases, the number of disabled people has markedly increased. Consequently, the number of individuals involved in caregiving, the duration of caregiving role and the type of caregiving tasks performed

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have also changed: for many caregivers such a role lasts for many years, even decades, and caregiving has become a very complex task.

Nevertheless, while most studies on the needs, quality of life and caregiving burden focus on different types of dementia, including Alzheimer’s and Parkinson’s disease [1–3], stroke [4, 5], cancer [6] and psychiatric disorders, such as schizophrenia or affective disorder [7, 8] less attention has been paid to family caregivers of persons who suffer from Huntington disease (HD). Thus, while most of research focuses on the impact of HD on the quality of life of the affected individuals and/or those at risk [9–12], informal caregivers of persons with HD (pHD) are sometimes referred to as ‘forgotten persons in the HD family’ [13] and ‘invisible patients’ as their needs are often overlooked by healthcare professionals. And although there are many measures to assess the needs, quality of life and caregiving burden [14], few address carers of pHD [15, 16]. Finally, while some research on HD carers was conducted in the UK [12, 16–20], including Scotland [21], the USA [22–27], Canada [28], the UK and the USA [29, 30], the USA and Canada [31–34], the Netherlands [35], Norway [36], Spain [37], Australia [11, 38, 39], South Africa [40] and even China [41, 42] little is known about informal carers of pHD in Poland [43]. Meanwhile, a comparative study between France, Italy, Germany, Poland and the USA [44] shows that Polish carers have the lowest level of resources use and spend the highest amounts of time looking after their HD relatives. It is of key importance as the Polish Minister of Health stresses the need to support patients with rare diseases [45].

Aim

The aim of the present paper is to review the literature on family caregivers of persons with Huntington disease. The PubMed database was searched between 2000 and 2013 for research and review papers on caregiving burden, (unmet) needs and quality of life of informal caregivers of pHD. The search terms included: ‘Huntington disease’, ‘family caregivers’, ‘caregiving burden’, ‘needs’, ‘quality of life’, ‘well-being’ and was limited only to articles published in English. The initial search has identified 84 articles. To provide more homogenous results, the author has decided to exclude papers about juvenile patients from the analysis, as jHD’s clinical view is somewhat different from the adult form of the disease (it lasts shorter and has more severe symptoms). Also, articles referring to some aspects of HD but not focusing on family caregivers or caregiving tasks were excluded. Among such papers were those that dealt with professional caregivers and those on psychosocial implications of predictive testing for at risk individuals and their families, clinical aspects of HD and their perception by either patients and/or their caregivers and those that aimed to validate some new instruments for the measurement of psychological stress of HD carers. Thus, only those papers that focused on informal, family caregivers of persons with diagnosed HD in advanced stage were analysed. After screening the titles and abstracts for relevance 20 papers were assessed for eligibility and included in the analysis. Additionally,
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A reference list of the articles identified was also reviewed which helped in locating additional 13 papers. In sum, 33 papers were included in the analysis, sixteen of them being quantitative studies [11, 15–17, 20, 24–27, 29, 30, 35, 37, 39, 41, 43] and fifteen qualitative [13, 18, 19, 21–23, 28, 31–34, 36, 38, 42, 46]. The last two papers [40, 47] present the theoretical aspects of taking care of pHD.

Clinical dimension of Huntington disease

Huntington disease, formerly known as Huntington chorea, is a rare, chronic, neurodegenerative disease of the brain characterised by progressive cell death primarily in the basal ganglia, especially in caudate nucleus and frontal lobes with an associated triad of symptoms: movement abnormalities and loss of motor skills, cognitive and affective disturbances and loss of behavioural functions that together cause severe disability. Its prevalence is estimated to reach 4–8 persons per 100 000 [45]. While clinical symptoms usually begin at the age of 35–40, there is a wide variation in age of the onset and subtle symptoms may be present up to decades before clinical diagnosis is met [19, 30]. For that reason, in contrast to many other types of dementia, HD has a long trajectory and carers of HD subjects are engaged in caregiving tasks for decades [30]. There is neither a cure for HD nor treatment that can cure, delay or slow progression of the disease and it is purely palliative, i.e. HD can only be treated to alleviate the symptoms. Consequently, death occurs on average 15–17 years after the onset of the disease.

