Senate Committee on Health and Welfare

Testimony in Support of H.46

(An act to establish a Rare Disease Advisory Council in the state of Vermont.)

Good morning Senators, my name is Sarah Elliott. I am a constituent from Glover, Vermont and a patient with a rare disorder. I want to thank you for considering this bill and for inviting advocates like myself to come in today to provide support for bill H.46 to establish a Rare Disease Advisory Council. Rare disease is a bipartisan topic; and it can impact individuals from all parties in the same way that disability can.

I grew up in rural Vermont with a rare skeletal condition, giving me valuable and unique insight and perspective into the various systems in Vermont as well as other parts of the country. My experiences with these systems have been varying, some not always that pleasant, but because of the work of our legislative body - you, our senators and the representatives over in the house, I have personally witnessed improvements in these systems that have increased my ability to access services and medical care to manage my rare disorder. I have a form of Diastrophic dysplasia which results in my stature being shorter than average, but taller than the typical Diastrophic dwarf. I also deal with a lifetime of chronic pain, respiratory issues, fatigue, and quite frankly, battles with mental health that can feel more difficult than the arthritis working its way through my spine and joints.

Vermont has made amazing strides in the programs and services available to individuals with some types of disabilities and more common disorders. Expanding these offerings to the rare community will continue to make our state a place that is accessible, safe and welcoming. Ensuring the RDAC is passed is a step in this direction.

An effort of the RDAC that I want to touch on is the newborn screening and consulting with experts to develop policy recommendations. In Vermont we currently screen for 35 genetic conditions on the newborn screening panel, and that puts us low on the list, as many other states are testing for many more, even double this number. Early testing is important as it allows families to make decisions about health care that might otherwise not be available and to connect with specialists and other families early on, which is truly priceless. I never met anyone else with

dwarfism until I was in my late teenage years and had to travel out of state for both building connections and for seeking expert medical care.

While there is no need to cure or treat dwarfism, individuals with various types can lead long fulfilling lives, there are some treatment options available that can ease symptoms. Currently they are for the most common form of dwarfism- achondroplasia- but these treatments need to be started early for optimal results. Historically, the “wait and see” approach was used with many forms of dwarfism, and this has resulted in more costly medical conditions later in life, and sometimes even death.

Personally, I was misdiagnosed and it took 25 years to meet with a genetics specialist because we didn’t know to question the diagnosis or that getting a proper diagnosis would be important. It then took another 10 years to reach the current clinical diagnosis that I have now, and that was only possible through my genetics team after I was pregnant, because I could not afford the out-of-pocket cost for the testing prior to this time. Having a proper diagnosis has been important to getting providers to listen to my needs as there is not much research that currently exists, so as I attend conferences and learn from experts in the field, I am able to bring this back as an advocate for my own needs.

Continuing to access medical care that helps me stay healthy while also considering the true developments of my body is why I support the establishment of the RDAC and advocate so strongly for the needs of the both the rare community and the disabled community.

Thank you for your time and support on this extremely important topic.