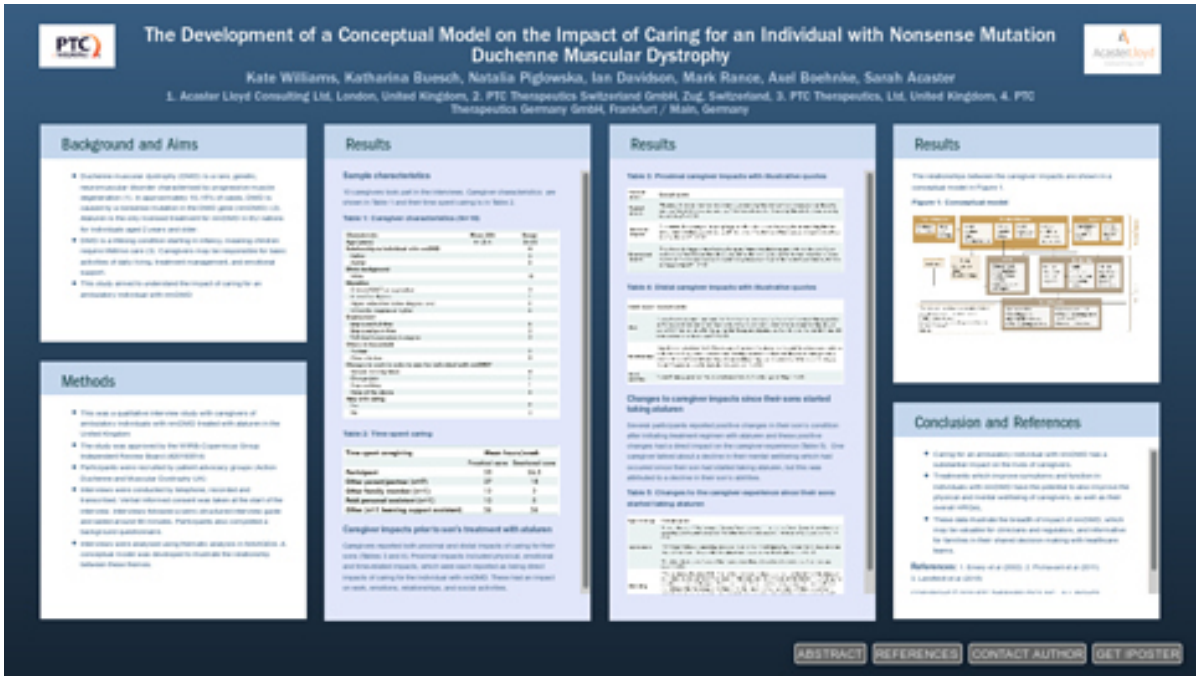
The Development of a Conceptual Model on the Impact of Caring for an Individual with Nonsense Mutation Duchenne Muscular Dystrophy



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BACKGROUND AND AIMS

- Duchenne muscular dystrophy (DMD) is a rare, genetic, neuromuscular disorder characterised by progressive muscle degeneration (1). In approximately 10–15% of cases, DMD is caused by a nonsense mutation in the DMD gene (nmDMD) (2). Ataluren is the only licensed treatment for nmDMD in EU nations for individuals aged 2 years and older.
- DMD is a lifelong condition starting in infancy, meaning children require lifetime care (3). Caregivers may be responsible for basic activities of daily living, treatment management, and emotional support.
- This study aimed to understand the impact of caring for an ambulatory individual with nmDMD

METHODS

- This was a qualitative interview study with caregivers of ambulatory individuals with nmDMD treated with ataluren in the United Kingdom
- The study was approved by the WIRB-Copernicus Group Independent Review Board (#20193514)
- Participants were recruited by patient advocacy groups (Action Duchenne and Muscular Dystrophy UK)
- Interviews were conducted by telephone, recorded and transcribed. Verbal informed consent was taken at the start of the interview. Interviews followed a semi-structured interview guide and lasted around 90 minutes. Participants also completed a background questionnaire.
- Interviews were analysed using thematic analysis in MAXQDA. A conceptual model was developed to illustrate the relationship between these themes.

RESULTS

Sample characteristics

10 caregivers took part in the interviews. Caregiver characteristics are shown in Table 1 and their time spent caring is in Table 2.

