Members of the House Commerce Committee,

I am a part of a group of parents who share the bond of being moms to children with a rare disease in the state of Minnesota. These children rely heavily on insurance to lead their best lives and participate in their communities. **The measures that are being proposed by HF 626/SF 464 and unrestricted access to services for the diagnosis and treatment of rare diseases is a vital resource needed for our children.**

First, I will share a bit about my journey with Leo.

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**Anne & Leo**

**Leo was born in May of 2016 and was diagnosed with infantile Pompe disease, a rare genetic disorder.** His body stores up glycogen in the muscles and the heart causing deterioration, muscle weakness, respiratory failure, cardiomyopathy, and more. The list goes on. Throughout his life, Leo has received support from quite a few different medical specialists and home care services. He has weekly enzyme replacement infusion therapies, monthly respiratory therapy visits to check his trach and ventilator, weekly physical therapy, weekly occupational therapy, weekly speech therapy, and even enteral nutrition support. That's not even the full list. Needless to say, he is a very complex patient, but at the end of the day he is still a child. And he wants to play, to run, and to keep up with his older brother, just like any other child. For that to continue, Leo needs to be granted access to care through an organization that understands the unique needs of a child like him. He needs access to care from providers that we choose and know understand our child's disease. Leo should not have to worry about network barriers and being told he can't see a provider because they are out of network. As Leo continues to grow his needs will change and those doctors may change as well. The ability to be nimble and adjust to fit his needs is one of the many reasons Leo is succeeding at home today. The specialist and medical team we have worked to create to treat Leo is why we have been able to avoid hospital admissions despite his complex diagnosis and care needs.

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**Now that you know a little more about Leo, I'd like to share why I support HF 626/SF 464 and why I want you vote in favor of this legislation to make sure individuals with a rare disease have access to a provider that is knowledgeable about their disease.**

A rare disease is defined as a condition affecting fewer than 200,000 people in the United States. Over 25 million Americans and more than 300 million people around the world are living with a rare disease and more than half are children. There are over 7,000 identified rare diseases. Of those defined as a rare disease only 10% have an approved FDA treatment. Furthermore of those rare disease patients, 30% will die before the age of 5 of these many life threatening diseases. This could be because, on average, it takes seven years for people with a rare disease to get a diagnosis in the U.S. In general, clinicians are likely to be very unfamiliar with most of the 7,000 identified rare diseases, and a limited number of healthcare professionals specialize in each rare disorder. Lack of appropriate disease-specific care contributes to delays and inaccuracies in diagnosis, delays in proper treatments, and sometimes

death. On top of these barriers, navigating insurance networks can be yet another challenge throughout a patient's journey to a rare disease diagnosis. And getting the proper treatment is literally a matter of life or death, the longer you wait.

**This legislation is important to me because we have personally dealt with insurance barriers to getting access to the proper specialists and treatments we need.** I will share with you just one of our insurance struggles. Leo's Pompe disease diagnosis only came because he almost died. At 4 months of age (and after seeing our experienced pediatrician many times for what we thought were feeding issues) we finally pushed providers to do lab tests. These labs came back normal - but the feeding issues and weight loss continued. Our pediatrician finally recommended that we go see a pediatric neurologist, but we needed to wait for approval from our insurance. **As I sat there and watched my beautiful baby get weaker and weaker I knew he could not wait the 3 to 6 weeks we were told it could take for insurance to approve the appointment.** On top of this, a limited number of specialist appointments meant we needed to wait the average of 2 months to just get into see the doctor. I called the neurologist and told them I would pay out of pocket. They told me they normally don't do that and I would need to give a credit card and it could cost up to $2,000 for the visit. They also said that, even if insurance did end up approving the visit and I had paid out of pocket, they could not go back and charge insurance or refund me. **I did not care and told them I would bring my credit card with me.** They then tried to give me a paid appointment in 4 weeks which I also told them I would not stand for. Luckily they had a pediatric neurologist physician assistant that I could see the next day for a little less- but it was still costly. We took that appointment and prayed. The next morning we checked in at our neurology appointment. I gave my credit card to the receptionist and we waited. I remember thinking this was it we were going to get the answers we needed. It would be worth the cost and Leo would be ok. Instead the Clinical Assistant came into the room, looked him over, listened to his heart, and then told us she was calling an ambulance and checking us into the hospital. When we got to the hospital they did an EKG and saw that Leo was in heart failure. Pompe disease causes the glycogen that cannot be broken down to be stored in the skeletal muscles and mostly the heart. His heart was so large it was crushing his left lung and he was also in the beginning stages of respiratory failure.