HD is a genetic condition caused by mutation in the HTT gene located on the short arm of chromosome 4. It is inherited as an autosomal dominant trait which means that a child of the gene carrier has a 50% risk of inheriting the HD gene and developing the disease. Moreover, HD does not skip generations, no one can be a carrier without developing the disease. Additionally, it may be present in multiple family members, thus limiting the amount of available help and support [30]. Since the gene mutation that cause HD was identified in 1993, predictive testing for those at risk is possible without cooperation of relatives.

One of the most characteristic symptoms of HD are progressive involuntary (choreic dancelike) movements of the body and abnormal gait. Nevertheless, cognitive and behavioural changes may occur much earlier, which makes this disease difficult to diagnose. Its early symptoms also include problems with concentration and memory loss, mood changes, including depression episodes that often lead to suicidal thoughts, aggressive and antisocial behaviours, problems with walking and clumsiness. With the progression of disease involuntary movements escalate and problems with speech and communication begin. Additionally, pHD suffer from impaired short-term memory, have difficulties with planning and problem solving. Mood swings and emotional liability become more frequent and due to the problems with swallowing patients experiences severe weight loss. Emotional symptoms include personality changes such as impulsiveness, disinhibition, depression, mood swings and aggression. As a conse-
quence, in later stages pHD require full nursing care [48]. Death occurs usually after 15–20 years and is caused by secondary illness, mainly aspirational pneumonia. Other causes of death include suffocation and suicides which are four to six times more common than in general population [49].

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Negative experiences related to healthcare services

The most common theme present in the literature on HD carers is related to their negative experiences with health and social care services. For example, a study by Skirton et al. [29] showed that both in the USA and UK access to health services for pHD and lack of knowledge on HD from health professionals constituted the biggest problem for many caregivers (UK = 82.4%, USA = 64.3%). Additionally, in both countries carers were concerned about insufficient number of resources available to support care (UK = 69.1%, USA = 62.2%), had difficulties with finding facilities with trained staff (UK = 64.9%, USA = 58.3%) and those that would accept the affected person (UK = 64.9%, USA = 49.2%). Caregivers also complained that healthcare professionals are not giving pHD the required health care (UK = 48.9%, USA = 34.9%), do not address their other health problems (UK = 41.6%, USA = 28.4%) and do not take into account their treatment preferences (UK = 34.9%, USA = 27.3%). At the same time, despite many similarities, UK carers were significantly more bothered with getting affordable quality care (UK = 79.9%, USA = 57.1%), finding appropriate activities (UK = 76.5%, USA = 57.1%), lack of HD education from health professionals (UK = 82.6%, USA = 64.3%) and difficulties with obtaining financial benefits from disability services (UK = 59.3%, USA = 34.2%). The same theme emerges in many other studies. A qualitative research on UK carers [18] showed that most frequently they complain about the lack of access to specialist services and report problems with access to routine medical care for pHD and residential care [27]. The reason why it is so is that due to a relatively young age of the onset of HD people who suffer from it are usually below the age at which geriatric services are available [29]. On the other hand, persons in preclinical phase of HD may not be active participants in health care, which may hinder receiving professional help [31]. In both cases pHD are often refused treatment, including additional rehabilitation [45]. What is more, such services are often available to them only in crisis situations [19, 21, 38]. Additionally, as HD is a rare disease it is not included in the lists of chronic diseases, which results in that the patient cannot use the preference prices for medications and their access to the most modern drugs for symptomatic treatment of HD is seriously limited [45]. Caregivers are also disappointed with the (lack of) support they receive from health care professionals and with their lack of practical knowledge on HD [18, 19, 27–29, 38, 45]. Research by Skirton and Glendinning [12] and Etchegary [28] also showed carers’ dissatisfaction with the coordination of care between families and professionals, and lack of awareness of the latter of the needs of caregivers. Another problem they face
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is related to lack of housing options for affected relatives, especially juvenile patients [27]. On the other hand, caregivers express negative experiences with unskilled stuff in nursing homes [38].

