Pediatric RESEARCH

INSIGHTS

Family reflections: MPS II

Jane Andrews[1]

Pediatric Research (2019) 86:134–135; https://doi.org/10.1038/s41390-019-0374-z

Andre is a 25-year-old young adult who has an MPS disorder known as mucolipidosis II (I-cell disease). It is a rare inherited metabolic disorder characterized by coarse facial features, skeletal abnormalities and mental retardation. It is a severe lysosomal storage disorder caused by defects in the α and β subunits of the hexameric GlcNAc-1–Ptase complex essential for the formation of the mannose 6-phosphate targeting signal on lysosomal enzymes. It has also been considered to result from a deficiency of the enzyme *N*-acetylglucosamine-1-phosphotransferase, which phosphorylates target carbohydrate residues on *N*-linked glycoproteins. The symptoms of I-cell disease are similar to but more severe than those of Hurler syndrome, which is also known as MPS I.

As a result of this disorder, Andre is now restricted to a wheelchair. He was able to walk until he was about 3 years old, at which time his growth stopped and he developed severe contractures in his arms and legs. Over time he has gradually lost the strength in his hands and is unable to feed, bathe or dress himself, as well as unable to do most of the activities of daily living. He is able to use the toilet, but requires assistance being placed. He is unable to stand or sit alone outside of his wheelchair. He does attend college and is able to use a laptop and mouse, but requires accommodations and assistance for note taking, holding his textbook, and anything that has to do with writing. As a result, he requires someone to be with him at all times.

Secondary to his I-cell, Andre has progressive restrictive lung disease, hypoventilation, obstructive sleep apnea, and reactive airway disease. He also has significant hypertrophic cardiomyopathy and aortic insufficiency. In addition, Andre has skeletal dysplasia, stenosis of the craniocervical junction, bilateral hearing loss and corneal clouding. His condition also significantly limits his dexterity and range of motion of all major and small joints. His wrists have gradually lost range of motion and his hands have become progressively more stiff and fixed in a claw-like flexion. This condition has global effects on his health, especially impacting his heart and respiratory system, as well as the bones and joints. He sleeps and travels with a biPAP machine at night and doesn't leave home without it. It is a necessity when we travel so that he is able to sleep.

The only treatments Andre has received are Pamidronate infusions. This was just started in 2018, as before that time it was not a procedure covered by the insurance.

Naturally, the big issues that we would like for the researchers to help us with would be to find a cure for the disease. We realize that his diagnosis is irreversible, but any type of research that would look into providing him with a good quality of life would be of tremendous help.

The biggest issue facing children with MPS is the fact that their disease will eventually take their life. To that end, clinical trials to help with any of their diagnoses could extend that time and provide them some level of comfort. The children suffer terribly with pain and Pamidronate has its own share of side effects, so looking in the direction of some type of pain management could provide some relief. My son has a significant airway disorder that prevents him from having almost any type of surgery, and I believe looking into a way that these children can be sedated without risk of life or ending up with the use of a trach would be something positive to look forward to. His neurosurgeon believes he is going to need an extensive surgical procedure if his spine becomes unstable, but, because at this juncture that surgery could impact his quality of life, we feel that it is better to live for the moment than possibly be worse off than he is, unless it becomes a case of life or death.

I would like to say to researchers in this area to look not only at the MPS disorders but also at mucolipidosis. From discussions with geneticists at various times, it is the belief that since their disorder is so complex it would require some type of a cocktail to attempt to find a drug that could possibly slow down the progression of the disease. I often failed to understand why even though they are not directly MPS diseases, they do pattern themselves in the same way, and perhaps with safety they too could participate in clinical trials that could help them significantly increase their quality of life. I have waited for 25 years and it seems like there may not be a significant movement in this direction in the future, so I would like to reach out to the researchers and ask them to at least give our disease a try. I believe it would offer a challenging experience, and the end result would be rewarding not only for you but also for our children and young adults. It is heartbreaking to see so many of them die, two who were close to us over the past year and one who has recently gone into hospice. He wants to be as happy and healthy as he can, so any research that could help with any of the diagnoses that he has would be better than none.

It is my hope to make Andre a happy child and to allow him all the possibilities that his disability may limit him from. We were told that he would never see his 3rd birthday, and so to love him and take pictures because that would be all we are left with. To that end I have been able to purchase a handicap-accessible van, so that I can take him everywhere he needs to go. I contact local states when we travel so that he can have accommodations to get around to do the things he likes to do. It is my goal for him to see all 50 states and we are slowly working on that plan for him.

For the next 22 years I have gone to conferences and meetings and everywhere I could to see if there was any information on the horizon, any chance or possibility that someone was willing to take an interest in the children with this disease. I have stood up at meetings and asked why, and am being told that the disease is too complex, but what does that mean for these children? The reason I thought of doing research was to find the answers to some of the questions and not just let it be left on the side to

[1]Washington, DC, USA
Correspondence: Jane Andrews (info@pedres.org)

Received: 13 March 2019 Accepted: 13 March 2019
Published online: 20 March 2019

pursue those diseases that might be easier. I think they should all have equal opportunity to be looked at, and it is my hope that one day it will happen.

As a parent I have been blessed and fortunate to have my son to live as long as he has. He has overcome the odds, but no one knows what that means. No one has any answers as to what causes his longevity, but I will not complain. Despite his illness, he has finished school with honors, was valedictorian of his middle school and ranked 6th in his high school class. He has been bowling for at least 10 years and serves as a Director for the local bowling association. As mentioned earlier, he attends a local community college and is majoring Accounting. He wanted to live on campus, but accommodations for him would be almost impossible. He works diligently at church, in Sunday School and ushers.

He has goals and aspirations and I don't have all the answers. He wants to be a parent and I definitely don't know how that can possibly happen. He wants to drive, but it would take a special kind of car that hasn't been invented yet. He wants to be able to go out and enjoy activities on his own, but there are really no facilities available to him. He wants to work and take care of himself and contribute to the household, but would need someone by his side to help him with his daily needs. There needs to be a book on what parents need to know when it comes to children with disabilities, but I have worked hard to try to make sure that he gets the benefits he needs, despite the roadblocks that we encounter. I am willing to make whatever sacrifice I can to make sure that he is happy and can look forward to each day of his life for whatever time we have. Research could go a long way to make some dreams a reality.