NRD-2283-RDAC-1Pager_Nov2022_v1 (1).pdf
Uploaded by: Clarence Lam
Position: FAV

RARE DISEASE ADVISORY COUNCILS (RDAC)

The National Organization for Rare Disorders® (NORD®) is working to establish high-functioning Rare Disease Advisory Councils in every state through Project RDAC. Learn more below and at ***rarediseases.org/projectrdac****.*

Currently, there are twenty-four states that have enacted legislation in support of their rare disease community and proven that the RDAC can be an invaluable resource.

In 2015, the first RDAC was created in North Carolina by patients, caregivers, families, and providers. Since then, rare disease advocates and stakeholders have sought councils in other states to help better represent their communities.

What is an RDAC?

An RDAC is an advisory body that gives the rare community a stronger voice in state government. RDACs address the needs of rare disease patients and families by giving stakeholders an opportunity to raise awareness and make formal recommendations to state leaders on the most important issues they face.

What is the purpose of an RDAC?

With over 7,000 known unique rare diseases, it is difficult for state government officials to have an in-depth understanding of the rare disease community's needs. This lack of awareness contributes to the obstacles that rare disease patients and their loved ones face. RDACs can use their diverse membership and broad community support to identify barriers and propose solutions to help the rare disease community.

How are RDACs organized?

RDACs are organized differently in each state. Some of the distinguishing features include the type of entity that houses the RDAC, the composition and size of the council, and the duties and accountability requirements of the council.

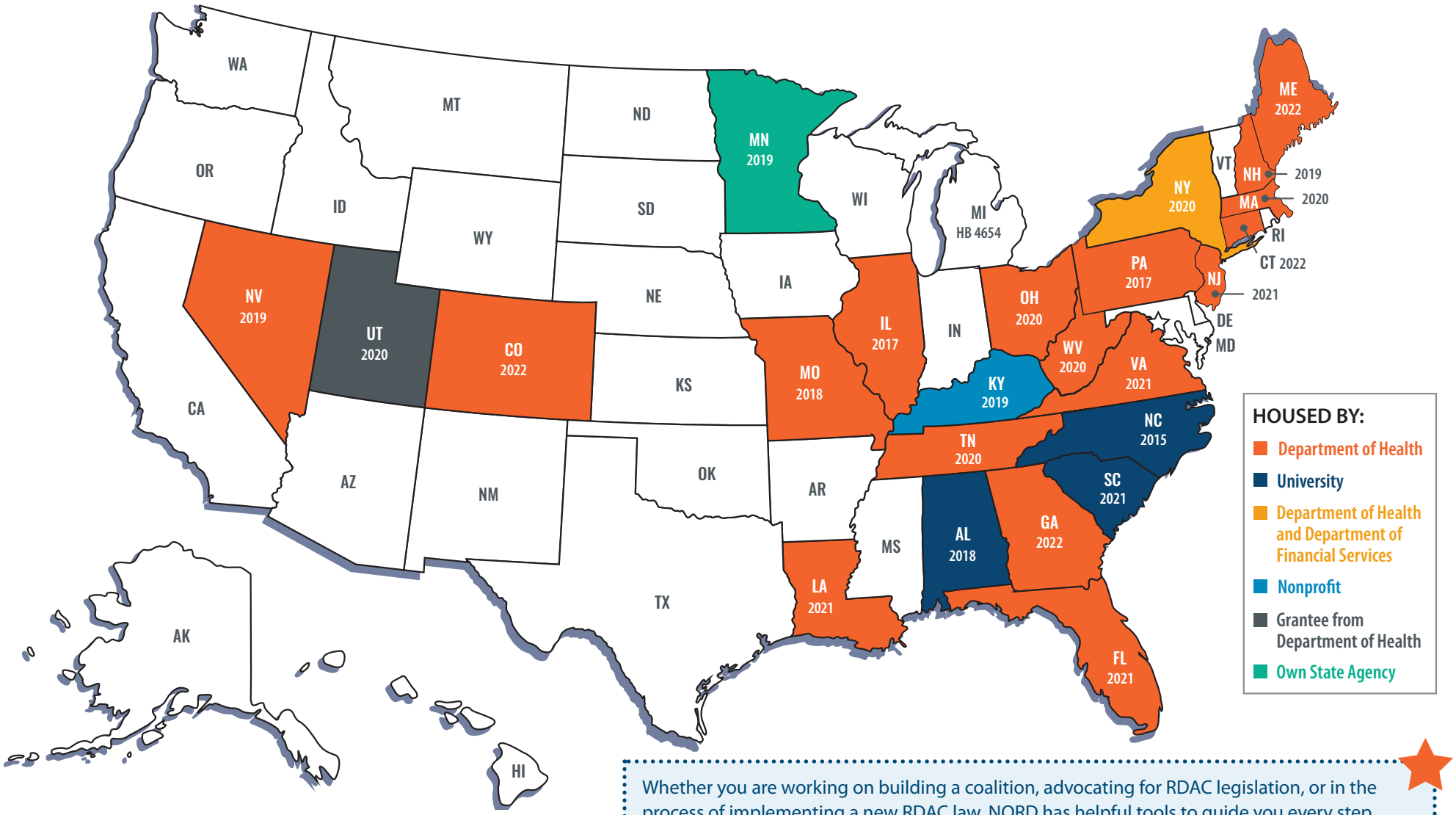
Who serves on an RDAC?

