Left Out in the Cold:

Barriers to Clinical Trial Participation and the Impact on Parents of Sons with Duchenne Muscular Dystrophy

Master’s Thesis

Presented to

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By
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Abstract

Left Out in the Cold: Barriers to Clinical Trial Participation and the Impact on Parents of Sons with Duchenne Muscular Dystrophy

A thesis presented to the Graduate Program in Genetic Counseling

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The primary goal of clinical trials is to advance our understanding of human disease, yet participants may have the potential to see direct benefits on their condition. Novel therapeutic clinical trials specific to Duchenne muscular dystrophy (DMD), a life-limiting condition, have begun to show promising results, but inclusion criteria for these trials often require ambulation, a criterion that the majority of older boys with DMD do not meet. The impact of ineligibility for eagerly anticipated clinical trials has never been examined.

The purpose of this study was to explore the feelings of parent caregivers of boys with DMD that are ineligible for novel therapeutic clinical trials for DMD. We interviewed seven parents of non-ambulatory boys with DMD using a semi-structured interview guide to assess the impact of exclusion from clinical trials on parental hopefulness, expectations for the future, and continued interest in clinical research. To find common and divergent themes, we used software to identify and code themes that emerged from the interview transcripts.
Four common themes emerged: (1) Ambulation represents a barrier to research participation, and parents are often frustrated by their exclusion due to this barrier. (2) Parents conveyed resignation to their son’s condition, his ineligibility, and the limitations regarding therapies to be developed during his lifetime. (3) Parents’ active interest in clinical research opportunities waned. (4) Parents still expressed hope for their sons, often related to the concept of emotional well being, but also to the possibility of a cure or slowing the progression of his condition. These results highlight how parents rationalize being left out of clinical trials while maintaining hope. These parents’ hope likely represents healthy coping with chronic disease. Genetic counselors and other providers should continue discussing research with older boys with DMD.

Keywords: Duchenne muscular dystrophy, clinical trials, ineligibility, exclusion, hope, ambulation
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Introduction

Therapeutic clinical trials are advancing our understanding of human disease. Though the goals of clinical trials are not the same as clinical care, participants have the potential to see direct benefits of their condition, should the therapy prove effective. Not all patients are able to participate in trials for any given condition, however. There can be many barriers to participation, some which can be individual-driven, such as lack of transportation to study appointments, and some can be scientifically driven, such as specific disease-related inclusion criteria. This is the case with Duchenne muscular dystrophy (DMD). DMD is a life-limiting neuromuscular condition that affects 1 in 3,500 males and is characterized by progressive muscle weakness, leading to loss of ambulation and full-time wheelchair use by age 13, onset of respiratory and cardiac failure, and premature death (Bushby, et al., 2010). Utilization of corticosteroids and assisted ventilation in the treatment of DMD has increased the average survival from late teens into the third and fourth decade of life and sometimes beyond (Eagle, et al., 2002; Bushby, et al., 2010). Even with these advances, DMD still proves to be a devastating condition with no cure.

Recently, however, novel therapeutic clinical trials for the treatment of DMD have provided some promising results. Techniques that aim to delay loss of ambulation by exon-skipping strategies and premature stop codon read-through technologies hope to yield improved clinical symptoms in this condition (Govoni, et al., 2013; Finkel, 2010; Fairclough, et al., 2013). These trials, however, have strict inclusion criteria that limit participation to boys who can walk at the time of the trial, as the main outcome measure used historically in DMD research is the
six-minute walk test (McDonald, et al., 2013). The use of this measure as the primary outcome to determine efficacy excludes the majority of patients who are older and who have already lost their ability to walk.

The experience of parents and families caring for boys with DMD has been well characterized in the literature, with many parents reporting increased stress; however, parents often report holding onto hope that a cure may be found in time for their son (Nereo, et al., 2003; Erby, et al., 2006). As their child’s condition progresses, parents of older boys with DMD tend to shift their priorities to preserving the psychiatric health and well-being of their child in the face of their late-stage condition (Bothwell, 2002). With the advent of new promising therapeutic clinical trials, however, the medical trajectory of DMD is potentially changing, with increased possibility for a treatment effect for the ambulatory boys that participate. Older, non-ambulatory boys have largely been unable to participate, and parental impact of this disqualification has never been studied.

Time is of the essence in this progressive condition, so inclusion-based barriers to participation may fuel some parents’ drive to find a trial that will allow their son to be included. Alternatively, this disqualification could cause their interest in clinical research to fade, causing some families to shift their focus away from all clinical trial participation. We interviewed parents of older, non-ambulatory boys with DMD who are currently ineligible to participate in many novel therapeutic clinical trials. The purpose of this study was to assess the impact of ineligibility from clinical trials on these parents’ hopefulness, expectations, and interest in clinical research. Understanding the perspective and needs of patients and families with advanced DMD is helpful for medical staff, including genetic counselors, as research opportunities are often discussed with families at clinic visits.
Methods

Methodology and IRB Approval

We utilized qualitative methods through phone interviews to gain an in-depth understanding of the participants’ individual experiences. The Brandeis University Institutional Review Board approved our study materials and protocol, which included our recruitment letter (Appendix A), interview guide (Appendix C), and a description of study procedures to be followed. Boston Children’s Hospital’s Institutional Review Board approved the recruitment letter, and we mailed this letter to families of boys who had previously participated in the Harvard Neuromuscular Disease Project at Boston Children’s Hospital and had indicated they could be contacted for future research projects.

Sampling Methods, Study Participants, and Informed Consent

To recruit for our study, we mailed recruitment letters to the family of individuals who had previously participated in the Harvard Neuromuscular Disease Project at Boston Children’s Hospital. Subject inclusion criteria were: being 18 years or older, being fluent in English, having a son with Duchenne muscular dystrophy who has lost his ability to walk, and being interested in clinical research for their son. The principal investigator, who is part of the research team at the Harvard Neuromuscular Disease Project, mailed these letters to potential study participants. Along with this recruitment letter, the principal investigator included an opt-out card for the participant to mail back to the principal investigator if they were not interested in being contacted for this study.
Two weeks after these letters were mailed, the principal investigator contacted by phone anyone who had not declined the study to assess interest, provide more information about the study if needed, and to explain that the student interviewer would call to discuss the interview further. The principal investigator then provided phone contact information of participants to the interviewer, and the interviewer contacted these participants via phone to verify eligibility and coordinate the informed consent process. Of seventeen potential study participants, five mailed back opt-out cards, three participants did not return phone calls, one participant’s phone number was not in service, one participant did not qualify for the study based on our eligibility criteria, and seven agreed to participate. The interviewer emailed the informed consent form (Appendix B) to the participants for review prior to conducting the phone interview. At the time of the interview, the interviewer ensured the participant had received the form, answered any questions the participant had, and obtained verbal consent to participate in this study. We audio recorded and transcribed this verbal consent, and kept a copy of this transcription as documentation of consent. We gave each participant a $25 gift certificate to thank them for their participation.

