PODCAST 3:

How has Myotubular Trust funding helped?

The Myotubular Trust has been fantastic and the support has been wonderful. The research that we did was I think early stage and at first more on the high-risk-high-reward side. Having support through the initial phase of it really enabled us to get to the part we are doing now: we've been through the initial discovery part, and having the support of the Trust enabled us to get to that place, so now we are really drilling down to the specific targets that we could potentially bring to clinical trial. That wouldn't have been possible without the support from the Myotubular Trust.

I think also one of the things that I really value about the interaction is that the projects are evaluated based on scientific merit. Even if I don't get a grant (and I had applied previously with a different project that wasn't funded) it makes me feel like the best projects are being funded: and not because of who I am or who I know, but rather because the work that I'm doing has been reviewed by experts in the field and thought to be really the most meritorious. I think that's a really important strategy, because I know in a lot of other rare diseases there isn't that opportunity, and it becomes a bit of a challenge to make sure that the best work is moving forward. I value that part as well. It probably takes a lot of work for the Trust to have a Scientific Advisory Board and to have people have to review grants externally and it would probably be easier in some ways just to decide 'okay, I like this person and I'm going to give them funding' but I think the fact that the Trust has stepped back and done it in that way is really outstanding. I appreciate it as a scientist, even though it may mean that I might not get funding at a time in the future, that's something I won't take personally, it just means that I will need to work harder to have a more meritorius project in the future.

Is there anything else that we can be doing as a patient community to help work like yours?

In terms of the patient community - I think the MTM patient community is amazing! Very communicative with the researchers and doctors; very interactive; very willing to work together; seems very mobilised. Which is, I think, amazing on two fronts: one is - that in itself it is great but also given the challenges of the condition and everything it takes on a day-to-day basis to take care of a child with MTM, the fact you guys are doing what you are doing is amazing. To take that extra time to have jobs and also take care of a child who has extra needs, and then to also be involved in clinical trials; or thinking about clinical research; supporting a Trust; fundraising - I mean these are all amazing things. I think you are not alone - I would say in other families I've met, and other MTM support foundations that I have been privileged to work with, it's been incredible in terms of the spirit of the families and the support of friends and relatives. I guess I would say that - continue doing what the community is already doing. As trials come, obviously participation is key. And always asking questions, I would say.