Duchenne’s Muscular Dystrophy

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Duchenne Muscular Dystrophy (DMD) is an X-linked, degenerative, neuro-muscular disorder with an estimated male birth incidence of 1:3800 to 1:6200. The disease is characterized by a progressive degeneration of muscle fibres resulting in muscle weakness and eventual loss of ambulation. Functional dependence typically occurs in the second decade of life with cardiac and respiratory complications often shortening life. Other types of muscular dystrophies, such as Becker’s muscular dystrophy (BMD) and limb-girdle muscular dystrophy (LGMD), have similar progression to DMD but a near normal life expectancy with symptoms appearing later, being less severe, and thus preserving ambulation often to mid-life.

psycho-social needs | muscular dystrophy | Duchenne’s muscular dystrophy | parents caregivers | family burden/care burden

1. Introduction

Duchenne Muscular Dystrophy (DMD), as the most common and severe form of the disease, is the focus for this review. Disruption to daily life with DMD can commence at an early age and not only impacts the child but also the family. The nature of the disease itself and care required places a heavy burden on parent caregivers for an extended period of their lives. Improvements in supportive care for children and young people with DMD have resulted in improved quality of life and life expectancy, but, until recently, drug therapies had seen little change. This situation is rapidly changing with the emergence of new therapies that address underlying genetic defects setting a change in course for DMD treatment [1]. Antisense oligonucleotides (ASO), are new therapies that modify disease pathways by targeting underlying genetic changes. There is now potential to prevent clinical features of disease occurring with early intervention [2][3]. Another new treatment is Ataluren (Translarna); this is not an ASO, but this drug demonstrates that the non-sense mutation a genetic defect causing some types of DMD, can respond to treatment [3]. In a disease that has been seen as incurable for so long, these novel treatments offer a much-improved outlook for the future.

2. Living with DMD

2.1. Psycho-Social Impacts

Parents reported having feelings of loss, sadness and depression [4][5] as they lived with their child’s condition but identified their main psychological issue as their distress and worry for the future [6][7]. These parents experienced

significantly greater stress than parents of healthy children and where their child also had difficulties in social interactions, this further increased stress levels [8]. Generally, parents felt they had coping mechanisms for day-to-day living but reported struggling, at times, with both interventions and behavioral changes [6]. In addition, parents not only worried for their affected child but also for the negative influence DMD had on the psychological well-being and social life of the siblings [5][7].

Although continually living with stress and worries for their child and family, peaks in parental stress levels were experienced at life-changing moments, notably the time of diagnosis, the point where disease progression rendered their sons immobile, when a powered wheelchair or non-invasive ventilation (NIV) became necessary, and at the death of peers [8][9][10]. These major events marked out disease progression with parents identifying their child's loss of ambulation as one of the most difficult challenges to cope with emotionally [8]. As parents struggled with the psychological, physical, and, for some, financial impacts of their child's disease, they often disengaged from their own hobbies and social activities [7].

Despite the many negative psychological impacts of being a parent of a child with DMD, parents repeatedly identified their experiences as positive [5][7][11][12]. Those most positive about their situation were long term caregivers and those who viewed their child as being both sensitive and talented: very few mothers reported any negative feelings about assisting their child [11]. The parents' experiences often changed their personal life values and increased their strength and courage in facing adversity [5]. Parents were themselves ageing as their child's dependence was increasing, but they felt increased confidence from having raised their children and satisfaction with their work as caregivers [12]. In contrast, some parents who had developed health problems themselves (sight issues, back problems and hypertension) had concerns about not being as able to care for their child as previously [12].

2.2. Physical and Financial Impacts

Home management of their child's care was a demanding role for parents and this physical care burden was perceived as greatest where the child had suffered with the disease longer, had lower functional ability, and was more dependent on caregivers [5][7]. The demands on parents were extensive with many reporting night-time wakening to give care and respond to equipment alarms, especially in the later disease stages due to the child's immobility and need for non-invasive ventilation (NIV). Malfunction or dislodgement of NIV can be fatal for the child should the care giver not intervene, and these nighttime care demands negatively impacted the quantity and quality of sleep for parental caregivers, with those less experienced being most adversely affected [10]. Perception of the extent of the care burden was linked with parental access to social contact and support from friends, family, and professionals, especially in emergency situations [5][7].

The nature of the disease meant there was a growing physical dependency on parents as the child matured and an increasing financial burden for some families [10][13]. As their child grew into a young man, parents anxiously anticipated their own ageing, retirement, and the changes in family relationships and structures as their other

children grew up and had families [12]. For parents, these events coincided with increasing care needs and financial burden for them alongside the loss of their own primary caregiver role [12].

