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I, Chandni Khudai , hereby submit this original work as part of the requirements for the degree of Master of Science in Genetic Counseling.

It is entitled:

A Descriptive Study on the Effect of Carrier Status on Mothers' Wellbeing and Adaptation to Duchenne and Becker Muscular Dystrophy

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2777

A Descriptive Study on the Effect of Carrier Status on Mothers' Wellbeing and Adaptation to Duchenne and Becker Muscular Dystrophy

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Abstract

Duchenne and Becker muscular dystrophy (DBMD) are progressive and debilitating neuromuscular disorders caused by X-linked recessive mutations in the dystrophin gene. Although improvements in the care of patients with DBMD have enhanced patients' quality and duration of life, there has been no effect on long-term prognosis. Little is known of the psychological morbidity associated with DBMD on families and patients. Birth mothers of children with DBMD, who are themselves carriers of DBMD, may have increased psychological burden. The purpose of this analysis was to describe differences in adaptation and wellbeing between mothers who were carriers and those who were not carriers of DBMD. Data was collected from mothers with biological children with DBMD using a mixed-methods web-based survey. The primary outcome variable, adaptation score, was generated for each participant using the Psychological Adaptation to Illness Scale. Various other scales and investigator-developed questions were also used to measure secondary outcome variables. One hundred twenty-five participants completed the questionnaire and 116 responses were analyzed. Fifty-one (44%) were carriers of a *DMD* gene mutation, 47 (40%) were not carriers, and 18 (16%) did not know their carrier status. The mean adaptation score was 3.68 (SD=0.9) for carriers and 3.25 (SD=0.9) for non-carriers. Carriers showed better adaptation and higher perceived control than non-carriers (pooled t-test, $p=0.02$ and $p=0.05$, respectively). These results were limited by several factors including small sample size, recruitment bias, and increased risk of type 1 error resulting from multiple tests on the data. An open-ended question completed by the carriers revealed various positive and negative effects of being a carrier. In conclusion, carrier status may affect mothers' adaptation to DBMD and perceived control in this population. A larger, more representative sample of participants may be beneficial in confirming these findings.

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I would like to acknowledge my research advisory committee for providing me with their insight and expertise. A particular thanks goes to Holly Peay for allowing me to be a part of this study, and my research advisor, Martha E. Walker, for all of her guidance throughout this process.

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Tables and Figures

Introduction

Duchenne and Becker muscular dystrophy (DBMD) are dystrophinopathies caused by mutations in the *DMD* gene on chromosome locus Xp21.2. The majority of cases of DBMD result from the X-linked inheritance of a *DMD* mutation from the mother, but approximately 1/3 of cases are caused by *de novo* mutations [1, 2]. Duchenne muscular dystrophy (DMD) is the most frequently occurring of the muscular dystrophies, affecting approximately 1 in 3,500 males [3, 4]. DMD involves rapid progression of proximal muscle weakness; intellectual impairments may also occur in affected males [5, 6]. There is often loss of ambulation in the first decade of life and death by the second or third decade [7, 8]. Becker muscular dystrophy (BMD) is a milder, more variable form of dystrophinopathy with later onset of symptoms and longer life expectancy compared to DMD [4, 7]. Improved quality of life and clinical outcomes for people with DBMD have been observed during the past decade due to the use of corticosteroids, advances in respiratory support, and early initiation of cardiac care, but long-term prognosis has not changed [9-12]. As a result, parents and patients with DBMD are faced with a progressive fatal disease which is responsible for significant psychosocial morbidity.

DNA analysis can be performed to determine the carrier status of the mother and other female relatives of a person affected with DBMD. Female gene mutation carriers of DBMD are typically asymptomatic, but a variety of symptoms has been reported [4, 13]. These include muscle weakness, myalgia/muscle cramps, and, less commonly, dilated cardiomyopathy, asymmetrical muscle weakness, and mild to severe BMD and DMD-like phenotypes [13]. Females carrying a *DMD* mutation have a 50% chance of passing the gene mutation to each of their offspring. With every conception, a female carrier has a 25% chance of having an affected male and 25% chance of having a daughter who is also a carrier. Females with a negative carrier

status have a 15-20% risk of passing DBMD to their offspring due to the underlying possibility of germline mosaicism [14].

Female carriers for various X-linked diseases often cope with guilt, stigmatization, and other negative emotions associated with the reproductive risks and associated family planning decisions [15-17]. Many believe they have a genetic responsibility to stop the recurrence of the condition. Kay and Kingston [16] studied the reproductive decision making of female carriers of the X-linked, disabling conditions DBMD, Lesch-Nyhans syndrome, Menkes syndrome, and Fabry disease and found that carriers with personal experiences with the disease were more concrete in their decision to avoid having affected children. Many of these women expressed guilt and assumption of responsibility for the birth of an affected child and the negative effects of the condition on the family. There were similar findings on reproductive guilt and decision making in female carriers of Fragile X syndrome [18]. Compared to female carriers of autosomal recessive spinal muscular atrophy types II and III, female carriers of the X-linked conditions, chronic granulomatous disease and DBMD, experienced more guilt over their carrier status. These women also perceived more blame from their child's father in comparison to the carriers of autosomal recessive conditions [15].

Learning that one is a carrier for a genetic disorder also has the potential to alter self-concept, self-efficacy, and coping [19-21]. In 2004, Daoud, Dooley and Gordon reported that parents of children with DMD had higher rates of major depressive episodes than the population rate, and had significantly lower self-esteem and lower mastery scores [22]. In Kenneson and Bobo's 2010 quantitative study of women caregivers of males with DBMD, approximately 50% of mothers had sustained a high level of stress and caregiving demands. Caregivers with a low-level of social support were four times more likely to report distress [23]. Research on chronic illness

and disability has shown that an improved quality of life may be an outcome of successful adaptation to the disorder [24]. Adaptation refers to the process of accepting the implications of the disorder and the outcomes of that process. Outcomes of successful adaptation include increased self-esteem and decrease in depressive symptoms [25].

Published literature indicates that being a female carrier for DBMD significantly affects one's life, especially in the areas of reproductive decision-making and emotional wellbeing. However, there is still limited information on these effects and their long-term consequences in families with DBMD. This study therefore sought to test the hypothesis that adaptation of mothers who are carriers would be different from those who are not carriers. Specific aims were to describe the effect of carrier status on adaptation, self-esteem, perceived effects on family, burden, perceived control, self-concept, and coping efficacy.

Methods

A five year longitudinal study titled "Assessing Wellbeing in Women Caring for Children with Duchenne or Becker Muscular Dystrophy" was initiated in fall, 2011 by Holly Peay, MS,CGC, Senior Director of Education and Outreach at Parent Project Muscular Dystrophy and Kathi Kinnett, RN,CNP, at Cincinnati Children's Hospital Medical Center (CCHMC). It is a longitudinal study with the purpose of informing the development of a set of interventions aimed at improving wellbeing in mothers of offspring with DBMD by examining predictors of wellbeing that may be modified through interventions. Participants will complete an online questionnaire annually for five years. The current study is a mixed-methods analysis of data from a subset of items in the year-one (baseline) questionnaire of the longitudinal study. The current study was approved by the Institutional Review Boards at CCHMC and the University of Cincinnati as part of the longitudinal study.

