**Massachusetts Rare Disease Advisory Council**

**Virtual Town Hall**

**Saturday, October 28, 2023**

**Special Report**

**Executive Summary**

The Massachusetts Rare Disease Advisory Council (RDAC) held its first virtual town hall on Saturday, October 28, 2023, to listen and learn from the public and the rare disease community. The meeting was open to the public, and stakeholders in the rare disease community were invited to provide testimony. The purpose of the town hall was to gather information about the availability of resources needed by those living with a rare disease in Massachusetts.

The central theme of the rare disease town hall testimonies focused on challenges in accessing therapies and social supports. Multiple testimonies referenced a lack of insurance coverage for costly treatments for people with rare diseases. Lack of access to home care was also cited as a major challenge to quality of life for people with rare disease.

These testimonies offered at the town hall will help the RDAC to make meaningful recommendations to the governor, the legislature, and the Department of Public Health about how to improve the lives of people with a rare disease living in Massachusetts.

**Oral Testimony**

*As a member of the Rare Disease Advisory Council member, I am sharing my knowledge and experience as a mother and caregiver to my three children who live with a rare disease. I have two sons with Deschenes Muscular Dystrophy (DMD) and one son with Primary Immune Deficiency (PID). I wanted to share what is going well so we don’t just talk about what is going wrong. My oldest son, Austin, is on the call and will speak shortly, but I wanted to talk about a wonderful resource that we just recently learned about. It’s the Complex Care Team at Massachusetts General Hospital. I learned about this program through this council, and it has been a great resource for us. Austin is a frequent flyer to Mass General Hospital, and this Complex Care Team has helped us navigate some of the challenges associated with regular admissions to the hospitals.*

*One of the biggest challenges we have faced is access to innovative medications and treatments. Although Massachusetts has a strong biotech presence, access to some of its innovative drugs is limited. For example, my two older sons are wheelchair users, and the only medications approved for DMD are not available for wheelchair users. We have been trying to get access to these innovative drugs and treatments for almost ten years without success.*

*Another big challenge for me as a caregiver is home care. My primary goal has been to keep my sons home, and home care is a critical piece of that. As a single mom who also has a full-time job, home care becomes one of our primary needs. Finding quality and reliable home care has been a real challenge.*

*Lastly, I want to say that I’m happy to be living in Massachusetts. It has a lot to offer as far as medical care, and I know we are very lucky, but improving access to critical resources and social supports is necessary to improve the lives of those living with a rare disease in Massachusetts.*

Jenn McNary, Saugus

*I am Austin, and I am living with a rare disease. I am twenty-four years old, and the most important thing to me is my independence. In order to have that independence, I need to rely on services like “the ride.” I have a lot of medical appointments, and “the ride” is not always reliable. It doesn’t always get me to my appointments on time. There often isn’t anyone available by phone to verify my pick-up time or even confirm that my ride is scheduled. This adds a lot of stress.*

*I rely on people almost 24 hours a day to ensure my independence. Personal Care Attendants (PCAs) are a critical part of my care team. I would like to say that PCAs don’t get paid much, and they often don’t stay at the job long because of the pay. This makes it hard for me. When I don’t have caregivers, I have to rely on my mom and my 16-year-old brother for my care. My brother has his own rare disease and should not have to care for me. He has his own life, and we are worried now because he is looking at going to college and, therefore, won’t be available for backup care. I now need a BiPAP (a non-invasive ventilator) at night. I’m still getting used to it. I had a near-death experience due to RSV, so I now need a BiPAP at night. I think I should be able to rely on nursing and home care to help me, especially at night, but insurance won’t approve it. It really bothers me that people think that my mom should take care of me. It’s not fair. I don’t understand why it is expected that my family takes care of me when I’m an adult. I hope people realize that nursing is keeping me out of the hospital, and I don’t understand why I can’t get approved for the nursing hours I need to stay healthy and active and out of the hospital. People seem to think I should be in a facility that is a lot more expensive than living at home, besides the fact that other people may be sicker and need that hospital bed more than me.*

