

Good morning, Chair Nosse, Vice-Chairs Goodwin and Nelson and Members of the Committee.

My name is Amanda Bain, my family and I live in Representative Owens's district in Baker City. Thank you for the opportunity to share my family's experience and express our strong support for newborn screening for Duchenne muscular dystrophy.

While it was my hope to speak to you in person, my son, Abel, was just diagnosed with Duchenne on February 3<sup>rd</sup> of this year and he will be receiving gene therapy this Friday at Stanford, so we're unable to speak with you in person today.

I want to tell you a little bit about Duchenne Muscular Dystrophy.

As Lauren mentioned, Duchenne is a rare pediatric disease that causes patients living with Duchenne to experience irreversible and progressive muscle deterioration. Without any intervention, patients living with Duchenne lose 50% of their functional muscle tissue by 5 years old. Abel will be 8 in December.

Sadly, 5 remains the average age of diagnosis for Duchenne despite significant advances in medicine and 8 FDA-approved treatments with many more in development. Many families experience a long and painful road to diagnosis that involves months and even years of doctor visits and unnecessary and sometimes counterproductive interventions. That was unfortunately my family's experience.

### **My Family's Diagnostic Journey**

Abel is 7 years old, and he is among the many patients living with Duchenne who received a delayed diagnosis.

I have two older children and with Abel, we noticed all the signs that something was different since he was a baby, besides the fact that he was seemingly the perfect baby, always happy, smiling and making everyone laugh. Honestly just the perfect person, which makes it all the more tragic that he has to go through all of this.

A few signs that should have pointed us to a diagnosis were, he never met physical and developmental milestones. He experienced delays in his speech, learning, and cognition. He had large calves and walked on his toes after a lot of physical exertion, (this was where we would lose the Drs and they would go straight to autism). His calf muscles would get tight and sore after certain activities. He struggled going up and down stairs, running, jumping and getting into the car.

Because of these delays, we went to extra well-child visits and met with different doctors to get answers but because he was so easy going and didn't complain or let any of this bother

him we never pushed for answers that hard, assuming it must not be anything that serious. During the pandemic, like everyone else, we didn't go to the doctors as often, and since Duchenne is a rare disease, our doctors in Eastern Oregon were unfamiliar with and unable to connect the symptoms to Duchenne.

Thankfully, a physical therapist came to Abel's school and assessed him. She wrote up a report for a pediatrician that was recommended to us in LaGrande, she noted that Abel's muscles—especially his core muscles—were extremely weak, she let us know that this was not normal or likely to be something we could help him correct and that we needed to continue to seek out a Dr. who could further assess him. Initially, our pediatrician didn't think of Muscular Dystrophy as she was very focused on autism. After we insisted the toe walking was not an autism sign but rather a sign of being sore after playing hard, she asked him to get up off the floor and when he displayed the gowers sign she later confirmed our physical therapist's assessment and tested Abel's blood to measure his Creatine Kinase levels, which generally rise when you have muscle damage. Normal CK range is between 97-100%, but Abel's were very high at 35,000.

## **Conclusion**

As a parent, you're faced with a seemingly endless series of decision points, and you just hope that you're making the best decisions to raise a happy, healthy child. For parents of children living with Duchenne, those decisions weigh heavier because each misdiagnosis and every minute that goes by without appropriate treatments is muscle lost forever and you can't help but wonder if there was anything you should have or could have done differently. Unfortunately I live with a lot of regret for not pushing for answers harder and sooner, but that is the problem with a disease that was randomly mutated. I never imagined it could be my child, my family that would have to endure this horrible disease.

Fortunately, with Abel's diagnosis, we finally have some answers. We've been able to connect with specialists and the broader Duchenne community. We've seen Abel improve on his steroid treatment and with new treatments like gene therapy, we have hope for Abel and future generations of children living with Duchenne.

I share our story with you today to request your support for adding Duchenne muscular dystrophy to Oregon's newborn screening panel. This will ensure that other families have more time and face fewer struggles trying to get to a diagnosis. For children living with Duchenne, "time is muscle." Adding Duchenne to Oregon's newborn screening panel will serve as an important tool to help parents pursue effective treatment for Duchenne as early in a child's life as possible, and I urge your support. Thank you for the opportunity to testify today.