Recruitment/Participants

Eligible participants were adult, English-literate females who had at least one living biological son with DBMD. They were recruited from the membership of the national advocacy and support organization Parent Project Muscular Dystrophy (PPMD) and its patient registry, DuchenneConnect. Recruitment was conducted through announcements in online forums, newsletters, and an online support blog for female caregivers, www.HerSelfFirst.com. Flyers describing the study were distributed in the Neuromuscular Care Center at CCHMC and through other clinics. Women who were interested in participating were instructed to contact Holly Peay by phone or email. Those who agreed to participate provided their name and contact information, which was stored in a password-protected electronic database administered by Holly Peay. For

tracking in the longitudinal study, a unique code was assigned to each participant and was used to generate a unique link to the survey. An email including the unique link to the SurveyMonkey® questionnaire was then sent to each participant.

Informed Consent

This study was exempt from using a signed informed consent. The collected data did not include sensitive health information or a review of medical records. The questionnaire began with a statement about the risks and benefits of participation and the option of withdrawing from the study. Each participant indicated her acceptance of the terms before responding to the questionnaire. Participants were encouraged to contact Holly Peay with any questions or concerns about the study.

Questionnaire

The questionnaire (Appendix A) was investigator-developed and adapted from several validated and semi-validated tools. In addition to some demographics, information was requested on the participants' children whom were affected and unaffected with DBMD, gene carrier status and carrier status-related feelings, self-esteem, perceived effects on family, burden, perceived control, self-concept, coping efficacy, and adaptation. The questionnaire collected additional information (such as perceived needs, motivation, home care and respite care) that will be analyzed in the longitudinal study but not the current study. Participants were instructed when to answer the questions with regards to themselves or with regards their oldest living child with DBMD. The questionnaire was constructed in skip-logic format and published online using the web-based survey software, SurveyMonkey®.

The primary outcome variable of this study was adaptation score. Adaptation was measured using the validated Psychological Adaptation to Illness Scale [26]. The adaptation scale consists of 20 statements regarding adaptation to illness. Participants used a five-point scale to show their level of agreement with each statement. An adaptation score was given to each participant by averaging the total score from their responses.

The secondary outcome variables included self-esteem, perceived control, self-concept, coping efficacy, burden, and two Likert scales measuring positive and negative effect of DBMD on the family, and were measured using Rosenberg's Self-Esteem Scale [27]; the revised perceived control measure [28]; the self-concept measure from Esplen et al., 2009, moderately revised for use in this population [29]; The Coping Self-Efficacy Scale (CSES) [30]; Zarit burden measure [31]; and investigator-developed questions, respectively.

Data Analysis

Responses to the completed questionnaires were downloaded from SurveyMonkey® into an Excel database. Data were cleaned by removing duplicate and blank entries prior to performing the analysis. The statistical software, SPSS, was used to generate scores for each outcome variable. The primary outcome was a score on the adaptation measure. The secondary outcomes include scores on the measures for self-esteem, perceived burden, control, self-concept, coping efficacy, and two questions regarding the perceived positive and negative effect of DBMD on the family . Before data analyses were performed, the distribution of the outcome variables in carriers and non-carriers was checked. All of the outcome variables showed approximately normal distribution. F-test was used to test whether the variances of the outcomes from carriers and non-carriers were equal. No significant differences were detected, thus pooled t-tests were

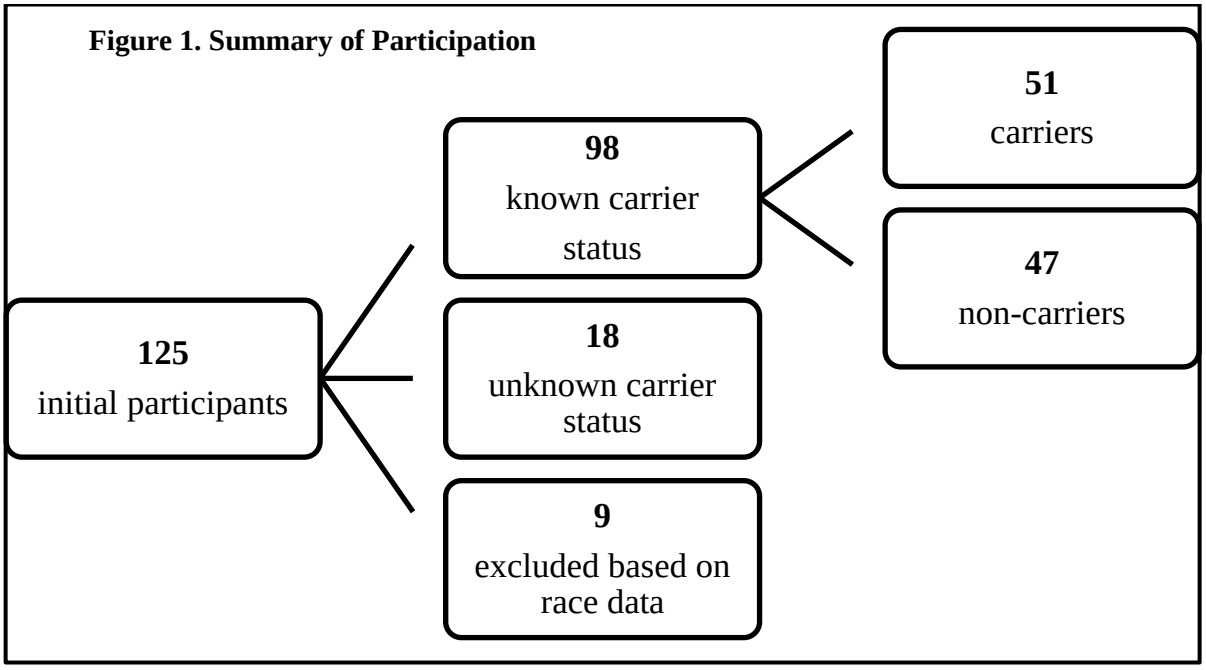
performed for all of the outcomes using Statistical Analysis Software (SAS), version 9.3 (SAS Institute Inc., Cary, NC). Eight t-tests were conducted. Bonferroni correction would require a p-value of 0.006 for significance; however, Bonferroni is conservative, especially in the context of correlated outcomes. Thus, we report uncorrected p-values in this manuscript. P-values between 0.05 and 0.006 are at increased risk of false positive association, and should be interpreted with caution. Descriptive statistics were performed on the demographic data of both carriers and non-carriers, the two items for those with unknown carrier status, and nine items specific to women who are carriers. Open-ended responses were analyzed for recurrent themes. The responses were coded and grouped accordingly to determine the frequency of each theme.

Results

Demographics of Participants

One hundred fifty-four women requested information about participation in the study and 125 women initiated the internet-based questionnaire. Nine participants (seven non-Caucasian and/or Hispanic respondents and two with incomplete race information) were excluded to eliminate race and ethnicity as possible confounders. Eighteen women reported that they did not know their *DMD* gene carrier status. Their responses to questions 2 and 3 under the section titled "Your Carrier Status" were described in this study but were otherwise excluded from the comparative analysis. Among the remaining ninety-eight participants, fifty-one were carriers and forty-seven were non-carriers; all of them were included in the comparative analysis of adaptation score and the secondary outcome variables (Figure 1).

