

Dear House Energy and Commerce Committee members,

Polycystic Kidney Disease (PKD) took my life by storm nearly 15 years ago when a week long hospital stay due to a severe kidney infection led to it's diagnosis. These past years I have lived my life with the knowledge that kidney failure is very likely in my future, that I might have inadvertently passed this disease onto one, or both, of my daughters, and that there is absolutely nothing anyone can do to stop this from happening.

In response to my frustration, I have made it my mission to do whatever I can to help improve not only my own quality of life, but that of the entire PKD community by participating in as many research programs as I can. I believe that research is our ticket to a better life for all us affected by this dreadful disease. Despite the fact that I live in Memphis TN, I have traveled to Emory in Atlanta, GA and the University of Maryland in Baltimore at my own expense over the course of a 12 year period in order to participate in both observational studies and clinical drug trials. The researchers whom I've met are caring individuals who are not only accessible to their patients, but work extremely hard at trying to find pathways to a treatment for us. They are so close to discovery, but will need much more help than they currently receive to continue to bridge the gap of knowledge and discovery in order to make treatments for PKD a reality.

All we ask is that you please help us find a way to live healthy 'normal' lives. Thank you for considering my plea to work with us to improve the quality of life for the thousands of Americans battling PKD.

Karyn [REDACTED]

[REDACTED]
[REDACTED]
[REDACTED]
[REDACTED]

*Please help make a difference in the lives of PKD sufferers.
Visit www.pkdcure.org for details. Thank you.*

Dear Chairman Upton and Congresswoman DeGette:

I am writing to express my support for the proposed “21st Century Cures” bill. It is my understanding you are seeking input from outside sources on how to promote “discovery, development, and delivery that promises new treatments to patients.” Through my own health experiences I have developed ideas that I respectfully request be considered for this bill. I would like to advocate for increased funding for research related to food and the impact it has on certain diseases. My request for greater funding is based on the following observations:

- · My Brother and I, like 1.4 million other Americans, suffer from Inflammatory Bowel Disease (IBD). After trying different medications and seeing little change in our blood work or symptoms, we looked for options other than medication. Our doctor suggested we explore the possibility of a diet called the Specific Carbohydrate Diet.1[1]
- · I believe that this diet has had a positive impact on the progression of my disease. In my quest to discover if other IBD patients have had similar experiences with dietary changes, I created a survey on my website, www.scdresearch.com. The website has over 8,000 hits and 170 people have completed the survey to date. The responses overwhelmingly suggest that diet has an impact on IBD disease activity.
- · Most doctors recommend expensive drugs with dangerous side effects to treat IBD. Unfortunately, due to the lack of research regarding diet, most in the medical community do not consider dietary change to be a viable treatment option.
- · I am not aware of any NIH funded studies related to food and IBD.
- · I know of two small studies2[2] that have been conducted in the United States on the relationship between diet and IBD, both of which have shown that changes in diet may be used as an alternative therapy to treat IBD.
- · Several countries are currently working on larger studies on the relationship between diet and IBD.

The focus of the 21st Century Cures bill seems to be on funding for research and development of medications. I would like to recommend that you include language supporting increased funding for NIH to conduct clinical research on whether dietary changes can be an alternative therapy for certain diseases. I would appreciate the opportunity to talk to you for a few minutes about my ideas. I will be in Washington DC on the 24th and 25th of July. Thank you,

Kate [REDACTED]
[REDACTED]

1 The Specific Carbohydrate diet is based on the research of Elaine Gottschall. You can find more information about the diet in her publication called, “Breaking the Vicious Cycle.”

2 University of Massachusetts study on the IBD-AID diet and The Seattle Children’s Hospital study on the Specific Carbohydrate Diet and its relationship with IBD.

I am writing because my son and I both have **Polycystic Kidney Disease** an incurable genetic disease. As you may be well aware, there is currently no treatment to slow or stop the growth of kidney cysts that plague generations of families suffering from **polycystic kidney disease (PKD)**. The only remedies for PKD patients once their kidneys fail are dialysis and transplantation. While these options are life-saving, having a treatment that preserves healthy kidney function is the best option.

Kate [REDACTED]
[REDACTED]

My journey through Polycystic Kidney Disease has been basically a “Don’t ask, don’t tell” adventure. Until the Affordable Care Act ObamaCare, I feared that my disease would be found out and that I would not be able to get individual insurance until age 65 and on Medicare. I would have naturally sought help sooner if there had been any kind of treatment available, but there isn’t. The only thing to do is wait until my kidneys fail and then with go on dialysis or get a kidney transplant.....both options are expensive for the government. Wouldn’t it be better to find a treatment so my kidneys could have functioned for the rest of my life? Dialysis is no way to live and kidneys for transplant are scarce can be hard to come by.