RDAC members typically include a variety of rare disease stakeholders, including patients, caregivers, health care providers, health insurers, biotech industry, researchers, patient advocacy organizations, and state government officials.

How do RDACs help the rare disease community?

RDAC responsibilities vary by state and should be tailored to meet state needs. RDACs help their local rare disease community in a variety of ways, including by conducting surveys to better understand common challenges rare disease patients or caregivers face, consulting with experts to improve access to and quality of care, and publishing a list of rare disease resources available in the state on their website.

STATES THAT HAVE ENACTED RDACS (AS OF NOVEMBER 2022)



Whether you are working on building a coalition, advocating for RDAC legislation, or in the process of implementing a new RDAC law, NORD has helpful tools to guide you every step of the way. Visit **rarediseases.org/project-rdac-resources** for toolkits, webinars, and more!

For more information on Project RDAC, please contact RDAC@rarediseases.org.

NORD: Fighting for the rare community every day for nearly 40 years. NORD is committed to the identification, treatment and cure of rare disorders through programs of education, advocacy, research and patient support services. NORD does not recommend or endorse any particular medical treatment but encourages patients to seek the advice of their clinicians. NORD is a registered 501(c)(3) charity organization. The mission of NORD's Rare Action Network® (RAN®) is to connect and empower a unified network of individuals and organizations with the tools, training, and resources to become effective advocates for rare diseases through national and state based initiatives across the United States. NRD-2283

NORD® RARE ACTION NETWORK®

SB0188_FAV_MTC_Public Health - Rare Disease Adviso
Uploaded by: Drew Vetter
Position: FAV

MARYLAND TECH COUNCIL

TO: The Honorable Melony Griffith, Chair
Members, Senate Finance Committee
The Honorable Clarence K. Lam

FROM: Andrew G. Vetter
Pamela Metz Kasemeyer
J. Steven Wise
Danna L. Kauffman
Christine K. Krone
410-244-7000

DATE: February 7, 2023

RE: **SUPPORT** – Senate Bill 188 – *Public Health – Rare Disease Advisory Council*

The Maryland Tech Council (MTC) writes in **support** of *Senate Bill 188: Public Health – Rare Disease Advisory Council*. We are a community of over 700 Maryland member companies that span the full range of the technology sector. Our vision is to propel Maryland to become the number one innovation economy for life sciences and technology in the nation. We bring our members together and build Maryland's innovation economy through advocacy, networking, and education.

Senate Bill 188 establishes a Rare Disease Advisory Council to study and make recommendations on matters relating to individuals with rare diseases in the State. We are strongly in favor of any efforts in Maryland to better understand the impact of rare diseases on Maryland residents, the needs presented to caregivers and health care providers, and the role that technology plays in addressing those challenges. MTC has numerous member companies actively engaged in researching these issues and developing solutions. We appreciate that the sponsor included seats at the table for industries engaged in this critical and lifesaving work. By bringing together this diverse group of stakeholders, we can ensure that the challenges faced by patients impacted by rare diseases are addressed in a comprehensive and coordinated manner. As a result of these efforts, we believe Maryland can be a national leader in developing solutions to the problems faced by those affected by rare diseases.

We urge a favorable report.

SB188_RareDisease_KennedyKrieger_Support.pdf
Uploaded by: Emily Arneson
Position: FAV

Kennedy Krieger Institute

DATE: **February 7, 2023** **COMMITTEE: Senate Finance**
BILL NO: **Senate Bill 188**
BILL TITLE: **Public Health - Rare Disease Advisory Council**
POSITION: **Support**

Kennedy Krieger Institute supports House Bill 188 - Public Health - Rare Disease Advisory Council

Bill Summary:
Senate Bill 188 establishes the Rare Disease Advisory Council to study and make recommendations on matters relating to individuals with rare diseases in the State.

Background:
Kennedy Krieger Institute is dedicated to improving the lives of children and young adults with developmental, behavioral, cognitive and physical challenges. Kennedy Krieger's services include inpatient, outpatient, school-based and community-based programs.