Figure 1. Summary of Participation



Ages for carriers and non-carriers ranged from 27 to 63 years with a median age of 41.5 years. The majority of women had a college degree or higher (70%), were married or in a long-term committed relationship (90%), were employed (77%), and had a household income of $50,000/year or greater (85%). The median number of total children was two. Twelve women each had two children with DBMD and the remaining eighty-six had one child with DBMD. A family history of DBMD was reported by twelve carriers (24%) and, as expected, by none of the non-carriers (Table 1).

Table 1. Demographics of Participants

Characteristics	Carriers	Non-carriers	Total
Age			
n, range	48, 27-63	46, 29-63	94, 27-63
median (IQR)	41 (37-48)	42.5 (39-51)	41.5 (37-48.5)
mean (SD)	42.2 (±8.5)	44.6 (±8.6)	43.4 (±8.6)
Number of Children			
n, range	51, 1-5	47, 1-6	98, 1-6
median (IQR)	2 (2-2)	2 (2-3)	2 (2-3)
Number of Children with DBMD			
n, range	51, 1-2	47, 1-2	98, 1-2
median (IQR)	1 (1-1)	1 (1-1)	1 (1-1)
Family History of DBMD (n, %)			
yes	12 (24)	0 (0)	12 (12)
no	39 (76)	47 (100)	86 (88)
total	51	47	98
Education (n, %)			
some college or less	16 (31)	13 (28)	29 (30)
college degree or higher	35 (69)	34 (72)	69 (70)
total	51	47	98
Marital Status (n, %)			
married	46 (90)	42 (89)	88 (90)
divorced	5 (10)	5 (11)	10 (10)
total	51	47	98
Employment Status (n, %)			
unemployed	13 (25)	9 (20)	22 (23)
employed	38 (75)	37 (80)	75 (77)
total	51	46	97
Household Income (n, %)			
less than $50,000/year	7 (14)	7 (16)	14 (15)
$50,000/yr or more	44 (86)	37 (84)	81 (85)
total	51	44	95

Adaptation and Secondary Measures of Control, Self-esteem, Burden, Coping, Self-efficacy, and Effect on Family

The mean scores of the primary and secondary outcome variables were compared (Table 3). Carriers may have higher scores than non-carriers in adaptation (p=0.02) and perceived control (p=0.05). Carriers and non-carriers did not differ significantly in other outcome variables.

Table 3. P-values, Means, and Standard Deviations of Outcome Variables

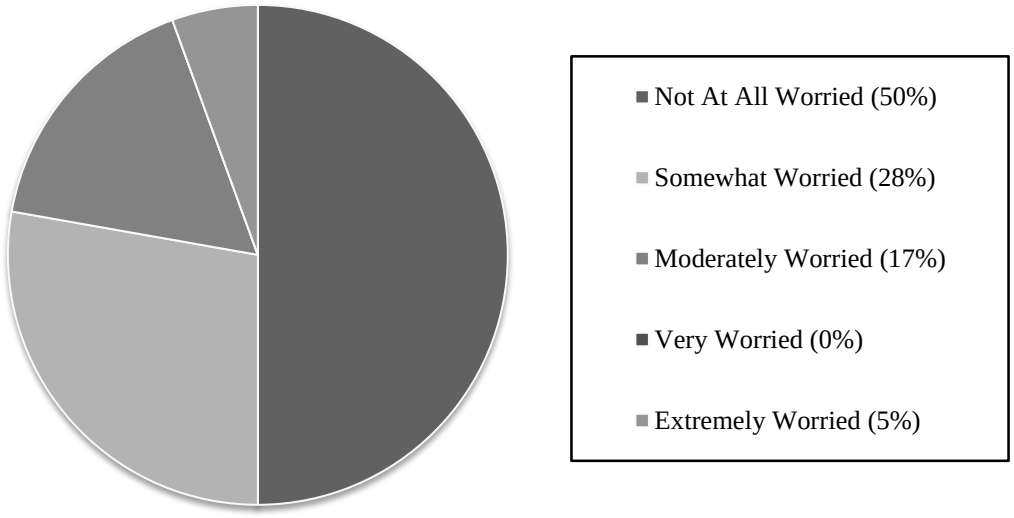
	P-value	**Carriers** mean [SD]	**Non-carriers** mean [SD]
Total Adaptation	0.02	3.68 [0.9]	3.25 [0.9]
Perceived Control	0.05	5.90 [1.9]	5.15 [1.9]
Self-esteem	0.29	21.33 [4.7]	20.21 [5.6]
Level of Burden	0.38	17.33 [8.2]	18.94 [9.4]
Coping Efficacy	0.27	159.42 [47.1]	148.11 [52.5]
Self-Concept	0.34	58.88 [15.4]	62.11 [17.1]
Positive Effect of DBMD on Family	0.23	6.85 [2.8]	6.12 [3.0]
Negative Effect of DBMD on Family	0.12	6.27 [2.8]	7.17 [2.7]

Investigator-developed Questionnaire

Responses from Women with Unknown DBMD Gene Carrier Status

Question numbers 2 and 3 in the section of the questionnaire titled "Your Carrier Status" were completed by women with unknown carrier status (Appendix B). Question 2 asked participants the reason for not knowing their carrier status. Six out of the eighteen women (33%) reported that they have no reason to know their carrier status and 5 women (28%) reported that insurance would not cover the testing or that testing was too expensive. The remaining seven participants were either waiting to get results (n=3), not ready to know their carrier status (n=3), or unable to get tested as a mutation was not detected in the child with DBMD at the time of his testing (n=1). In response to question 3, nine of the women with unknown carrier status said they were "Not at all" worried regarding the possibility of being a carrier. Only one woman reported being extremely worried (Figure 2).

Figure 2. Worry Regarding the Possibility of Being a Carrier (responses from women with unknown carrier status; N=18)



Responses from Women who are DBMD Gene Carriers

Items 4-9 in the section titled "Your Carrier Status" were completed by fifty women with a positive carrier status for DBMD (Table 2). Twenty-five of these women (50%) reported that their carrier status had changed their lives by indicated "agree" or "strongly agree". Twenty of the twenty-five women did not have a family history of DBMD. With regard to the way they see themselves as mothers, 42% (n=21) believed their carrier status had an effect and 38% (n=19) believed that it did not have an effect. The majority of women did not believe that being a carrier for DBMD had an effect on their relationship with their partner, child with DBMD, or their daughter(s). However, fourteen women (28%) reportedly felt less connected to their partner due to being carriers. Two of these women were divorced from their partner and three of these women reported a family history of DBMD. Thirteen women (26%) felt more connected to their child with DBMD after learning that they were carriers.