Austin LeClaire, Saugus

*I’m Criss Quigley, I’m here representing people with rare craniofacial disorders. They are dentinogenesis imperfecta, amelogenesis Imperfecta, and ectodermal dysplasia. We are a small group, and our primary challenge is insurance coverage for treatment. To give some background about this birth defect, it is a birth defect that affects the teeth, soft tissues of the mouth, and the jaw. Teeth become nonfunctional over time. Kids have trouble eating and often need a liquid diet. The social stigma and isolation that occurs is very damaging. It also affects one’s ability to get a job and be productive. Most treatments are not covered by insurance. Parents often learn that the treatment will cost them thousands of dollars. Research shows that treatments for these conditions can cost as much as half a million dollars. Parents go to extreme measures to find the funds to pay for these treatments.*

*Fifteen years ago, when my daughter’s out-of-pocket expenses exceeded $150,000, I decided to try to do something. I found another family who had a child with a similar condition, and we worked with our legislator to get a bill written that would ensure insurance coverage for these conditions. A similar bill was presented at the same time for people with cleft lip and palate. The bills traveled together from 2009 to 2013, when the bill for cleft lip and palate passed but ours did not. The bill has been refiled seven times and still has not passed. Our families have met with many advocacy groups, hospitals, and dental schools and have testified in front of the legislature many times. Our children deserve treatment as much as others. We are a small group, and we hope that the Rare Disease Advisory Council can help us.*

Criss Quigley, Springfield

*My name is Jillian, and I am the mother of a little girl who has a rare disease. She has OCNDS or Okur-Chung neurodevelopmental syndrome. She presents with epilepsy-type symptoms, and her condition is considered ultra-rare. Because of that, most doctors, nurses, teachers, and caregivers do know about her disease. I find that I have to make up education sheets to help people understand her condition. I believe that one of the biggest problems of having a rare disease is education. I feel very lucky that we have access to services like early intervention, Physical therapy, occupational therapy, etc, but it’s really hard explaining her condition to everyone.*

*It is heartbreaking to hear that kids are not getting the treatments they need due to costs, but I know it’s a reality. All kids should get access to the medications and treatments they need without worrying about costs.*

Jillian Kavanaugh, South Hamilton

*My name is Tai. I am a member of the RDAC, and I became a member to contribute as a researcher, but recently, my mom has shown signs of cognitive decline. With a family history of a rare neurocognitive condition, we became concerned. My mom recently moved from Connecticut to Massachusetts to be close to us. She had to change all of her doctors. We have had a very difficult time finding specialists for her to see. The wait times to get into a neurologist are really long. Medicare tried to find a plan that would allow her to go back to Connecticut to see some of her doctors in the meantime, but the plan they picked was not a good fit. We also realize that doctors don’t know about this rare condition. She needs to see a specialist, but it’s almost impossible to find a specialist near us. We are not sure if my mom has the condition at this point, and we don’t know when we may need to travel to find a doctor who can diagnose and treat her. It’s really hard to know when to do this. It's crazy that insurance is preventing us from accessing the right doctor for her.*

*As a supporter of a single-payer healthcare system, I know that one of the biggest complaints is long wait times, but we have long wait times now. There needs to be a better system. People should be able to get the care they need without worrying about networks, formularies, and other restrictions that are insurance-driven. Thank you.*

Tai Pasquini , East Longmeadow

*I am a mom and caregiver of a little boy with three rare diseases. One of the challenges I have experienced is the difficulty of getting a diagnosis. A lot of times, you can’t access services if you don’t have a diagnosis. We need to fix that. It sometimes takes years to get to a diagnosis, yet the child and family need help while trying to figure out the diagnosis.*

*The other topic I’d like to talk about is home care. As stated earlier, home care is critical. The PCA program sounds great in theory, but it’s broken. PCAs are great when you find a good one, but at $18/hour with no benefits, there aren’t a lot of people wanting to do this work. Personally, I don’t know why a family can’t be paid as a PCA. Currently, my 80-year-old mom is providing these services for my son, but it’s really hard. Many families can’t work because they need to stay home to care for their child. If they could be paid as a PCA, it would help.*

Ali Mahady, Westford

**Question and Answer Session**

Question: Coverage for treatment and equipment continues to be a challenge for many with a rare disease. Is it MassHealth or private insurance?

*My daughter currently needs treatment, and both dental and health insurance have denied her to get this treatment which will cost over $100,000. Most of the families we talk to have private insurance, but I can honestly say that I don’t know one family that has got insurance coverage for their treatment. To answer the question, I don’t know if it’s a MassHealth or a general insurance issue.*

Criss Quigley, Springfield

Question: If you could remake “the ride” what would it look like? Have you ever used Uber, if so, does that work?

