Understanding anxiety experienced by young males with Duchenne Muscular Dystrophy: a qualitative focus group study

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Highlights

- The first qualitative study of the experiences of anxiety in young people with DMD.
- We identified six anxiety characteristics describing the anxiety phenotype in DMD.
- Anxiety in DMD is influenced by external factors and endogenous anxiety traits.
- Standard anxiety scales may underestimate anxiety symptoms in the DMD population.
- Many parents were dissatisfied with existing mental health support for their child.

Abstract

In this multi-methods study we explored the characteristics, causes and impact of anxiety in Duchenne Muscular Dystrophy (DMD) from the perspective of young males with DMD and their parents. Eight young males with DMD (7-18 years) and 14 parents participated in separate focus groups. Perspectives on anxiety were explored using semi-structured interview schedules. Themes were identified using Framework Analysis. Neurodevelopmental, emotional and behavioural symptom scores were obtained using standard instruments including the Strengths and Difficulties Questionnaire and Revised Children's Anxiety and Depression Scale. We identified six common anxiety characteristics: Catastrophic conclusions; Rigidly-held anxieties; Extreme distress; Social anxieties; Physical changes/needs; Unexpected/unfamiliar. Four further themes described influential systemic factors:

Individual, Family, and Social responses and Physical environment and service contexts. All DMD participants had significantly higher total difficulties, emotional problems and impact scores than population norms. The Revised Children's Anxiety and Depression Scale showed low sensitivity in identifying anxiety symptoms. Fifty-seven percent (8/14) of parents who had wanted help for their son's anxiety were dissatisfied with the available support. In conclusion, anxiety can severely impact wellbeing and functioning of individuals with DMD. There are important nuances to consider when managing DMD-associated anxiety. We highlight the importance of multimodal assessment considering the multiple contexts within which anxiety arises.

Keywords

Duchenne muscular dystrophy; Anxiety; Qualitative Research; Focus Groups; Psychometrics.

Abbreviations

ADHD - Attention deficit hyperactivity disorder; ASD - Autism spectrum disorder; CBCL - Child Behavior Checklist; CNS - Central nervous system; DMD - Duchenne muscular dystrophy; DSM - Diagnostic & Statistical Manual of Mental Disorders; ICD-10 - International Classification of Mental and Behavioural Disorders 10th revision; RCADS - Revised Children's Anxiety and Depression Scale; SCAS – Spence Children's Anxiety Scale; SCDC – Social Communication Disorders Checklist; SDQ - Strengths and Difficulties Questionnaire; SMA - Spinal muscular atrophy; UCL - University College London.

1 Introduction

Duchenne muscular dystrophy (DMD) is a life-limiting, X-linked muscle-wasting disorder affecting 1:5000 male births [1], caused by mutations in the *DMD* gene encoding the protein dystrophin. Lack of dystrophin causes progressive muscle-wasting, cardiomyopathy and can be associated with central nervous system (CNS) disturbance [2]. With current standards of care, average lifespan has increased from two to three decades with further increases likely in future with improving therapies focused on the progressive muscle weakness [3, 4].

The lack of brain dystrophin proteins is associated with higher rates of intellectual disability, autism spectrum disorder (ASD), attention deficit hyperactivity disorder (ADHD), behavioural problems and anxiety disorders in individuals with DMD than the general population [2, 5]. With improvements in physical health, optimising mental health is increasingly important as poor mental health can significantly affect wellbeing, social participation and adaptive functioning. Recent meta-analyses confirmed an anxiety disorder prevalence of 24.0% [5] in DMD compared to 7.2% in typical 5–19-year-olds, and 19.1% in young people with life-limiting conditions [6].

The 2018 DMD care guidelines recommend mental health screening at neuromuscular clinic visits and a specialised mental health professional within the neuromuscular team [4], reiterated in a 2021 report by the advocacy group Muscular Dystrophy UK [7]. However, in the UK people with neuromuscular disorders have struggled to access appropriate psychological support [8]. Due to the complexities of the condition, anxiety may present differently in DMD compared to typically developing individuals, and some anxiety screening instruments may not adequately capture the symptoms experienced in DMD [9]. Two instruments widely used in UK

practice are not validated in DMD: the Strengths and Difficulties Questionnaire (SDQ) [10] and the Revised Children's Anxiety and Depression Scale (RCADS) [11].

Anxiety has multi-factorial causes, including interactions of genetic and environmental risk. Evidence from DMD animal models and psychophysiological tasks in young males with DMD suggests a neurobiological contribution, with dysfunctional anxiety-related fear responses directly linked to dystrophin deficiency in the brain [12, 13]. Social and environmental responses towards an individual with DMD compared to an unaffected child may also influence anxiety. Additionally, stress and emotional responses are common parts of adjusting to chronic, life-limiting conditions, risking the pathologising of normal responses to difficult situations and of coping behaviour [6, 14].

To explore the anxiety phenotype in DMD, we conducted a qualitative focus group study to gain insights into the nature and causes of anxiety in DMD and its impact on these children and their families, and a quantitative analysis of parent-report anxiety questionnaires.

2 Materials and methods

This multi-methods study of anxiety in young males with DMD incorporated qualitative and quantitative data obtained during focus groups at UCL Great Ormond Street Institute of Child Health. Ethical approval was granted by UCL Research Ethics Committee (Project ID 12307/001).

2.1 Participants

Participants were recruited via DMD advocacy groups and a research database. Eligibility criteria were: child/youth participants – aged 7-18 years, male with a

diagnosis of DMD; parent participants – having a child with DMD under the age of 18 years. Eight young males with DMD were recruited to two groups of four (Child: 7-11 years; Youth: 12-18 years); 18 parents of young males with DMD were recruited into three groups of six, of whom 14 parents attended (13 female, 1 male). There were six parent and child participant pairs from the same family and only one parent from each family took part. Group sizes and the duration of sessions were informed by previous literature [15, 16].

Prior to commencement of the focus groups a group discussion took place to explain the study procedures and ground rules for the focus groups. Individual consent discussions then took place in private with each participant or parent/child pair. All parents gave informed written consent, either as participants or as parents of participants, child/youth participants under 16 gave informed written assent and youth over 16 years gave informed consent, using ethically-approved informed consent/assent forms. All participants agreed to audio- and video-recording of the groups, to be used only for the purposes of transcription and not for publication themselves.

2.2 Qualitative study design – focus groups

A flowchart giving an overview of the study design is shown in Appendix B (Fig. B1). Focus group discussions followed semi-structured interview schedules and covered: features of anxiety; impact of anxiety (child/family); responses to anxiety (Appendix B). Additional creative methods were devised for child and youth groups to encourage discussion, drawing on ideas from child focus group literature [17, 18]. These included a card-sorting task comprising pictures of different situations to sort into ‘big’, ‘middle-sized’, and ‘little’ worries (Appendix Fig. B.2). The schedules were independently reviewed by two clinical psychologists (WM & JP). Focus groups were

audio- and video-recorded and facilitated by a trainee clinical psychologist (RT) and a neurologist (KM), with clinical experience in child and adolescent mental health and young people with DMD respectively.

2.3 Quantitative data collection

Parents, including those who participated in focus groups, provided anonymous data regarding their child (demographics, previous neurodevelopmental or mental health diagnoses, provision of mental health support), and consented to publication of anonymised data.