**Interviews and Data Management**

We designed a semi-structured interview guide to elicit the experiences of our participants in a conversational yet consistent format. This guide included closed-ended questions related to demographic and background information, as well as open-ended questions. We created this interview guide based on past research experiences and perspectives from the entire committee. This guide and the subsequent interviews included topics such as the parents’ experience and perceptions of clinical research, how parents get their information regarding DMD and clinical research, communication with their affected son regarding clinical research, and hope and expectations for the future. Prior to conducting interview with participants, this
interview guide was pilot tested with committee members who have personal and professional experience with clinical research in DMD.

Interviewees were assigned unique identifiers to ensure confidentiality. To keep consistency, one interviewer conducted all interviews using the same guide, but each interview was unique in the exact questions asked, the order of the topics, and the amount of time spent per topic. All interviews were audio-recorded with consent from the participant, and interviews lasted between 30 and 60 minutes. A professional transcriptionist transcribed these audio recordings. Transcripts, audio recordings, and any documents with identifying information were stored on a secure, password-protected laptop.

Data Analysis

We uploaded our transcripts into Atlas.ti (version 7.0) for qualitative analysis of the data. We coded the interviews by reading the transcripts and assigning codes to sections of text, paragraphs, and sentiments expressed by the interviewee based on expected codes derived from interview guide questions as well as new themes that emerged from the interviews. An original set of 253 codes was obtained after coding the interviews, and these were focused and merged to a set of 45 codes, which were further organized into eight code groups. Within these eight code groups, four major themes emerged: more active interest in research in the past, ambulation representing a barrier to research, resignation about disease progression, and hope for the future. We analyzed these interviews for commonalities within these themes, as well as divergent opinions or responses.
Results

Demographic Information and Resource Identification

Six of the participants in this study were mothers, and one interviewee was a father. All of the participants had at least one son with Duchenne muscular dystrophy; one mother had two sons with DMD, and one mother had another son with Becker muscular dystrophy. In the instances where participants had multiple affected children, the interview was focused predominantly on their experiences with the older affected son.

The participants’ sons’ ages ranged from 13 to 25 years of age. All of the boys with DMD were wheelchair-dependent, and began using wheelchairs full-time at ages that ranged from 9.5 to 15.5 years of age. None of these children had been diagnosed with any learning disabilities or autism. The details of our participants and their sons are listed in Table 1.

Table 1: Demographics of participant and affected son

<table>
<thead>
<tr>
<th>Participant’s Gender</th>
<th>Participant’s current age</th>
<th>Son’s current age</th>
<th>Son’s age at DMD diagnosis</th>
<th>Son’s age at full-time wheelchair use</th>
</tr>
</thead>
<tbody>
<tr>
<td>Female</td>
<td>60</td>
<td>21</td>
<td>3</td>
<td>10 ½</td>
</tr>
<tr>
<td>Female</td>
<td>65</td>
<td>25</td>
<td>6</td>
<td>10 ½</td>
</tr>
<tr>
<td>Female</td>
<td>50</td>
<td>21</td>
<td>2</td>
<td>9 ½</td>
</tr>
<tr>
<td>Female</td>
<td>47</td>
<td>21</td>
<td>2 ½</td>
<td>12</td>
</tr>
<tr>
<td>Female</td>
<td>47</td>
<td>25</td>
<td>8</td>
<td>15 ½</td>
</tr>
<tr>
<td>Female</td>
<td>51</td>
<td>17</td>
<td>6</td>
<td>9</td>
</tr>
<tr>
<td>Male</td>
<td>53</td>
<td>13</td>
<td>2</td>
<td>11</td>
</tr>
</tbody>
</table>

We also asked participants about where they primarily obtained their information regarding clinical trials and research for DMD in order to understand the resources these families were accessing for information. Notably, many parents stated their doctors used to discuss
research opportunities with them, but these physicians have not discussed research with them recently. Some parents also cited the fact that they used to be the ones to ask their medical staff about research opportunities in the past, but this practice has slowed or even stopped as their son has gotten older and his disease has progressed giving them many more health concerns. The details of these sources are shown in Figure 1.

**Figure 1: Primary source of information for DMD trials and research**

![Bar chart showing primary sources of information for DMD trials and research]

Through our interviews, four major themes emerged: more active interest in research in the past, barriers to research participation, resignation, and hope for the future.

**Theme: More research interest in the past**

Our interviews began by discussing the participants’ interest or experience with DMD clinical research both past and present, if applicable. This discussion included questions about their level of interest in research, their son’s interest in research, whether these perspectives have
changed over time, what clinical trial participation means to their family, and what motivates the participants to look for research opportunities or trials.

Overall, parents reported being more actively interested in clinical research in the past than they are at present. Within this theme of increased interest in the past, three subthemes emerged that contributed their current level of interest: hesitation to participate for fear of worsening conditions or adverse side effects, other priorities due to complicated medical management of advanced disease, and discouragement with their perception of lack of progress in research.

**Hesitation to participate**

Of the seven participants interviewed, six discussed factors that contributed to their hesitation to participate in clinical research. Many of these participants cited the risk of side effects of research medication as a factor that contributed to this apprehension especially in the face of the complex medical care needed in advanced DMD. As one mother illustrates:

“I think [research] is very, very important, but at the same time it’s a scary thing because you never know what could happen with these clinical trials. You don’t know if it could have any side effects… so it’s a little scary.” –Participant 4

Another mother discussed her thought process for considering research trials and opportunities, citing her fear of causing her sons any additional pain from side effects of trial drugs:

“It depends… I don’t want anything to hurt them or make them worse. Do you know what I mean? As long as there is no risk in hurting them or pain for them…. Because I do not want them to be in any pain.” –Participant 5

Three participants discussed their hesitation to participate stemming from the desire to avoid multiple visits or significant travel to research sites, as it would take away from their son’s other activities, interests, and educational goals. One mother described her desire to help her sons
have full lives rather than spend time and energy looking for trials in desperation, especially at this point in their lives:

“I feel I would rather teach my sons how to live as full a life as possible, even if they only have 20 or 25 years of life, versus trying to drag them around the country for clinical trials and have them almost lose their childhood.” –Participant 6

**Lack of Research Progress Discouraging**

Four participants cited the lack of progress and slow speed of research to be significant factors that contributed to their lack of interest in clinical research. Three participants cited the lack of trials they were eligible for and shortage of positive results from other ongoing trials as factors that discouraged their interest in clinical research. In discussing her communication with her son’s medical team about clinical trial opportunities, one mother highlighted her perception of a lack of ongoing studies as a factor that impacted her interest:

“Well, we used to ask a lot [about trials], and now I’ve just stopped asking. I don’t know. Because there is very little happening, it seems to me.” –Participant 1

The lack of positive results from existing clinical trials in combination with their medical team’s perspective was discouraging to parents’ and sons’ level of interest in findings clinical trials in which to participate. As one mother described:

“When push comes to shove, they’re not getting FDA approval, they’re not moving forward, and so our doctors are not necessarily saying, “You need to jump on the bandwagon” because they’re just not moving forward… And I think if there was solid research that was getting results and our physicians were saying, “Look at what is happening. This trial or this new drug is having a great success. I think you should try it.” If the physicians were giving us that kind of feedback that maybe the boys would be more interested… I mean there are no boys that I know of who haven’t died from Duchenne muscular dystrophy at a certain point. There are no boys jumping out of their wheelchairs and getting cured. So, if things were more positive then I think [we] would be more interested.” –Participant 6

One mother cited the speed of research to be a factor as well:

“Well, most of [the trials] aren’t ready for people yet, especially in the United States… A lot of them go nowhere.”–Participant 3
One father discussed frustration with the speed of research:

“It can be a little frustrating to see studies take many, many years with various little setbacks and moving forward a few steps and back a step.” –Participant 7

While participants cited the lack of progress as discouraging, some participants discussed their understanding of the scientific process and the slow pace of research.

Other Priorities

Parents often brought up their focus on other priorities at this stage of their son’s life rather than concentrating their efforts into finding clinical research for their son. While research is still important to these parents, there are other more pressing priorities that have emerged at this point in their son’s life and condition.

Parents cited other medical issues as more of a concern as opposed to considering research options at this point in their son’s life. As they age, these sons have become progressively more disabled, and as this occurs, their care becomes these parents’ primary focus. A mother described the fact that the focus in their family is less on research opportunities and more on advanced care planning and medical decisions:

“I think we’re at a point where what we’re discussing now is what would happen if he had to have a breathing apparatus or stomach feeding tube because of the stomach issues, so those are our major concerns.” –Participant 4

Another priority that parents reported was their focus on present and spending quality time with their son. As one mother describes, she has put a premium on focusing on the “now” and improving his life at this point rather than the future, including searching for research opportunities at this point in his life:

“We focus on making life as good as it can be. And I think we are trying to be prepared without really dwelling on the future. We’re trying to dwell on now.” –Participant 2
Education was also a priority that parents cited as more pressing than searching for research opportunities. When asked about their level of interest in clinical research, one mother describes that in the past they were more dedicated to research, but their priorities shifted towards education and achieving academic goals:

“It would be great to have lots of time and be able to write your Senator and this and that… Our project right now is to get him through college… which is more than a full-time job.” –Participant 1

When a child has a progressive disability, parents must dedicate more time and energy to his care over time, further shifting the focus away from having time for clinical trials.

**Theme: Exclusion Criteria and Barriers**

During the interview, we asked participants about their familiarity with the use of ambulation as an endpoint in clinical research for many DMD clinical trials, their opinions about these criteria, and whether these criteria represented barriers to clinical trial participation. From these questions, five participants were familiar with ambulation as a study endpoint and exclusion criterion in many DMD trials, one participant was unfamiliar with ambulation as a criterion, and one was unfamiliar with any criteria for research or the definition of exclusion criteria in general. Four participants also cited age as a criterion for exclusion from many clinical trials for DMD. Regarding ambulation as exclusion criteria for clinical trials, certain sub-themes emerged, including parental perception of the criteria, ambulation as a barrier to research, and their son’s specific mutation as a barrier to current research opportunities.
Parental perception of criteria

Of the five participants who were familiar with the use of ambulation as exclusion criteria for clinical trials for DMD, three stated that they believe these criteria to be fair and that they understand from the researchers’ standpoint why it studies are often designed to only include boys who can walk independently. Parents often cited their understanding of the measurement tools used in the studies and the goals of the experimental drugs, and that children who are non-ambulatory cannot demonstrate the effectiveness of the drugs through walking. While they scientifically understood the criteria as logical, parents appeared to reluctantly accept these criteria as fair. One father described his interpretation of why researchers set up criteria to deem non-ambulatory boys ineligible and how he had to accept this fact:

“Well, given the level of the studies, the very constrained things they are trying to discover and how early it is in the treatment for this... I figure that [researchers] have their reasons and you have got to take this one step at a time, not to make a bad pun.” – Participant 7

Despite their understanding and resignation to the criteria, three participants stated that being ineligible from these studies is still frustrating. As one mother illustrates this combination of sentiments:

“Well, you know obviously it’s frustrating when your kids are not able to be included, but it makes total sense to me... but, it makes me sad, because it makes you face reality. It makes you face reality that your son has progressed... beyond the medical community being able to improve their physical ability. So, we come face to face with that reality, but we come face to face with that reality every day as we watch our boys lose function.” – Participant 6

Their frustration with the criteria was clear, and for this reason we aimed to further explore these parents’ perception of ambulation as inclusion criteria, whether it represents a barrier in their way towards research participation.
Ambulation as a barrier to research

Parents next spoke about their perception of ambulation as a barrier to clinical trial participation. Four participants stated that they felt ambulation was a barrier to research for their son, often stating it is a barrier that they had come to understand for the status of medical research at this point. As one mother describes, “Yes, [ambulation] is a barrier, but it is a barrier that I understand.” –Participant 6. Also echoing this sentiment, one father states his perception and scientific understanding of this barrier:

"A lot of the studies require being ambulatory, so it is a barrier. But, I don’t know if it’s an unnecessary barrier. I think it does make sense that it’s a lot easier to measure strength and to measure results in terms of how many steps someone can take or how much they can walk. Once they can’t walk I can imagine it’s much more difficult to do concrete measurements.” –Participant 7.