*First, I think that the ride should be free. Second, there should be some dependability. When I have an appointment, I need to get there on time. Also, sometimes, the drivers will just leave us, even if we missed our appointment. Unfortunately, accessible Uber rides are hard to find.*

Austin LeClaire, Saugus

Question: We heard that specialists are hard to find. Do you know if there is a way to find specialists in other areas?

*My daughter’s specialist is at Boston Children’s Hospital, so we are very lucky. Although we are close to her specialist, there are other healthcare providers we need to see, and they know nothing about her diagnosis. Bringing a fact sheet whenever we go to a new provider has been the best solution for us.*

Jillian Kavanaugh, South Hamilton

Question: Can you speak to the PCA program? What would you like to be improved about it?

*The first thing would be to pay. PCAs are getting roughly minimum wage. The work is hard. People need to be responsible. It’s not babysitting. My son needs to be spoon-fed close to 15 times a day. He is blind, so he needs ongoing assistance. PCAs should get benefits. Other states allow parents or other family members to be paid as a PCA. It’s also hard to get the number of hours you really need. Also, nursing hours are important. MassHealth seems to be cutting nursing hours when they are clearly needed.*

Ali Mahady, Westford

Question: How does the PCA program work?

*The PCA program is a state program of MassHealth. There is a fiscal agent called Tempus who pays the bills, but it is up to me to interview, hire, fire, and manage the schedule and timesheets of the PCA. The family is also responsible for training the PCAs. It is often a full-time job, just managing the PCAs and the schedules. There are many barriers to the coordination of care when using PCAs.*

*The home care situation is a significant problem for those living with a rare disease in Massachusetts. The PCA program is broken and needs revisions. We know there are many issues related to home care to address, and we hope that the RDAC can tackle some of them.*

Jenn McNary, Saugus

*I know there is a PCA union, but I’m not sure how much they have been able to change for PCAs in Massachusetts. I think the pay has gone up a little but not enough to make a difference. MassHealth breaks down to minutes you get for the year. If your child needs to have a diaper changed and be fed, the PCA isn’t going to come just for an hour. MassHealth needs to relook how they calculate the time that a child needs. My friend, who is in another state, has to calculate how much time it takes to feed her child. As we all know, feeding a child one day may take 20 minutes, and feeding on another day could take 40 minutes. I believe that MassHealth is going to start requiring PCAs to clock in and clock out when they come to provide services.*

Ali Mahady, Westford

**Written Testimony**

***Please provide testimony on the most important healthcare resources and social supports that currently exist and how you found them to be valuable (up to 500 words).***

*I am responding as a friend of several Massachusetts ALS patients. Two of them have died during the several years I have been involved in ALS advocacy. ALS is a devastating diagnosis. Families impacted by ALS deserve and require extensive support. We are fortunate in the greater Boston area to have support from CCALS and ALSA, along with drug research by TDI and area hospitals. However, many regions in MA lack these resources. For patients with ALS, access to trained home health care aides and adaptive equipment should be available statewide.*

Jennifer Wright, Wakefield

*I am specifically responding with second-hand (as an advocate, not a patient or caregiver) to advocate for change in the treatment/care of patients with ALS. I am a member of the Massachusetts ALS Champions (www.palsmac.org), a non-affiliated group of local advocates for change in the ALS landscape. My perspective is shaped by watching Becky Mourey and Cathy Nally, and their families, fight hard and struggle through the challenges of ALS. I understand that dedicated ALS clinics provide top-notch care but are not always accessible/available to patients from outside the areas where these clinics are located. Organizations such as CCALS and the ALS Association (ALSA) provide fantastic support and care services to families dealing with ALS, such as equipment (power wheelchairs, vans, communication tools such as Tobii, etc.). ALS organizations, including CCALS, ALS One, I Am ALS, and ALSA provide networks and social supports.*