Parents completed the following questionnaires about their child(ren) with DMD: The SDQ [10]; RCADS [11]; Social Communication Disorders Checklist (SCDC) [19]; Child Behavior Checklist (CBCL) [20]; Spence Children's Anxiety Scale (SCAS) [21]. This range of scales was chosen to enable comparison of data between different population and DMD cohorts to situate the sample and also to act as a stimulus for further discussion in focus groups allowing a qualitative evaluation of the scales. The SDQ and RCADS are recommended instruments used in NHS child and adolescent mental health services, the SDQ is recommended for use in young people with DMD [4, 9]. Our group has previous experience of using the SCDC in a large DMD cohort, and this is also validated in typical UK populations [2, 22]. A recent systematic review of psychometric instruments in DMD has suggested that the CBCL is less well-suited to the DMD population [9], however we included this scale and the SCAS to enable comparison of these data with data from our studies on other DMD cohorts which have used these instruments [2, 13]. As not all the scales were available for the younger age group in self-report format and to reduce the burden on child and youth participants, only parent-report questionnaire data were gathered.

2.4 Data analysis

Audio- and video-recordings were transcribed and processed using NVivo 12 software (QSR, 2018). Transcriptions were evaluated using Framework Analysis to identify themes [23]; a thematic framework applied to the data, to enable indexing, charting, mapping and interpretation. Coding validity was reviewed with four parent and two child participants and an independent researcher (RT). Audio/video recordings were deleted following transcription, and quotes assigned with anonymised identifiers.

Pre-morbid neurodevelopmental and mental health diagnosis data were extracted from baseline demographic information questionnaires. These were compared with previous prevalence data from other DMD studies and typical population data by calculating Z-scores (Appendix, Table A.2). The DMD prevalence data were weighted proportions derived from a meta-analysis conducted by our group [24], which only included studies using direct assessment/formal diagnosis as outcome measures, as it was not possible to calculate Z-scores using the available data from another published systematic review and meta-analysis [5]. Typical population prevalence data was taken from national population surveys of children and adolescents in England, where possible using population data for males only for the most accurate comparisons [25-27].

Parent-report questionnaire data were scored according to the instrument guidelines. Scores were compared against data from previous DMD cohorts studied by our group using independent samples *t*-tests in IBM SPSS v.24.0, to determine whether this study cohort was similar to other DMD research cohorts. Quantitative data were examined alongside the qualitative data and integrated where appropriate.

3 Results

3.1 Neurodevelopmental and neuropsychiatric diagnoses and symptom profile

Parent-report questionnaire datasets were gathered for 18 DMD children, which were completed by 16 parents (two of whom had multiple children with DMD) (Table 1). Previously diagnosed neuropsychiatric and neurodevelopmental conditions are shown in Table 2. Parents of two youths included in the cohort reported their child took medication for a diagnosed condition: one for anxiety and one for ADHD.

The prevalence of anxiety diagnoses of 11% in this cohort was non-significantly lower than anxiety prevalence reported in a recently published meta-analysis (24%) [5] or our own group's meta-analysis data (22%; Table 2 and Appendix Table A.2; p=0.27). However, anxiety-specific symptom scores in the current study cohort were significantly higher in the Youth group than in previous DMD cohorts tested using the same anxiety measure (the SCAS; Appendix Table A.1), with trends of higher scores on the other instruments used.

Within our cohort, the percentage of participants with significant social communication problems (41%) and ASD diagnoses (28%) were significantly higher than the published 7% prevalence in DMD [5] and our meta-analysis DMD prevalence of 17% (Table 2; Fig. 1A). Of the 41% (7/17) boys scoring in the highest range for social communication problems (which suggests a likely diagnosis of ASD) three had a prior diagnosis of ASD, two had suspected or 'borderline' ASD and two had no prior diagnosis.

The comparison of prevalences of other previous neurodevelopmental and mental health diagnoses in our cohort with previously reported DMD population cohorts, included a lower proportion of diagnoses of intellectual disability, depression and ADHD, and a similar prevalence of obsessive-compulsive disorder diagnosis (Table 2). Compared to the typical child/adolescent male population, our study cohort had significantly higher diagnoses of ASD, ADHD and OCD (Table 2).

Emotional and behavioural data from parent-report questionnaires (shown in Fig. 1A) assessing behaviours, emotional symptoms and relationships identified ‘very high’ SDQ emotional problems scores in 19% (3/16) (vs. 5% in typical population), and ‘clinical anxiety’ range scores on the RCADS in 6% (1/18) (vs. 2% in the typical population). The SDQ data also showed significant differences when compared with the typical paediatric population: ‘very high’ scores on ‘Total Difficulties’ subscale in 25% of our cohort (vs. 5% in the typical population), and ‘high’ or ‘very high’ Impact Scores in 69% (vs.10% in the typical population).

3.2 Characteristics of anxiety and relevant contextual factors

Using Framework Analysis, we identified six key characteristics of anxiety and four influential systemic factors incorporating individual, family and social responses and physical environment and service contexts (Table 3). Participant quotes illustrative of each theme are shown in Table 4.

Three anxiety characteristics related to internal states and their expression: catastrophic conclusions, rigidly-held anxieties and extreme distress. **‘**Catastrophic conclusions’, such as worries about extreme eventualities (tsunamis, burglary, terrorist attacks, fatal accidents), were reported by 57% of parents and all young

DMD participants. Rigidly-held anxieties were reported by 57% of parents, including fixating on and repeating particular worries, and inflexible thinking about them. Whilst young participants did not reflect on these thinking patterns, we observed repetitive conversations during their focus groups. Young males experiencing extreme distress, or “meltdowns”, in response to anxiety were identified by 93% of parents, describing big reactions to seemingly trivial triggers and greater difficulty controlling emotional responses when compared to peers. Two Child participants also described extreme fear-distress responses. However, comments from Youth participants and parents of adolescents indicated that the frequency and severity of difficulties with unfamiliar and unexpected situations, and the occurrence of ‘meltdowns’, reduced for some children as they got older.

The remaining anxiety characteristics concerned external situational factors: anxieties about physical changes or needs, social situations, and unexpected or unfamiliar situations. Anxieties about physical changes and needs were reported by 100% of young males and 86% of parents. Child participants mentioned this theme 2.3 times more frequently than parents. DMD participants connected their anxieties about wheelchair use with accessibility issues, the responses of other people, and their physical vulnerability. Several parents and youth described anxieties about physical decline and the future implications of this, including worries about death (parent-reported) and employment (youth). Child participants did not mention physical decline or death. All DMD participants and 57% of parents described social anxieties, including worries about doing activities which highlight their physical disability in front of others, for example younger children playing sports, or youths being hoisted into a swimming pool. Despite the relatively less advanced features of DMD in younger children, this still provoked anxiety for some of the Child

participants, whereas anxieties relating to visible differences were referred to more often by the Youth than Child participants. Unexpected or unfamiliar situations triggering anxiety and distress was recognised by 64% of parents, and half of child participants identified experiencing anxiety in such situations in the card-sorting task.

We identified four broad systemic factors that could both influence and be influenced by anxiety: Individual, Family and Social responses, and Physical environment/ service contexts (Table 3).

Many individual factors connected with anxiety were described by parent and young participants. Physical functions including sleep, toileting, and vomiting were prominent and existed in circular relationships with anxiety for some children. For example, poor sleep quality due to worries at night led to further increased anxiety due to tiredness. One parent described her son vomiting every morning on the way to school due to extreme school-related anxiety.