Kennedy Krieger treats more than 50 rare diseases in pediatric patients, including leukodystrophy, muscular dystrophy, Kabuki syndrome, Rett syndrome as well as other disorders that impact the nervous system. The Institute is home to several rare disease programs that also have been designated as Centers of Excellence by various organizations, such as the Center for Genetic Muscle Disorders, the Moser Center for Leukodystrophies, the Rett and Related Disorders Clinic, and the SYNGAP clinic.

Rationale:
For decades, medicine has offered little hope to patients with rare genetic diseases such as DYT1 torsion dystonia and X-linked adrenoleukodystrophy. There were too few patients with any one rare disease to be able to study it thoroughly, and it wasn't always possible to tell what disease a patient had. But this is just the sort of challenge that Dr. S. Ali Fatemi and members of Kennedy Krieger's neurogenetics research team take on.

Increased awareness about rare diseases through the proposed legislation is an effective approach to educating the broader community about the real-world experiences of patients living with rare diseases and their families. This approach will also help to educate individuals throughout Maryland about rare diseases and increase the services and resources available to them.

Kennedy Krieger Institute requests a favorable report on Senate Bill 188.

Emily Arneson – AVP Government Affairs – arneson@kennedykrieger.org or 443-631-2188
707 North Broadway Baltimore, Maryland 21205 (443) 923-9200/Telephone (443)923-9125/Facsimile

Oral_RDAC Testimony_Jeneva Stone_2.7.23.pdf
Uploaded by: Jeneva Stone
Position: FAV

SB0188: Public Health/Rare Disease Advisory Council, 2/7/23
Jeneva Stone, Rare Disease Caregiver (Oral Testimony)

My son Rob Stone, 25 years old, an artist and advocate, has an ultra-rare disease, dystonia 16. He's the only reported case in the U.S., among only 20 cases worldwide. Dystonia 16 is so rare that we spent 14 years searching for a diagnosis that is still only available through genome sequencing—a test that analyzes the more than 20,000 protein-encoding genes in DNA.

Rob also has disabilities, and while Maryland has strong supports for disabled persons, the needs of the rare community do not fully align with the disability community. Our family struggles with prior authorization for life-saving medications, with coverage for the high-dose supplements research has shown are essential to his well-being, with continuity of care, and with access to Medicaid services.

Rob was not enrolled in the Rare and Expensive Case Management program (REM) until he was 15 due to wait lists and lack of a diagnosis. There are 302 qualifying diagnoses for REM, yet there are 7,000 rare diseases—REM serves less than 1 % of the rare disease population. Furthermore, of those qualifying diagnoses, 68% end at the age of 21. Rare adults are often left out in the cold.

While I applaud Maryland's progress with newborn screening, it would not have diagnosed Rob. Nor would it my dad and sister, diagnosed with rare diseases at the ages of 70 and 40, or my hair stylist's mother, diagnosed at 60.

Rare Marylanders need access to genetic sequencing, adult services, experimental treatments and fair insurance guidelines. We need a Rare Disease Advisory Council so we may have a voice in shaping the policies that impact our lives.

Written_RDAC Testimony_Jeneva Stone_2.7.23.pdf
Uploaded by: Jeneva Stone
Position: FAV

SB0188: Public Health/Rare Disease Advisory Council, 2/7/23
Jeneva Stone, Rare Disease Caregiver (Written Testimony)

My son Rob Stone, 25 years old, an artist and advocate, has an ultra-rare disease, dystonia 16. He's the only reported case in the U.S., among only 20 cases worldwide. Dystonia 16 is so rare that we spent 14 years searching for a diagnosis that is still only available through genome sequencing—a test that analyzes the more than 20,000 protein-encoding genes in DNA.

Dystonia affects muscle contraction. Our muscles work in pairs: one contracts (flexes) and the other relaxes (extends). When someone with dystonia tries to initiative movement, both flexor and extensor muscles tense up at the same time. Dystonia can be focal (affecting only certain parts of the body), or global (affecting the entire body). It can be painful—writer's cramp is a mild, incidental form of dystonia. There are dozens of dystonias, linked either to a genetic mutation or to defined clinical symptoms.

As a result of his rare disease, Rob also has disabilities, and while Maryland has strong supports for disabled persons, the needs of the rare community do not fully align with the disability community. Our family struggles with prior authorization for life-saving medications, with coverage for the high-dose supplements research has shown are essential to his well-being, with continuity of care, and with access to Medicaid services.