In discussing barriers to clinical research participation, four participants brought up the fact that past or current clinical trials were not applicable to their sons’ specific gene alteration, further excluding them from participation. Some parents talked about their interest and excitement when they heard about a new trial, and the letdown they felt when their son’s specific mutation is a limiting factor to his participation. As one mother describes:

“In the past we were so excited, when the stop codon thing came out for some of the boys, which is, what, like seven percent of the boys have a stop codon in the mutation. And I was so excited that day and I thought that [my son], just maybe he would have that. But, he didn’t. It’s an out of frame and he has an in-frame, or something like that.” – Participant 3

Interestingly, mutation status impacted how at least one parent perceived barriers to research. One father recounted conversations he had with his young son about barriers to research, and described that while ambulation did feel like a barrier to research, the fact that his son’s specific mutation type was not being investigated influenced his perception of this barrier, that his son was not missing out on an applicable opportunity:
“It’s mainly that they’re not making something for [his type of mutation], not as much as that he can’t walk… I’m sure [ambulation] would seem like more of a barrier to us if he couldn’t participate.” – Participant 7

**Theme: Resignation**

An overarching theme among the participants was the idea of being resigned to their son’s progressing condition and the decreasing chances of being involved in future trials or therapies. This theme was especially prevalent when we asked if clinical research would be a part of their future plans. Throughout the interviews, four of our seven participants alluded to this theme of resignation, and described their understanding of their son’s condition and what they feasibly believed they could expect in the future. Parents spoke of understanding the reality of their son’s condition, most notably about the fact that the muscle loss could not be reversed, that there would not be a new therapy in their son’s lifetime, and that there would not likely be a clinical trial that would include their son.

**Cannot reverse muscle loss**

Four participants discussed their understanding and resignation that once the muscle was lost in individuals with DMD, it cannot be regained or reversed. They indicated that they did not expect their son to walk again or for their condition to be cured, which they often discussed as a part of their understanding of why non-ambulatory boys are often ineligible for clinical trials. As one mother describes about being resigned to the limitations of improving her sons’ condition:

“They’re never going to get a child out of a wheelchair… never going to take muscles that have been atrophied for a number of years and breathe new life into them, it’s never going to take the bones that have weakened without use. So, I know my boys are not going to get up out of their wheelchairs, no matter what clinical trials move forward and no matter how successful they are… They’re not going to be able to get back that function.” – Participant 6

Another parent was resigned to the fact that it is impossible to reverse muscle loss, citing the only hope for muscle recovery would be a miracle:
“If you think about it, they’re not going to be able to reverse it, they’re never going to be able to reverse it because of the wasting of the muscles, they’re so wasted after [time]. There is nothing they can do unless there is a miracle.” –Participant 4

No future therapy for their son

Three participants discussed the fact that while they were interested in research, they did not realistically feel like there would be a novel therapy to help their son in his lifetime. Some parents described this perspective with a relatively positive angle, that previous advances in therapies had helped him to exceed these parents’ expectations already.

“I pray for other kids that they come up with cures and solutions, but I don’t believe there is anything they can do for [my son]. I mean, the surgeries that they have done in the past have expanded his life expectancy a lot, and even putting in the trach and vent have helped, I mean because he wouldn’t have made it with the lung capacity he was at, let alone for a few surgeries they didn’t expect him to survive. It’s just, at this point, every day is an extra day you’ve been given.” –Participant 3

Another parent described the fund-raising and awareness they raised in the past for DMD research, that it may not prove helpful to her son’s health specifically, with a bit more frustration about seemingly fruitless efforts.

“So [fund-raising] was great, but we never have gotten anything from it, and it looks like we’re not going to in the long run.” –Participant 1

Clinical trial unlikely to include their son

We then asked parents if clinical research was going to be part of their future, most parents indicated their understanding of their sons’ exclusion from research trials, for better or worse. Most parents stated that they would potentially be open to research opportunities if they arose, especially if they were noninvasive, but that they were not “holding their breath” for anything and did not expect to be included. In response to whether research would be a part of their future, one mother described:

“It could be, but… I’m not very optimistic about it.” –Participant 4.
By parental report, their sons often shared this perspective as well. Many parents stated that their sons did not often discuss research options with parents, as they were also resigned to the reality of the state of clinical research. As one mother illustrated regarding her son’s interest:

“I mean, he feels as though there is [no available research] out there for him right now, anyway.” –Participant 4

Overall, parents expressed their reluctant acceptance and resignation to their son’s condition, that they were aware of the limitations of medical science and what could feasibly be expected for their son’s future.

**Theme: Hope**

The final segment of our interviews consisted of questions aimed at parents’ hope for the future, for both their own sons and the DMD community in general. In most interviews, the first hope to be shared was almost always for a cure, and oftentimes this hope for a cure was intended as an obvious, understood response, as exemplified by one mother:

“Well, *obviously* we would all like a cure. Nobody wants their son to have an incurable disease.” –Participant 6

Another mother describes this sentiment:

“I wish they could cure it and no child would have to live through it, or any disease to be honest with you. I mean, things like this shouldn’t happen to innocent children.” –Participant 3

Apart from this universal desire for a cure, certain sub-themes arose as well, namely hope to stop the progression of the disease, hope for their son to have a full life, hope for other families to have support and adequate information, and one parent’s hope to keep their son optimistic about future walking ability.
Stop progression of the disease

The majority of the affected boys in this study were over the age of 17, and most were severely affected by the condition. All of the affected boys required assistance with the majority of their activities of daily living, needing help with bathing, dressing, oftentimes feeding. Most lacked both upper and lower body mobility, but most of the affected boys retained the ability to move their hands, for example to use a mouse on a computer as their main independent ability. The one outlier was the youngest of the group. He was reported as independent in eating and writing, which made him comparatively less affected than the other boys in this study.

Even with parents dealing with the almost complete loss of their child’s mobility, six parents spoke about their hope for researchers to at least stop the progression of the disease, to retain what little mobility their son currently had. Many parents illustrated their desire to keep the precious hand mobility that they see slipping away over time, as one mother stated:

“I wish they would find something for the hands and help them at least keep their hands going, because everything is run by computers now anyway and those 10 digits become very, very important in his world.” –Participant 3

Another mother echoed this similar desire, as well as frustration she feels that she cannot herself stop the progression of the disease:

“I just hope they have something to stop the madness. That’s what I call it. Just stop the disease, to help it. Being a mother, to watch and there is nothing you can do to stop it, because your role as a mother is to protect and you can’t protect them from anything from this disease.” –Participant 5

Parents also discussed their desire for researchers to be able to prolong heart function, respiratory function, and to extend the life expectancy.