Varied individual coping strategies and responses were reported, such as "*taking it* [anxiety] *out on the ones you love*" [Child], and some parents reported their son avoiding information about DMD, withdrawing from social situations, sabotaging potentially exposing physical activities, and concealing signs of their condition. Helpful coping responses reported by young participants included using humour, playing computer games to calm down, focusing on the present rather than the future, confronting others who stare or ask personal questions, and "*instead of thinking about what you don't have, thinking about what you do have*" [Child].

Family factors included the emotional impact of the child's anxiety on family members, such as parental feelings of helplessness, and finding the frequent questioning and difficulties with spontaneity "*wearing at times*" [Parent], which

caused daily problems for some families. Some parents worried that their own feelings about their son's physical and mental health, such as guilt, helplessness, anxiety, sadness, and physical and emotional exhaustion, further impacted upon his anxiety. Parents reported that their strategies for responding to anxiety were less effective in their son with DMD than his siblings.

Social contexts could have positive and negative impacts on anxiety, depending on whether social responses were experienced as supportive. Unsupportive responses were mentioned by 75% of young participants, with examples of bullying behaviour including peers throwing things out of their reach, switching on their wheelchair, and threatening them. Only one parent mentioned their child being *"picked on"*, suggesting that young males were not fully disclosing their experiences to their parents, or they did not view it as bullying. Supportive and understanding responses from teachers, strangers, fellow DMD parents and children and online communities were reported to reduce anxiety. Examples included additional help with schoolwork, adults and other children understanding their condition without needing an explanation (young participants), people taking steps to include them, and receiving information and moral support from social media groups (parent participants). Some participants described positive benefits from taking part in the focus group discussions: "*At school I normally get a bit scared when someone … asks me questions but today everyone's made me feel like I don't need to do that I can just tell exactly what it is.*" [Youth]; "*I feel like I've had a therapy session*." [Parent].

The final influential systemic factor was physical environment and service contexts. Amongst young participants, wheelchair accessibility was the most reported subtheme (63%), citing the high prevalence of non-wheelchair accessible environments as increasing anxiety although this was mainly noted in the Youth

group rather than the Child group. This was noted less by parents (29%), although only three parents had a wheelchair-dependent child. Parents' most identified environmental/service domain was access to and quality of schooling and healthcare (79%), describing both positive and negative experiences of accessing educational and healthcare services. Fourteen parents had requested mental health support for their son, of whom 8/14 (57.1%) felt dissatisfied with the support received (Figure 1B), and several commented on a lack of professional therapeutic support available for parents, which affected their capacity to support their son.

4 Discussion

This is the first formal qualitative study investigating anxiety symptoms with young males with DMD. Our findings support the hypothesis of multiple interacting factors connected to anxiety in DMD, summarised in Fig. 2. This proposes that anxiety in DMD is an interaction between biological, social and environmental factors. Whilst this simplified model cannot map the full extent of the complexity of anxiety, it provides a framework for professionals and those living with DMD to identify and address areas of need, and a set of testable hypotheses to guide future research.

Causes and consequences of anxiety are difficult to untangle, as anxiety symptoms can have reciprocal relationships between the four systemic factors (Individual, Family and Social responses, and Physical environment/ service contexts), both influencing and being influenced by them. Systemic factors have the potential to both negatively and positively affect anxiety: social and environmental contexts may increase anxiety through poor accessibility, bullying and discrimination; inclusive school cultures, wheelchair-friendly environments, and others' understanding may reduce anxiety. Systemic factors are also inter-related, for example individual and

family responses to anxiety may be influenced by the availability of social or professional support.

Identifying the systemic factors influencing anxiety symptoms can help to define the most appropriate interventions. Cognitive behavioural therapy (CBT), commonly the first-line psychological intervention for anxiety, is typically more focussed on individual factors rather than targeting relational and systemic factors such as those we have identified in this study. For example, we observed a therapeutic value in the focus groups themselves, with some parent and young male participants reporting a positive impact of having taken part in a facilitated reflective peer group discussion. This highlights peer support groups as a potentially effective intervention for young people with DMD and as an indirect intervention with parents. Previous studies have also shown the benefits of peer support and social networks for individuals with DMD and their parents [29, 30].

Many of these factors may affect individuals with other life-limiting conditions and physical disabilities, such as anxieties about physical needs and the impact of physical and environmental contexts, rather than being DMD-specific. In spinal muscular atrophy (SMA), another life-limiting, disabling childhood-onset muscle wasting disease, a study using structured psychiatric interviews did not find any difference in internalising or externalising symptoms between SMA and control groups [31], although a more recent study in SMA children found a 40% prevalence of anxiety symptoms using a screening questionnaire [32]. Having a life-limiting condition is associated with a higher prevalence of anxiety and depression; a recent meta-analysis found a pooled anxiety prevalence of 19.1% in young people with different life-limiting conditions [6], which is nevertheless still lower than the 24% prevalence from a meta-analysis in DMD [5]. Social factors such as bullying are also

more common in children with chronic illnesses or physical disabilities, and lead to their own mental health burden [33].

However, in addition to risk conferred by the physical disability and life-limiting nature of DMD, we suggest that individuals with DMD have a greater predisposition to anxiety disorders. We recently showed that physiological startle responses are increased in young males with DMD compared to controls, which correlated with increased anxiety symptoms, suggesting there are underlying neurobiological factors increasing the risk of anxiety in DMD [13], paralleling preclinical studies that have clearly highlighted exaggerated fear response as an integral feature of the condition in the *mdx* mouse [12]. In the current study, we also identified anxiety characteristics, such as extreme distress to change, social anxieties, rigid-thinking and inflexible adherence to routines, which suggest important nuances in the DMD anxiety phenotype. This DMD anxiety phenotype overlaps with features of ASD [34], and whilst 41% of participants in our study had high social communication problems scores, these characteristics were not limited to children with ASD diagnoses. Autistic traits are more common in DMD than the general population [2, 5], however it is not possible from this study to determine the extent to which the presence of ASD influences anxiety symptoms, or whether there is a more general ASD-like phenotypic spectrum in DMD which includes heightened anxiety. This warrants attention in future studies.

Given the complexities outlined above, screening for anxiety and other mental health problems in DMD is challenging. The SDQ is a suggested screening tool in the DMD care guidelines and from a recent systematic review [4, 9]. In our quantitative analysis, the SDQ was more sensitive than the RCADS in identifying significant emotional difficulties, in line with evidence for the presence and impact of anxiety

from the qualitative data. Furthermore, the SDQ Impact Score measures the impact of emotional and behavioural difficulties, which can be more effective than measuring symptoms alone [35], and 69% of our cohort scored in the high range on this subscale. The standard RCADS is less effective at identifying anxiety in the context of ASD, such that a modified version is now used to evaluate anxiety in young people with ASD [36]. Therefore, as there was a significant degree of ASD (28%) and autistic traits (41%) within our sample, this might account for the poor sensitivity of this scale in our cohort. Whilst this effect may have been more extreme in our study, given that in the wider DMD population clinical ASD diagnoses are seen in at least 7%, and clinical range autistic traits in 21% [2] of individuals with DMD, we suggest that whilst the RCADS may not be appropriate for use in DMD, other assessment methods should be considered to ensure that children requiring additional support or interventions are appropriately identified. Our results support the use of the SDQ as a useful screening tool in DMD, although ideally a validation study in DMD should be conducted.