Table 1: Caregiver characteristics (N=10)

Characteristic	Mean (SD)	Range
Age (years)	44 (5.4)	26-52
Relationship to individual with nmDMD		N
Father		5
Mother		5
Ethnic background		
White		10
Education		
O level/GSCE or equivalent		3
A Level or Highers		1
Higher education below degree level		0
University degree or higher		6
Employment		
Employed full-time		6
Employed part-time		2
Full-time homemaker/caregiver		2
Others in household		
Partner		9
Other children		8
Changes to work in order to care for individual with nmDMD*		
Reduce working hours		4
Change jobs		1
Stop working		1
None of the above		5
Help with caring		
Yes		7
No		3

Table 2: Time spent caring

Time spent caregiving	Mean hours/week	
	Practical care	Emotional care
Participant	33	26.3
Other parent/partner (n=7)	27	18
Other family member (n=1)	12	2
Paid personal assistant (n=1)	10	5
Other (n=1 learning support assistant)	36	36

Caregiver impacts prior to son's treatment with ataluren

Caregivers reported both proximal and distal impacts of caring for their sons (Tables 3 and 4). Proximal impacts included physical, emotional and time-related impacts, which were each reported as being direct impacts of caring for the individual with nmDMD. These had an impact on work, emotions, relationships, and social activities.

RESULTS

Table 3: Proximal caregiver impacts with illustrative quotes

Proximal impact	Example quotes
Physical impacts	*"Physically it had an impact on me because I was carrying him a lot, so I was carrying him up the stairs, carrying him out of the school, carrying if we were out as a family carrying him out of the car, so doing lots of lifting"* – C101
Emotional impacts	*"Just constantly worrying if I'm going to get a call he's been rushed into hospital or something like that, just my mind overthinking, overrunning all the time, thinking that something's going to happen, and always thinking the worst"* – C105
Time-related impacts	*"I would say the biggest impact is hospital appointments and physio appointments. You have to fit your work around these because they all occurred during the working day and in the week, Monday to Friday. So, lots of time was spent going to hospitals or getting cover to go to the hospital appointments, that was the biggest impact"* – C110

Table 4: Distal caregiver impacts with illustrative quotes

Distal impact	Example quotes
Work	*"I would work very minimum hours, I think at the time I was working in a school as a lunchtime supervisor so I'd maybe do two and a half hours a day just purely because I needed to be around for him, he was like a 24/7 type of job with him, giving him the support physically and mentally so it meant that I couldn't have a career or a full-time job"* – C108
Relationships	*"Myself and my husband had different ways of approaching things, so his would have been very much live in the here and now, where I was constantly looking towards the future and things that could go wrong and I think we just had different ways of approaching things…there was a few instances where we just, we didn't agree on what the best plan forward was"* – C102
Social activities	*"I wasn't feeling great so I would cancel social events, I wouldn't go to things"* – C101

Changes to caregiver impacts since their sons started taking ataluren

Several participants reported positive changes in their son's condition after initiating treatment regimen with ataluren and these positive changes had a direct impact on the caregiver experience (Table 5). One caregiver talked about a decline in their mental wellbeing which had occurred since their son had started taking ataluren, but this was attributed to a decline in their son's abilities.

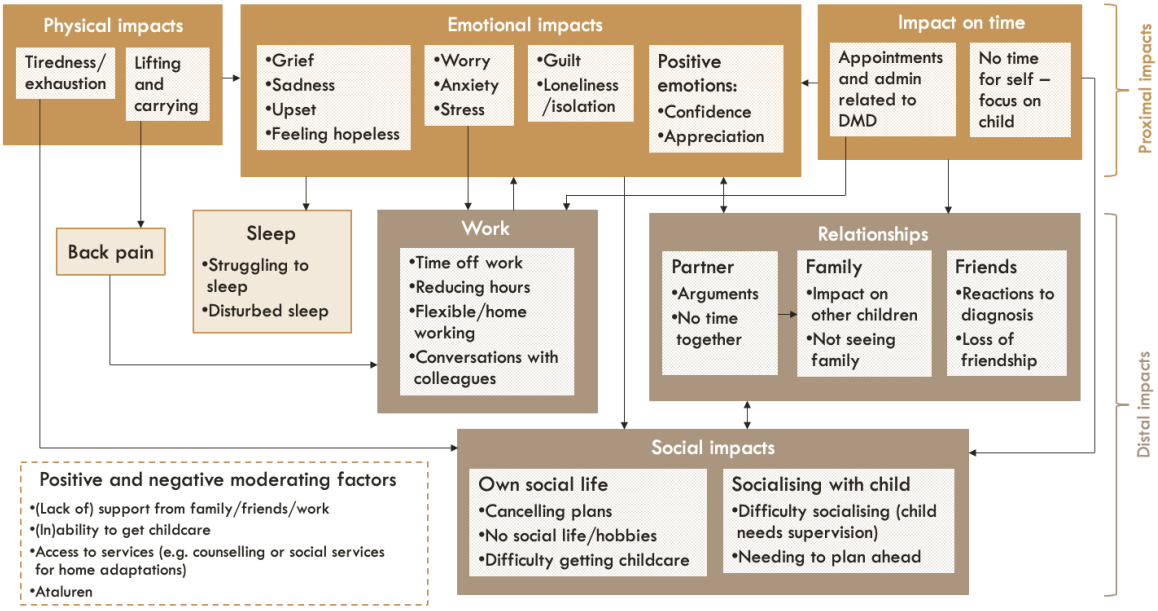
Table 5: Changes to the caregiver experience since their sons started taking ataluren