Parents admitted harboring some regrets for the life constraints that DMD had imposed on them, some of which were financial [12]. Some studies have demonstrated that families with a child with DMD have a lower than national median income with many costs associated with care provision resulting in substantial economic burden for families [13][14], and, in low socio-economic countries, the impact on families of the disease was even greater [9]. Economic worries were real, and families found it difficult to escape poverty or even think about how to increase their income [12].

2.3. Building Resilience

2.3.1. Adapting

Resilience is not just the ability to bounce back from adversity but the process of adapting [15]. There is a need to foster the mother's resilience using psycho-social interventions aimed at improving acceptance by identifying the positive aspects of living with DMD rather than just the burden and deficit [5][16][17].

Psycho-social support should start when the child is young, and those involved in caring for families should assess unmet needs. Being proactive in identifying the need for help and understanding the family's fears and uncertainty also helps in the identification of resources needed to prioritize and customize interventions building on family strengths [18].

2.3.2. Wellbeing

Parental health is seen as a necessity for family adaptation [5][17][19]; however, the requirements for good parental health are varied. The family environment can contribute to or mitigate burden [12][18]. Family and partner support are seen as important [18]. It has been found that an intact family structure may influence family hardiness and the provision of emotional support [18][19][20]. If the parent is single, or the child not of school age, there is greater need for social and professional support to manage care and minimize burden [5].

Parents of children with DMD experience higher levels of stress than other parents [17][21], and it is reported that distressed or depressed parents may become frustrated and see their children as more of a burden [17]. Where stress is related to their child, or problem behaviors, such as clinginess and poor socialization skills, interactions between mother and child also become more stressful [21]. This correlation between parental stress and psycho-social adjustment can lead to a decline in good parenting skills and poorer coping mechanisms [17].

Good parental health and management of stress is reliant on responsive community services [4][5][19] and supportive health professionals [18]. Attendance at support groups, where parents can discuss their fears and anxieties, gain advice on resources, and expand their knowledge, is seen to reduce levels of stress and aid good psycho-social adjustment [17].

2.3.3. Socializing

Creating opportunities for socialization is an important intervention, not just for the young person but the parent [4][22][23]. Social support is frequently used by caregivers as a coping strategy [23], and, whilst it can be challenging, the benefits are clear [22]. Parents found that engaging in support groups gave them access to practical advice, emotional support, and helped them to understand their child's condition better [23]. Sharing and improving knowledge is linked with active coping [24], giving parents power and the voice to advocate for their child [4].

Access to social support and the opportunity to socialize outside of the home can allow the caregiver to escape (24) or get away (8) from caregiver responsibilities. This becomes more important as DMD progresses as there is a potential for caregivers to reduce social engagement to cope with caring responsibilities and, thus, further increase burden. To avoid this vicious cycle of events, there is a need to build upon existing social supports to improve the psychosocial outcomes for families coping with the effects of DMD.

2.3.4. Escaping

There was acknowledgement that parents could benefit from having some time-out from their role and responsibilities. Escaping the ongoing daily pressures could provide some well needed relief for parents. So, paid employment may not only have financial benefits but also could be a means of escaping care burden even if only for short periods of time [5][19]. This time out is important.

Parents also identify exercise and self-care activities as being necessary for their overall health [18]; however, many neglect hobbies [5] and spend little time on social activities and rest [20]. Over time, as their child matured and became more dependent, the parents' neglect of their own well-being often increased [7], and their quality of life deteriorated [25]. Respite care can improve caregiver burden [19], but uptake is low [17].

What factors influence accessibility of psycho-social interventions for these caregivers?

Accessibility of psycho-social interventions for caregivers was not a central focus of the studies; therefore, this research question was not fully addressed, indicating a need for further research in this area. Researchers noted variation in the level of professional and social support for parents within their studies, but factors influencing this situation were not explored. For example, it was noted that uptake of respite care is low [17], but the reasons for this are not clearly established. It is, however, suggested that it could be 'too time consuming to organize' or due to maternal anxiety about relinquishing care [18]. It is possible there are many issues underlying the decision making in accessing respite services; therefore, to fully understand this, future research needs to concentrate on uncovering the potentially complex influencing factors.

Similarly, there is recognition that good parental health and management of stress is reliant on responsive community services [4][5][19] and supportive health professionals [18]. However, some families struggled to access resources and had to fight for services, identifying a lack of joined up thinking as a barrier to consistent care [4].

Without a fuller understanding of these critical access issues, it is difficult to draw any conclusions regarding factors influencing this situation.

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