We observed some differences in themes between the Child and Youth groups, many of which related to the degree of physical differences in the DMD participants in relation to their peers, dependant on their age and stage of their condition. For example, social anxieties were experienced by all DMD participants, but in the younger group this more often related to the less visible nature of their DMD, whereas anxieties relating to visible differences were more prominent in the older group. Similarly, wheelchair accessibility was often a significant concern for older children and their parents. Extreme distress or ‘meltdowns’ were reported as being less prominent in the older DMD age group. We also noted some disparities between parent and child perspectives, for example, 75% of young participants described

social responses such as bullying, whereas this was only mentioned by one parent participant. This highlighted that multi-informant reports from young people with DMD and others such as their caregivers and teachers are important, as also emphasised in a recent systematic review of behavioural and psychosocial instruments in DMD [9].

We found evidence from within our DMD cohort which reflects the wider problem of limited access to psychological and psychiatric care for people with neuromuscular disorders. This has been recognised both in the UK, in an All Party Parliamentary Group report in 2019 [8], and in the United States, from an advocacy group survey of clinicians and parents [37]. The anxiety symptom screening questionnaires were suggestive of higher anxiety traits in our cohort than in comparable DMD cohorts, whilst the prevalence of diagnoses of anxiety disorders within our cohort were lower than the reported DMD literature. This discrepancy might reflect a bias in parent reporting, such as a possible tendency to over-report anxiety symptoms. However as we also found qualitative evidence for significant anxiety symptoms within this cohort, we suggest that this reflects a disparity between the number of children suffering from anxiety symptoms and the number with a formal diagnosis. This corresponds with the finding that over half of parent respondents who had wanted help for their son's anxiety had been dissatisfied with their access to professional support, suggesting there is an unmet need in the current assessment and management of anxiety in young males with DMD. The current lack of systemic availability of mental health team resources within neuromuscular services is preventing the DMD standards of care for the management of psychosocial problems being met [4].

The quantitative aspect of the study was designed primarily to compare the yield of structured interviews with existing questionnaires validated in other populations, however the data on the prevalence of different pre-existing neurodevelopmental and mental health condition diagnoses and symptom scores also provided some context to the qualitative data. The neurodevelopmental and neuropsychiatric profile of our young DMD cohort had some differences when compared to larger DMD cohorts, including a greater prevalence of ASD and a trend of higher anxiety symptom scores. This may reflect some bias within the sample however it does not detract from the primary aim of the study and in fact this enrichment of our sample may have enhanced the extraction of meaningful qualitative data. Our cohort also had a higher prevalence of ASD/autistic traits compared to previously tested DMD cohorts. This is not unsurprising, as anxiety disorders affect approximately 40% of young people with ASD [28] therefore the higher anxiety scores seen in this cohort may in part reflect the co-morbid higher rate of ASD/autistic traits. A potential limitation of this study was the risk of selection bias, as some participants were self-selected and may have been drawn to the study if they were affected by issues surrounding anxiety symptoms in DMD. As discussed above, the neurodevelopmental and neuropsychiatric profile of our cohort was not completely representative of the typical young DMD population. However, as the study was designed to explore anxiety symptoms in DMD and was not a prevalence study, this bias should not detract from our findings and may have actually enriched our data. Another limitation is that there was a bias in the parent respondents/participants as most parent participants were female and had DMD children under 12 years. We acknowledge that the parent-group findings may have skewed towards younger children and the perspectives of mothers, and that we did not have representation

from families with DMD boys with intellectual disability, which may have influenced the themes extracted from our data. However, as this was an exploratory study to understand the characteristics, causes and impact of anxiety in DMD rather than a prevalence study, we suggest that this does not impact significantly on our results.

5 Conclusions

In summary, this study highlights the importance of better understanding both the symptoms and impact of anxiety within the DMD population, and the multiple interacting factors involved, including normal adjustment processes in response to a chronic and fatal illness. It additionally highlights the significant influence on wellbeing and functioning that anxiety can have within this group, and that group interventions can be a useful approach to support individuals with DMD and their families. We have shown that the anxiety phenotype in DMD may not always be adequately identified by current standard screening instruments and support the use of multi-informant screening and the validation of instruments in DMD populations. Consideration of mental health screening methods in DMD are important in identifying those in need of support and intervention, to optimise the wellbeing and functioning of young people with DMD.

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Declaration of interest

KM, RT and WM report no conflicts of interest. FM declares conflict of interests outside the scope of this study (Scientific Advisory Board consultancies from Pfizer, Dyne Therapeutics, PTC Therapeutics and Sarepta) and involvement in UCL grants for studies on DMD/dystrophinopathies from the EU (Horizon 2020 BIND study) and from Sarepta Therapeutics (D-Brain study).

Appendices

Appendix A: Supplementary Tables A.1, A.2.

Appendix B: Figure B1 (Study procedure flowchart), Interview schedules and card-sorting task overview.

Appendix C: Additional supporting quotes from focus groups.

Tables

Table 1: Participant demographics.

Parent ID no.	Age	Gender	Ethnicity [a]	Child ID no.	Group (Child 7-11y; Youth 12-18y)	SDQ Total Difficulties score [b]	SDQ Impact score [c]	RCADS Total Anxiety score	Support satisfaction level [d]
P01	46-50	F	Br (W)	-	Child	10	0	36	n/a I have not required support
P02	41-45	F	Br (W)	C03	Child	7	1	31	Somewhat dissatisfied
P03	41-45	F	Br (As)	-	Child	13	4	51	Very satisfied
P04	41-45	F	Br (W)	C07	Youth	8	3	68	Somewhat dissatisfied
P05	41-45	F	Eur (W)	-	Child	16	2	51	n/a I have not required support
P06	36-40	F	Br (W)	C01	Child	15	2	36	Neither satisfied nor dissatisfied
P07	41-45	F	Br (W)	-	Child	11	2	42	Somewhat dissatisfied
P08	46-50	F	Br (W)	-	Youth	4	0	45	Very dissatisfied
P09	36-40	F	Br (W)	C02	Child	21	6	47	Very dissatisfied
P10	46-50	F	Br (W)	C04	Child	14	1	51	Very satisfied
P11	46-50	F	Br (W)	C06	Youth	25	7	46	Very dissatisfied
P12	46-50	M	Br (W)	-	Child	9	1	38	Very satisfied
P13	26-30	F	Other	-	Child	18	3	46	Neither satisfied nor dissatisfied
P14	41-45	F	Br (W)	-	Child	9	2	56	Very satisfied
n/a	-	-	-	C05	Youth	26	8	75	Very dissatisfied
n/a	-	-	-	C08	Youth	29	2	50	Somewhat dissatisfied
P04*	-	-	-	-	Youth	-	-	11	-
P08*	-	-	-	-	Child	-	-	33	-

	Total		Child	Youth	Total
Parent participants	14	Young DMD group participants	4	4	8
Parent questionnaire respondents	16	Young DMD questionnaire responses	12	6	18

Parent-reported demographics for parent and child/youth participants, including information about satisfaction with level of support for mental health problems in their child with DMD. * denotes parents who completed questionnaires for two children with DMD. [a] Ethnicity (self-reported): Br (W) = white British; Br (As) = Asian British; Eur (W) = white European; Other (Arab/African). [b] SDQ Total difficulties score: 0-13 = close to average; 14-16 = slightly raised; 17-19 = high; 20-40 = very high. [c] SDQ Impact Score: 0 = close to average; 1 = slightly raised; 2 = high; 3-10 = very high. [d] RCADS total anxiety score: $T \geq 65$ = borderline clinical; $T \geq 70$ = clinical threshold. [d]Support satisfaction level = Level of satisfaction with access to support around understanding and managing their son's anxiety.

Table 2: Prevalence of neurodevelopmental and mental health co-morbidities in study cohort compared with estimated prevalence in the wider DMD population and typical young male population.

