

October 26, 2023

Senate Aging Committee Hearing

My name is Maureen. I'm 50 years of age and I live in Shelton, Pennsylvania.

I have lived with Galactosemia for my entire life.

Shortly after my birth in 1972, I began to show the classic hallmark signs of failure to thrive. Jaundice, repeated vomiting and weight loss. Seven days later, I was diagnosed as having classic galactosemia upon release from the hospital.

My mom and dad were given a 32 page booklet entitled A Parents Guide to a Galactose Restricted Diet. They were instructed to follow this booklet as I would need to be on a lactose free, dairy free diet for the rest of my life at that time. The only treatment for galactosemia. I doubt my parents knew that many more challenges would lie ahead in the following years to come then just for having me follow a dairy free diet.

It seemed like everything was working fine for me at the time, but I was getting older and it made me feel very left out when I could not eat the same way as my peers. Without the modern advances we have in nutrition today, I cannot tell you how isolating this was for me. There were many social events that I could not go to. My mom tried so hard, always sending me to school with a special treat – not just enough for me but for everyone to share. As a child this was really difficult.

I also could not process information the same way. I was always a low average student, and math was always my most difficult subject and still is today, but not in the way other people say that. I struggle with making change, leaving tips at restaurants, figuring out percents when stores have a percentage off sale. I cannot process it at the moment. Basic life circumstances became a struggle and only when I met other people living with Galactosemia did I realize that the comprehension issues were similar for all of us.

Just as I was slow academically, I was also slow in developing physically. While my friends were going through puberty and talking about it, I was not having any of these issues or normal signs such as late development and still looking like I was in grade school physically as opposed to being in high school. This continued the sense of isolation that having galactosemia makes me feel. After going through puberty, I began to have unusual symptoms such as hot flashes, mood swings and night sweats. After about a year of experiencing these symptoms and having blood work, I was diagnosed at the age of 18 as having premature ovarian insufficiency. Which is a direct result of Galactosemia. This is something that I continue to struggle with today.

As a result of this diagnoses, I became very reclusive. I wanted no part of dating or socializing like any other 18 year old would be interested in the impact of POI. Secondary to having galactosemia, this was very difficult in my 20s. I just wanted to be like any other woman my

age, socializing and having fun. However, I was depressed and had a difficult time making friends and meeting new people. There were many nights where I cried myself to sleep. At a church event in 1999, I met a man named Bill who showed interest in me and started spending time with him. I was very apprehensive about getting involved in a relationship, even though I was 26 years old. The fear of falling for someone only to have to tell him that I was infertile and taking a chance on losing this relationship weighed heavy on my mind. However, I finally got the nerve up to tell him. Bill is a pharmacist and his response was priceless, not caring about my diagnosis and instead advising me to take my calcium because I was such a calcium slacker. What a huge weight off my shoulders and we celebrated our 21 years of marriage this past May.

I was told long ago that having galactosemia was like looking at everyone at a party through a glass window and watching others being a part of eating foods I could not have. I could see them through the window and through the glass, but could not break it or join them. The same is true for infertility. I can see moms and children through the glass, but cannot experience what it is truly like to be a mom. All my life I longed to be on the other side of that window.

There could finally be a treatment for people like me. And even if it is too late for me, it could work for younger ones living the way I grew up. I spoke at the Patient Focused Drug Development meeting, but our community never heard from FDA after that. They had no questions, no clarifications and that made us worry they had not taken the time to learn from our work before reviewing a drug. Will they understand that speech issues don't matter to us the same way as cognition and developmental delays do? Some of us have even more scary symptoms than the ones I have had to live with, like seizures, kidney issues, tremors, cataracts, neurological impairments, and intellectual impairments.

Will they understand that if they ask for another trial, our community won't have enough patients for another trial? Will they understand that if the drug is approved for something else rare, our doctors have seen the improvements in our community and will want to prescribe it for us too. The drug is already being studied in other rare diseases. These days we have to fight for approval but then fight insurance too. The Galactosemia Foundation is working hard on both of these things and I hope you can help us. Today a bill is being introduced in the House by Representatives Dunn and Matsui that will help us with the insurance side. I would like my Senator and the others in the room to look at this bill too. Please help us with this bill.

I'm trying to learn about what places like FDA and CMS and Congress can mean to me and to the others whose lives are impacted by diseases like Galactosemia. I don't pretend to understand it all. But I know even one person can make a difference and I want to help the little girls with Galactosemia going through the isolation, the puberty, the depression, the fear, the inability to understand and process. If all of you can take my story and help them, it will mean so much.