Rob's throat muscles are affected, and he has life-threatening gastroesophageal reflux disease (GERD). He cannot swallow, and needs an omeprazole solution that can go through his G-tube. Keeping this prescription approved is an ongoing Medicaid nightmare. Research is being done on Rob's cells, and we have discovered that high-dose biotin, thiamine, and luteolin (vitamins and a flavonoid) stabilize him and appear to be preventing his disease from progressing. Luteolin slows and stops cell apoptosis, or cell death. Maryland Medicaid constantly erects barriers to these therapies in the form of bureaucratic harassment.

Services are also a problem. Rob was not enrolled in the Rare and Expensive Case Management program (REM) until he was 15 due to wait lists and lack of a diagnosis. There are 302 qualifying diagnoses for REM, yet there are 7,000 rare diseases—REM serves less than 1 % of the rare disease population. Furthermore, of those qualifying diagnoses, 68% end at the age of 21. Rare adults are often left out in the cold.

While I applaud Maryland's progress with newborn screening, it would not have diagnosed Rob. Nor would it my dad and sister, diagnosed with rare diseases at the ages of 70 and 40, or my hair stylist's mother, diagnosed at 60.

Rare Marylanders need access to genetic sequencing, adult services, experimental treatments and fair insurance guidelines. We need a Rare Disease Advisory Council so we may have a voice in shaping the policies that impact our lives.

SB 188_testimony_JPayne 2.7.2023.pdf
Uploaded by: Jennifer Payne
Position: FAV

In Support of SB 188: Establishing a Rare Disease Advisory Council (RDAC)

Honorable Senator Melony Griffith
3 East
Miller Senate Office Building
Annapolis, Maryland 21401

Re: Written testimony of Jennifer Payne 2/7/2023 at 1pm.

Dear Chair Griffith and Members of the Senate Finance Committee,

My name is Jennifer Payne, and I am a constituent of District 12. As an ambassador for the greater Maryland rare disease community, I thank you for the opportunity to submit my testimony in support of SB 188. Not only it is a critical imperative for Maryland leadership to engage - *as partners with rare disease patients*, for shaping policy development relevant to the unmet needs and concerns on equitable, quality, and affordable access to specialty treatment, but it is a moral imperative to identify and eliminate the inappropriate and discriminatory barriers preventing individuals diagnosed in the State of Maryland from obtaining the continuity and quality of care across all stages of life.

Every baby born in the United States is screened for the early identification of phenylketonuria]PKU] to prevent severe disability. Hence, this is my story, being among the first diagnosed with PKU [1973] in the early days of Maryland's newborn screening program. And, 50 years later [as 2023 marks my milestone birthday], I can tell you I have been a lifelong, proactive champion on medical nutrition equity for inclusion on the whole health equation - to the level of the courts. My whole health, mentally, physically, and spiritually has been sacrificed at the expense of having [and continuously fighting] singlehandedly - over the last 7 years and ongoing, - for ending discriminatory provision of health care services by insurers using PKU - and my age - as arbitrary and exclusionary criteria prohibiting access and coverage on the treatment I need to survive and thrive; – even into adulthood. My PKU story of prevention has become a story of social injustice and discrimination.

And, I am not alone. Many in the rare disease community share such common struggles and obstacles that are often 100% preventable and socially unjustifiable, and result in unnecessary suffering and disease progression. I wholeheartedly welcome support for the establishment of an RDAC to serve as a health advocate for all Marylanders, like myself, in need of representation and a special conduit to amplify vulnerable voices in health matters that have direct, and more often than not, negative implications and risk. Now is the time for us all to partner and be change ambassadors in this cultural shift that embraces diversity, equity, inclusion, and accessibility in the ways we live and the ways we care – for rare disease as part of the whole health equation. We must ensure the necessary improvements to Maryland's life saving newborn screening program stay strong. We must educate, as I have done with University of Maryland medical students - the future doctors of America - to think out of the box and equip them, the greater public, and all stakeholders – with the requisite tools to recognize, diagnose, facilitate, and optimize treatment specific to rare diseases. We can establish a published list or compendia to allow for transparency, accessibility, and education in the process. Knowledge is power, and patients are often the experts in their own rare diseases. Will you listen? We need novel

reimbursement payment models that allow coverage for specialty treatment accordingly – and equitably – and with inclusion keeping pace with innovative technology platforms. Rare disease patients only ask for a fair shake of such readily available resources, not to be targets as demonstrated by my example. The RDAC serves to empower rare disease patients as partners – from point of the research bench (informing future studies), to diagnoses, to getting the treatment into the hands of those that need it most and at the right time - for life. Until there is a cure, for change ambassadors and partners with RDAC, the story [as with my PKU story] does not end at diagnosis.