Barbara Kipp, Bedford

*We are primary care pediatricians who have a combined 60 years of caring for infants to young adults with complex and chronic illnesses here in the Commonwealth. Many of our patients have so-called rare diseases or congenital issues or conditions that have yet to be named or fully understood. Like many in this forum, their family experience is made more challenging by the lack of reliable sources of information and the sea of misinformation and questionable assertions online, including social media. We have shared the learned lessons of what works better and, at times, even best. We learned this early: parents are often the true experts in the particularities of their child’s health and offer critical insights to make optimal shared plans of care. In turn, a family and care team must learn to work collaboratively to seek the best evidence and weigh the risks and benefits of treatments. While the provider and care team relationship may be important in promoting wellness and preventing illness, it is by no means the only source of support for a family. Other parents, peer mentors, health coaches, and online networks can offer the benefits of community and shared experience. From referrals made by palliative care providers to online platforms like the Courageous Parents Network and more, caretakers may develop friendships and associations that provide a crucial and rare layer of support and perspective. We cannot say enough about the excellent work of organizations in the Commonwealth like MassFamily Voices or from the Federation of Children with Special Needs who can help families address whatever their evolving needs may be, including their physical, emotional, financial and spiritual health over time. While in the past few decades, there have been great advancements in the care of children with disabilities allowing them to live outside the hospital and for longer periods of time, supports for families have not been adequate nor equitable. Longer survival times have not necessarily resulted in a better quality of life for patients and their families. Families are burdened by hidden costs and challenges to even health-literate patients and families, including service gaps and the opportunity costs faced by caretakers foregoing employment opportunities to administer care for their child and/or to navigate health systems. As a result, these children may now be outliving their parents and will very likely live beyond their ability to be cared for by them. We now have a moral obligation to patients, their families, and providers to develop infrastructure to support in-home and alternative settings for this growing population. We want to use our testimony to draw attention to some of the great resources like MassFamily Voices or from the Federation of Children with Special Needs that exist for families of children with rare diseases and raise awareness of our collective need to better support the families impacted by what can be devastating or costly conditions that affect entire households. We also want to advocate for sustainable strategies for aging parents to support their families for the years to come.*

Jack Maypole, Waban

*MA offers good Medicare options. We also have excellent medical institutions that not only provide excellent care but also do incredible research in the rare disease space. The life science industry here is second to none.*

Steven Kowalski, Boston

*In October 2020, my sister and best friend Cathy Nally was diagnosed with ALS. Cathy faced ALS with the same love of life, determination, courage, and energy that marked her impactful 62 years on this earth. Widowed in 2001, Cathy was fiercely independent and raised two amazing children on her own. That independence served her well as she battled ALS; she tried to identify and address her ever-increasing needs by herself. It soon became clear to those of us who loved her that such an approach was not sustainable. Living in the north end of Boston, she was blessed to be so close to MGH and the Healey Center, where she received much of her world-class care and support. Most other ALS patients in MA are not so fortunate. But as ALS progressed, Cathy and I learned that she couldn’t do it by herself. A dear friend who lost his brother to ALS connected me with CCALS based here in Massachusetts. They were an incredible resource to my sister and our family, and I have recommended their services and programs to other families suddenly facing the uncertainty that comes with an ALS diagnosis.*

Ed Hurley, West Yarmouth

*The most important healthcare resources in our 30-year experience as an active organization supporting our rare disease community are proactive, well-organized, and well-funded nonprofits that are formed to work closely with healthcare providers and other stakeholders to assess community member's needs and do all it can to fulfill them, socially, financially and otherwise. ‘It takes a village’, as they say, and nothing could be more true than in the rare disease community. With rare disorders, it’s important for the community (patients, caregivers, care providers, industry partners, governmental agencies, and other nonprofits) to come together, stay connected, share information, support each other, and advocate for one another. Coping with a rare disorder is hard enough in and of itself. Remaining positive, managing the disorder on a daily basis, and continually advocating for oneself/family while planning for the future is even harder. Meeting people “where they are” at and helping them improve their health care is essential to ensure a healthy community. NECPAD often receives referrals from local clinics or individuals who reach out to us directly. Some just need someone to talk to about their diagnosis (not from a clinical perspective, but practical) and what the next step is in terms of how, in practice, they will address the diagnosis and ongoing care of an individual; others need some help financially to afford deductibles, copays or medically prescribed items not covered by insurance, others need help fighting for, and affording, insurance coverage (this is huge), others need help purchasing essential items to help them manage their care such as an extra refrigerator to store medically prescribed nutrition or transportation costs to clinic visits, others need legal advice, others need assistance with other basic life needs that are related to their disorder, and yet others just want to be connected to a community which “gets it” and can be there to lend advice, support and frankly, some respite! All are equally important and essential to help live, as well as possible, with a rare disorder. NECPAD strives to achieve this and inspires other stakeholders to work together to do the same. Thank you for the opportunity to comment and please do not hesitate to reach out to me so that we may continue this important discussion and work together to define and implement ways to improve the lives of those living with rare disorders in the Commonwealth of Massachusetts (and beyond) as Massachusetts has always been a leader in this regard (e.g. first state to implement Newborn Screening Program (NBS) and leader in healthcare and biotech industry)!*