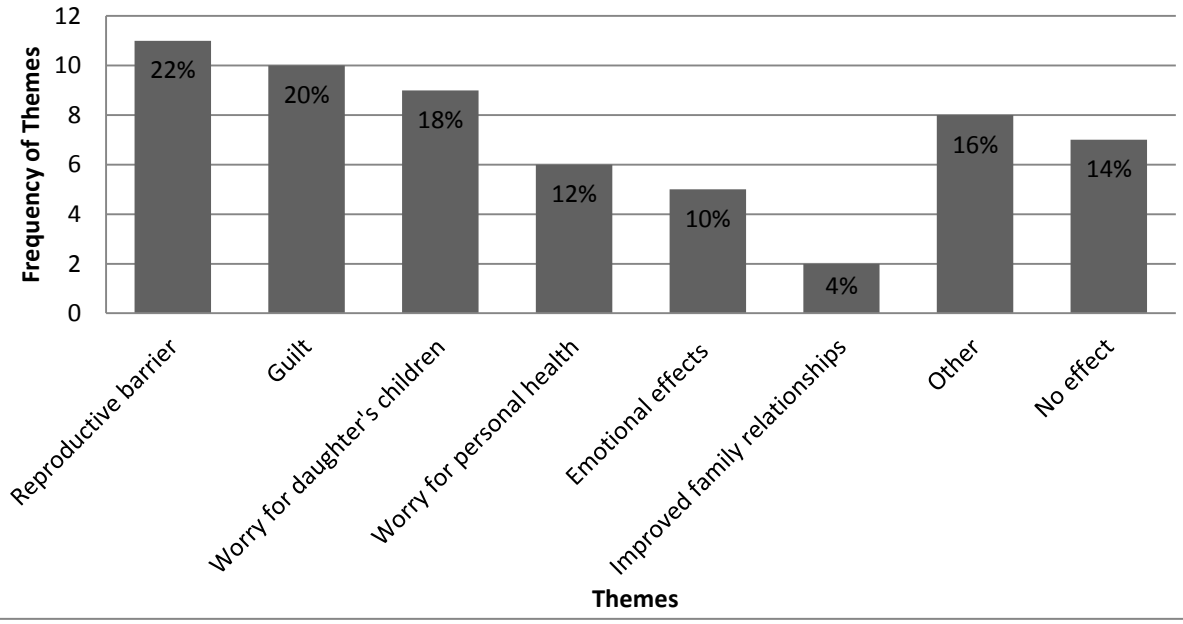
Table 2: Description of Items 4-9 under "Your Carrier Status" Section (n=50)			
	Answer Choices	**n**	**%**
Finding out that I was a carrier changed my life.	Strongly disagree	4	8%
	Disagree	6	12%
	Neither agree or disagree	15	30%
	Agree	10	20%
	Strongly agree	15	30%
Finding out that I was carrier changed the way I think of myself as a mother.	Strongly disagree	10	20%
	Disagree	9	18%
	Neither agree or disagree	10	20%
	Agree	13	26%
	Strongly agree	8	16%
How did finding out you were a carrier affect your relationship with your **partner**?	Much less connected	3	6%
	Somewhat less connected	11	22%
	Relationship did not change	32	64%
	Somewhat more connected	0	0%
	Much more connected	2	4%
	I do not have a partner	2	4%
How did finding out you were a carrier change your relationship with your **child with DBMD**?	Much less connected	0	0%
	Somewhat less connected	0	0%
	Relationship did not change	37	74%
	Somewhat more connected	9	18%
	Much more connected	4	8%
How did finding out you were a carrier change your relationship with your **daughter(s)**?	I have no daughters	21	42%
	Much less connected	0	0%
	Somewhat less connected	0	0%
	Relationship did not change	22	44%
	Somewhat more connected	4	8%
	Much more connected	3	6%

Responses to the Open-Ended Question

Question 9 was an opened-ended question asking the participants how a positive carrier status has most affected their lives (Figure 3). Seven women stated that their carrier status had no effect on their life, 11 women reported that their carrier status was a barrier to having more children, and 10 women mentioned guilt associated with being a carrier. Other themes identified in the women's responses were worry for daughter's children (n=9), worry for personal health (n=6),

emotional effects such as anger and depression (n=5), and improvements in family relationships (n=2).

Figure 3: Aspects of Life Most Affected by Positive Carrier Status: Responses to an Open-ended Question (n=50)



Discussion

This study surveyed mothers of children with DBMD to determine whether carrier status had an effect on their wellbeing and adaptation. The Psychological Adaptation to Illness scale was used to measure how well the women in this study had adapted to their child's diagnosis of DBMD. Carriers were compared to non-carriers in adaptation and several other variables which measured wellbeing. In this study, carriers had higher adaptation and perceived control than non-carrier. Carriers and non-carriers did not differ in any other outcome variables. These findings must be interpreted with caution since multiple tests on a data set can increase the potential for false positive results. Using a Bonferroni corrected significant p-value of 0.006 or less would allow for more definitive conclusions to be made. Also, other factors affecting wellbeing and adaptation, as well as small sample size and recruitment bias should be taken into consideration when assessing these results.

Although some carriers experience guilt and worry associated with their carrier status, their adaptation to their child's condition may not be directly affected by these feelings. A 2008 study by Wheeler and Bailey found that higher levels of stress and depression were predictors of a low quality of life in mothers of children with Fragile X syndrome and the general population [32]. Therefore, factors directly affecting the stress level of these mothers, such as problematic behavior [33], may have a greater impact on adaptation than carrier status.

Financial resources may also be a source of parental stress. Parents of children with DMD highly value services, such as physical and occupational therapies, which are aimed at prolonging their child's ambulation [34]. Inaccessibility to these and other health-related services due to a lack of financial resources could also be stress inducing for parents. The majority of participants in this

study were college educated, married, and had a household income of $50,000/year or greater. Thus, the population of women who participated in our study may have access to greater resources than the general population (2010 US Census data).

It is also possible that a more representative sample or larger sample size would confirm the carrier status effect on adaptation and perceived control with a p-value of 0.006 or lower. Higher perceived control in carriers may be attributed to having more control over recurrence, but further research would be required to determine how being a carrier increases one's adaptation. Twenty-five of the carriers reported that being a carrier had changed their lives, the participants, however, were not able specify if this change was negative or positive. Overall, the majority of the carriers surveyed did not report their carrier status as having a negative effect on their relationship with their partner or child(ren). In this population, the greatest effects of being a carrier were guilt and the limitations placed on family size due to recurrence risk. One participant wrote "I feel like I am causing my children to have a disease…" while another wrote, "I wanted to have more children and after finding out I'm a carrier, know it's not possible." Worry for future generations was also a concern for many of the women with daughters. Positive effects of being a carrier were reported by three of the women in this study. One participant reported that having a being a carrier helped create a sense of community with other female carriers in her family.

Half of the women with unknown carrier status indicated some level of worry regarding the uncertainty of their carrier status. The questionnaire did not gather qualitative data on what these women were most worried about. When asked why they did not know their carrier status, six women reported that they had no reason for knowing. One area of interest is whether the women who were not worried about carrier status were aware of the potential for health risks,

specifically cardiomyopathy, associated with being a carrier. A research study on this topic revealed that only 46.9% (n=192) of women with unknown carrier status were aware of the risk for cardiomyopathy in female carriers if DBMD [35]. This data reveals a gap in the counseling provided to the mothers of children with DBMD.