Previously diagnosed co-morbidity	Study cohort %prevalence (n/N) [a]	Published DMD %prevalence [b]	DMD meta-analysis %prevalence [c]	Study cohort vs. DMD Sig., *p*	Typical population %prevalence [d]	Study cohort vs. typical population, Sig., *p*
Intellectual disability	0% (0/18)	11.4 - 40.5% [2, 38]	23%	***0.02***	2.5% [26]	*0.5*
Autism spectrum disorder	27% (5/18)	7% [5]	17%	***<0.001***	1.9% [25]	***<0.001***
Anxiety disorder	11% (2/18)	24% [5]	22%	*0.27*	5.4% [25]	*0.29*
Depressive disorder	0% (0/18)	11% [5]	12%	*0.12*	1.4% [25]	*0.61*
Attention deficit hyperactivity disorder	11% (2/18)	18% [5]	27%	*0.13*	2.6% [e] [25]	***0.03***
Obsessive-compulsive disorder	6% (1/18)	12% [5]	6%	*0.87*	0.5% [25]	***0.001***

[a] Parent-reported previously diagnosed co-morbidities in their sons. Two parents gave information on more than one son with DMD, resulting in data from 18 DMD boys. 5/18 boys had confirmed diagnoses of ASD, and 2 further parents reported their sons had 'borderline' or 'suspected' ASD; [b] Studies in children and adolescents with DMD using standardised assessment methods; [c] data from a meta-analysis conducted by our group[24]; [d] General population prevalence data from national surveys of children and adolescents aged 5-19 years in England (prevalence data for boys only apart from intellectual disability which is for both genders combined).[e] ADHD prevalence based on ICD-10 (International Classification of Mental and Behavioural Disorders 10th revision) diagnostic criteria. Using DSM (Diagnostic & Statistical Manual of Mental Disorders) criteria prevalence estimated 3-9%[27].

Table 3: Themes identified from Framework Analysis of focus group discussions.

	THEME	SUBTHEME	PARENTS (%)	BOYS (%)
CHARACTERISTICS OF ANXIETY	Internal factors	**1. Catastrophic conclusions**: "*Worrying about the worst situation*"	57	100
		2. Rigidly-held anxieties: *"Going round in circles"*	57	13
		3. Extreme distress: *"Total meltdown"*	93	38
	External factors	**4. Anxieties about physical changes/needs**: *"We're the most vulnerable"*	86	100
		5. Social anxieties: *"Will I fit in?"*	57	100
		6. The unexpected and unfamiliar: *"Not in the plan"*	64	50
INFLUENTIAL SYSTEMIC FACTORS	**Individual**	**Individual responses**	86	75
	Family	**Family responses**	100	0
	Social	**Stigma, misunderstanding, and unhelpful responses from others**	36	88
		Support and solidarity	64	50
	Physical environment and Service Contexts	**Access and quality of schooling and healthcare**	79	13
		Wheelchair accessibility	29	63

Table 4: Example participant quotes organised by theme.

THEME		SUBTHEME	SAMPLE QUOTES ‡
CHARACTERISTICS OF ANXIETY	Internal factors	**1. Catastrophic conclusions**: "*Worrying about the worst situation*"	"*[X]* does a lot of scenario planning in his mind and he's constantly asking "but what if this happens?", and much more in a way that you wouldn't expect from a seven- or eight-year-old boy, such as, "Will the airline have enough fuel to get us across the water?"" *(Parent)* "…when you go out somewhere and your parents are at home and you think, "what would happen if I'm out here and they die or something?"" *(Youth)* "… you start worrying, you get to like extreme conclusions, so like 'if this happens, this happens, this might happen to me and I might be ill' and stuff like that, so like jumping to bad conclusions." *(Youth)*
		2. Rigidly-held anxieties: *"Going round in circles"*	"Once he's got a worry in his head he'll just go over and over and over it to the point where I don't know what else to say to him, but he just can't shake it out of his head, he just goes on and on and on about it." *(Parent)*
		3. Extreme distress: *"Total meltdown"*	"*[X]* is very emotionally immature, he has meltdowns like crying, screaming. You tend to think 10-year-olds would know, but he just can't seem to control that emotion. And he recovers quite quickly, but for that moment, it's like the world is ending." *(Parent)* "It's usually something quite trivial that's caused this massive meltdown." *(Parent)*
	External factors	**4. Anxieties about physical changes/needs**: *"We're the most vulnerable"*	"*[X]* talks about dying and stuff and he doesn't want to die, he wants to live to 100 and he gets really anxious about it. This time he couldn't do half of the things they *[physiotherapists]* wanted him to do and he got a bit anxious over it because he knows what's coming which is really difficult." *(Parent)* "What are we going to do for a job because we've got weak muscles and we can't do jobs that other people do." *(Youth)*
		5. Social anxieties: *"Will I fit in?"*	"Stuff that you have to do in front of other people, like in the swimming pool, being hoisted in and everyone is looking at you." *(Youth)* "Or like getting out of your wheelchair and looking a bit weird because you can't get up yourself." *(Youth)* "My son hates any signs of disability, he really struggles with it…and that is a source of anxiety for him" *(Parent)*
		6. Unexpected and unfamiliar: *"Not in the plan"*	"If I pick *[X]* up from school, and then I need to stop at the shop, that's a big deal. But it's not in their plan, and it could be something trivial, but that is distressing for them." (Parent) "Going somewhere new is a big worry." *(Child)*

THEME		SUBTHEME	SAMPLE QUOTES ‡
INFLUENTIAL SYSTEMIC FACTORS	**Individual**	Individual responses	"I've destroyed a lot of things…It's like you don't stop, you just carry on going and then after you've done it or after your parents start crying you're like 'oh, what have I done?' *(Youth)* "He bottles it all up inside *[the anxiety]* and then it comes out in anger or he mucks around." *(Parent)* "I understand that sometimes I'm not going to be able to do everything…, that sometimes it's ok to let people do stuff that you can't do." *(Youth)*
	Family	Family responses	"You feel helpless, you can't switch it off, you can't tell him to stop *[feeling anxious]*. It's something else that is out of control as well as all the stuff with Duchenne." *(Parent)* "In our house there's a low level of anxiety every minute of every day and every night." *(Parent)* "The anxiety is more disruptive to our day-to-day life than Duchenne. It's every day." *(Parent)* "We try to get things across without making him worried but parental anxiety comes into all of this and how you engage with your son." *(Parent)*
	Social	Stigma, misunderstanding, unhelpful responses	"People say 'if you run me over then I'll pull you out the wheelchair'." *(Youth)*
		Support and solidarity	"Other kids are scared of doing tests but I think *[X]* has had to have a lot more support to get to those deadlines and not feel completely terrible about them. I suppose they're just managed by people being aware of his condition." *(Parent)*
	Physical environment and service contexts	Access and quality of schooling and healthcare	"I think there's no support for parents in terms of helping you deal with your own anxieties to then help the boys." *(Parent)*
		Wheelchair accessibility	"It's hard as a parent because you can't go "well it's all going to be fine" [accessibility of somewhere], because you don't know. Other people's idea about accessibility is very different." *(Parent)*

‡ Abridged, anonymised quotes from parent, child and youth focus groups. Full quotes are available in Appendix C.