Living a full life

While many parents spoke about their medical hopes, five participants discussed their hope for their son from a more holistic perspective, and commented on their desire for their son
to live a full life and reach milestones of individuals without disabilities. One mother described the hope she felt when she saw a friend with muscular dystrophy fulfill typical adult milestones, wishing for the same case for her two affected sons:

“My hope for him is that he can live as independently as possible without me, that he can direct his own caregiving… I would love it if he met a beautiful young lady and were to get married. We went to a wedding of my younger son’s drum teacher [who has Becker muscular dystrophy], and they are expecting their baby in June… And so I have hope that they may get married and have a child someday and be able to support a family.” – Participant 6.

Parents also spoke about their desire for their son to achieve goals he sets for himself unrelated to his diagnosis of DMD, and to spend time doing things he likes doing in the future. One mother discussed her son’s pursuit of a graduate degree, that her hopes were more in the realm of his achieving his own goals:

“I hope that he is able to achieve his goal of getting a graduate degree and find satisfying work and have as full a social life as he wants, and to keep doing the activities he enjoys.” – Participant 2

This mother also discussed her son’s personal hopes and wishes, both past and present, which tended to revolve around being a typical young man:

“And what his fondest wish has always been, is to be a part of the group, to do what the other kids were doing as much as he could… He wants to do what everyone else is doing… He’s a young man like the rest of them.” – Participant 2

Interestingly, when asked about their hope for the future for their son, only one parent stated that he hopes to keep his son optimistic that he might benefit future therapies. This parent was the father of a 13 year old, who discussed the conversations he has with his son regarding research and the potential for a future therapy for him:

“They are building a new high school in our town… and I think he is getting more anxious about how is it going to work… Part of that is, ‘Am I going to be able to walk in five or ten years? Maybe I won’t need this wheelchair once they build the new school.’… So I think he is just wondering ‘What are the chances I’m going to be walking again in
three or four years when high school starts?’ Stuff like that, normal kid stuff. So, we try and keep him hopeful but not give him any false hopes.” –Participant 7

This parent also illustrated the optimism he and his family had demonstrated throughout his son’s life, which had been noticed by outside organizations. Through the MDA, he and his family had often been a resource for families struggling with diagnoses of DMD to serve as a positive outlook and optimistic perspective. He was the only father we interviewed and also had the youngest son in this study, which may be two interesting areas for future research.
Discussion

In this study, we interviewed seven parents of non-ambulatory boys with Duchenne muscular dystrophy to gather information about their interest in clinical trials for DMD and their perspectives on trial participation. Through these interviews, we identified themes that represent the feeling of frustration with being left out of clinical research opportunities, the rationalization of ambulation as a barrier to research, a shift in priorities as their son ages, and maintained hope for their son that these parents experienced. These themes highlight the fact that these parents feel excluded from the research community and employ coping mechanisms in order to process their loss of access to research opportunities and possibly the DMD community. These themes also emphasize ways in which medical professionals can best keep parents informed about research through open discussions about trials with all families, regardless of their child’s age, disease state, or study eligibility.

Barriers to Research

Of the parents that were aware of walking ability as inclusion criteria for clinical research for DMD, the majority stated they considered ambulation a barrier to research. Through our interviews, these parents often reported feeling emotionally impacted by this exclusion, feeling frustrated and sad. By describing the negative effect of these barriers, parents indicated their feelings of exclusion from clinical trials; that they feel left behind by research since their sons cannot participate.

In further discussions regarding this barrier to research, however, parents were quick to rationalize these feelings of exclusion by saying that they are aware of the scientific and clinical
reasons for the barrier’s existence. Parents believed that they fully comprehend how the research trials were being conducted using walking ability as a measure of efficacy, and that their exclusion made logical sense due to these necessary study parameters. These parents were resigned to ambulation as a barrier, and rationalizing it was their way of handling feeling left out, by trying to make sense of it. When individuals are faced with anxiety regarding serious illness and life-limiting situations, rationalization is a coping mechanism that people can use, to strive to make sense and logically understand one’s given circumstances as a way to handle the negative emotion (Gershuny & Burrows, 1990). These parents are rationalizing their loss of access to clinical trials by citing their technical understanding of the state of DMD research as a way to cope.

Through the acceptance of the six-minute walk test as the measurement tool in these trials, the parents demonstrated their interpretation or belief that this study endpoint is the only available method to measure trial efficacy, possibly for the foreseeable future. While the six-minute walk test is historically the most widely used FDA-approved method, other means of measurement are in development, specifically aimed at non-ambulatory boys. Devices designed to measure hand grip strength, pinch strength, and repetitive finger tapping have been developed and shown to be accurate, comparable, and reproducible as measures of strength in non-ambulatory boys with DMD (Servais, 2013). These alternate means of measuring outcomes could allow even more individuals to participate in research if they garner FDA approval, a process that can be expensive and time-consuming. It is possible that if healthcare providers kept parents aware of research advances, including other methods that could be used in studies in the future, parents may interpret this barrier differently, perhaps making them more hopeful and interested for their son to participate in research, despite his loss of walking ability.
**Shifting priorities**

With their son’s progressed disease, his physical condition had slowly deteriorated, but his other abilities had developed similarly to an unaffected individual. As these parents recognized their son’s progressing limitations, these parents’ priorities shifted away from looking for clinical trials to instead focusing on his emotional quality of life, his drive to attain goals that incorporate his personal interests, and maintaining his connection to the world as many of his physical abilities wane. This shift in priorities over time is supported by previous literature, which illustrates that the focus of parents of older boys with DMD changes from exclusively being directed towards physical implications of the disease and moves towards their child’s internal well-being (Bothwell et al., 2002; Samson et al., 2009).

In addition, caring for their son’s physical needs became more involved and time-consuming, with parents often assisting with and performing many of these boys’ activities of daily living including bathing, eating, and dressing. Children with disabilities require a significantly greater level of care and time from their parents than do children without disabilities, and this parental aid does not decrease as the child ages, as is the case for unaffected children (Curran, et al., 2001). As parents devote more and more time to caring for their child with DMD, they have less time to research and participate in clinical trial opportunities.