Moreover, in most studies caregivers also complain about the amount and kind of information they receive from physicians. And while some carers are dissatisfied with too much information following the diagnosis, in most cases they are stressed with general lack of knowledge by health professionals on HD, its symptoms, treatment and support. They are also dissatisfied about the amount of time they have had to spend searching for such information [19, 28, 31, 38]. Additionally, caregivers express their concern over low levels of practical support in coping strategies and problem solving and feel lack of general understanding in the medical community [27]. Such results are confirmed by the findings from a study by Soltysiak, Gardiner and Skirton [19] who suggest that care for pHD should be more individualized. At the same time, they believe that in contrast to many other neurodegenerative disorders general public lacks basic knowledge on HD and that there is not enough media attention [19, 30, 36, 38]. All in all, many caregivers have a general sense of struggle with the system for the costs of medications, equipment, transport and home modifications [21, 38].

Caregivers’ feeling of role overload

Another problem described in the literature is related to carers’ dissatisfaction with the caregiving role. A study by Roscoe et al. [27] showed that over half (65%) of caregivers perceive their role as ‘very stressful’. Such stress results mainly from the neuropsychiatric character of HD which enables the care recipient to complete his/her tasks independently and leads to emotional, cognitive and behavioural changes. Role reversal from a spouse or a child to a caregiver is also very distressing [32, 33], especially that many carers do not feel prepared for the caregiving task and have problems with adjusting to the new roles, responsibilities and schedules [31]. Consequently, they feel role overload.

Due to an early onset of HD caring for pHD is particularly problematic for middle-aged people who just begin to live independently after their children are mature and live outside the home. Thus, at the time when a person could pursue one’s own interests and dedicate her/himself to professional career and strengthening bonds with the spouse, one may feel burdened by the new role. On the other hand, as the onset of HD begins when the family life cycle is at its most complex moment (when a person has child-rearing responsibilities and develops his/her professional career) the imperative of caring may negatively affect one’s ability to perform the parental role and produce adverse parenting behaviours [23, 39]. Consequently, caregivers often report conflicting responsibilities of caring for pHD, raising children and taking care over the household [21, 32, 36]. Caregivers’ negative emotions are also caused by their feeling that providing care is a kind of ‘duty’ that is placed upon them [18]. Especially female carers and daughters in particular, are more pressured into caregiving and are expected to
abandon their plans for a professional carrier whereas sons often leave home to pursue their academic and professional aspirations. Consequently, they often feel as being restricted by their caring responsibilities and are concerned over the loss of former roles and relationships [23, 27, 30, 38]. Apart from these important gender differences, caregivers in general feel that they have lost their lives, identity and personal independence [13, 16, 21, 22, 30, 32, 36, 38]. Those who continue to work professionally are also concerned about leaving the affected person at home without adequate support [32]. Such stress is further increased by the fear that with age one’s ability to take care will decrease [29]. Finally, a financial burden is also mentioned [24, 30, 32, 33, 36].

Also, teenage carers who are sons and daughters of pHD report problems that arise from their unwillingness to accept the caregiving role. Many researchers show that caring responsibilities may have a devastating impact on their health, psychological and emotional development, education and social life [33, 46]. Folstein et al. showed that 48% of the offspring of a parent with HD suffered some kind of psychiatric disturbance, including affective, personality and behavioural problems [39]. Juvenile carers are concerned that the new role will deprive them of normal life and making plans about the future [33, 34, 36], especially that due to their at risk status they are ‘waiting in the shadow of HD’ [33] and can do nothing to alleviate the symptoms. Other problems result from the fact that due to the existing law children cannot undergo predictive testing while being pre-symptomatic, which prevents them from planning their future. For all these reasons, they feel alone in their genetic reality and not understood by others, including healthy parents. Nevertheless, also adult carers often feel trapped in the caregiving role and express a wish to escape from it. They also believe that their own needs are neglected [17]. Finally, some are ashamed and blame themselves that such feelings make them disloyal to their relative. All in all, caregivers struggle with tiredness, exhaustion and lack of time. They also experience depression, frustration, anger and guilt [25].

