

Appendix

DMD-adult and Parent quotes on which the analysis / conclusions are based.

■ **Quote 1 (referenced in manuscript but not quoted)**

DMD-adult: '...Yes well, I think I would try to get more information regardless (even if) it is morbid. I ... like to take responsibility for my body. It is my body. What happens? What is possible? What worries me is realising that something could happen ... I would rather know about something than thinking at the time, 'Oh, I should have thought about that earlier'. I like to be proactive and this is my (being) proactive...'

■ **Quote 2 (quoted in manuscript)**

DMD-adult: '...To me it is important to know all the facts about all the treatments, because you need to know what to do to try and live longer. Because you need to have the right treatment at the right time, because it can be poorer treatment if you get it wrong...'

■ **Quote 3 (referenced in manuscript but not quoted)**

DMD-adult: '... From my experience, cause obviously because I am in a different area than some of you, I think a lot of the issue seems to be that a lot of the NHS treatment is a postcode lottery for what sort of treatment you get. Obviously, the heart is important, so breathing seems to be the biggest difficulty for people with our condition. It is keeping the lungs clear, because that is the most vital thing, obviously apart from eating to survive. I think, that is probably part of it, because different cardiologists offer different treatments. There is no standard treatment, but then the issue is there are no full studies which prove conclusively – whatever... that is the difficulty there. But personally, I would rather that some treatment was done in the earliest instance. There is a risk of your heart might stop anyway, I much rather have

the risk of being shaken quite badly [by an ICD shock therapy restarting my heart] than obviously it not starting at all. I mean – that is just my perspective on there ...’.

■ **Quote 4 (excerpts referenced in manuscript)**

DMD-adult: ‘...It is hard obviously. You know there are different people and different family circumstances as well. My experience has been that my mother was quite honest with me, but still when you are very young, I don’t think it would help to know about the heart as well. When you ... are dealing with the whole idea of not being able to walk, for a start. So, there is a time and a place. I think honesty is important but ... it is difficult, isn’t it ... because obviously when parents are told as well, this idea is quite difficult ... this idea that you are not going to survive past a teenager. Obviously, [it needs to be] acknowledged that this might unfortunately be the case of some people, but they also might live long past that. That’s what I found most ... in terms of what you don’t want to know - you don’t want a ‘sell-by’ date given. The whole idea ‘you will not survive past that date’, because I find that more depressing than anything else. I’d rather people were more honest and [treatments] invasive ... not necessarily invasive, but if active treatment would begin earlier, if they thought it would make a difference ...’.

■ **Quote 5 (referenced in manuscript but not quoted)**

Parent: ‘...He gets to live longer. If you reach that point where you are having that discussion, then you are having that discussion, because you know it’s on the cards that your heart could be the thing that could end your life. And that maybe, you know, you can have a bit longer, that you would have a choice. I mean, the heart beats no one knows how long, because ultimately, you are already doing it, you recognize it: you are on a slippery slope... and there are big unknowns...’.

Parent: 'I think it would be reassurance that the heart would keep beating and if there was ... it would kick in... and it would give reassurance that it is going to sort the heart out. Obviously, there will come a time when it's going to stop working and everything will shut down, we know that.'

Parent: 'I think it gives you hope'.

Parent: 'Yes, a bit longer, bit more quality. It would be reassurance, I think'.

Parent: 'You know that you exhausted everything...!'

■ **Quote 6 (quoted in manuscript)**

DMD-adult: '...I think for me, it is not straight forward, because obviously you got the risk of the procedure, but you also got the risk of not having the procedure, and then, you got the risk of what happens if you get to a certain point when you can't actually have the surgery, and that is another risk ...'.

■ **Quote 7 (referenced in manuscript but not quoted)**

Parent: '...It is always risk and benefit, it is always the balance, isn't it? So, what I want, and I think what [my son] wants – because we are both pretty logical when we think about things – is, you'd want that written down: "Right, we can do this, these are the risks, these are the complications because of Duchenne, this is what..." ... because, I don't think that they'll be able to be say, "this is the percentage chance it is going work." I think it is going to be a bit, "this is what we hope, this is our experience with it, this is what happens in other conditions, is it worth of you going through all of this? But a potential benefit, it might give you another year, might give you another five years, you don't know" ... and then they have to decide, because that is all they can do. You will not have this black and white... it will never be that black and white..'