The findings in this study have generated discussion for future research in this population. These findings, however, were limited by several factors. Participants reflected a population of carriers and non-carriers that are mostly working, middle-class women. Since a majority of the women were married and recruitment was primarily through PPMD, participants in this study may have access to more social support than the general population. The high response rate (81%) indicated that this was a very motivated population, and those who participated may be different from those who did not participate in a way that may have skewed the results. Therefore, this sample may not be representative of the majority of mothers of children with DBMD. Dividing recruitment efforts more evenly between multiple neuromuscular clinics and PPMD may be helpful in achieving a more representative sample. A larger sample size may also contribute to a more representative sample and produce more confirmatory results. Analysis with pooled t-test limited the ability to find predictors of adaptation. The longitudinal study will have a larger sample size and will analyze the data using a regression model to look for predictors of adaptation.

Conclusion

DBMD is a severe neuromuscular disease that causes significant psychological morbidity in patients and their families. The purpose of this study was to contribute to the medical knowledge in the area of carrier status effect on adaptation and wellbeing of mothers of children with DBMD. In this population, carrier status had an effect on adaptation and perceived control. These findings, however, were limited by factors including using uncorrected p-values, small sample size, and recruitment bias. This study was able to confirm previous findings on some psychological effects of being a carrier and generated new ideas for future research in the area of adaptation and wellbeing of these mothers. Studies have shown that disease-specific interventions may be useful in raising the moral and adaptation of affected families [36]. By contributing to the literature on the effects of carrier status, physicians and genetic counselors will be better informed to provide these families with counseling and anticipatory guidance.

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Appendix A: Questionnaire

Thank you for participating in this study about the feelings, thoughts, and needs of mothers caring for children with Duchenne or Becker muscular dystrophy (DBMD). This is the first survey. You will receive follow up surveys about every 12 months for the next 5 years. This survey may take you about 45-60 minutes to complete. You do not have to finish all of the survey at one time, but please do answer all of the questions. There are no correct or incorrect answers.

While we use the term "child" in this survey, we understand that some participants have children who are teens or adults. We are interested in the thoughts and feelings of mothers of adult children with DBMD as well as mothers of younger children.

We appreciate you sharing your thoughts and experiences so we can create interventions that will help mothers' wellbeing.

Demographic Questions

Please answer the following questions about yourself.

1. What is your year of birth?

2. What is your race/ethnicity? Choose all that apply.
    - Caucasian/White
    - African America
    - Asian/Pacific Islander
    - Native American
    - Other

3. Do you consider yourself to be of Hispanic origin?
    a. Yes
    b. No

4. What is the highest level of education that you have completed?
    - Grade school or middle school
    - High school diploma/GED or high school equivalency
    - Some college or technical training
    - College degree
    - Post-baccalaureate degree

5. What is your current marital status?
    - Single, never married
    - Married or long-term committed relationship
    - Separated
    - Divorced
    - Widowed

6. What is your current employment status?
    - All of my time is spent caring for my family and my home
    - I have a **full-time** job or schooling program
    - I have a **part-time** job or schooling program

7. What is your household income?
    - Less than $50,000
    - $50,000 - $99,999
    - $100,000 - $149,999
    - $150,000 - $199,999
    - $200,000 - $249,999
    - $250,000 or more

8. What state do you live in?
    a. [list of states]

About Your Children

1. Please describe your children, from OLDEST to YOUNGEST.

	Year of birth	Sex M/F	Does this child have Duchenne or Becker? Yes, Duchenne/Yes, Becker/Yes, intermediate /No/Don't know	Is this your biological or adopted child? Biological/Adopted	Is this child still living? Y/N
Child 1					
Child 2					
Child 3					
Child 4					
Child 5					
Child 6					
Child 7					
Child 8					
Child 9					
Child 10					

About your Child with DBMD

1. At what age was your child with DBMD diagnosed? (If you have more than one child with DBMD, please answer for the oldest child.)

- Prenatally (before birth)
- 0-3 years
- 4-7 years
- 8-11 years
- 12 years or older

2. Prior to your child's diagnosis, did you know of any blood relatives with DBMD?

- Yes
- No

3. Chose the option that best describes your child's physical abilities today. If you have more than one child with DBMD, please answer this question about your **oldest** child. Every child is unique, and may not match the descriptions perfectly. Please select the answer that is **the best fit**. My child with DBMD:
    a. Presymptomatic – Has no symptoms
    b. Early-ambulatory – Walks with an unusual gait but is able to climb stairs
    c. Late-ambulatory - Walks with more difficulty, sometimes uses a wheelchair, is losing the ability to get up from the floor and climb stairs
    d. Early non-ambulatory – Is unable to walk alone but can still sit and stand, uses a non-powered wheelchair on his or her own
    e. Non-ambulatory I- Uses a powered wheelchair but is no longer able to use a non-powered wheelchair on his or her own; is showing limited arm strength though is able to raise hands to mouth
    f. Non-ambulatory II– Is no longer able to raise hands to mouth but is able to hold a pen or to move powered wheelchair
    g. Non-ambulatory III- Is no longer able to use his/her hands to hold a pen

4. Do you often think about the life expectancy of your child with DBMD?
    a. Never
    b. Very little
    c. Occasionally
    d. Very often

5. If you do think about life expectancy, what is your feeling about your child's life expectancy? Most often I feel that my child will:
    a. Live 30 years or less
    b. Live more than 30 years
    c. N/A because my child with DBDM is older than 30 years

Your Carrier Status

The next set of questions is about your carrier status for DBMD.

1. Are you a carrier for DBMD?

- Yes, I am a carrier. [answer questions 4-9]
- No, I am not a carrier or I am extremely unlikely to be a carrier. [skip to end of section]
- I do not know if I am a carrier. [answer questions 2 and 3 and then skip to end of section]

2. If you do not know your carrier status, what is the reason for not knowing?

    - A healthcare provider told me but I do not remember.
    - I am waiting on carrier test results.
    - My health insurance will not cover the test or it is too expensive.
    - I am not ready to know my carrier status.
    - I have no reason to know my carrier status.
    - I do not ever want to know my carrier status.
    - Other (please specify):

3. How worried are you that you may be a carrier for DBMD?
    - Not worried at all
    - Somewhat worried
    - Moderately worried
    - Very worried
    - Extremely worried

    [Skip to next section]

    [Q4-Q9 for those who answered “yes” to Q1]

    If you are a carrier: Please indicate your agreement with each of the following statements or questions.

4. Finding out that I was a carrier changed my life.

    - Strongly disagree
    - Disagree
    - Neither agree or disagree
    - Agree
    - Strongly agree

5. Finding out that I was a carrier changed the way I think of myself as a mother.

    - Strongly disagree
    - Disagree
    - Neither agree or disagree
    - Agree
    - Strongly agree

6. How did finding out you were a carrier affect your relationship with your partner?
    - I feel much less connected to my partner.
    - I feel somewhat less connected to my partner.
    - My relationship with my partner did not change.
    - I feel somewhat more connected to my partner.
    - I feel much more connected to my partner.
    - I do not have a partner.