Denise Queally, Chilmark

***Please provide testimony on the healthcare resources and social supports that are lacking or are difficult to access and how they would improve the lives of people with rare diseases (or their families if they did exist) (up to 500 words).***

*As mentioned above, access to support for families coping with ALS varies widely in MA. Non-profits can help with adaptive equipment to some extent. The biggest problem is a lack of home healthcare aides specifically trained to assist ALS patients. Care often falls to family members, with little or no respite. And even if qualified help were available, most families do not have the financial means to pay for such support. Patients with ALS should be provided with trained, well-paid home health support funded by private or government insurance.*

Jennifer Wright, Wakefield

*Despite some of the great resources mentioned above, it seems that ongoing care for people living with ALS (PALS) still falls mostly on family members. PALS should not have to rely on family members (who are also trying to take care of other family members and support themselves) for their care. While insurance (including Medicare) sometimes covers nursing/home health care, many/most of these workers are not skilled or trained in the needs of PALS, which includes dealing with respirators, feeding tubes, electronic communication equipment, patient lifts, etc. Some of these specialized skills apply to patients with other diseases too. Concerted efforts to increase the number of providers with these skills is needed, including better pay for the workers and specialized training (perhaps even “certifications” in specific skills). I understand that funding is available for this, but it has not been applied in a disciplined and focused way. I think the government should partner with the aforementioned ALS organizations to make this happen and should be held accountable. For people who don’t live near established ALS clinics, programs need to be developed/expanded to enable PALS and their families to participate in clinical trials and be treated by world-class clinicians. This will necessitate funding for care (including insurance coverage), transportation, and temporary housing arrangements. This greater access will help ensure that the care (and clinical research) surrounding ALS is representative of both the full disease population and the diversity of our country (including ethnic and socio-economic diversity).*

Barbara Kipp, Bedford

*As an affected constituent and parent of an affected child with rare dental disorders, ensuring access to medically necessary treatment for individuals with these conditions is an essential step toward achieving equitable healthcare, and I would greatly appreciate your support and advocacy. I am 49 years old and have seven dental implants that my parents were able to help pay for over the years. I am extremely fortunate to have parents who worked hard and were able to help pay the large dental bills for me. My son Jeremy (see pictures below) is 20 and has just completed his first major procedure. Although he does not have implants yet, what you see in the picture will cost my family $27,000 dollars, and this is just for the front teeth. He is a junior at Boston University (he has siblings at UMASS Dartmouth, a senior in high school, and a freshman in high school), so we will be helping to pay college tuition bills for years to come. This extra dental bill is a tremendous burden for our family. Any help getting this bill passed is greatly appreciated. Children and adults with rare craniofacial disorders affecting the mouth, jaw, and teeth experience pain, difficulty with eating and speech, bone loss, infection, jaw dysfunction, and psycho-social difficulties due to their facial differences. Although physicians categorize treatment as medically necessary, health insurance companies currently use loopholes to deny coverage to this group. These insurance denials result in individuals or families facing catastrophic out-of-pocket medical expenses for treatment, upwards of hundreds of thousands of dollars. Patients with cleft lip and palate are currently covered for dental treatment in Massachusetts, but those with more rare craniofacial disorders are not. This is not only unjust but also counterproductive to the overall well-being of our community. By providing insurance coverage for rare dental disorders, we can ensure that affected individuals receive timely and appropriate treatment without the heavy or impossible financial burden that often accompanies these conditions. This legislation would not only alleviate suffering but also promote better oral health outcomes, reduce the long-term healthcare costs associated with untreated dental conditions, and ultimately enhance the quality of life for those affected by these rare disorders. According to the most recent CHIA study, the cost to the insured population is pennies per month.*