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Appendix A: Supplementary Tables

Table A.1: Anxiety scores in child and youth groups compared to previous DM studies

		DMD Anxiety study		Other DMD studies		
Scale	**Age group**	**Mean *(SD)***	**n**	**Mean *(SD)***	**n**	***Sig., p***
CBCL DSM-Oriented Anxiety Problems	Child (7-11 yrs)	6.1 *(3.2)*	12	4.6 *(3.1)*	12	0.25
	Youth (12-18 yrs)	7.4 *(4.6)*	5	4.0 *(3.0)*	10	0.11
SCAS Total Anxiety	Child (7-11 yrs)	27.6 *(12.2)*	12	19.7 *(15.1)*	45	0.10
	Youth (12-18 yrs)	32.6 *(24.0)*	5	16.1 *(13.1)*	27	**0.03**
SDQ Emotional symptoms subscale	Child (7-11 yrs)	4.6 *(1.6)*	11	5.7 *(4.8)*	35	0.50
	Youth (12-18 yrs)	5.0 *(4.0)*	5	4.2 *(4.6)*	23	0.72

Comparison between mean anxiety/emotional problems scores on three parent-report scales in two different DMD cohorts to determine the representativeness of this cohort against a larger DMD cohort: this study (DMD Anxiety, n=18) and a combined cohort from two other DMD studies from our group (Ricotti *et al*., 2016[2]; Maresh *et al*., 2023[13]; total n=72). Data were compared from three parent-report scales used in all the studies (CBCL, SCAS and SDQ) using independent samples *t*-tests. The larger combined DMD cohort recruited participants from a clinic-based paediatric DMD population and is therefore more likely to be representative of a typical DMD population. Where the same individual took part in more than one study, only one dataset was used. SD = standard deviation. Sig.= significance level, *P*, with alpha level of *P*=.05. P-values <.05 are highlighted in bold.

Table A.2: Z-scores to compare neuropsychiatric co-morbidities between study cohort and DMD and typical populations

	DMD Anxiety study cohort			DMD population (meta-analysis)			*DMD Anxiety cohort vs. DMD population*		Typical population		*DMD Anxiety cohort vs. typical population*		*DMD population vs. typical population*	
	n	**N**	**Prop-ortion**	**n**	**N**	**Prop-ortion**	***Z-score***	***p-value***	**N**	**Prop-ortion**	***Z-score***	***p-value***	***Z-score***	***p-value***
Intellectual disability	0	18	0	320	1289	0.23	*-2.31*	***0.02***	6,839 ,000	0.03	*-0.07*	*0.5*	*47.1*	***<0.001***
Autism spectrum disorder	5	18	0.28	81	482	0.17	*42.36*	***<0.001***	3851	0.02	*7.85*	***<0.001***	*16.82*	***<0.001***
Anxiety disorder	2	18	0.11	45	196	0.22	*-1.1*	*0.27*	3851	0.05	*1.05*	*0.29*	*9.4*	***<0.001***
Depressive disorder	0	18	0	18	150	0.12	*-1.56*	*0.12*	3851	0.01	*-0.51*	*0.61*	*9.59*	***<0.001***
Attention deficit hyper-activity disorder	2	18	0.11	144	505	0.27	*-1.51*	*0.13*	3851	0.03	*2.22*	***0.03***	*22.75*	***<0.001***
Obsessive-compulsive disorder	1	18	0.06	3	59	0.05	*0.17*	*0.87*	3851	0.01	*3.22*	***0.001***	*4.57*	***<0.001***

Appendix B: Interview schedules and group tasks

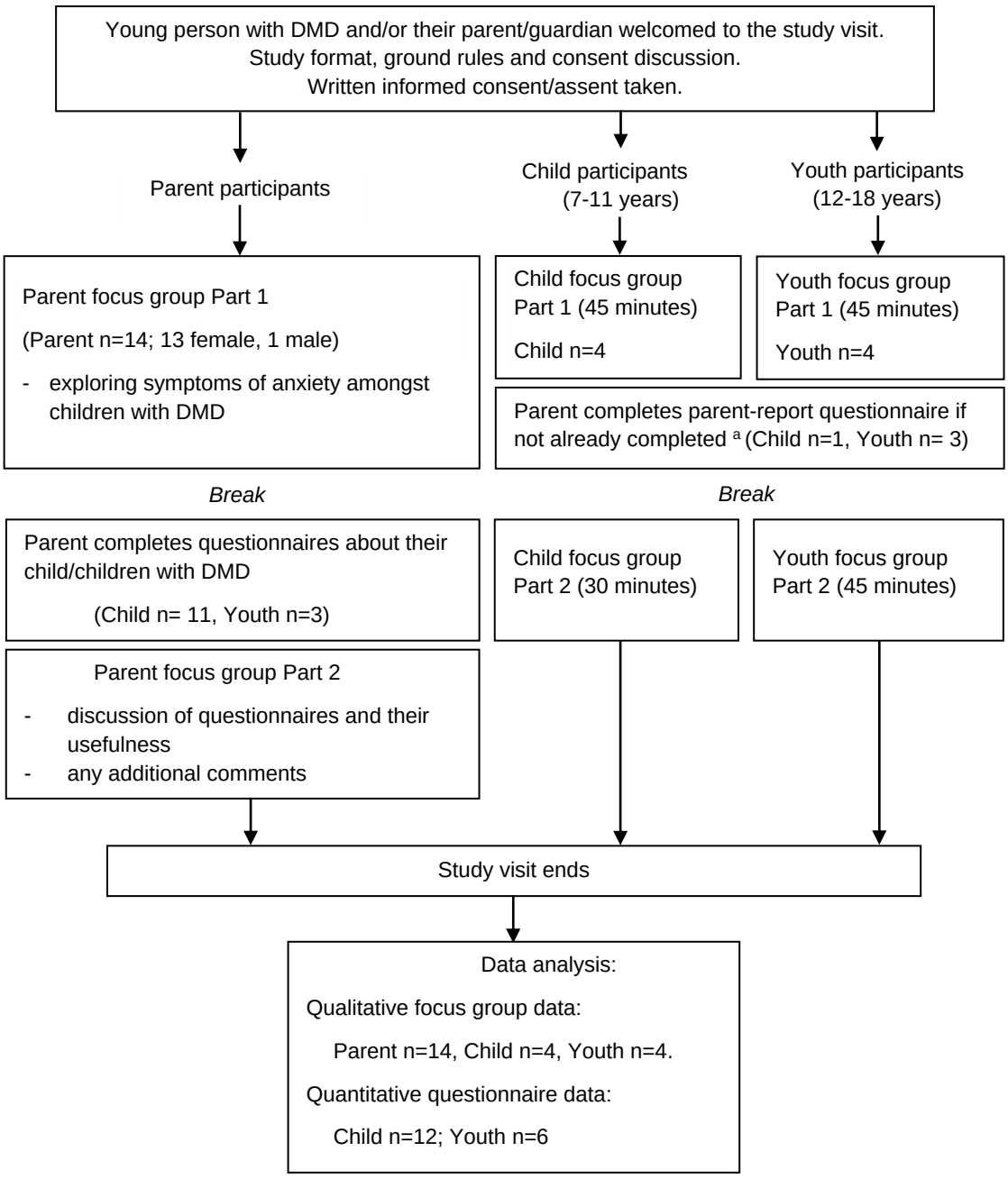


Figure B1. Flowchart of study procedures. Parent focus groups and child/youth focus groups took part on different days but are shown here in parallel.

[a] If parent had already completed questionnaire at a parent focus group (these were conducted first) they did not need to repeat this, except for those with >1 child with DMD in which case they had the opportunity completed an additional questionnaire for their other child.