This disease is rampant in my family. Now I have to worry about my kids and grandkids. I pray that there will be a treatment for them.

Sincerely,

Kathleen [REDACTED]

Please continue to support Arthritis research and drug development. Also educate doctors for earlier diagnosis of this debilitating disease.

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Kathy [REDACTED]

Hello

My name is Kathy [REDACTED] I am a 55 year old woman with inherited polycystic kidney and liver disease (PKD/PLD). This disease was passed on to me via my mother (she had it) and her side of the family (her sister, brother and mother had it). All of my relatives died in their early 60's from the disease. The exception was my mother; she was able to survive on dialysis for several years before passing away at the age of 72. The impact of this disease on my life is enormous. My liver is at least 4 times the size of a normal liver. While my liver function is normal for now, the size of this organ impacts the rest of my body. For example, the enlarged liver puts pressure on my chest making it difficult to breath properly, puts pressure on my stomach causing acid reflux and stomach upset, and puts added strain on my back. My center of gravity is compromised and I have to be very careful how I walk, where I walk etc so that I do not fall (it happens!). My two kidneys are also affected - the right kidney is so large that it cannot be visualized by ctscan anymore. The kidneys are 2 to 3 times the size of normal kidneys and their function is in decline. I have daily pain and discomfort in my abdominal cavity as well as lower back pain from the excess bulk I carry. Several hours of continuous sleep is a luxury. There is no cure for this disease and patients such as myself face a shortened life and a compromised quality of life. Kidney transplant is available for some patients but I was told that there is no room in my abdominal cavity to implant a new kidney. When my kidneys fail, I must make the choice of life on dialysis or succumb to the disease. There is no treatment for the liver except transplant in very unique circumstances.

My husband and I had to make life decisions based on my condition. We decided not to have children to stop the generational progression of PKD/PLD in my family. It pains me to have had to make this sacrifice. I have a PhD in immunology and I had a very successful career in industry R&D but we decided to cut this short to lessen the stress of working on my body. We both became ardent savers so that we would have money when I could no longer work. I was fortunate enough to be able to take an early retirement package from my company but of course our earning and saving potential has changed.

I have been making cash donations to the PKD foundation since the mid-1990's and I will continue to do so in the hopes that the money will be used to help find a cure for this disease. There are no drugs to slow or stop the growth of cysts on the kidneys or liver, there are no drugs to reverse the effects of cysts on these organs. The only treatments available are transplant for the kidney or liver or dialysis for the kidney. Organ transplants are fraught with issues from availability to rejection and dialysis is expensive and very stressful to the patient and the family.

As a scientist I know that federal funding of biological research in the US has been severely cut due to the economy. Private funding of research is also on a tight leash. Viable research proposals that would have been funded several years ago are no longer getting any money. Only the very biggest initiatives are getting funded. Scientists are leaving their fields since they cannot support themselves or their families. The state of science and biological research in the US is poor and this has to be reversed in order to find cures or better treatment options for diseases like PKD/PLD. I do not have a solution to this problem but I urge Congress to look at ways that research collaborations are being hampered by regulations; can this be alleviated? I also urge Congress to take a careful look at how money is spent in government research programs - eliminate the pet projects and funnel the money to real problems. I also urge Congress to encourage the development of centers of excellence for research and treatment of different diseases - how can this be made financially viable and attractive at local levels?

Let me end this letter by stating that I consider myself a very lucky woman. I have a loving and devoted husband and loving family and friends. For all of my physical problems I enjoy life and look forward to

every day. Being born and raised in the US has given me advantages and opportunities that I know I would never have had elsewhere. I truly hope that the cures initiative is successful.

Regards

Kathy [REDACTED]

[REDACTED]

NEWS Flash... Aduro BioTech, Inc. has received a “breakthrough designation” after positive clinical evidence in the treatment of pancreatic cancer. A breakthrough designation is reserved for drugs that would treat a serious or life threatening condition and preliminary clinical evidence shows great potential for improvement over available therapies, the FDA states. The San Francisco Times reported that the FDA's action could result in drugs being approved in as soon as 60-days, but it does not guarantee approval of the therapy.

If I had cancer instead of a devastating Myalgic Encephalomyelitis/Chronic Fatigue Syndrome (ME/CFS), the FDA would be willing to let me have treatment.

Drugs with risks of fatal autoimmune (Yervoy) and other extreme adverse events are perfectly justified if I might live a month longer – yet being sick for more than two decades and unable to participate in life from a disease that costs this nation more than \$22 billion a year warrants nothing! There are no approved therapies for ME/CFS.

