

# The patient's view on why Respiratory Health in Duchenne Muscular Dystrophy (DMD) matters

## Key learnings from a qualitative study led by Santhera Pharmaceuticals.

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### 01 Introduction

#### What is Duchenne Muscular Dystrophy (DMD)?

DMD is a genetic, degenerative disease that is inherited in an X-linked recessive mode with an incidence of up to **1 in 3,500** live born males worldwide. <sup>(1) to (4)</sup> DMD causes a progressive loss of muscle strength since boys diagnosed with DMD have a lack of a protein called dystrophin. Dystrophin protects muscle fibers from breaking down. Usually the leg muscles are affected first, resulting in children losing the ability to walk by the age of **7-13 years**. As boys grow older, the heart and respiratory muscles are affected<sup>(2)</sup>.

#### Why is respiratory health important in DMD?

Duchenne affects muscles — including those important for breathing. Lung muscles will get weaker as the patient grows older. Patients with DMD are at high risk of respiratory complications as their clinical condition deteriorates due to progressive loss of respiratory muscle strength. Indeed, respiratory complications are a major cause of morbidity and mortality in DMD. <sup>(4) and (6)</sup>

#### Which respiratory symptoms do DMD boys experience?

**a)** Inability to cough properly; **b)** Fatigue and/or problems concentrating; **c)** Frequent chest infections; **d)** Morning headaches.

If a DMD patient experience any of these warning signs of respiratory dysfunction, a pulmonologist should be notified.

#### What was the purpose of this project?

Although there is currently no cure for Duchenne, high quality care can significantly improve both quality of life and life expectancy of patients. This project aims:

- To provide a deeper understanding about the impacts of respiratory problems associated with DMD from patients' own perspective, as well as from the perspective of their carers;
- To understand the strategies used by patients and carers to prevent and overcome the negative impacts of RFD.

### 02 Materials and Methods

#### Qualitative interviews, six participants from two countries, (3 from the UK and 3 from Italy).

- All participants had direct experience with DMD as patients (n=5) or as carers (n=1);
- The ages of the participating patients was between 15-40 years.

#### Methods - Qualitative research:

1) A patient interview guide was designed;

Identification of Interview Concepts	Topics for Exploration
Symptoms and impacts	<ul style="list-style-type: none"><li>• Symptoms experienced.</li><li>• Impacts of reported symptoms.</li><li>• Most bothersome/important symptoms.</li></ul>
Strategies to address the challenges associated with Respiratory Function Decline (RFD)	<ul style="list-style-type: none"><li>• Strategies to minimise the impact of respiratory problems.</li></ul>

2) The data were collected;

3) Questionnaire data were analysed using descriptive statistical presentations, with the assistance of the software Survey Monkey.

### 03 Results: Impacts of DMD on people living with the condition

#### Respiratory problems are one of the most salient symptoms experienced by participants in this study.

“Right now my personal experiences that I've found hard is to turn in bed when I sleep at night, to have a shower for which I need of course some help, and to cough, which is virtually impossible. I have recently had quite a remarkable cough attack, which I overcame thanks to the respiratory machine. Interviewee | 5”

### 04 Results: Impacts of respiratory problems on people with DMD

#### Experiences with respiratory problems can be so intense that, when they are not well managed, they can cause anxiety throughout the life of patients with DMD. The need for respiratory assist devices were as well identified during interviews.

“I was waking up a lot more during the middle of the night. (...) I was being monitored for my breathing and I got referred to a clinic. (...) They did a sleep study as well and found that the CO2 levels were increasing so they gave me a ventilator to try. (...) I was quite aware of what to watch for and I had read the care guidelines for Duchenne, I was seeing a specialist (...). So I think I felt quite confident about what to look out for and I think I started ventilation before I was really getting any problems from it. Interviewee | 1”

#### Negative consequences of respiratory problems for DMD patients:

Respiratory function decline may lead to people with DMD becoming unable to pursue their life plans and goals. As one participant reported, respiratory disease can be so acute that it forces people to withdraw from social and educational activities and can even lead to a situation of social isolation.

