My name is Tai Pasquini, and I am a Steering Committee member of the Massachusetts Rare Disease Advisory Council (RDAC). The MA RDAC was established in 2021 with 29 members representing the diverse stakeholders of the rare disease community.

Our Council holds full council meetings every other month. We also have a Steering Committee and focused working group sub-committees. In 2024, the RDAC held a total of 25 meetings including participation in a Rare Disease Day event and a legislative briefing. Our current established subcommittees include a Policy Working Group, a Research Group, and a Community Engagement group.

Through our meetings in 2024, the RDAC focused on five areas that impact the lives of people with rare diseases: 1) newborn screening, 2) medical nutrition, 3) legislation/policy, 4) economic burden, and 5) community engagement.

Since the formation of our RDAC we have generated a report estimating the prevalence of rare diseases within the state, launched an online survey to understand the needs of the community in MA, provided recommendations to improve newborn screening in MA, and heard from many members of our community related to their experiences living with rare diseases which further exemplifies the needs that our RDAC hopes to highlight and address.

Project identification within our RDAC has been primarily driven by the legislation. We also decided to utilize the National Organization for Rare Disorders (NORD) policy report card to flag additional areas of improvement.

One of the major strengths of the RDAC is the ability to bring together the diverse stakeholders within our state. Like many members of the RDAC, I came to the committee with a few relevant “hats” -researcher and loved one. I have a PhD in public health which I pursued to address some of the pervasive challenges families face, such as navigating the U.S. healthcare system for access to critical treatments and healthcare resources.

This led to my current role as the Chief Research Officer for Congenital Hyperinsulinism International, a patient organization that represents individuals living with hyperinsulinism, a rare disease that is categorized by severe persistent hypoglycemia, which, if left untreated, can lead to brain damage or death. The disease requires swift action within the first few days of life to prevent the avoidable impact of prolonged neurologic damage. It is characterized by the need for constant vigilance to prevent hypoglycemia and current treatment options are subpar.

At face value, this has little relationship to Progressive supra-nuclear palsy (PSP), a rare neurodegenerative condition that is often misdiagnosed as Parkinsons'. However, like many rare diseases, in both, the diagnosis is challenging and often delayed; most people

have not heard of the condition, individuals struggle to find specialists, treatment options are limited and costly, and families are overwhelmed as they try to find a new normal. In the last year, I have watched the diagnosis of PSP impact my mom as this powerful woman who was once a second-degree blackbelt now struggles to walk. A woman who earned 2 master's degrees, can no longer read to her grandson and struggles to formulate sentences.

The RDAC has an opportunity to shine a light on these concerns. We have had opportunities to collaborate across disciplines and with the changemakers within the state. We are finding ways to make sure that this community has representation in programs and policies that will impact individuals' daily lives.

Beyond our state lines, it has also allowed members of our team to connect with others in other states. We are learning from each other's wins and finding the "low hanging fruit" of meaningful change. Chances are that everyone knows someone who is impacted by a rare disease or someday will. I am proud that MA made that commitment to our families and our neighbors and I encourage Vermont to make that same commitment.