■ **Quote 8 (referenced in manuscript but not quoted)**

DMD-adult: "...Yes, I mean you just have to be advised – I mean any surgery – about the different risks that you'd been under. It would also be a personal choice to each individual whether they think the risks are worth it. As [DMD adult] was saying, you got a risk of infection, risk of being under general anaesthetic, you know. I suppose, but they are risks that everyone is exposed to, again, with our condition it is just a greater risk. So, you got to weigh up, what the best time is. But from a psychological angle – obviously we are in (this discussion group) about this – personally when it comes down to it, it is not very nice having a condition like ours, but I think once you had it for a little bit and get used to the consequences ... obviously it is scary, when you hear about the risks of different things but it is much better to be aware of the different treatments available; because there is no point in being scared and in the dark, when you might be scared informed ...".

■ **Quote 9 (excerpts quoted in manuscript)**

DMD-adult: '...Yes, I think, quality of life is quite subjective. You can adapt quite well to whatever is thrown at you. In a way, I just want to prolong live as long as I can and then work out afterwards what my quality of life is...'.

DMD-adult: '..Yes, I think, I agree with that. That is a good point...'.

DMD-adult: '..I think also the medical profession may view our quality of life has been worse than what it is ...'.

DMD-adult: '...We come across non-feasible people. They'd say, 'I could never have your condition' and 'I'd rather kill myself than live with that'. But when you actually have it, I think, you see this complete differently...'.

DMD-adult: '..Sometimes even I look at people, thinking their lives are worse than ours sometimes...'.

■ **Quote 10 (referenced in manuscript but not quoted)**

Parent: ‘...No, it is different. I think by the time you are talking about that – we all got anxiety all the time – you learn to deal with it. It comes up and it goes down again, you manage it and every now and then it comes up and gets you in the throat...’.

■ **Quote 11 (excerpts quoted in manuscript)**

Parent: ‘...I think, I am in denial. You know, I am blocking it all. So, yes, I am like that, I am scared. I don’t want him to go... I am scared...’

Parent: ‘...No, no, no... you don’t know what happens in two weeks. I am in denial. I think that is a form of protection for me. I am quite happy like that to be fair...’

Parent: ‘...I think because, I don’t know, over the years of seeing how things can just change so quickly. He can become ill, he’d get seriously ill very quickly, you sort of just live for the day ... you don’t plan years ahead You just carry on the way you do. Get on with life every day and not sort of focus too much about everything that is going on. You just have to enjoy the time that you are together and...’

Parent: ‘... you are right...’.

Parent: ‘... Yes, and then you go to [a specialist] and they say, “your boy is living until they are 50.” And you are like, “shit... that is really good news, but... shit, because I’ll be 80...”.

Parent: ‘...Yes, that is my thinking, too, if [my son] keeps going, I’ll be very old...’.

Parent: ‘...And the game plan changes, because suddenly you’re like now, I’ve got a good few years. Whereas my brother died at 21 and [my son] is 25 and his life expectancy has been 28 or 30 if we make it... but now...’.

Parent: ‘... but now it is higher and now, I am thinking, selfishly, I do feel so pleased but our lives are on hold. I mean, I have got a new husband and we are married for three years and he wants to go out and go abroad and do that...’.

Parent: '[...] But as parents, we are just parents, we are... we are parents, nurses, carers, pharmacists, physiotherapists, psychologist. It's... everything is on us. We then go out to work and we try to have a family life and a social life, it is all on us...']

■ **Quote 12 (referenced in manuscript but not quoted)**

DMD-adult: '...Personally, I cannot envision the situation, where I get to the point that I want it to be turn it off. But having said that, I am saying that when I am not in pain or anything like that...']

DMD-adult: '...If my kind of life would be miserable and I was in pain all the time, I think, yes, I maybe would consider that. But the thing is, it would have to be pretty bad for me, and often doctors underestimate the quality of life, so I think, ... even if I am in a little bit of pain, if I am still getting enjoyment out of my life, then I would want to hold to life as long as possible...']