These parents’ new focus has put the pursuit of research opportunities on the backburner of their priorities, which can be interpreted as another way these parents are compartmentalizing and making sense of their feeling of being left out of research. These parents may view their more involved caretaker role as prohibiting them from having the time to search for research opportunities, even if their son could be eligible. This makes it important for health care providers to offer research updates to these boys and their families at medical appointments, as it
may be their only opportunity to learn about this topic. Allowing parents the opportunity to be kept informed about research is important for their full understanding of advancements in the field of DMD research and potential future treatment, and may keep them feeling connected to the larger DMD community.

**Maintaining hope**

Overall, parents were clear about their resignation to their son’s progressed condition and their realistic expectations for the future, that this disease cannot be cured or treated, that muscle loss cannot be reversed, and that their sons are unlikely to be included in research. Stemming from this reluctant acceptance of these factors, most parents indicated they hope for their son’s development from a holistic perspective, aimed at aspects aside from their physical wellbeing. This relationship is natural, that with resigning themselves to a devastating circumstance such as their son’s disease progression, further hope would lie in what remaining abilities their son still retains, such as his cognitive, social, and emotional abilities.

Alongside these hopes for their son to develop fully as an adult, however, parents also almost uniformly spoke of their desire to stop the progression of their son’s disease and even of finding a cure, often expressed with a sense of “wishful thinking”. This desire for their son to medically benefit from a trial or treatment is something that appears to be in conflict with their aforementioned resignation to the condition and the unavailability of future treatment for their son. This hope shows these parents’ inner desire to change the course of his condition, however far-fetched they indicated this hope to be. While the desire for a cure appears to be in direct conflict to the concept of resignation, these parents’ ability to distinguish between their hope and realistic expectation for their son regarding his condition is supported by recent studies, in the context of parents whose children have been involved in a novel therapeutic clinical trial for
DMD (Peay et al., 2014). Researchers report a disconnect between these parents’ cognitive understanding of research and their emotional hopes for their son, and suggest this is a mechanism for coping with their son’s condition (Peay et al., 2014).

Having hope for the future is essential for caregivers’ coping and adaptation to illness, especially in the setting of a life-limiting condition such as DMD (Gelling, 1999; Samson et al., 2009). While these parents state that they understand their hope for treatment and cure may realistically be unattainable in his lifetime, the fact that they still have this hope for their son also alludes to their wish for their son to see some medical benefit in his lifetime. Parents state wanting to preserve the function he had remaining and to stop the progression of the disease, indicating that they have some interest to learn about research updates, even if only minimal benefits are feasible. This further emphasizes the role of healthcare providers to continue discussing research with families of all boys with DMD, regardless of their eligibility for trials, as they have a wish for something medically beneficial to become available for their son, and research will shed light on these possibilities.

**Role of Genetic Counselors**

This study illustrates that parents of non-ambulatory boys do feel excluded from clinical trials, but many rationalize these feelings with logic and science. Knowing their child is ineligible for many potentially beneficial research trials often makes parents frustrated and sad, and their justification of their exclusion potentially represents a coping mechanism to manage feeling left out of research opportunities. These parents cited a general lack of enthusiasm regarding ongoing research studies, and also referred to the feeling that not much was going on in the field of DMD research, which is currently booming with opportunity. This perspective may be a reflection of the level of information they are receiving from their medical staff at this
late stage in their son’s condition. In the past, parents reported receiving the majority of their news regarding research from their medical team, but that this practice has slowed or stopped as their son has gotten older and his physical abilities have declined (Figure 1). Their son’s medical team used to be the source of information as well as excitement for research, so it stands to reason that their enthusiasm and interest in research would wane as their physician’s discussions of research have decreased. If their doctors or genetic counselors are not discussing research updates or opportunities with them, these parents may feel more discouraged and inadvertently believe nothing is happening in the field of DMD research.

Clinical research in the field of DMD is expanding exponentially as of late, with more opportunities being available to non-ambulatory boys than previously. Physicians, by training, are focused on their patient’s physical well-being, especially older boys with DMD, with their complicated medical needs at this stage of their life. Due to their focus on medical care, many physicians let discussions about research take a backseat when boys with DMD are older. This highlights a role for genetic counselors to be the providers that continue this discussion of research with these older boys and their families so that they are aware of what is going on in research, which is a precursor step to any new treatments that may become available. While these discussions can be difficult to initiate as older boys with DMD currently may be ineligible, it is still important to continue the dialogue, and genetic counselors are specifically trained to be able to discuss difficult topics. This ability will be very useful, as it will be essential to tailor any discussions of research to each individual family’s goals and desires, so customizing these conversations for each family is critical. Some families may wish to focus on other aspects of their son’s care and may not have an interest in research, but every family deserves the opportunity to discuss and learn more about research no matter their age or their level of
eligibility. This will help foster the inner hope these families evidently have, but also keep them grounded and not give any false hope. As genetic counselors, being open and honest about research is crucial, and these practices will allow genetic counselors to provide critical care for these young men and their families.
**Conclusion**

We interviewed seven parents of non-ambulatory boys with Duchenne muscular dystrophy to assess the impact of exclusion from clinical trials on these parents’ hopefulness, expectations, and interest in clinical research for their sons. Through these interviews, three major themes emerged. A first theme that emerged was that parents had a more active interest in research the past. Parents named various factors contributing to their level of research interest, including risk of side effects, their perception of a lack of progress in research, and a shift in priorities. Parents’ focus appears to shift from concentrating on the physical implications of their son’s condition to primarily on his internal well-being and development. The second theme that emerged was the perception of ambulation as a barrier to research participation. Parents state that it is a barrier that they understand, citing their scientific understanding with how research trials for DMD are conducted, but many parents report they are frustrated by this barrier and their exclusion. A final theme that emerged from this study was these parents’ maintenance of hope for their son, despite their resignation to their son’s condition and their reluctant acceptance of his ineligibility for trials. One parent of a younger son hoped to keep their son optimistic for future therapies, which was a striking difference from the perspectives of parents of older, more severely affected sons.