“I've heard of several people that when they were younger they weren't on ventilation and they ended up getting quite ill. (...) Then they ended up in the hospital and they got the ventilator (...) and recovered but haven't always gone back to the university or college. So, it's around (...) the early twenties when this seems to happen, [and it] interrupts people's lives and experiences at university and college and they have ended up staying at home. Interviewee | 1”

“(...) But I've been quite lucky in the sense that I've been relatively healthy. I've really just gone on to non-invasive ventilation. (...) But obviously that could change when I get a chest infection. That's probably one of my biggest fears, because I've had some really bad experiences. (...) The longest I'm alone is 5 minutes (...), but I don't like being left because of the times that I've been bad. (...) I'm not anxious all the time but I do get anxious about it. Interviewee | 2”

### 05 Results: Impact of DMD on carers

#### Providing care to people living with DMD puts a considerable demand on carers' time and energy and it can become a full-time activity. This can impact both carers, who experience exhaustion as well as patients, who generate feelings of guilt.

“We have family members that live 24 hours at home because they need to be there with their sons. Interviewee | 4”

“I have someone who comes in three times a week. (...) And on the other days I can manage pretty well. But increasingly I'm thinking that I need someone with more time and with flexibility, [to] not having to rely on my family all the time, which can be quite burdensome. Interviewee | 6”

### 05 Results: Impact of DMD on carers

#### Negative consequences of respiratory problems for carers:

Respiratory problems can cause a lot of distress to carers especially when they need to manage emergency situations for which they feel unprepared. These events can be so intense that they generate feelings of panic, which can hinder the ability to provide assistance.

“(...) At the time we had never faced any sort of emergencies (...). It had all been like kind of a slow decline. (...) And she [caregiver] really panicked, and she was just kind of screaming very loud. Interviewee | 2”

### 06 Results: Strategies to address the challenges associated with respiratory function decline

#### Acknowledging the need to be proactive:

“Prevention is not everywhere. (...) Families don't realise the importance of prevention, (...) that they need to use the ventilator every night, even if the boys are well. Interviewee | 4”

#### Acting to prevent respiratory problems:

“I'm engaged in the respiratory gymnastics, so to speak. (...) Actually every day I do twelve exercises with the respiratory machine, in series of five. (...) I do this as a precaution, to be prepared for future problems. Interviewee | 5”

“The only problem (...) is when you have to cough. And it's bad because I can't breathe on my own, so I need like a push on the diaphragm [abdominal thrust movement]. Interviewee | 3”

#### Providing information about how to obtain respiratory devices.

Respiratory problems can cause a lot of distress to carers especially when they need to manage emergency situations for which they feel unprepared. These events can be so intense that they generate feelings of panic, which can hinder the ability to provide assistance.

“We suggest to other families to be very persistent when they ask for the cough assist [machines], because (...) every single director decides if they want to give you the cough machine or not. (...) We suggest to them to ask the government official to sign a paper where it is written that, if he says “no”, he will be responsible for every problem. Interviewee | 4”

### 07 Resources

#### To learn about respiratory symptoms, management & care

##### Organisations around the world play a key role in providing information to people affected by DMD.

Patient organisations' preferred tools and materials to provide information about respiratory function decline include social media, scientific conferences, newsletters, group meetings, online forums and websites. Other resources are:

##### Duchenne Pulmonary Awareness Video Series at:

[www.parentprojectmd.org/duchenne-pulmonary-awareness-video-series/](http://www.parentprojectmd.org/duchenne-pulmonary-awareness-video-series/)

**DMD Clinical Trial Updates** — Clinicaltrials.gov is a good and trustworthy tool to look at ongoing studies in DMD.

### 08 Conclusions

Respiratory problems are one of the most important symptoms in DMD.

### 09 References

(1) Mendell JR & Lloyd-Puryear M. Muscle Nerve 2013; 48:21–26. (2) Passamano L, et al. Acta Myol 2012; 31:121–125. (3) Bushby K, et al. Lancet Neurol 2010; 9:77–93. (4) Timpani CA, et al. Med Hypotheses 2015 ; 85:1021–1033. (5) <https://takeabreathdmd.com> accessed 20 February 2020. (6) S. Khirani, A. Ramirez, G. Aubertin, et al. Respiratory muscle decline in Duchenne muscular dystrophy Pediatr Pulmonol, 2014; 49; 473-481.

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