Child focus groups semi-structured interview schedule

- **7-11: Part 1: 45 mins; Part 2: 30 mins**
- **12-18: Part 1: 45 mins Part 2: 45 mins**

Intro (5 mins)

Warm up game (5 mins)

Exercise (35 mins)
This is Josh. Josh is 8 years old and he has Duchenne Muscular Dystrophy. Josh is feeling worried. What are some of the things he might be feeling worried about? (*Picture of "Josh" printed onto piece of A0 paper. One facilitator writes each suggestion into a thought bubble above Josh. The other facilitator writes down each suggestion onto A5 coloured card for card sorting exercise in Part*)

Prompts:

- **What if Josh is at school. What might he be worried or afraid about there?**
- **What if Josh is playing with others…**
- **What if Josh is at home…**
- **What if Josh is trying to get to sleep…**
- **What if Josh is at the hospital?**

- I wonder if any of you have ever felt worried or afraid in these places? (***Prompts: can you tell me more about that. What did you worry would happen?***)

- Are your worries similar or different to Josh's? (***Prompts: tell me more about that, can you think of an example? What about at school, do you have worries that are similar or different to Josh's? What about at home? What about when you're with your friends? What about at the hospital? What would be the worst thing that could happen? What would be the best thing that could happen?*** *One facilitator to write down any new worries shared on coloured card for later exercise*).

Further prompts: How often does that happen? Is that similar or different for others? How worried does it make you feel (use fear thermometer).

- What happens when you feel worried or afraid? How would other people know you are feeling worried or afraid? (*Can use an example that has come up lots. Use printed A3 body outline and one facilitator write ideas onto it.*)

Prompts

- **What do you do when you are worried or afraid? How is that different to normal?**
- **What do you say? Who do you say it to?**
- **Would they notice any changes in your body? E.g. heart beating faster**
- **What do you feel like inside when you are anxious?**
- **What sort of thoughts go through your mind? What things do you think that other people can't see?**

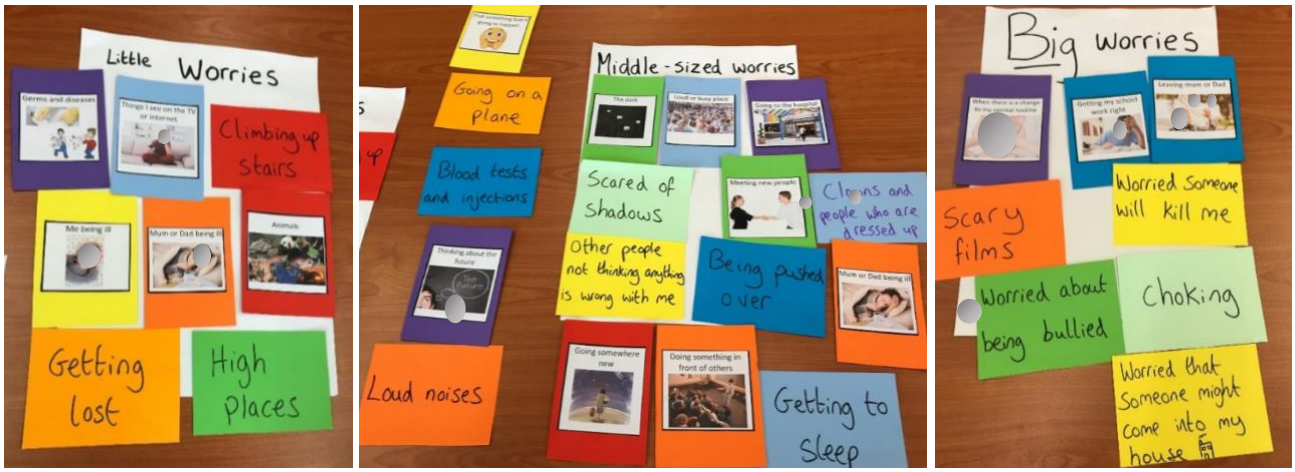
Exercise (20 mins):
(*Use worries that have been written on coloured A5 card plus pre-made worry cards*).

Ask group to put all cards into three different groups, (each with a piece of A3 paper): one for 'big worries', one for 'middle-sized worries' and one for 'little worries'. Also ask them to put the cards in an order from most worrying to the least worrying within each category. (*Aim: to stimulate interesting discussions about the different things making the participants feel anxious*) **Follow up questions e.g. why did you decide to put X there and not there? Did anybody disagree with that? Why is X a little worry? E.g. because it doesn't happen very often or because it doesn't matter very much? How big is the biggest worry? (Can use fear thermometer again) Is that similar or different for others? Why is that such a big worry?**

Finishing up (5 mins):

- Vouchers as a thank you
- Any questions for us?
- What did you like about today? Were there any bits you didn't like?
- Reiterate speaking to us if they are feeling worried or upset about anything they have heard today.

Figure B.2. Examples of card sorting exercise from child focus groups



Parent focus groups semi-structured interview schedule

Intro (5 mins)

- Is anxiety something you have ever noticed your son experiencing?

- How do you know when your son is feeling anxious? What do you notice?

  a) ***Prompts to put forward if they don't come up naturally:***
     - **What do you notice him doing? What sort of behaviour do you see? E.g. becomes shy/quiet, becomes demanding or angry, becomes controlling**
     - **What does he say (to you/to others)?**
     - **Do you notice any physical signs in his body or does he complain of any? E.g. trembling, tense, tummy ache, eye contact reduces**
     - **What do you think is going through his mind? What is worrying him?**
     - **Are there any behaviours that you understand happening as a result of your child's anxiety but which you think could be interpreted differently by people who know your child less well (e.g. aggression or attention-seeking behaviour)?**

- What does your child feel anxious about? Or worry about?
  a) ***Prompts***
     - **Unexpected changes**
     - **Separating from you**
     - **Being around others/crowded places**

- What is often happening around your child when your child becomes anxious? Are there any particular triggers you are aware of?
  a) ***Prompts:***
     - **Particular places / situations / events**
     - **Times of the day / week**
     - **In relation to particular people**
     - **Any other things that seem to trigger anxiety in your son*?***

- Are there situations or triggers which you actively avoid because your son would find it very difficult to tolerate due to anxiety?

- How does your son manage his feelings when he is experiencing anxiety?

- What do you do as a parent to try to prevent or avoid him feeling anxious?

- What is it like for you as a parent when your child is anxious?

- Is there an impact of your child's anxiety on the family?
  a) ***Prompts***
     - **Are there things it stops you or your family doing?**
     - **Does it impact siblings?**

Appendix C: Additional supporting quotes from focus group transcripts

Selected, anonymised full quotes from the focus group supporting different themes, abridged versions of which have been included in Table 3.

Characteristics of Anxiety

1. Catastrophic conclusions: "*Worrying about the worst situation*".

"*[X]* does a lot of scenario planning in his mind and he's constantly "but what if this happens?" and much more in a way that you wouldn't expect from a seven- or eight-year-old boy "Will the airline have enough fuel to get us across the water?"" *(Parent)*

"Some of the questions they come out with, you think you're going to crash the car!" *(Parent)*

"I always have a feeling I'll get lost." *(Child)*

"I get worried I will get on the wrong plane." *(Child)*

"…when you go out somewhere and your parents are at home and you think, 'what would happen if I'm out here and they die or something?'" *(Youth)*

"… you start worrying, you get to like extreme conclusions, so like 'if this happens, this happens, this might happen to me and I might be ill' and stuff like that, so like jumping to bad conclusions." *(Youth)*

2. Rigidly held anxieties: "*Going round in circles*".

"Once he's got a worry in his head he'll just go over and over and over it to the point where I don't know what else to say to him, but he just can't shake it out of his head, he just goes on and on and on about it." *(Parent)*

(Child 1): "…someone might start a fire and how are you meant to get out of there?