Overall, these parents feel left behind and frustrated by research due to their son’s ineligibility, and they rationalize their loss of access to research opportunities by exercising their cognitive and scientific understanding of the limitations of research. This rationalization and
their distinction between hope and realistic expectations represent coping mechanisms used to process this feeling of exclusion and adequate adaptation to their son’s deteriorating condition. These parents’ maintenance of hope highlights an area of improvement for medical professionals, where providers such as genetic counselors should continue to discuss research advancements with all families of boys with DMD, despite their loss of walking ability, as research opportunities in DMD are expanding more than ever before. It is important to balance these parents’ dreams of treatments or a cure to avoiding giving any false hope to families, and to customize these conversations for every family’s individual needs and goals for their son. While it can be difficult to bring up exciting research that may be out of reach of some individuals, these conversations are important to make available nonetheless, as every individual deserves the opportunity to learn more about research and potential advances in therapies.
Limitations

Limitations to this study included small sample size and lack of diversity among participants, as six participants were female and one was male. In addition, another limitation is in lack of diversity in ages of participants’ sons, as one parent of a child under the age of 15 was included in this study and had markedly different answers than the six participants with older sons who are more significantly affected. Furthermore, the participants were recruited from one institution and had participated in a research study previously, perhaps adding bias to the interest in research.

Future directions

Stemming from these limitations, a potential direction for future research would be to compare perspectives of parents of younger, non-ambulatory boys with DMD to parents of older, non-ambulatory sons with DMD, to evaluate their level of optimism, realistic expectations, hope for the future. In addition, evaluating potential gender differences in these perspectives would be an interesting avenue to pursue. Lastly, discussing mutation status as a barrier to research could also provide insight into how their son’s specific genetic alteration impacts their perspective on their son’s condition as many newly emerging therapeutic trials are focused on specific types of genetic alterations.
References


dystrophy. *Cellular and Molecular Life Sciences: CMLS* 70, 4585-4602.


Appendix A: Recruitment Letter

Family of Patient 1
123 Main Street
Anytown, US 90210

Dear Family of Patient 1,

My name is Lauren Murray and I am a graduate student of Genetic Counseling at Brandeis University. I am collaborating with the Harvard Neuromuscular Disease Project in the Program of Genetics & Genomics at Boston Children’s Hospital for my thesis project. As a participant in the Neuromuscular Disease Project, you gave the researchers permission to contact you for future studies. The team is now contacting you to determine if you would be interested in participating in my research study, entitled “Left Out in the Cold: Barriers to Clinical Trial Participation and the Impact on Parents of Sons with Duchenne Muscular Dystrophy”.

The goal of my thesis project is to discuss feeling and attitudes of parents when their son with Duchenne Muscular Dystrophy, has lost his ability to walk, and thus does not meet criteria to participate in novel therapeutic clinical trials currently recruiting participants. We aim to better understand how this exclusion impacts their hopefulness, expectations, further interest in clinical research, and discussions of the future with their son. This information will help genetic counselors and other medical professionals better understand how to help families cope with difficult medical challenges, experiences and situations.

Participation is voluntary and open to parent/guardian(s) who:
- are 18 years of age or older
- are fluent in English
- have a son with Duchenne Muscular Dystrophy who has lost his ability to walk
- are interested in clinical trials for their son

Participation will consist of a phone interview, which will be audiotaped and is expected to last about 30-45 minutes. Participants will receive a $25 Amazon.com gift card to thank them for their time and insight adding to this project.

We would like to contact you regarding this project, but do not want to inconvenience you if you are not interested. If you do not want to be contacted, please return the enclosed “opt-out” card in the prepaid envelope, indicating you do not want to be contacted for this study. Allowing us to contact you does not mean you have to participate, but only that you would like to hear more information about the study.

Please feel free to contact us with any questions regarding this study. Thank you for your time.

Sincerely,

Elicia Estrella, MS, CGC, LGC
Harvard Neuromuscular Disease Project
Program in Genomics
Boston Children’s Hospital
Elicia.Estrella@childrens.harvard.edu

Lauren Murray, BS
Genetic Counseling Graduate Student
Brandeis University
Waltham, MA
lmurray1@brandeis.edu
Appendix B: Informed Consent

BRANDEIS UNIVERSITY
DEPARTMENT OF BIOLOGY
GENETIC COUNSELING GRADUATE PROGRAM

Informed Consent to Participate in Research

Left Out in the Cold: Barriers to Clinical Trial Participation and the Impact on Parents of Sons with Duchenne Muscular Dystrophy

Principal Investigator: Elicia Estrella, MS, CGC, LGC
Student Researcher: Lauren Murray, BS

INTRODUCTION
Lauren Murray is a graduate student at Brandeis University pursuing a Master’s in Genetic Counseling. Elicia Estrella is the study coordinator of the Harvard Neuromuscular Disease Project at Boston Children’s Hospital and a licensed genetic counselor. We are conducting a research study by interviewing parents of non-ambulatory boys with Duchenne Muscular Dystrophy (DMD) about research participation. Specifically, we are interviewing parents who are interested in clinical research for DMD, but their son’s loss of ambulation has become a barrier to many of these recent opportunities. We want to learn if this impacts their hopefulness, expectations, further interest in research, and discussions of the future with their child.

Taking part in this research study is completely your choice. You should not feel any pressure to participate. You can decide to stop taking part in this research study at any time for any reason.

Please read all of the following information carefully. Ask any questions that you have about this research study. Do not consent to this study unless you understand the information in it and have had your questions answered to your satisfaction.

If you decide to take part in this research study, you will be asked to give verbal consent at the time of the interview. This verbal consent will be audiotaped and transcribed for documentation. You should keep a copy of this consent form for your records. It has information, including important names and telephone numbers, to which you may wish to refer in the future.

PURPOSE OF STUDY
The purpose of this study is to investigate the experiences of parents of non-ambulatory boys with DMD who cannot participate in novel clinical trials due to their loss of walking ability. We want to learn if this barrier to research impacts you and your family emotionally, how it impacts your interest in future clinical trials, and how you communicate with your son regarding research. This information will help genetic counselors and other medical professionals understand these experiences from the parent’s perspective.

PROCEDURES TO BE FOLLOWED
The study will involve a phone interview that will last 30-45 minutes. We will ask you some questions about your family, your perceptions of the clinical research community, as well as
what you perceive to be your son’s perceptions. We will also ask you some questions regarding your experience with the DMD research community; particularly about clinical trial interest, how you learn about research opportunities, how you communicate with your son regarding research, how being non-ambulatory effects your efforts to join research, and how this barrier to clinical trial participation effects your expectations and hopes for the future. The interview will be audiotaped and transcribed for analysis. Audiotaping the interview is required for analysis, so if you do not wish to be audiotaped, we will unfortunately be unable to proceed with the interview. If you choose not to answer a particular question, please let us know and we will move on to the next question. Participation is voluntary.