The doctors raced to do blood work and to stabilize Leo, but still were unsure what was causing this. They did blood work on me and my husband which we later paid for out of pocket since it was not approved or as insurance said "needed" for Leo's diagnosis - even though most rare diseases are genetic diseases and parents are carriers of these diseases. Leo was rushed into emergency heart surgery and had a trach placed later since after surgery he could not breathe on his own. After 2 weeks, the results of the large and expensive genetic panel came back and so did the Pompe disease diagnosis. While we waited for these results many experienced pediatric doctors and specialists had come to review Leo's case and they all had different educated guesses as to what was wrong with him. The only one who "guessed" correctly was a pediatric intensivist who had over 35 years of experience working in 5 different cities. He told us that he thought it was Pompe disease. When we asked follow up questions he said he had only seen 3 other cases in his over 35 years and did not know much about the disease. It was becoming very clear to us that these doctors who we look to to save our children did not have much knowledge about rare diseases. After the diagnosis, we were given some good news - that Pompe disease did have a treatment but not a cure. The treatment had only been around since 2016 and it was only meant to stabilize him. The drug they used cost on average $400K per year and also would require a month of chemotherapy treatments to break down his immune system to accept the new drug. They

advised us we needed to call our insurance or start the medical assistance application process before they could fully authorize monthly treatments. **We reached out to our insurance right away and were told they needed more documentation from doctors to approve**. Luckily, we were in the hospital and had many doctors who helped us work with insurance to get approval. Our insurance at first denied the large amount of medicine that Leo needed since they said it was only approved at half the dose per their guidelines. More paperwork, more begging for approval, and more watching our son lay in a hospital bed watching time go by and praying he could get the medicine that would help him. Things did work out in the end. We were able to appeal to the Electrical Union board for fast approval and payment for Leo's treatment. It took 6 months in the hospital to stabilize Leo and to make him strong enough to go home. He still receives weekly heart infusion of his medicine at the larger dose and will for the rest of his life. We also have 7 specialty doctors ranging from a geneticist to a gastroenterologist who we have to see each year and get approval from our insurance for their continued essential care to Leo's complex rare disease.

We are one of the lucky rare disease families out there. We had good private PPO insurance when we entered that hospital. We had the ability to afford out of pocket doctors to be seen quickly which literally saved Leo's life. Our child was so sick, near death, and was physically in the hospital such that we had access to specialists who we could pressure to write and call our insurance providers for us to breakdown the insurance barriers. We were lucky that we were in a larger metro area and had access to experienced doctors who had seen some other rare disease cases even though they still had limited knowledge on treatment. But you know who was not lucky - Leo. **During those 4 to 6 months we waited for diagnosis and then treatments, the glycogen continued to build up in his skeletal muscles. Today, Leo is in a wheelchair and requires intense weekly therapies just to be able to sit up, feed himself, and speak.** We eventually applied for the medical assistance TEFRA program where we pay a monthly fee based on yearly income so that we can have a secondary instance to cover the expensive costs of all his specialist appointments and therapies. **If the insurance barriers for rare disease patients were not so complicated, the state of Minnesota would save money on those families who are forced to apply for medical assistance - even while they have their own primary coverage.** The insurance companies would cover these essential costs to treat rare diseases and not make the state medical assistance programs do it.

As a parent and a citizen of Minnesota I am asking for your support of HF 626/SF 464. I don't want the other families of rare disease children to have to go through what we went through. I want all the rare disease children who come after Leo to have access to a provider that is knowledgeable about their disease. I want insurance companies to know that while rare disease are rare, there is still a possible diagnosis for some patients. I want the medical community to understand that time is essential to treating these rare diseases. I want insurance companies to not stand in the way of a child's life or future because it needs a prior authorization.

Thank you for your time and attention to this matter.

Sincerely,

Anne St. Martin | 630-670-6569

A Concerned Parent of Children with Rare Disease