(Child 2): What if you're on your own? Like for like two minutes. If your mum's in the toilet and she doesn't even know what's going on or something

(Child 1): Yeah or they're asleep and you're awake

(Child 3): Yeah and something happens. Someone comes into the house and they're like, obviously my mum doesn't know what's happening and then I'm downstairs and someone comes up to me and something."

3. Extreme distress: "*Total meltdown*".

"I think *[X]* is very emotionally immature, he has meltdowns like crying, screaming. You tend to think 10-year-olds would know, but he just can't seem to control that emotion. And he recovers quite quickly, but for that moment, it's like the world is ending." *(Parent)*

"He stopped being sick because he doesn't go to school, but it was a fantastic performance the sickness, I mean it was projectile, he was literally destroying the car. We used to give school a bucket, this bucket so he could be sick in the bucket but it got to the stage where, personally, I couldn't watch him being so anxious, getting him to school, if it was his brother I would have travelled without the car but he was like putting himself on the floor of the car and stuff so it got to the stage where we thought, we need to stop." *(Parent)*

4. Anxieties about Duchenne-related change, needs, and medical procedures: *"We're the most vulnerable."*

"He is afraid that some classmates will run into him because he has a lot of accidents happen, that they push him, he can't stay properly because we know his muscle is not like ours." *(Parent)*

"[X] talks about dying and stuff and he doesn't want to die, he wants to live to 100 and all this and gets really anxious about it. This time he couldn't do half of the things they *[physiotherapists]* wanted him to do and he got a bit anxious over it because he knows what's coming which is really difficult." *(Parent)*

"What are we going to do for a job because we've got weak muscles and we can't do jobs that other people do." *(Youth)*

5. Social anxieties: "Will I fit in?"

"Stuff that you have to do in front of other people, like in the swimming pool, being hoisted in and everyone is looking at you." *(Youth 1)*

"Or like getting out of your wheelchair and looking a bit weird because you can't get up yourself." *(Youth 2)*

"my son hates any signs of disability, he really struggles with it and that is a source of anxiety for him" *(Parent)*

"Meeting new people [is a worry] because they don't know what's wrong with you." *(Youth)*

"If he *[Josh, fictional character]* goes in *[to hospital]*, he knows he has something, but then when he goes in, he's like…people look at him like he's not got anything wrong with him." *(Child)*

6. The unexpected and unfamiliar: "*Not in the plan*".

"If I pick *[X]* up from school, and then…I need to stop at the shop, that's a big deal. But it's not in their plan, and it could be something trivial but that is distressing for them." *(Parent)*

"Going somewhere new is a big worry." *(Child)*

[When there's a change to my normal routine] "That one's going on a 'big worry'." *(Child)*

"We tried to get him into new social groups, which took a lot of building up to because of the anxiety thing" *(Parent)*

7. Individual responses

Youth 1: "I've destroyed a lot of things…It's like you don't stop, you just carry on going and then after you've done it or after your parents start crying you're like 'oh, what have I done?'
Youth 2: And then you feel awful about it often"

“He bottles it all up inside [the anxiety] and then it comes out in anger or he mucks around.” *(Parent)*

“…even when my parents are over, he had to take the meds in another room, he doesn’t want them to see it. And when he had his biopsy scars, he was going a week later on a camp, I had to go in and talk to them, it was good friends he was sharing a room with, and say, “He’s just had some little cuts on his arms, don’t mention it.” So he’s happy that they know, but he doesn’t want to have to talk to them about it and he does not want to talk about Duchenne.” *(Parent)*

“I understand that sometimes I’m not going to be able to do everything, that sometimes it’s ok to let people do stuff that you can’t do.” *(Youth)*

8. Family responses

“You feel helpless, you can’t switch it off, you can’t tell him to stop [feeling anxious]. It’s something else that is out of control as well as all the stuff with Duchenne.” *(Parent)*

“…it’s hard not to sometimes let them know that you get fed up with it and that obviously has an impact because I think we’re anxious as well about some things.” *(Parent)*

“In our house there’s a low level of anxiety every minute of every day and every night.” *(Parent)*

“The anxiety is more disruptive to our day-to-day life than Duchenne. It’s every day.” *(Parent)*

“And I think with the others (siblings) I would have just walked away and said ‘this is what’s happening, we’ve talked about it’, and it would have been fine, whereas it just seems to go on *[with X]*, we don’t get to do it.” *(Parent)*

“I wonder if *[X]* felt much more relaxed at that point of understanding that there was a difference, there was an explanation for it *[DMD]*, that we [parents] seemed to understand what it was, we didn’t seem overly worried about it, you know, you try and get that across without making him worried. “Parental anxiety comes into all of this and how you engage with your son.” *(Parent)*

Social Responses

9. Negative social responses, stigma and misunderstanding: "They don't understand."

"People say 'if you run me over then I'll pull you out the wheelchair'" *(Youth)*

"People always push me over… I got bullied by this kid … he kept bullying me because I was slow. I can't help that." *(Child)*

"[At school] there's just bullying in general, people just take things off you and like throw it where you can't reach." *(Youth)*

*"*What happens if they hit you because you're just, you can't run away from them and you just sit there and let them." *(Youth)*

*"*If you snitch on them [bullies], they'll know it's you that snitched on them because we're in a wheelchair and you can just spot us every day." *(Youth)*

"There was a kid at my old school and he took it [direction stick on electric wheelchair] and he threw it in the bush and I told him to go and find it and he was like 'why?'" *(Youth)*

"He gets so anxious about going to school because there was a kid last year who picked on him." *(Parent)*

10. Positive social responses, support and solidarity: "You're not on your own."

"I normally get a bit scared when like someone comes up to me and asks me questions *[about my condition]*, but today everyone's made me feel like I don't need to do that, I can just tell them exactly what it is." *(Child)*

"Other kids are scared of doing tests but I think that he has had to have a lot more support to get to those deadlines and not feel completely terrible about them. I suppose they're just managed by people being aware of his condition." *(Parent)*

Physical environmental and service contexts

11. Access and quality of schooling and healthcare "A postcode lottery."

"I think there's no support for parents in terms of helping you deal with your own anxieties to then help the boys." *(Parent)*

12. Wheelchair accessibility

"I took *[X]* to *[a city]* on my own earlier in the week and it was really stressful because you're going to a hotel, you don't know what it's going to be like, you don't know what there's…and in fact, it was a bit of a nightmare for me because it was a lot of physical work and he's anxious about being hoisted and everything in the new place, but yeah, so I can understand how anxious you'd be, all that kind of stuff, and it's hard as a parent because you can't go "well it's all going to be fine" because you don't know. Other people's idea about accessibility is very different." *(Parent)*

Medication

"When he is on *[steroids]*, you can't say anything. You must be aware of what you talk about with him, how you talk with him because he is very…he starts to cry and he makes…. you know…and we know that he is on steroids. "Sshh! Zip it!" We are much quieter…We do not say "you cannot go", because he will get angry and say "ooh why not?" And the school, they notice as well. When he is on *[steroids]*, he is… "I want to do that, I want to do that, I want to do that." When he is not on *[steroids]*, "ahhh, no, no", he is very quiet." *(Parent)*