**Progression of disease**

The third most common theme refers to the changes in pHD including alterations in cognition, behaviour and instrumental activities of daily living, such as work responsibilities, handling money and driving [31, 32, 37]. Caregivers from most of the studies report problems with patients’ moodiness, irritability, depression, anger, loss of memory and suicidal ideations. Interestingly, while HD patients and their partners do not differ significantly in their beliefs on the duration, consequences, causality and curability of the disease, the latter attribute more symptoms to HD than the patients [35]. Moreover, they perceive HD as less controllable and have more negative beliefs about it. It may result from the fact that as HD may manifest itself many years before the patient meets the official diagnosis, which makes it very difficult to understand the emotional, cognitive and behavioural changes in their affected relative [30, 31, 36, 38]. It should be also noted that caregivers’ interpretations of their caring experience may
have more significant impact on their well-being than the objective characteristics of that experience [27].

**Concerns over inherited character of HD**

As HD is one of a few genetic disorders that are inherited in a typical Mendelian style, carers of pHd are burdened by the fact that HD may develop in other family members. More importantly, what they find the most stressful is that while caring for the sick, one anticipates the disease in future generations, i.e. children [13, 16, 25, 36, 39]. For that reason, they feel guilt and blame for being responsible for passing the disease to their offspring [13, 19, 21, 24, 27, 31, 32, 36, 38–40]. Significantly, such stress is often accompanied by symptom-spotting in children. Additionally, they are strained by the responsibility to inform others about the risk of having HD [32, 40]. Another source of carers’ anxiety is their fear that they will have to take care of multiple generations and that such a role may last for decades [16, 18, 21, 30, 38]. At the same time, a study by Williams et al. showed that some caregivers are afraid that they may develop HD themselves and that it will impair their ability to care for pHd. On the other hand, those carers who are tested negative often experience survivor guilt [38].

**Loss of meaningful relationship with affected relative**

Like in other types of dementia, HD carers find it difficult to maintain a meaningful relationship with the affected relative and talk about ‘the end of mutual friendship replaced by relation of care’ [32, 33, 36]. Especially spousal carers experience loss of their marital relationship and feel sad that they had to replace romantic love they once felt with a ‘mother-child relationship’ [13, 16, 22, 23, 32]. At the same time, they have problems with distinguishing HD symptoms as separate from the person with HD. Consequently, caring for pHd may lead to family breakdown [22]. Also, teenage carers express their sadness not only due to loss of relationship with the affected parent but also with the non-affected one who is absent due to professional and caring responsibilities [33]. In both cases, such feelings are strengthened by their fear of sudden, premature death of pHd and are often associated by anticipatory or ‘dis-enfranchised grief” [12, 18, 32, 36, 38, 39] which it is not acknowledged by society as social rituals around it are missing. Such grief, which cannot be publically mourned, makes caregivers feel even more isolated and lonely. Thus, while they are prepared for the worst, they frequently talk about pHd as if he/she is already dead [32].

**Negative impact of HD on family system**

Additionally to losing a good friend and supportive companionship, HD carers often report that HD seriously impedes one’s relationships within and outside the family [31]. They feel as not being understood by others [27, 30, 38] and describe how HD
negatively influences family cohesion and expressiveness and becomes a source of conflicts. Consequently, they experience family separation and extreme isolation both in terms of discussion about HD and support [30, 31, 36, 38, 39]. One of the reasons for being so is that having an HD relative is often felt as embarrassing and shameful. Especially children of pHD feel that disease impedes their peer interactions [33]. As a result, they often avoid contacts with the affected parent [20, 30].