Type of change	Example quotes
Improvement	*"I mean physically it's had an impact because I'm not carrying him as much so I'm not having to give him quite so much physical support, so my health is better in that sense because I'm, my back isn't giving out so much"* – C101 *"With [son]'s behaviour improving and his ability to control himself improving, it meant that the daily stress and worry of how [son] interacts with other children and his brother was lifted significantly"* – C110 *"I'm able to have more of a social life, I can do more things. He can be left alone for you know hours and hours"* – C108
Worsening	*"We [partner] both found 2018-19 ish quite a hard year emotionally with some of his decline in his abilities. I have seen a psychologist as well, the same one that [son] sees. My eldest son saw her because he was struggling as well, so it's not only the impact it's having on [son], it's the impact it's having on my older son as well. And then obviously that has an impact mentally on me. So, I don't mind saying to you that when I was seeing the psychologist, she said to me that I was moderately depressed and I have started taking medication for that"* – C107

RESULTS

The relationships between the caregiver impacts are shown in a conceptual model in Figure 1.

Figure 1: Conceptual model



CONCLUSION AND REFERENCES

- Caring for an ambulatory individual with nmDMD has a substantial impact on the lives of caregivers.
- Treatments which improve symptoms and function in individuals with nmDMD have the potential to also improve the physical and mental wellbeing of caregivers, as well as their overall HRQoL.
- These data illustrate the breadth of impact of nmDMD, which may be valuable for clinicians and regulators, and informative for families in their shared decision making with healthcare teams.

References: 1. Emery et al (2002). 2. Pichavant et al (2011). 3. Landfeldt et al (2018)

AUTHOR INFORMATION

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ABSTRACT

Objectives

Duchenne muscular dystrophy (DMD) is a rare genetic neuromuscular disorder characterised by progressive muscle degeneration and weakness. Loss of muscle function starts early and worsens over time, resulting in loss of motor function and premature death. Individuals with DMD can require 24-hour care, but little is known about the extent to which this impacts the lives of their caregivers. This study explored the impact of caring for an individual with nonsense mutation DMD (nmDMD) before complete loss of ambulation.

Methods

Qualitative interviews were conducted with caregivers of individuals with nmDMD treated with ataluren in the UK. An interview guide was developed with input from clinical experts and patient advocacy groups and included open-ended questions to elicit information on the impact of caring for an ambulatory individual with nmDMD. Interviews were conducted by telephone, recorded and transcribed. Data were analysed using thematic analysis and saturation was recorded. A conceptual model was developed to illustrate the reported caregiver experiences based on the data.

Results

Ten interviews were conducted with parents of individuals with nmDMD aged 4-19 years at various stages of ambulation. Caregivers reported both proximal and distal impacts. Proximal impacts were physical (e.g. lifting their child), emotional (e.g. anxiety/worry/stress) and time-related (e.g. administrative tasks). These were associated with a range of more distal impacts including the impact on work (e.g. time off work due to back pain), relationships (e.g. with partner) and social life. These impacts and relationships between them were illustrated in a conceptual model.

Conclusions

Caring for an individual with nmDMD has a substantial multifaceted impact on the lives of caregivers and those around them. Treatments for individuals with nmDMD which have the potential to improve symptoms, reduce loss of function, or delay progression, may also have a positive impact on the quality of life of caregivers.

REFERENCES

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