Gregory Gay, Taunton

*Lack of insurance coverage for treatment for rare craniofacial disorder: My 12-year-old daughter, Elayna Melendez, born July 2, 2011, has been diagnosed with Amelogenesis Imperfecta (AI). AI is a condition that causes teeth to be unusually small, discolored, pitted or grooved, and prone to rapid wear and breakage. As of right now, I have already started to pay out of pocket for crowns for her teeth in the back of her palate that have been through rapid breakage. I am emotionally stressed and worried about how much I would have to pay in the future for a new set of teeth for my daughter as well as the maintenance of costs. Since Kindergarten, Elayna has been stressed, sad, worried, and upset, and I can name many more emotions that she feels about not having normal teeth. When people without a teeth disorder hear the word SMILE, it’s a positive thing. But for Elayna, it’s not. Smiling with AI teeth has made Elayna an easy target to get bullied. As a mother, I try my best to stay strong and tell her one day we will fix her teeth, and each time I tell her that, I wonder if it will be fixed or if I will have the funds to fix her teeth or smile when that day comes. My daughter bed wets still, and I believe her stress with her teeth may play a big role in it. My child isn’t allowed to eat certain things because of her condition; her teeth are so sensitive that sometimes she tells me she prefers not to eat. Parents normally worry or must plan to save for college for their children’s future. For parents with children with AI, it’s stressful thinking about saving for new teeth for their child. Or if that’s even possible. I’ve learned and heard of people paying $100,000 for new teeth, and that’s not including the maintenance. I hope this testimony gives you a realistic idea and understanding of what it’s like to have AI and also to be a parent to a child who has AI and the uncertainty of how much it’ll cost for treatment, the worrisomeness of just going to a dental appointment and just helping your child handle bullies, insecurities, and much more.*

Jasside Carvalho, Agawam

*I have been receiving treatment for my rare craniofacial condition, dentinogenesis imperfecta (DI), since I was two. People with DI have abnormally formed teeth — small, with tiny roots, grayish-brown, with no hard-white enamel so that they wear down or chip away without treatment. The condition is very painful and patients lose teeth easily. Patients may have difficulty eating or speaking, and the jaw may become wasted or deformed. In some cases, the jaw may even lock and not be able to open. These are serious, debilitating disabilities and treatment is complex, requiring a team approach. Without early treatment, my teeth would have worn down to gum level from normal chewing. I was fortunate that my parents were able to pay for a full mouth reconstruction at 16, and since then, I’ve continued to require treatment for infections, an open bite, broken teeth, loose and broken prosthetics, and jaw pain. The cost has been over $150,000 so far, almost all out of pocket, because Massachusetts law contains a loophole where insurers can deny coverage for birth defects that affect the teeth, even though the patient’s nutrition, speech, psychological well-being and quality of life are impacted by the disorder. Although the cost has been a huge burden on my family, I am fortunate compared to many who can’t afford the type of treatment I’ve had. I’m 35 now and just beginning another full mouth reconstruction, which will mean more oral surgery, bone and tissue grafting, implants, and all new crowns. It’s not uncommon for people with DI to need ongoing treatment throughout their life. The cost for this phase of treatment is estimated at least $110,000, and both my medical and dental insurance have already denied coverage. I delayed treatment much longer than was recommended, hoping that legislation mandating insurance coverage for these rare conditions would pass, and meanwhile dealing with broken crowns, an uncomfortable temporary denture, an increasingly limited diet, the risk of bone loss, and anxiety about the cost of treatment and my future. I am part of the Massachusetts advocacy group that has been working to pass legislation to mandate insurance coverage for patients with these rare disorders for the past 15 years. Since 2012, patients with cleft lip and palate have had mandated insurance coverage for dental treatment related to their craniofacial birth defect, but rare disease patients were not included in that mandate, so families like mine continue to face hundreds of thousands of dollars in out-of-pocket costs. Health insurance should be inclusive and not discriminate based on diagnosis or the prevalence of a condition. Mandating insurance coverage for these rare disorders would ensure that children and adults with these rare disorders have equal access to necessary medical care. Society as a whole benefits when all its members have improved health outcomes and quality of life.*