All in all, it can be said that HD affects every aspect of family life. It challenges the stability of the family system by changing family positions, roles and functions. Family breakdown often occurs and due to hereditary nature of the disease it becomes a source of secrets, which hinders family communication, sharing of feeling and reinforces each family member’s emotional isolation [21]. And while most HD carers eagerly accept their new role, many offer their help at a considerable cost of their own health [32]. Skirton and Glendinning [12] found that 20% of caregivers of pHD suffered from stress-related illness. Another study by Pickett, Altmairer and Paulsen showed that while no demographic characteristics were significantly related to caregivers’ depression, subjective caregiving burden (CB) was strongly related to the number of years the pHD has been diagnosed and to a greater length of time spent on caregiving [25, 27]. Additionally, it was associated with the carer’s perceived sense of control and problem-solving confidence. Also, pHD’s physical functioning and depression influenced their feeling of burden [25, 37]. On the other hand, one of the few Polish studies conducted by Banaszkiewicz et al. [43] showed that CB was mainly influenced by patient’s depression and decreased motor skills. Yet another study by Ready et al. [26] on 22 patient-caregivers dyads showed that patient functional and cognitive capacities had the biggest impact on subjective perception of the quality of life of both persons with HD and their carers.

Caregivers’ strategies of coping with HD

Among the strategies used by caregivers to deal with HD, Williams et al. have identified the following: appreciation of positives, anticipatory mourning, setting boundaries and using medications [32]. Other studies report that spirituality and religion may be an important coping resource as it helps carers to find meaning in times of hardship and to accept thing as they are [18, 27, 32]. Research also shows that many carers adapt to the changes caused by HD as they arise and do not plan long-term solutions [21, 32]. Nevertheless, one of the most frequent coping strategies of dealing with HD is denial [21, 23]. Although carers experience serious distress they avoid talking about HD both within and outside the family [13]. Frequently, they deny the early signs of the disease and pre-diagnosis and avoid getting into any situations in which HD may express itself [21]. Such a strategy is especially common among families with children who are at risk, where parents protect the children by separating them from the affected parent so they would not have to watch him/her ‘fall apart’ in front of their eyes [21, 23, 33]. Nevertheless, while in some cases avoidance may be effective, in the long run
it increases carers’ anxiety, negatively influences one’s relationships with relatives and may prevent them from seeking and/or accepting external help. It also impairs one’s ability to plan ahead and make anticipatory arrangements for the future [21]. Thus, it can be said, that while HD is rarely spoken about, subconsciously it is always present.

Discussion

The available literature suggests that there are many similarities between caring for pHD and person suffering from other neurodegenerative diseases [30, 50]. As pHD gradually loses his/her psychomotor and cognitive functions, HD changes the relations within the family and allocation of daily tasks between its members. It influences the nature of relationship with the affected relative and becomes a source of emotional burden which includes role changes and loss of intimacy with the loved one. Additionally, as caregivers are on ‘on-call’ for 24 hours, seven days a week, they have little time for themselves and experience disintegration of one’s life [19, 32]. They are often forced to abandon their leisure activities, give up vacations or hobbies, have little time for other family members and spend less time with their friends. Studies also report their financial burden and lack of support. Moreover, like other carers, they experience deterioration of health. And while some studies emphasize the positive aspects of caregiving as HD may bring one’s family closer and can give a carer a feeling of mastery [18, 27, 39] most studies show that they experience feelings of helplessness, loneliness, anger and frustration [19, 27, 28]. Consequently, carers have problems with maintaining normal life and describe it as ‘being lost’ [32, 38]. Such feelings are the strongest among teenage carers who are forced to take many adult responsibilities for which they are not prepared. As a result, they feel overwhelmed by the caregiving role and wish to live a ‘normal life’ and have a possibility of ‘being truly oneself’ [33, 46]. All in all, like in other diseases, HD carers’ quality of life is seriously impaired in terms of burden [16, 22], social isolation [30, 38, 40], ability to cope [21], financial costs [30, 32], access to healthcare services [19, 29], lack of knowledge form health professionals [11, 12, 18, 19, 28, 29, 38] and decline in health [12, 25, 32].

