

Interviewed by Shelley Bowen, Director of Family Services and Advocacy
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MEET JOHN

I love the photo of you holding the TAZ mutant mouse in your hands. Tell me a little about that moment.

At first, I thought what a cute little mouse. Then, I thought deeper about all the work that went into making this animal model with Barth syndrome and how important it is to helping scientists better understand Barth syndrome which may one day help myself and others who have Barth syndrome. It was a pretty special moment.



Tell me about John.

I have a sister. Growing up, I'm sure it was difficult for my sister to have a brother with special needs. I'm not sure. I didn't notice because I was the object of the attention so that worked for me. Seriously though, she's great. I enjoy spending time with my sister, her husband and my niece.

I enjoy football and basketball. I am a Nebraska Cornhusker fan. I enjoy going to the games, but I also enjoy listening to the games on the radio while I am tinkering about on my model trains. Sometimes I travel with my family to out of town games. I stay in the room and watch the game on the television and serve as the color commentator.

I fix computers and I volunteer with the Barth Syndrome Foundation. I like flying flight simulators on my computer. I am a model railroader. I have a layout in my basement. I have been doing that for as long as I can remember.

Computer repair comes naturally. My dad always had computers. My uncle taught me how to take them apart and put them back together. I like to figure out how things work. Mostly I do tech support and help people with their software program challenges. My clientele is typically over the age of fifty. I enjoy helping my clients. Many of them have my name and number taped to their monitor.

Tech Support requires a great deal of patience.

It does; I always try to be patient. Maybe my capacity to be patient comes from having rare genetic disorder. You have to be patient with that. It just kind of bleeds over into other things in life. I do admit, it's not always easy to be patient when I am tired or hungry. But I try my best.

What happens when you are tired or hungry?

No good comes of trying to do anything when I'm tired or hungry because I get cranky. When I hit the limit, I know I am done. I have to rest when my people skills are depleted. That's when it's time to go back into my little shell.

Tell me a bit about your blissful little shell.

I live by myself. I like my little shell. That is where I am most comfortable. I don't date and don't have a need to date or be in a relationship. I don't have a need for constant companionship whether it's with a person or a pet. I'm doing well to take care of myself. Relationships and people in general are tiring. It takes a lot of energy to be social.

What happens when you reach your threshold with fatigue?

What is most bothersome is my inability to concentrate or keep my eyes open when I am tired. Thinking makes me tired. For example, if I know I have something coming up I will feel more tired before and after. I can push myself, but I'll be tired, really tired afterwards. I had something I had to do last weekend. I felt really tired leading up to it and afterwards but during the hours when I had to be there, I was able to get through it. I can push myself, but I will crash when I get back home.

Do you find fatigue has affected you more that you are getting older?

Leaving the house has become more of a problem as I have gotten older. I worry about things. always worry I will forget something even though I don't ever forget anything because I worry so much about it ahead of time. Thinking and worrying is exhausting. I have found that it is helpful to take a photo about things that I will worry about later like "Did I close the garage door?" When those thoughts come, I can just pull out my phone and see it closed, which puts the worry to rest.

I do think the fatigue is getting worse. I don't have as much energy as I used to have. For some reason every time I have my pacemaker changed, I find I get more tired and never seem to get back to the same place, in terms of energy, as I was before the procedure.

What is it about model railroads that interests you and so many other kids who have Barth syndrome?

I think it's a curiosity about what makes things "go". Having a hobby in model railroad is accessible. You can do it in a compact space. You don't have to go outside. It involves a lot of different skills that I enjoy like electrical, carpentry, building, painting, planning and computer programming. Sometimes it does just go in a circle but a lot of it is being creative and realistic. It's fun to figure out how to get them from one point to the other which is what trains do to make money and transport goods. You can be creative and hide fun little things like superman among other things to make it unique.

Tell me about your interest in aviation.

I think it falls in line with the railroad curiosity about big things that "go". Flying is fun. The mechanics of flying is fun. The fact that it takes you somewhere else is fun which is usually to see some nice people or go someplace interesting. I've never flown outside of being a passenger, but it seems interesting and fun.

I've noticed that you and your compadres seem to have a natural talent in mechanics with a common interest in figuring out how things work. Would you agree?

I liked Legos when I was little and still do. I liked building blocks when I was little and the Brio Trains with the little magnets. I think dexterity is a skill that we share. I think putting hands on things and putting it together to figure out how things work is something I enjoy. I also think having my hands on something helps me figure out how things work.

Do you find your natural curiosity and talent in figuring out how things "go" has helped you to better understand how Barth syndrome affects your body?

No. Well maybe a little bit but I find biochemistry and the physiology of how the body works much more complicated. It's harder to understand than mechanics which is interesting because as you say it comes down to understanding how things work. I have a harder time grasping life-sciences. Maybe because with mechanical stuff you can experiment with it. You can get your hands on it and play around, if you break a part you can go out and get a new one. You can't go rummaging around in someone's mitochondria and risk breaking it.

Maybe we should tell the doctors to describe Barth syndrome in Tinker Toy terms during the conference.

That would be helpful.