Thank you,

/s/

Jennifer Payne, PharmD, MAPP

Independent Advocate, Adult with PKU
7115 John Calvert Ct.
Elkridge, MD 21075
443-535-5322
pkupioneer@gmail.com

story-oraltesitmony.pdf
Uploaded by: Kristin Anzelc
Position: FAV

Hello Chair Griffith and the and the Senate Finance Committee. My name is Kristin Anzelc and I was born May 2, 1985 in Austin, Texas. My mom reported that I had a normal birth - much like my neurotypical Brother and Sister (I am the Middle of 3 kids). However, immediately after birth I was sent to the NICU for 6 weeks. I never really learned how to latch correctly, so had to use a special bottle (could never be breastfed). I was said to be neurologically at-risk and Failure to Thrive. I was always considered a mystery to doctors.

Fast forward to 1995 when we moved to Baltimore, Maryland for my dad's job. Due to us wanting answers we were immediately followed at Johns Hopkins. May 1996 I will forever remember this weekend. I was attending a Camp sponsored by Hopkins and it was at this Camp where I met Dr. Eric Wolfsburg and where I first heard of Kabuki Syndrome. I had many of the classic symptoms and there is only 100 Cases in the World (at that time – now it is thought that 1:32000 births are affected). From 1996-2002 I saw Dr Wolfsburg but in 2002 we stopped due to nothing new ever happening.

From 2002 - 2012, a lot happened to me medically and personally. I was still being followed for my ITP (diagnosed in 2001 and resolved in 2003). I also had 3 knee surgeries (2002, 2003, and 2005). Between 2002 and 2012, I managed to graduate from High School, attend Community College, AND Graduate from Stevenson University.

July 2012, my mom hears from someone in her Pilates class at our local gym that Johns Hopkins (Hopkins) was doing the testing on the specific genes for Kabuki now that they had been identified in 2010. We immediately got an appointment with Genetics at Hopkins. The new geneticist asked us who my Immunologist was and I had none, but my allergist did the Testing of my Immune System – and the results that came back from that testing were surprising - it showed that I had no Immune System (which was when we were followed by an Immunologist at Hopkins - which was the beginning of my CVID Diagnosis – a serious immune issue). We realized after the testing of the Immune System that is why I was constantly getting sick as a child – but no one had ever even THOUGHT to check my Immune System until this Geneticist asked us that question/ Additionally, I had the Genetic Testing done to confirm the Kabuki diagnosis and I tested positive for type 1. Additionally, Kabuki can lead to other

health issues - like heart conditions, orthopedic issues, low muscle tone and sometimes problems with communication. Those with Kabuki have mild - extreme cognitive delay (mine is more mild - my cognitive age is very similar to my biological age but some with Kabuki struggle more cognitively).

Additionally, those with Kabuki can have breathing issues. I remember in 2013, I was having issues with breathing, so my Immunologist connected me with Pulmonology at Hopkins. I had various tests done that Summer and into the Fall but all that showed us NOTHING. November 2013 I had a standard follow up with my Pulmonologist and at this point I was very close to death (due to not being able to get a deep breath and being sick for a month). From his office he admitted me to the Hospital in Critical Condition. After a few more days of testing (and me being on IV Antibiotics) my doctor made the decision to put me on Rituximab. This was a battle between my Pulmonologist and the "insurance company". Every day we were hearing that "the insurance company will not approve it" (and this is AFTER my dad had talked to the person at the Insurance Company - he negotiated the contracts for the teachers for his job AND the Insurance Company had talked to the doctor directly at that point). We found out later that it was Hopkins Pharmacists that did not want to approve it, but ultimately it was approved.

May 2015, I attended the first ever Kabuki Research Conference held at Hopkins (where I was also on the Young Adult Panel). This was my first time ever meeting others with Kabuki. 2 years later we heard that 2 adults I had met at the Kabuki Conference in 2015 had passed away due to Lung Issues. I know had I not been where I am at – there would have been a 3rd – ME. I consider myself lucky that I live about half an hour from Baltimore – and therefore close to many prestigious medical centers (Johns Hopkins and University of Maryland).

Much of the time those with Kabuki can have a shortened lifespan because of all we go through. I consider myself lucky that my dad's previous company kept agreed to allow me to have my own Insurance through them because of being a Dependent with Disabilities. Having been a Warrior from birth till now and going through multiple medical issues - I have seen how incredibly Vital Healthcare access is. Today I ask you to support a rare disease advocacy council

to support both myself and others like me with a Rare Disease. Thank you for your time.