7. How did **finding out you were a carrier** change your relationship with your child with DBMD?

    - I feel much less connected to my child.
    - I feel somewhat less connected to my child.
    - My relationship with my child did not change.
    - I feel somewhat more connected to my child.
    - I feel much more connected to my child.

8. How did **finding out you were a carrier** change your relationship with your daughter(s)?

    - I have no daughters.
    - I feel much less connected to my daughter(s).
    - I feel somewhat less connected to my daughter(s).
    - My relationship with my daughter(s) did not change.
    - I feel somewhat more connected to my daughter(s).
    - I feel much more connected to my daughter(s).

9. In what way has your gene carrier status most affected your life?
[open field for responses]

Your Needs

This section of the survey is about needs that some mothers have told us they have. Please choose an option that tells how much each item applies to you.

This is what the numbers mean:

1. *I don't have this kind of need or it is not relevant to me.*
2. *I used to have this kind of need but my need has been met.*
3. *I have a low need for this type of help.*
4. *I have a moderate need for this type of help.*
5. *I have a high need for this type of help.*

I would like to learn more about:

1. Specific ways to cope with being a mother of a child with DBMD.
2. Specific ways to manage my sadness related to my child's DBMD.
3. Specific ways to manage my fears related to my child's DBMD.

4. Specific ways to deal with uncertainty about my child's future.
5. Whether the way I feel is normal.
6. Relaxation techniques.
7. Better ways to get the support I need from others.
8. How to take time for healthy life choices (such as diet and exercise).
9. Ways of self-care that improve my sense of wellbeing and happiness.

****

10. What specific things do you do, if any, to help keep up your own wellbeing? For example, some mothers talk about exercise, taking brief time-outs for alone time, or speaking to or meeting with friends. [open text field]

Your Home

Please tell us whether you agree with the statements below, which are about your home.

This is what the numbers mean:

1 =*strongly disagree*

2 =*disagree*

3 =*neutral*

4 =*agree*

5 =*strongly agree*

1. I feel unable to get out of my house because of my care responsibilities.
2. I find my home environment pleasant.
3. There is a place in my house where I can go to relax and be calm.
4. When I have the opportunity to take time out of the house to myself, I find that it isn't worth the effort.

Finances

How much do the financial (money) aspects of caring for your child with DBMD worry you?

Marking 0 on the scale means you feel that you have no worries, while marking a 10 means you feel that you are extremely worried, and marking a 5 means you have a medium amount of worry.

0	***5***	***10***
Not worried at all		Extremely worried

Respite Care

Please tell us whether you agree with the statements below, which are about respite care.

This is what the numbers mean:

1 *=strongly disagree*

2 *=disagree*

3 *=neutral*

4 *=agree*

5 *=strongly agree*

N/A= my child with DBMD is independent and does not require respite care

1. I have all of the information I need on how to find respite care.
2. I regularly use respite care.
3. Finding respite care is more trouble than it is worth.
4. I am worried about allowing someone else to care for my child.
5. My child is willing to be cared for by someone else.
6. I could benefit from a break from caring for my child.

7. I don’t deserve a break from caring for my child.

Caring for Your Child with DBMD

This section is about things that help you give optimal care to your child with DBMD. How much do you agree that these things help you care for your child with DBMD?

This is what the numbers mean:

1. not at all
2. a little bit
3. somewhat
4. very much

1. My relationship with my partner helps me give the best care I can give to my child with DBMD.

I don’t have a partner.

2. My relationships with family members help me give the best care I can give to my child with DBMD.

I don’t have any relationships with family members.

3. My relationships with friends help me give the best care I can give to my child with DBMD.

I don’t have any relationships with friends.

4. My faith/spiritual beliefs help me give the best care I can give to my child with DBMD.

5. Support from some of my child’s healthcare providers helps me give the best care I can give to my child with DBMD.

6. My child with DBMD’s approach to life helps me give the best care I can give to my child.

7. My child with DBMD has behavior or psychological issues that make it difficult for me to give the best care I can give.

8. If I had more money I could better care for my child with DBMD.

9. I feel more protective toward my child with DBMD than I would toward another child his/her age who does not have DBMD.

10. My relatives understand what it is like to care for a child with DBMD.

11. My relatives are supportive of the way I care for my child with DBMD.

Care During Difficult Times

Think back to the last really difficult time you had caring for your child with DBMD. Please indicate how much each of the items listed below keep you going during hard times.

This is what the numbers mean:

1. *Not at all*
2. *a little bit*
3. *somewhat*
4. *very much*

During difficult times when caring for my child with DBMD, the following things keep me going:

1. Love for my child
2. My duty to care for my child
3. There is no one else who can care for my child like I can
4. The importance of being a good mother
5. Believing that things will get better
6. Taking it one day at a time

Right now, what **one** thing about DBMD makes it most difficult for you to care for your child? [open ended]

Your Outlook on Life

This section includes questions about your general life expectations. Please answer the following questions by choosing the number that shows how much you agree or disagree with each statement. There are no right or wrong answers.

This is what the numbers mean:

1 *=strongly disagree*

2 *=disagree*

3 *=neutral*

4 =*agree*

5 =*strongly agree*

1. In uncertain times, I usually expect the best.
2. It's easy for me to relax.
3. If something can go wrong for me, it will.
4. I'm always optimistic about my future.
5. I enjoy my friends a lot.
6. It's important for me to keep busy.
7. I hardly ever expect things to go my way.
8. I don't get upset too easily.
9. I rarely count on good things happening to me.
10. Overall, I expect more good things to happen to me than bad.

Your Feelings about Yourself

This section includes questions about your feelings about yourself. Please answer the following questions by choosing the number that shows how much you agree or disagree with each statement.

This is what the numbers mean:

1 =*strongly disagree*

2 =*disagree*

3 =agree

4 =*strongly agree*

1.	I feel that I am a person of worth, at least on an equal plane with others.
2.	I feel that I have a number of good qualities.

3.	All in all, I am inclined to feel that I am a failure.
4.	I am able to do things as well as most other people.
5.	I feel I do not have much to be proud of.
6.	I take a positive attitude toward myself.
7.	On the whole, I am satisfied with myself.
8.	I wish I could have more respect for myself.
9.	I certainly feel useless at times.
10.	At times I think I am no good at all.

Your Resilience

This section includes questions about how you have feel about yourself. Please answer the following questions by choosing the number that shows how much you believe each statement is true.

This is what the numbers mean:

1. *Strongly disagree*
2. *Disagree*
3. *Neutral*
4. *Agree*
5. *Strongly agree*

1. I enjoy being with other people.
2. It is easy to be flexible in social situations.
3. I have friends/family members who appreciate my abilities.
4. When I have a goal, I do my best to attain it.
5. I establish friendly relationships easily.
6. I enjoy being with my family.
7. I communicate well with new people.
8. When in difficult situations, I know there is a better future.
9. There are strong connections among my friends.
10. I laugh easily.
11. I can discuss personal issues with friends/family members.
12. I believe in my abilities.
13. There are family members/friends who help me.
14. I know how to achieve my goals.
15. My family agrees on important affairs in life.
16. I can solve my personal problems.
17. Regular rules make my daily life easier.
18. It is easy to find subjects to talk about with other people.
19. I know I will succeed if I keep trying.
20. I have friends/family members who encourage me.
21. I prefer to have plans for my activities.
22. My family is optimistic in difficult situations.
23. I trust my judgments and decisions.
24. There is always someone who helps me when needed.
25. I am quickly informed when a family member has a problem.
26. I have strong connections in my family.
27. A good future awaits me.
28. My family is honest with each other.
29. I maintain daily rules even in difficult situations.
30. My family enjoys finding a chance to do things together.
31. I always find a way to solve problems regardless of what happens.
32. I have realistic plans for the future.