DMD adult: '...At the moment, I rather just concentrate on getting everything I can to prolong my life. [...']

■ **Quote 13 (referenced in manuscript but not quoted)**

Parent: '..It is about treating you like... you got to offer to people with cardiomyopathy, so why don't you offer it to them as well and let them decide. And it does make it hard, but there are so many hard, complicated decision..']

Parent: '..So many decisions throughout life that it is not going to make a difference .. one more...']

■ **Quote 14**

DMD-adult: '... I kind of suspected that, but no one's told me that before. I knew that the heart can still deteriorate. I did find often that specialists are quite vague about it. In fact, I did not really know until my last appointment how my heart was relative to a normal person's heart. They always say, "*you are doing really well*", but they mean,

“*you are doing really well relative to Duchenne*”. So, I did not even know from an objective position where my heart function is at. A lot of the time they just don't give you very much information on this ..'.

■ **Quote 15**

DMD-adult: '...Yes, what I wanted to say is generally - if you choose the right treatments than you can live longer ... if you get the right treatments at the right time before the deterioration of the condition. If you get it slightly wrong, then you got a big problem ... '.

■ **Quote 16**

DMD-adult: '...I think they should definitely be talking about it. I mean, it is just another guideline in treating Duchenne. That really is the role that specialists and consultants who are working out (?) how people can manage their own condition and empowering people with information. I don't think there is any reason they should not. ... Obvious, they'll be sensitive to each patient and when is the right time to talk about things. For me, it's always been a case of the more information the better... '.

■ **Quote 17**

Parent: 'Only if he'd [my son] wanted it. Because you have the conversation. I hope I'd be part of the conversation. I'd find it extremely difficult if... [...]'

Parent: '[My son] is like that. He just wants to make sure he does everything at the right time ... that's what he ... sort of realised ... throughout his life: there is a time to do everything; like you have time to do your spinal surgery before that window [closes]. Get on with it, [my son] is like that... '.

Parent: '..Same with [my son.].. '.

■ **Quote 18**

DMD-adult: ‘.. I am not really worried about having it or anything, because to me, I am already having a tracheostomy and how that is going to be done, (I’m worried) more, because I don’t want to be going through the same procedure every six years...’.

DMD patient: ‘...Yes, and if it helps. If it is going to have enough benefits - then fair enough. It is more the changing of it, like an operation every six years. If I need, it I will do it, but yes, that’s all...’.

DMD patient: ‘...Yes, after surgery I get really worn out, like sometimes even ill and stuck in hospital. It is a proper procedure every six years...’.

■ **Quote 19**

Parent: ‘...It is a difficult place though, because they are having to decide. Whereas if they did not have it, they did not have to decide. If it did happen, that’s it and they would let go and there would have not been a choice in that, it would just have happened...’.

Parent: ‘..It is hard..’.

Parent: ‘...Again, I forgot. I forgot that you can turn them off...’.

Parent: ‘...That he has got to make a massive decision, that for most of us we don’t have to think about it. We’ll go and that’ll be it...’.

Parent: ‘... eventually...’.

Parent: ‘... we actually rather not make that decision...’.

Parent: ‘... and we are forcing them... forcing them into... to make a decision... whereas the other turn is accepting that and that is a whole different thing, to actually having to say “I had enough.”.

■ **Quote 20**

Patient/Carer: ‘...I can see the advantages for the family...’.

DMD-adult: ‘...I think even though I don’t have a tracheostomy, the non-invasive intubation is still always there, like the wheelchair is always there as well. I don’t think I would be too bothered about being reminded of feeling it there, unless it is painful, that might be a different matter. Or if, I don’t know, I would regularly get a shock, maybe that would be more affecting my quality of life, but then probably my quality would be affected if I did not have it either...’.

DMD patient: ‘...Yes, but I think ... it would have to have quite a big impact on my quality of life. I don’t think I would be bothered by the more psychological aspects in terms of what I might or might not have to think about...’