Ann Quigley, Chilmark

My name is Karen Tenner. I was diagnosed with Amelogenesis imperfecta, a condition in which enamel or the hard outer layer of teeth does not form, leading to soft misshapen yellow teeth. *As a result of this, I have dealt with countless psychological, emotional, functional, and financial challenges. I am now a practicing primary care physician in the Commonwealth and know firsthand the impact poor dentistry has on many aspects of my patient's health. Early on, my teeth were brittle and brown, crumbly. I was lucky my family sought out consultation from experts across the world, and I was diagnosed with AI at age 2. As an elementary school child, I still remember the taunts, the questions, “Don’t you brush your teeth,” and the discomfort and pain of eating that came with having enamel-less teeth. As a teenager, I spent a summer recovering from a bone graft to prepare my mouth for essentially a full set of implants. During my first two years of high school, before this drastic intervention, I was missing 2/3 of my expected adult teeth. I remember hiding my smile, ashamed of the gaping holes in my mouth. I am one of the lucky ones. I speak before you after almost every single adult tooth has been replaced. I have almost an entire set of dental implants. While my bone graft was partially covered by my medical insurance, there was essentially no other coverage. Over the course of my childhood, my parents estimate that they spent over 250,000 dollars on my dental work. Even with all of these interventions, my problems continue today – in the last three years, I have had three teeth pulled due to AI-related complications and implants placed. Now as an adult with excellent dental coverage, this work has still cost thousands of dollars out of pocket. This work is not just cosmetic. Teeth are essential for speech, sustenance, and our ability to function in the world. I am almost certain that had I not been born into a family that was able to afford this care out of pocket, I would not be the same contributing member of society I am today. As a patient and a physician --- I ask for your support in this essential bill to guarantee coverage so that all patients, regardless of their financial status, can get the care they so badly need and deserve as constituents in the commonwealth with these challenging craniofacial diseases.*

Karen Tenner, Arlington

*Increasing Access to Telehealth Services: Virtual care improves patient access to high-quality care well beyond the COVID-19 pandemic. More communities than ever have experienced telehealth’s powerful impact in bridging gaps in care, especially underserved and rural communities that historically have had limited access to specialty care can now see top specialists due to expanded access to telehealth. Banning the Use of Quality-Adjusted Life Years (QALYs) and Equal Value Life Year Gained (evLYG): QALY and similar cost-effectiveness metrics are discriminatory towards those with disabilities and rare diseases like ALS. State governments should ban the use of discriminatory QALY and evLYG metrics when determining coverage of existing and new ALS therapies. Eliminating or Decreasing Durable Medical Equipment (DME) Taxes: In general, in the U.S., tangible personal property (i.e., items you can see, touch, and interact with) is considered taxable when sold at retail. However, states can determine what is and is not taxable within their borders, and many states make exceptions for necessities like medical equipment. Taxes on DME unnecessarily increase the financial burden of people living with ALS. Guaranteeing All Copays Count Towards the Cost of Drugs (Banning Copay Accumulator Adjustment Programs): Many insurers have disallowed copay assistance (also known as copay savings program, copay coupon, or a copay card) from counting towards a patient's annual deductible or out-of-pocket maximum. As a result, people living with ALS can face unexpected costs of thousands of dollars to get the medicines they need. Reforming the Prior Authorization Process: As a cost-control measure, health insurance plans often require physicians and other health care professionals to obtain advanced approval from a health plan before a specific service or treatment is delivered to the patient to qualify for payment coverage. Prior authorization delays treatment and negatively impacts health outcomes. Combating the use of "Fail First" Policies: Often, insurers use fail first policies (also known as Step Therapy) that require patients to fail on the insurer's preferred drug(s) before a patient can take the drug originally prescribed. People living with ALS should have barrier-free access to the medications as prescribed by their providers.*

Steven Kowalski, Boston

*I was honored to help to provide care and support for my sister during her battle with ALS. My experience highlighted- something that was reinforced as I met other families facing similar challenges -that the care for individuals living with ALS falls primarily on immediate family members. There are limited specialized medical resources available for families and an acute shortage of trained individuals to support the medical and emotional needs of ALS patients. What CCALS does every day for families is amazing and very much needed. But as a lifelong resident of the Commonwealth, I would like to see our state take more of a leadership role in working with ALS-focused organizations and the medical community to provide needed programs and medical support for families facing the emotional and financial burdens of an ALS diagnosis. I promised my sister, before she passed in August 2022, that I would do everything I could to continue her powerful legacy of being an advocate and work to increase ALS awareness with the hope of positively impacting the lives of current and future ALS patients. I stand ready to help the future efforts of RDAC and others in MA to shine a bright light on the needs of an ever-growing number of families in my home state. Thank you very much for providing the opportunity for me to share my personal experiences with ALS*