Effects of DBMD on Your Family

This section is about the effects of DBMD on your family. Please answer each question. Marking 0 on the scale means you feel that there is no effect, while marking a 10 means you feel that there is a very large effect, and marking a 5 means you feel there is a medium effect.

1. How much of a **positive** effect does your child's condition have on your entire family?
2. How much of a **negative** effect does your child's condition have on your entire family?

Effects of DBMD on You

The questions below reflect how people sometimes feel about **their child with DBMD**. After each statement, please choose the response that best describes how often you feel this way. There are no right or wrong answers.

DO YOU FEEL:

	Never	Rarely	Sometimes	Quite frequently	Nearly always
1. …that because of the time you spend with your child with DBMD, you don't have enough time for yourself?	0	1	2	3	4
2. …stressed between caring for your child with DBMD and trying to meet other responsibilities (work/family)?	0	1	2	3	4
3. …angry when you are around your child with DBMD?	0	1	2	3	4
4. …that your child with DBMD currently affects your relationship with family members or friends in a negative way?	0	1	2	3	4
5. …strained when you are around your child with DBMD?	0	1	2	3	4
6. …that your health has suffered because of your involvement with your child with DBMD?	0	1	2	3	4
7. …that you don't have as much privacy as you would like because of your child with DBMD?	0	1	2	3	4

8. …that your social life has suffered because you are caring for your child with DBMD?	0	1	2	3	4
9. …that you have lost control of your life since your child's diagnosis of DBMD?	0	1	2	3	4
78. …uncertain about what to do about your child with DBMD?	0	1	2	3	4
79. …you should be doing more for your child with DBMD?	0	1	2	3	4
80. …you could do a better job in caring for your child with DBMD?	0	1	2	3	4

About You and DBMD

This section is a list of statements that mothers of children with DBMD sometimes make about themselves. Please choose how much you agree with each item below. If the statement does not apply to you please choose 8 for "not applicable." Please answer each question.

This is what the numbers mean:

1. *Strongly disagree*
2. *Disagree*
3. *Somewhat disagree*
4. *Neither agree nor disagree*
5. *Somewhat agree*
6. *Agree*
7. *Strongly agree*
8. *N/A*

1. I feel I have lost my sense of privacy.
2. I am able to deal with my child's illness.
3. I distrust my ability to care for my son.
4. I feel isolated because of my child's illness.
5. I know my child's illness well.
6. I feel labeled.
7. I am worried that my child's symptoms will suddenly get worse.

8. My child's illness gets in the way of who I really am.
9. I have become more secretive.
10. I feel burdened by knowledge about my child's condition.
11. I feel different from parents who have healthy children.
12. I am hopeful for my child in the future.
13. I feel my life isn't fair.
14. I am worried that my child's doctor will find something wrong during appointments.
15. I feel cursed because of my child's illness.
16. I feel like at any moment I could fall apart.
17. I am in control of my child's health.

Your Control Over DBMD

This section asks you about how much control you or others have over certain aspects of your child's DBMD. Marking 0 on the scale means you feel that you have no control over a particular aspect, while marking a 10 means you feel that you have complete control, and marking a 5 means you have a medium amount of control.

Please answer each item.

1. In general, how much control do you feel you have over your child's DBMD?

No Control 0 1 2 3 4 5 6 7 8 9 10 Complete Control

2. How much control do you feel you have over your child's daily symptoms?

3. How much control do you think you have over the long-term course of your child's DBMD?

4. How much control do you think you have over the medical care and treatment of your child's DBMD?

5. How much control do you think that others (a spouse, doctor, God, etc.) have over your child's DBMD?

Your Worry about DBMD

This section is about your worries. Please choose the item that best describes your worries. There are no right or wrong answers. Please answer each question.

1. How worried are you about your ability to care for your child with DBMD?
   Not at all Somewhat Moderately A Great Deal Extremely

2. During the past week, how often have you worried about your ability to care for your child with DBMD?
   Never Rarely Sometimes Often All of the time

3. How *bothered* are you by thinking about your ability to care for your child with DBMD?
   Not at all Somewhat Moderately A Great Deal Extremely

4. How worried are you about your relationship with your partner?
   N/A Not at all Somewhat Moderately A Great Deal Extremely

5. During the past week, how often have you worried about your relationship with your partner?
   N/A Never Rarely Sometimes Often All of the time

6. How *bothered* are you by thinking about your relationship with your partner?
   Not at all Somewhat Moderately A Great Deal Extremely

7. How worried are you about your family's wellbeing?
   Not at all Somewhat Moderately A Great Deal Extremely

8. During the past week, how often have you worried about your family's wellbeing?
   Never Rarely Sometimes Often All of the time

9. How *bothered* are you by thinking about your family's wellbeing?
   Not at all Somewhat Moderately A Great Deal Extremely

Your coping with DBMD

Many mothers use different ways to cope with caring for a child with DMD. This section asks you to rate how well you can perform different types of coping. Choose the number (1-11) that best shows to how confident or certain you are that you can do what is described ***to cope with your child's DBMD****.*

When things aren't going well for you, or when you're having problems, how confident or certain are you that you can:

1. Break an upsetting problem about DBMD down into smaller parts

Cannot do at all	1	2	3	4	5	6	7	8	9	10	11	Certain can do

2. Sort out what can be changed and what cannot be changed about DBMD

Cannot do at all	1	2	3	4	5	6	7	8	9	10	11	Certain can do

3. Make a plan of action and follow it when confronted with a problem related to DBMD

Cannot do at all	1	2	3	4	5	6	7	8	9	10	11	Certain can do

4. Leave options open when things related to DBMD get stressful

Cannot do at all	1	2	3	4	5	6	7	8	9	10	11	Certain can do

5. Think about one part of a DBMD problem at a time

Cannot do at all	1	2	3	4	5	6	7	8	9	10	11	Certain can do

6. Find solutions to your most difficult DBMD problems

Cannot do at all	1	2	3	4	5	6	7	8	9	10	11	Certain can do

7. Resist the impulse to act hastily when under pressure

Cannot do at all	1	2	3	4	5	6	7	8	9	10	11	Certain can do

8. Try other solutions to DBMD problems if your first solutions don't work

Cannot do at all	1	2	3	4	5	6	7	8	9	10	11	Certain can do

9. Talk positively to yourself

Cannot do at all	1	2	3	4	5	6	7	8	9	10	11	Certain can do

10. Stand your ground and fight for what you want

Cannot do at all	1	2	3	4	5	6	7	8	9	10	11	Certain can do

11. See things from another person's point of view during a heated argument about DBMD

Cannot do at all	1	2	3	4	5	6	7	8	9	10	11	Certain can do

12. Develop new hobbies or recreations

Cannot do at all	1	2	3	4	5	6	7	8	9	10	11	Certain can do

13. Make unpleasant thoughts about DBMD go away

Cannot do at all	1	2	3	4	5	6	7	8	9	10	11	Certain can do

14. Take your mind off unpleasant thoughts about DBMD

Cannot do at all	1	2	3	4	5	6	7	8	9	10	11	Certain can do

15. Stop yourself from being upset by unpleasant thoughts about DBMD

Cannot do at all	1	2	3	4	5	6	7	8	9	10	11	Certain can do

16. Keep from feeling sad about DBMD

Cannot do at all	1	2	3	4	5	6	7	8	9	10	11	Certain can do

17. Keep from getting down in the dumps about DBMD

Cannot do at all	1	2	3	4	5	6	7	8	9	10	11	Certain can do

18. Look for something good in a negative situation

Cannot do at all	1	2	3	4	5	6	7	8	9	10	11	Certain can do

19. Keep yourself from feeling lonely

Cannot do at all	1	2	3	4	5	6	7	8	9	10	11	Certain can do

20. Visualize a pleasant activity or place

Cannot do at all	1	2	3	4	5	6	7	8	9	10	11	Certain can do

21. Pray or meditate

Cannot do at all	1	2	3	4	5	6	7	8	9	10	11	Certain can do

22. Get friends to help you with the things you need

Cannot do at all	1	2	3	4	5	6	7	8	9	10	11	Certain can do

23. Get emotional support from friends and family

Cannot	1	2	3	4	5	6	7	8	9	10	11	Certain

do at all can do

24. Make new friends

Cannot do at all 1 2 3 4 5 6 7 8 9 10 11 Certain can do

25. Do something positive for yourself when you are feeling discouraged

Cannot do at all 1 2 3 4 5 6 7 8 9 10 11 Certain can do

26. Get emotional support from community organizations or resources

Cannot do at all 1 2 3 4 5 6 7 8 9 10 11 Certain can do

Impact of DBMD on You

This section is about the impact that having a child with DBMD has on you. Read and answer all the statements, even if you are not completely sure about your answer. For each statement choose the number that best described how much you agree.

This is what the numbers mean:

1. *Not at all*
2. *A little bit*
3. *Somewhat*
4. *Quite a bit*
5. *Very much*

Being a parent to an individual with Duchenne or Becker muscular dystrophy has…

	Not At All	**A Little Bit**	**Somewhat**	**Quite a Bit**	**Very Much**
Helped me accept the way things work out	**1**	**2**	**3**	**4**	**5**
Helped me learn to deal better with uncertainty	**1**	**2**	**3**	**4**	**5**

Taught me how to adjust to things I cannot change	**1**	**2**	**3**	**4**	**5**
Helped me take things as they come	**1**	**2**	**3**	**4**	**5**
Helped me to look at things in a more positive way	**1**	**2**	**3**	**4**	**5**
Helped me learn to handle difficult times	**1**	**2**	**3**	**4**	**5**
Helped me become more comfortable with who I am	**1**	**2**	**3**	**4**	**5**
Helped me become a stronger person	**1**	**2**	**3**	**4**	**5**
Helped me feel better about my ability to handle problems	**1**	**2**	**3**	**4**	**5**
Helped me become a better person	**1**	**2**	**3**	**4**	**5**

	Not At All	**A Little Bit**	**Somewhat**	**Quite a Bit**	**Very Much**
Helped me know who I can count on in times of trouble	**1**	**2**	**3**	**4**	**5**
Makes me more willing to help others	**1**	**2**	**3**	**4**	**5**
Helped relationships become more meaningful	**1**	**2**	**3**	**4**	**5**

Helped me become closer to people I care about	**1**	**2**	**3**	**4**	**5**
Helped me become more aware of the love and support available from other people	**1**	**2**	**3**	**4**	**5**
Helped me learn my life is more meaningful	**1**	**2**	**3**	**4**	**5**
Given me a greater appreciation for life	**1**	**2**	**3**	**4**	**5**
Helped me develop a deeper sense of purpose in life	**1**	**2**	**3**	**4**	**5**
Helped me feel peaceful	**1**	**2**	**3**	**4**	**5**
Helped me find strength in my faith or spiritual beliefs	**1**	**2**	**3**	**4**	**5**

Thank you for completing the survey. If you have any questions or concerns, email Holly Peay at holly@parentprojectmd.org or call 443-791-5927. Please expect to receive the next survey in about 12 months. The next survey will only be about half as long as this survey. You will notice that some of the questions are the same, which allows us to look for change over time.

If you move or change your email address or phone number, please let Holly know. We appreciate your continued involvement in this project.

Appendix B: Carrier Status Section

Your Carrier Status

The next set of questions is about your carrier status for DBMD.

1. Are you a carrier for DBMD?

    - Yes, I am a carrier. [answer questions 4-9]
    - No, I am not a carrier or I am extremely unlikely to be a carrier. [skip to end of section]
    - I do not know if I am a carrier. [answer questions 2 and 3 and then skip to end of section]

2. If you do not know your carrier status, what is the reason for not knowing?

    - A healthcare provider told me but I do not remember.
    - I am waiting on carrier test results.
    - My health insurance will not cover the test or it is too expensive.
    - I am not ready to know my carrier status.
    - I have no reason to know my carrier status.
    - I do not ever want to know my carrier status.
    - Other (please specify):

3. How worried are you that you may be a carrier for DBMD?
    - Not worried at all
    - Somewhat worried
    - Moderately worried
    - Very worried
    - Extremely worried

    [Skip to next section]

    [Q4-Q9 for those who answered “yes” to Q1]

    If you are a carrier: Please indicate your agreement with each of the following statements or questions.

4. Finding out that I was a carrier changed my life.

    - Strongly disagree
    - Disagree
    - Neither agree or disagree
    - Agree
    - Strongly agree

5. Finding out that I was a carrier changed the way I think of myself as a mother.

    - Strongly disagree
    - Disagree
    - Neither agree or disagree
    - Agree
    - Strongly agree

6. How did finding out you were a carrier affect your relationship with your partner?
    - I feel much less connected to my partner.
    - I feel somewhat less connected to my partner.
    - My relationship with my partner did not change.
    - I feel somewhat more connected to my partner.
    - I feel much more connected to my partner.
    - I do not have a partner.

7. How did **finding out you were a carrier** change your relationship with your child with DBMD?

    - I feel much less connected to my child.
    - I feel somewhat less connected to my child.
    - My relationship with my child did not change.
    - I feel somewhat more connected to my child.
    - I feel much more connected to my child.

8. How did **finding out you were a carrier** change your relationship with your daughter(s)?

    - I have no daughters.
    - I feel much less connected to my daughter(s).
    - I feel somewhat less connected to my daughter(s).
    - My relationship with my daughter(s) did not change.
    - I feel somewhat more connected to my daughter(s).
    - I feel much more connected to my daughter(s).

9. In what way has your gene carrier status most affected your life?

[open field for responses]