BIO Letter of Support MD SB 188 - RDAC .pdf
Uploaded by: Laura Srebnik
Position: FAV

Biotechnology Innovation Organization
1201 New York Ave., NW
Suite 1300
Washington, DC, 20005
202-962-9200

February 6, 2023

The Honorable Melony Griffith
Chair, Senate Finance Committee
Miller Senate Office Building, 3 East Wing
11 Bladen St., Annapolis, MD 21401

Re: Testimony in Support of Senate Bill 188: Public Health - Rare Disease Advisory Council

Submitted By: The Biotechnology Innovation Organization (BIO), Washington, DC

Dear Chair Griffith and Committee Members,

The Biotechnology Innovation Organization (BIO) thanks the committee for the opportunity to comment on our support for SB 188 (Lam). This legislation to establish an advisory council on rare disease would give a strong voice to the rare disease community in Maryland.

BIO is the world's largest trade association representing biotechnology companies, academic institutions, state biotechnology centers and related organizations across the United States and in more than 30 other nations. Our members are committed to advancing science and improving the health and well-being of our planet using biotechnology.

Of the more than 7,000 known rare diseases, approximately 80% are genetic. Fifty percent of all rare diseases affect children, while 30% die before the age of 5 years.[1] Only 5% of all rare diseases have treatments available to patients.[2] Rare disease patients typically have complex conditions that come with their own unique set of challenges that they must face from testing and disease management, as well as insurance coverage difficulties.

The creation of an advisory council on rare diseases will give patients and caregivers affected by rare diseases a unified voice. These individuals will finally be provided a forum to make recommendations about pressing health care issues of rare disease

[1]National Institutes of Health, https://www.nichd.nih.gov/newsroom/resources/spotlight/020116-rare-disease-day. Accessed: December 1, 2019.
[2] https://innovation.org/about-us/commitment/research-discovery/rare-disease-numbers

patients. This advisory committee would give the state a compelling ability to improve knowledge, awareness, and management of rare diseases in Maryland, and bring together various stakeholders in the healthcare ecosystem to improve public policy regarding rare diseases. The results will be a great aid to patients, their families, and caregivers.

Thank you for the opportunity to comment on this legislation and for your support of SB 188. Please do not hesitate to contact us for any further information.

Sincerely,
/s/
Laura Srebnik
Director, State Government Affairs – Eastern Region
The Biotechnology Innovation Organization (BIO)
1201 New York Ave., NW
Suite 1300
Washington, DC 20005
206.293.1195 (mobile)

MD SB 188_RDAC_ALSA Written Letter of Support.pdf
Uploaded by: Lindsay Gill
Position: FAV

February 6, 2023

The Honorable Melony Griffith
Chair, Senate Finance
Room 3 East Wing, Miller Senate Office Building,
11 Bladen Street, Annapolis, MD 21401-1991

Re: S.B.0188: Public Health - Rare Disease Advisory Council

Dear Chairwoman Griffith,

On behalf of The ALS Association and the roughly 224 families we serve in Maryland, we urge you to swiftly move SB 188 through your committee. SB 188 establishes a rare disease advisory council (RDAC) within the state, which if passed, would help to give a voice to the 1-in-10 individuals living with a rare disease in the state of Maryland.

Any conditions that affect fewer than 200,000 Americans are considered rare. Rare diseases are present across a broad spectrum of medical conditions, including amyotrophic lateral sclerosis (ALS). ALS is a fatal progressive neurodegenerative disease that slowly robs a person's ability to walk, talk, eat, and eventually breathe. There is currently no cure or significantly effective treatment options for ALS. But, like other rare diseases, ALS patients face many unique challenges every day, from obtaining an accurate diagnosis and accessing medical specialists with knowledge of their condition to battling for fair insurance coverage for their treatment and care.

However, due to small patient populations and the large variety of rare diseases, it can be difficult for state governments to have an in-depth understanding of the rare disease community's needs. This lack of awareness often contributes to the obstacles faced by rare disease patients and their loved ones. Creating an RDAC in Maryland will raise awareness and give rare disease patients a unified voice in Maryland state government. Additionally, the RDAC will be a valuable advisory body to elected officials and other state leaders on rare disease research, beneficiary access to treatments, and best practices for the care of those with rare diseases.

The RDAC represents enormous value to our organization and the community we serve by allowing them to hear directly from a diverse group of stakeholders interested in identifying and solving pressing challenges. In addition, the RDAC would help relieve some of the burden from the state by expeditiously delivering direct feedback, solutions, and resources with one community voice.

In creating this council, Maryland would join twenty-four other states that have already enacted similar legislation in support of their rare disease community and proven that the RDAC can be an invaluable resource. Those states include Alabama, Colorado, Connecticut, Florida, Georgia, Illinois, Kentucky, Louisiana, Maine, Massachusetts, Minnesota, Missouri, Nevada, New Hampshire, New Jersey, North Carolina, Ohio, Pennsylvania, South Carolina, Tennessee, Utah, Virginia, and West Virginia.

