Conflict of Interest Disclosures

In relation to this presentation, I declare that there are no conflicts of interest.

Background

- Duchenne Muscular Dystrophy (DMD)
  - XL progressive neuromuscular condition
  - Loss of ambulation (12-13yo), shortened lifespan
- Novel therapeutic clinical trials
  - Mutation-specific
  - Eteplirsen, Ataluren
  - Initial results promising
- 6 minute walk test
  - Reproducible
  - Requires ambulation
  - Exclusion
Background

- Parents of boys with DMD
  - Increased stress
  - Hope for cure
  - Shift in priorities with son’s age
- Medical trajectory potentially changing with trials
- Inability to participate in research
- Assess impact of ineligibility on parents of non-ambulatory boys with DMD


Methods

- Qualitative interview study
- IRB Approval
- Criteria:
  - 18 years or older
  - Have son with DMD who is non-ambulatory
  - Interest in clinical research
  - Fluent in English
- 7 participants — Telephone interview
  - Participants: 6 mothers, 1 father
  - Sons: 17-25yo; 13yo
  - Audiotaped, transcribed interviews
  - Coded for common themes (Atlas.ti v7.0)

Interviews

- Demographic information
- Experience and perception of clinical trials
- Parent’s information gathering: trials/results
- Exclusion criteria
  - Understanding of criteria? Impact of criteria?
- Hope for the future
  - For research? For son?
Results - More research interest in past

- (6) Hesitation to participate
  - Side effects, travel time, multiple visits
- (4) Lack of research progress
  - Med team not encouraging, lack of progress
  - When push comes to shove, they’re not getting FDA approval, they’re not moving forward, so our doctors are not necessarily saying, “You need to jump on the bandwagon” because they’re just not moving forward... 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.

- Other priorities, focus on now
  - We focus on making life as good as it can be... without really dwelling on the future. We’re trying to dwell on now.

Results - Perception of exclusion criteria

- (5) Familiar with ambulation as exclusion criterion
  - (4) Age
  - Understand criteria, reluctantly accept
  - Frustrated, sad with ineligibility
  - Well, you know obviously it’s frustrating when your kids are not able to be included... it makes me sad, because it makes you face reality that your son has progressed beyond the medical community being able to improve their physical ability. So, we came face to face with that reality, but we came face to face with that reality every day as we watch our boys lose function.

- (4) Barrier to research, but they understand
  - A lot of studies require being ambulatory, so it is a barrier. But, I didn’t know if it’s an unnecessary barrier. I think it does make sense that it’s a lot easier to measure results in terms of how many steps someone can take... Once they can’t walk, I can imagine it’s much more difficult to do concrete measurements.

Results - Resignation

- (4) Cannot reverse muscle loss
  - No realistic expectation for walking, cure
  - 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 [fix] 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...

- (3) No future therapy for son
  - 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]... it’s just, at this point, every day is an extra day you’ve been given.

- Future plans for research
  - Potentially be open
  - Not invasive
  - Not “holding their breath”
  - Unlikely to include him
Results - Hope

- Cure
  - Intended as obvious, understood response
  - Well, obviously we would all like a cure. Nobody wants their son to have an incurable disease.

- (5) Living full life
  - 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.

- (6) - Stop progression of disease
  - Most >17 yo, significantly affected, hand mobility
  - 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.

Discussion - Coping with exclusion

- Parents feel excluded from research
  - Coping mechanisms
  - Barriers to research
    - Rationalize barrier
    - Other ways to measure - Impact on hope?

- Shifted priorities
  - More involved care, time-consuming
  - Only opportunity for updates?

- Hope
  - Emotional growth - healthy coping
  - Cure, stop progression
  - Keep families informed

Discussion - Role of GCs

- Lack of enthusiasm about research by medical team
  - "Not much going on in research"
  - Research expanding
  - Reflection of medical staff’s perspective?
  - Advanced disease - physicians’ focus on medical care
  - Research discussions decrease
  - Role for GCs
    - Continue discussion with all families
    - Precursor step to therapies?
  - Tailor conversation, balance
  - Keep families connected to DMD community
Acknowledgements

- NSGC
- Thesis Committee
  - Elicia Estrella, MS, CGC
  - Holly Peay, MS, CGC
  - Brian Denger
- Study participants
  - Leslie Mandel, PhD
- Brandeis GC Program
  - Judith Tsipis, PhD
  - Gretchen Schneider, MS, CGC
  - Beth Rosen Sheidley, MS, CGC
  - Missy Goldberg

References


References