RISKS
There is minimal risk to you as a participant. The only risk to you is if you become anxious or upset as a result of our discussion. If this should occur and you would like to contact the genetic counselor on-call through Boston Children’s Hospital for support, their contact information is included at the end of this document.

BENEFITS
There will be no direct benefit to you for your participation in this study. We hope that in the future information obtained from this study will help us gain a better understanding of the impact of barriers to pediatric clinical trial participation from the parent’s perspective. This insight will help genetic counselors and medical staff be prepared to help families in similar circumstances in the future.

ALTERNATIVES
An alternative is to not participate in this research study.

PRIVACY AND CONFIDENTIALITY
Each study participant will be assigned a unique identifier, void of any personal information. The only information linking the identifier and the participant will be stored temporarily on an encrypted laptop and in written form in a secure filing cabinet within the Brandeis University Genetic Counseling Program offices. All study information will be destroyed once interviews are finished and evaluated.

PAYMENT
You will receive a $25 gift certificate to Amazon.com for participation in the research study as a gesture of appreciation for your time and expertise.

COST
There will be no monetary cost to you to participate in the study.

WHOM TO CONTACT
If you encounter any problems related to study participation or have questions about the study, you may contact the Student Researcher, Lauren Murray, at xxxxxxxxx@xxxx.

You may also contact the Principal Investigator for this project, Elicia Estrella, MS, CGC, LGC, at xxxxxxxxx@xxxx.
For additional support throughout the study, you may contact the on-call genetic counselor from Boston Children’s Hospital Genetics Department at xxx-xxx-xxxx.

If you have questions about your rights as a research study subject, contact the Brandeis Committee for Protection of Human Subjects by email at irb@brandeis.edu, or by phone at 781-736-8133.
Appendix C: Interview Guide

Interview Guide

Thank you for agreeing to be interviewed today, I appreciate you taking time out of your schedule to speak with me. Your experiences and stories are valuable and will allow the medical and clinical research communities to become more aware of a parent’s perspective on barriers to clinical trial participation.

Participation in this study is voluntary. I will be recording the interview so that I can refer back to our conversation during the analysis portion of the study. I will keep all responses confidential, your name or identifying information will not be shared with anyone. This study will aim to uncover common themes shared from parents, rather than specific experiences of one person in particular.

I’m going to ask you questions regarding your son’s condition, your experiences with the research community, yours and your son’s opinions about clinical trials, and your hope for the future. I aim for this to be more of a conversation about your family’s story so that I can get a grasp of what your experiences have been like, especially pertaining to your family’s reaction to your son’s DMD diagnosis and your family’s experience with research.

I will now be turning on the audio recorder so I don’t miss anything you say. [Switch on recording]. Did you have a chance to look over the informed consent form? Do you have any questions about this form or this project? Do you voluntarily agree to participate in this study? Thank you very much, now we can begin, when you’re ready.

Demographic information

- Background on participant
  - Mother/father/guardian?
  - How old are you?
  - What is your occupation?
  - Race?
  - Marital status: Married, divorced, single?
  - Are you the main caregiver for your affected son?

- Background on their son’s condition
  - How old is your son?
  - How old was your son when he was diagnosed with DMD?
  - How old was he when he began using a wheelchair full-time?
  - Psychological function: Ever been diagnosed learning difficulties or autism?
  - Schooling/Education level
    - Mainstream school, IEPs, special education, high school, college?
  - What is his level of ability (ADL)?
    - Dressing, eating, bathing, showering with assistance?
    - Does he require a ventilator?
    - Any other medical complications?
      - Surgeries? Cardiac/respiratory difficulties?
  - Do you have other children:
    - With DMD?
Experience with Clinical research

- Tell me about your interest in clinical research.
  - Always had an interest? Newly interested?
  - Participated before?
- What does clinical trial participation mean to you and your family?
  - Important/unimportant/neutral?
- What motivates you to look for research for your son?
  - Probes: Cure for him? Other families? Science in general?

Parent’s information gathering

- Tell me about how you get your information about DMD in general.
  - Probes: Medical staff? DMD foundations (PPMD, MDA?) Internet (i.e. clinicaltrials.gov)? Social media?
- Tell me about how you get your information about clinical research.
  - Probes: Medical staff? DMD foundations (PPMD, MDA?) Internet? Social media?
- Tell me about how your son’s medical team communicates with you about clinical research.
  - Do you bring up research? Does his medical team tell you about specific trials?
  - Do they give you ample opportunity to ask questions and explore research options?
  - Who from his medical team talks to you about clinical research?
    - Physicians?
    - Nurses?
    - Genetic Counselors?

Inclusion/Exclusion criteria

- Do you know what inclusion/exclusion criteria mean in research trials?
- What is your understanding of the inclusion criteria for participation in a trial of a novel therapeutic drug for DMD? (i.e. Eteplirsen, Ataluren)
- How do you feel about the 6MWT requirement for participation in novel therapeutic clinical trials as a measure of study success?
  - Probes: Criteria fair/unfair?
- Do you feel ambulation status is a barrier to allow you to participate in research?
- If yes: How does this make you/your son feel?
  - Probes: Worry? Stress? Neutral?

Communication with their son

- Can you tell me about how you talk to your son about different stages in his condition?
  - Loss of ambulation?
  - Surgeries?
  - Future: college? Living independently? Ventilation?
- Do you discuss research with your son?
  - Who tends to bring it up, you or your son?
  - Do you discuss barriers to participation? Can you tell me about those conversations?
- How does your son view participation in clinical research?
  - Interested/Uninterested? Always been interested/uninterested?
• Have his views changed over time? How?
  • How do your son’s views on clinical trial participation make you feel?
    o Probes: Sympathetic to his views? Upset by discordance?

**Hope for the future**

• Tell me about your hopes for the future for:
  o Your son?
    ▪ Probe: Hopeful? Discouraged by these barriers?
  o DMD community as a whole?
    ▪ Probe: Other families in similar situation? Federal funding for research?
• Looking to the future, do you see clinical research fitting into your plans? How?
  o Probe: Continue looking for research? Not anymore?
• Looking to the future, do you see the DMD community events/support networks fitting into your future? How?
  o Probe: Continue participating? Not anymore?

**Finishing up**

• Is there anything else you’d like to add for genetic counselors to know?
• Is there anything else you’d like to add for families in a similar situation to know?
• Could I contact you by email or telephone if any questions come up when I review our conversation?
• Would you like to know the results of our study?
• Thank you for participating!