Once again, on behalf of The ALS Association and people living with ALS and their families in the state of Maryland we thank you for considering SB 188 that would enable the creation of a Rare Disease Advisory Council. Please support this legislation to give a voice to Maryland residents living with rare diseases.

For any questions, please feel free to contact Lindsay Gill with The ALS Association via email at Lindsay.gill@als.org Thank you for your consideration.

Sincerely,

Lindsay Gill
Managing Director, Advocacy
The ALS Association
30 W Gude Dr, Suite 100
Rockville, MD 20850
Lindsay.gill@als.org

SB 188 - Public Health - Rare Disease Advisory Cou
Uploaded by: Mary Morlino
Position: FAV

SB 188 - Public Health - Rare Disease Advisory Council
Sponsored by:
Senators Lam, Rosapepe, Salling, Muse, Carozza, Kramer, and Benson
In the Senate - Hearing 2/07 at 1:00 p.m.

Thank you for the opportunity to speak with you today.

My name is Mary Morlino. I have been advocating, volunteering and working professionally in the rare disease space for over 16 years. I am the Patient Services manager at Global Genes, a global non-profit advocacy organization for individuals and families fighting rare and genetic diseases. I am also the (FSR) Foundation for Sarcoidosis Research, Global Sarcoidosis Clinic Alliance support leader at Johns Hopkins.

Previously, I worked at the EveryLife Foundation for Rare Disease, a nonprofit, nonpartisan organization dedicated to empowering the rare disease patient community to advocate for impactful, science-driven legislation and policy that advances the equitable development of and access to lifesaving diagnoses, treatments and cures.

In 2021, I co-created MarylandRARE, a support and advocacy organization for the rare disease community of Maryland. but I need help. We (the Maryland rare disease community) need help, we need more support, more research, more champions and more action to help improve the lived experience for the Maryland Rare Disease community.

I am both motivated by and frustrated with the lack of information, research and access to effective medical treatments available for the rare disease community. I am a rare disease patient, for over 16 years I have been living with chronic Sarcoidosis, a debilitating multisystem rare disease of the immune system. In 2007, I was a very healthy and active mother of two young children.

While visiting my sister's family in CA, I unexpectedly collapsed and lost consciousness and got my first ride in an ambulance. 3 weeks and 3 different hospitals later, after extensive testing and evaluations I was told I needed a pacemaker because my AV node was severed, the electrical system of my heart was broken. No explanation, no diagnosis, but a solution. I recovered and restarted my life with Vivian. I named my pacemaker Vivian, her name means ‘full of life’.

Fast forward about 6 years later, My heart function was starting to rapidly decline. My cardiologist began preparing me for a heart transplant which included visiting all the transplant teams within 4 hours from where I was living. I’m wearing an external

defibrillator vest 24/7. Things are pretty scary. After another test, a lung biopsy, I finally got a diagnosis, Cardiac Sarcoidosis. Within 1 week I was upgraded to an ICD - pacemaker/defibrillator. Vivian II.

6 months later I was about to drive my 15 year old daughter to her friend's house and I had a cardiac arrest in my kitchen. I was shocked, literally. I came back, I came back to my daughter screaming ‘Mom!”. Had it not been for the diagnosis, I would not have gotten the upgraded ICD and I would have died for good in my kitchen that day.

Had I been diagnosed and received effective treatment in 2007, or even 2008, this disease would not have had 7 years to spread throughout my body and do irreversible damage to my heart and other organs. I am rare, but I am not unique in that there are hundreds of thousands of Maryland constituents living with rare and undiagnosed diseases, facing medical financial bankruptcy, devastating families, and increasing the economic burden of our communities.

This is why we need a rare disease advocacy council in Maryland. We need a task force, a team, a council who works together to improve the time to diagnosis, who works to increase research opportunities, who works collaboratively, efficiently and effectively to improve access to treatments as well improving the lived experience for the rare disease community of Maryland.

I respectfully ask all of you to support Senate Bill 188, and to champion the establishment of the Rare Disease Advocacy Council for the betterment of the Maryland Community.

Thank you for your time, and your attention.

SB 188 testimony.pdf
Uploaded by: Miriam Blitzer
Position: FWA

UNIVERSITY of MARYLAND
SCHOOL OF MEDICINE

Department of Pediatrics
Division of Human Genetics

Administrative Office
655 W. Baltimore Street, Rm. 7-037
Baltimore, MD 21201-1559
Phone: (410) 706-3480 Fax: (410) 706-6105

February 6, 2023

Re: **SB 0188**
Public Health – Rare Disease Advisory Council

Chair Griffith, Vice Chair Klausmeier, and Members of the Committee:

I am writing in support of SB0188 *with amendments*. For the record, this testimony is my own and does not reflect my institution. For background, I am a board-certified clinical biochemical geneticist and professor at the University of Maryland School of Medicine. I was on the Maryland State Advisory Council on Hereditary and Congenital Disorders from 2004-2013, serving as chair from 2010-2013.