At the same time, it is important to emphasize that there are some factors associated with HD that are not present in other neurodegenerative diseases [16, 18, 25, 26, 30, 32]. First of all, as the onset of HD begins much earlier, approximately at the age of 35–40, carers of pHD are also relatively younger than those caring for patients with Alzheimer’s or Parkinson’s disease [32]. Consequently, such carers may still have young children to look after and it may be difficult for them to combine the duties of a double carer, especially that at the same time they carry out their professional responsibilities [21, 32]. Secondly, as HD is characterised by slower progression of symptoms, it has a longer trajectory [32]. Thus, caring for pHD lasts much longer, typically 30 years. It also involves activities that are unique to HD [38].

Thirdly, as HD is a hereditary disease, it puts other family members at risk [16, 21]. For that reason, carers are anxious that they will have to take care of multiple
generations of sufferers and that their children may develop HD in the future [16, 21, 24, 25, 27, 30–33, 36, 38, 40]. Such knowledge is especially stressful to sons and daughters of pHD who are ‘watching and waiting’ [33, 39]: while caring for the sick, they observe what may happen to him/her [25, 36]. As Kessler [13, 16] observes, it is not uncommon that a person with HD nurses a parent, then an older sibling and finally develops HD him/herself at the same time worrying all the time that he/she might have passed the disease to his/her children. Consequently, as is estimated that for every person affected with HD there are another 10 people who suffer from its consequences, including those at risk, the unaffected spouse and other family members [13, 16, 21, 22, 36, 38, 40]. HD is a prime example of family disease.

Finally, both society and health care professionals lack basic knowledge on HD and are not aware of the needs and problems of pHD and their carers [17–19, 27–30, 38]. Moreover, other types of dementia are not only known better but health and social services for such patients are also less limited, better organised and adjusted to their needs [11, 29, 36].

Conclusions

While some limitations may exist in this paper as the numbers of eligible studies accumulated may not be sufficiently large for a comprehensive analysis, some advantages should also be acknowledged. First of all, it shows that while HD carers have many similar needs and problems to those faced by caregivers of patients with other neurological disorders social implications of HD are much more serious and far-reaching [47, 50], which makes looking after pHD more demanding and stressful. It also emphasizes that as most of care for pHD is provided by family caregivers, they are in need for health assistance and policies that will support their (unmet) needs. It seems that recommended strategies to help such carers would include: assessment of their health, planning care for the affected person, providing carers with adequate information on HD, its symptoms and their management and available treatment and psychological support. At the same time, it is important to note that such supportive care is needed from the time of the diagnosis throughout the prolonged trajectory of HD which mainly requires psychological and practical support, including adequate information on HD and financial help [32, 38]. Additionally, as some authors suggest, interventions which aim to help caregivers do not have to be focused on the caregiving itself, but may concentrate instead on problem-solving strategies, i.e. helping carers to plan strategy to deal with general life stressors and those associated with caregiving [25]. For that reason one must agree with Zielonka, Marcinkowski and Klimberg who claim that ‘Apart from changes in the regulations, more intense educational activity is necessary concerning the demand for the organization of rehabilitation, logopaedic, dietetic exercises, spreading a ‘protective parachute’ over the families of the patients, organization of support groups for families, in which Huntington’s disease occurred’ [45]. It is important as most HD carers feel let down by the system and express general
lack of trust in the standard of care [21, 38]. Meanwhile, carers who experience supportive social context tend to perceive the caregiving task more positively and have an increased perception of personal control [25, 27]. For that reason, the paper stresses that the view of caregivers should be instructive to those providing professional care and counsel. Thus, while in medical setting the HD patient is the one who draws professional attention, it is often the unaffected family carer who needs the most attention, support and help. For that reason, health professionals should monitor caregivers’ mental health, identify the sources of their distress and support effective strategies to cope with stress. It is of key importance, as for the most carers psychological factors, i.e. social support and coping strategies play a more crucial role in their well-being than the medical ones [19].

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References


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