

Report of my first Patient Advisory Board meeting in San Antonio, Texas, with Audentes

I was quite anxious about travelling to the USA on my lonesome and wasn't looking forward to the long flights. But all the worry was wasted, the flight went well. My anxiety about seating, leg room, airplane toilets was dealt with by giving me a whole row of seats to myself, and I bothered some kind-hearted passengers to assist me to get up from the awkward seating and lend me a shoulder to get to the toilet.

When I arrived in San Antonio I found that a two-week long festival was coming to its climax and there were going to be parades on the Friday and Saturday. I met up with Allan Muir and Allyson Lock and, as we had a few spare hours, we attended the festival together. I hired a scooter, without which I would not have even attempted to walk around. The atmosphere, the crowds, the floats and all the brass bands were fantastic and thoroughly entertaining.

It was my first visit to America and I was blown away with how accessible the whole city was. I could go into any shop, any restaurant, as all entrances had ramps into the property. It really put the UK's disabled accessibility system to shame.

The reason I was in the USA was to attend the Audentes Patient Advisory Board meeting, and after breakfast on the Saturday we started bright and early at 9am. The first part of the meeting involved gathering people's perspectives and opinions about Pompe and our outlook on the future of Pompe.

Afterwards a couple of the Audentes professionals did a small talk about Audentes and gene therapy. There wasn't a lot of new additional information, they said there would be more details released in the future.

I felt very honoured to represent the UK Pompe community and before I left for the USA, I had sent a large group of UK Pompe patients a message asking them for their own opinions, perspectives and if they had any questions they wanted to ask.

I will run through a few of the questions and answers below. Audentes weren't able to answer everyone's questions as they just don't know yet themselves.

1. Will gene therapy correct any damaged muscle?

I cannot give you a direct answer on this as they don't know. They are hoping that Gene therapy stops the progression of the disease, and they are still not totally sure whether we will be able to rebuild damaged muscle. They are also unsure whether it will be a one stop shop injection or whether there will need to be a top up once every 5-8 years. A lot of uncertainties are still hanging around and only once the trials have started will they be able to answer these questions.

2. Will they extend trials to the UK and Europe?

They are hoping to have trial centres in the UK. They have not been determined yet, so they are still not sure of number of centres. We will know this come Autumn time.

3. How long will the trial be and when will they start recruiting?

The length of the trial was not talked about. They are hoping to start recruiting for this trial in Autumn 2018.

4. I understand it's open to people on Myozyme to decide whether they continue to take it during the gene trials, will that be the case with new trial participants?

As far as I understood the trial will be run alongside people who are on ERT.

5. Can people who are CRIM and have reactions to ERT have this gene therapy?

There was some talk about how they are going to transport the new gene into the body, (excuse my laymen's way of explaining it), but I don't fully understand all the words and the in-depth scientific part of the treatment. But they did mention using a virus (AAV8?), and if we have had this strain of virus before then we may be ineligible for the trial. But they also said that they are working on method of suppressing the antibodies if this is the case. They estimated that around 20-30% of patients may have already had this virus. But again, it was all very uncertain, once they release the details of the trial criteria we will know more.

6. Are there any group of patients excluded for safety or effectiveness reasons?

This (and question 5) are things that will be better answered once they start recruiting for the trials.

In the later part of the meeting we discussed what was thought to be the most important outcomes of the trials. In our group, I made sure all the points that had been passed onto me were recorded, and if they weren't put on our board I made sure they were recorded on other groups boards. There has been some positive feedback from Audentes, so I thank you all for your messages, etc.

I didn't know what I was expecting beforehand, but it was an interesting experience, they took lots of information from us and they explored that information. When there are 12 patients and family members there are a lot of opinions to explore and record. But other patients and family members who had been to these kind of meetings before said that they thought Audentes were very good at exploring and asking the right kind of questions, and the best thing was that everyone got a chance to speak.

It was a privilege to represent the UK Pompe community and it is very exciting to see this happening and to be very close to reality for the Pompe patients.

There is supposed to be a press release sometime in May from Audnetes, I am sure it will be shared online once it is released. If there is any other news passed our way, I will share it around immediately.

Ben Parker

Pompe Support Team member