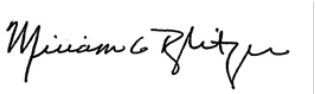
I support the creation of a Rare Disease Advisory Council (RDAC) for the state of Maryland. However, SB0188, as written, requires some amendments and clarification.

Specific concerns:

- 13-4803 (A) (5) – should be deleted. All aspects of newborn screening, an essential public health service, are addressed by COMAR 13-101 to 13-110, which established by statute the Council on Hereditary and Congenital Disorders with mandated composition and responsibility to review conditions screened in newborns, methods, and the newborn screening follow- up program. The proposed RDAC does not need to duplicate this activity.
- The composition of the RDAC is confusing as written. We recommend that this committee
  - specify one representative from each of the two academic medical centers in Maryland: Johns Hopkins University and University of Maryland. It is not clear whether the additional geneticist, physician, and scientist are separate or can be embodied in one individual (easily feasible in our State).
  - specify that the representative from the academic medical center(s) be a board-certified clinical geneticist.
- Council on Hereditary and Congenital Disorders is also mandated to have a Senator and Delegate as members. From my involvement with this Council, it has been challenging to have a Senator or Delegate participate consistently. We are concerned about creating another with these requirements (although participation in both activities would be wonderful).

I would be pleased to work with the bill sponsors to modify this bill or share my thoughts.

Sincerely,



Miriam G. Blitzer, PhD, FACMG
Professor of Pediatrics; Obstetrics; Gynecology & Reproductive Sciences,
Biochemistry & Molecular Biology, and Pathology
Division of Human Genetics, Pediatrics

5 - SB 188 - FIN - MDH - LOI.docx (1) (1).pdf
Uploaded by: State of Maryland (MD)
Position: INFO

February 7, 2023

The Honorable Melony Griffith
Chair, Finance Committee
3 East Miller Senate Office Building
Annapolis, Maryland 21401

RE: SB 188 - Public Health - Rare Disease Advisory Council – Letter of Information

Dear Chair Griffith and Committee Members:

The Maryland Department of Health (MDH) respectfully submits this letter of information for Senate Bill (SB) 188 - Public Health - Rare Disease Advisory Council. SB 188 establishes the Rare Disease Advisory Council to study and make recommendations on matters relating to individuals with rare diseases in the State. This bill requires the Council to maintain a website, develop and distribute educational material, operationalize the procurement and distribute the Council's funds, and submit an annual report. MDH must provide staff for the Council.

SB 188 requires the Council to "evaluate and make recommendations to implement necessary improvements to state newborn screening programs." MDH notes that this requirement is duplicative of the requirements of the established State Advisory Council on Hereditary and Congenital Disorders in Maryland, as stated in Health-General §13–108 (2).[1] This advisory council is staffed by the Newborn Screening-Follow up program in the MDH Prevention and Health Promotion Administration/Maternal and Child Health Bureau/Office for Genetics and People with Special Health Care Needs. An example of an implemented recommendation from this Council includes the addition of Spinal Muscular Atrophy (SMA) screening to the Maryland newborn screening panel. In March 2018 the Advisory Council recommended adding this item and the recommendation was implemented on May 30, 2019. To date, 15 babies have been identified with SMA in the newborn period.

A rare disease is defined as a disease or condition that impacts less than 200,000 people in the US.[2] According to the National Institutes of Health, there are approximately 7,000 known rare diseases, and collectively in the US, approximately 30 million people have a rare disease.[3] Examples of rare diseases include Thyroid Eye Disease and Gallbladder Cancer. Currently, there is no office in MDH that provides information on rare diseases. While many of the 50+ newborn

[1] Health-General §13–108. https://mgaleg.maryland.gov/mgawebsite/Laws/StatuteText?article=ghg&section=13-108&enactments=False&archived=False
[2] Orphan Drug Act. Public Law 97-414- Jan. 4 , 1983. https://www.fda.gov/media/99546/download
[3] NIH. About GARD. https://rarediseases.info.nih.gov/about. Accessed 24 January 2023.

screening metabolic diseases are rare diseases, they make up less than one percent of the total number of rare diseases. Rare diseases affect both pediatric and adult populations.

If you would like further information please contact Megan Peters, Acting Director of Governmental Affairs at megan.peters@maryland.gov or (410) 260-3190.

Sincerely,

Laura Herrera Scott, M.D., M.P.H.
Acting Secretary