Ed Hurley, West Yarmouth

*As a pediatrician practicing for over 35 years in Western Massachusetts and for the past 25 years at Baystate Children’s Hospital in Springfield, I would like to wholeheartedly support the statement of my colleague and Chair of the Massachusetts Chapter of the American Academy of Pediatrics Committee on Children with Disability, Dr. Jack Maypole. Great advances have been made for children with rare diseases since I started practice in the 1980s; unfortunately, there has been a progressive contraction of supports available to families. Fortunately, the Massachusetts Department of Public Health Division for Children & Youth with Special Health Needs (DCYSHN), Community Case Management, Family Voices, and the Federation of Children With Special Needs have bravely provided lifelines to our families. Our families of children with rare diseases are in crisis. There is a current shortage of many services in Western Massachusetts; these include Home nursing shortage, Durable Medical Equipment shortage, Limited availability of pediatric specialists with long wait times, Pediatrician shortage, Nursing shortage, Pediatric hospital bed shortage, Lack of respite services outside of eastern MA Complex prior authorization process of life-saving medication and treatments Medication cost – specifically the new biological agent's shortages of generic medication Limited supply of influenza and covid vaccines Hopefully as Mass Cares for Kids starts this January, we will be able to help comfort families, but until funding is devoted to this population we will continue to deprive families of the resources they need to thrive. This week the American Academy of Pediatrics announced a major new policy statement on Medicaid and CHIP, calling for bold changes in the program and a vision for what Medicaid and CHIP could be. I urge you to look at this resource, “Medicaid and the Children’s Health Insurance Program: Optimization to Promote Equity in Child and Young Adult Health,” in the November issue of Pediatrics as a blueprint for positive change.*

Matthew Sadof, Springfield

*For our nonprofit, we are all volunteers, and it is very difficult to dedicate the time and resources needed to properly perform outreach and address all the needs that are out there in our rare community. It would be great if we could actually have enough funds to pay for staff, and also to be able to do things like raising awareness and funds, marketing, communications, interactive website, survey capabilities, etc., so we can reach and support people! Also, in the PKU and Allied Disorder world, what is lacking is getting on the same page to share information, discuss our needs with one another and other rare disease communities and stakeholders, and identify resources that are either needed or available to us, locally and nationally. What is also lacking is accessing adequate health care coverage for our essential treatments. In the PKU example, we are blessed to have the Newborn Screening Program (NBS), which successfully identifies our disorder at birth, we are in the minority of rare diseases (less than 5%) which has a treatment (we actually have a few treatments) available to us that will allow us to live as healthy a life as possible, but yet we do not have sufficient health care coverage to be able to afford and fully access our medically prescribed treatments. Thus, for PKU, we have actually lost HALF of our community to care, meaning half of our community of 16,500, about 8,000, are “lost” to care, meaning they are not under the care of a metabolic clinic being followed by professionals and are not living to their full potential (lack of executive functioning, not able to hold a job or maintain a healthy relationship, depression/mental illness and, much worse, may not survive). Here we have NBS treatments but no access to living our best lives! I do not know another rare disorder, or other non-rare disorder, that has lost HALF of its own to care. It is atrocious! We have been fighting down in DC since 2009 for proper healthcare coverage, and we still do not have it. It is truly frustrating, time-consuming, and counterproductive. What good are treatments and cures if we do not have people well enough in the clinic to hear about them, participate in clinical trials, and benefit from these advances? First things first! Legislators need to intervene to ensure we have proper private and public health care coverage so that the rare disease community can live its best life according to the medical advances we have out there (such as NBS and proven treatments). Thank you for the opportunity to comment, and please do not hesitate to reach out to me so that we may continue this important discussion and work together to define and implement ways to improve the lives of those living with rare disorders in the Commonwealth of Massachusetts (and beyond) as Massachusetts has always been a leader in this regard (e.g., first state to implement the Newborn Screening Program (NBS) and leader in healthcare and biotech industry)!*

Denise Queally, Chilmark