Ampligen is the only treatment that has positively shown to help those with ME/CFS – and has been provided to patients via an open label trial for more than a decade – clinical trials covering 90,000 doses. It is deemed safe for approval by the top experts in the field and by the FDA advisory committee (Dec. 2012), yet the FDA continues to deny patients the opportunity for treatment.

Why? because they say they are unsure of its efficacy although they admitted after denying approval that they did not understand the disease.

THERE IS NO JUSTIFICATION FOR FAILURE TO PROVIDE TREATMENT.

FDA has the power to approve Ampligen with conditions. FDA is to protect public health not deny it. Give us the right to choose our care. We want our lives back. This isn't a game – it's the lives of more than 1 million Americans.

Sincerely, Kati [REDACTED]

Sick for the last 6years

June 5, 2014

Energy and Commerce Committee
United States House of Representatives
Chairman Fred Upton

Dear Chairman Upton and Rep. DeGette:

I am writing to express my concern about the current gap in our health care system to access treatment for PKU. I am a mother of a one year little boy with PKU. PKU has been successfully treated in the United States far more than 50 years, yet many children and adults cannot access the treatment needed to manage the disorder. We must ensure that everyone with PKU has access to the treatment they need for this rare genetic disorder.

Every baby born in the United States is screened for the early identification of PKU as a public health activity to prevent severe disability; the untreated effects of PKU can be devastating. The treatment for PKU includes the daily use of medical foods and foods modified to be low in protein that must be continued for life. However, this treatment is out-of-reach for most patients with PKU because of lack of insurance coverage. Providing coverage for medical foods for the treatment of PKU is medically supported, cost-effective, and the right thing to do. I am writing to ask you to pass H.R. 3665, the Medical Foods Equity Act, so that federal health programs provide medical foods coverage for the treatment of Phenylketonuria (PKU). This will be a significant step forward in closing the gap in coverage.

- Medical evidence has demonstrated the safety and efficacy of medical foods as treatment for PKU for more than 50 years. Just recently, the American College of Medical Genetics and Genomics issued the first-ever treatment guidelines for PKU that confirms the necessity of medical foods treatment for PKU for life.
- The impact of this lack of coverage on patients with PKU is disastrous and expensive. The average family cannot afford to pay for medical foods without insurance coverage. This includes my family.
- The long-term costs to the government for the care of untreated children and adults with PKU far exceed the cost of providing this essential treatment.

Decades ago, before the implementation of newborn screening and treatment with medical foods, children with PKU were doomed to a life of intellectual disability and costly institutionalization. Now, because of mandatory newborn screening and the proven treatment with medical foods, children and adults with PKU can lead normal and healthy lives. Don't put these lives at risk.

Please ensure that medical foods for the treatment of PKU are provided by the federal health programs and pass H.R. 3665, the Medical Foods Equity Act, so that everyone with PKU can grow up and become healthy and productive citizens of this country.

Sincerely,

Katie [REDACTED]
[REDACTED]
[REDACTED]
[REDACTED]

To Whom It May Concern:

I am writing in regards to the "21st Century Cures: A Call to Action". My daughter is 10 years old and was diagnosed with juvenile rheumatoid arthritis 2 years ago. The biggest struggles for our family have been access to medications that could help her to get better. She has been denied access to certain medications because the FDA has not approved their use in children due to lack of studies. Hundreds of thousands of children struggle with this illness and, therefore more studies need to be conducted to determine the effectiveness of medication for both adults AND children. Additionally, many of the medications she takes are prohibitively expensive. Fortunately, our insurance company covers the cost of many of these drugs, but I can imagine how difficult it must be for the uninsured to gain access to these necessary medications. It is my hope that more research can be done to find suitable medications for children that will be approved by the FDA and covered by insurance and that the cost of these medications is addressed in order to allow all those who need them to have access to them. Thank you for your time.

Kelly [REDACTED]
[REDACTED]
[REDACTED]

Dear Energy and Commerce Committee,

I am a mother of a child who has PKU or Phenylketonuria. I am a teacher and my husband is a state trooper. When we found out our son was born with PKU we were devastated. What is even more frightening though is how we receive no help purchasing his medical food. Parents have asked me, do you really believe he has this diet? Isn't it just like a Gluten Free diet that parents choose! The answer is a big NO! My son has to eat the medical food that is not available in most stores because if he doesn't it will have severe health issues. The financial burden that most families go through to purchase medical food is outstanding. I spend approximately \$200 a month just on his medical food and he is only six. When he becomes a teenager with a healthy appetite, I do not know how much more we could afford. If most states see doing the Newborn screening for PKU as important, then why would they not see the treatment as just as important?

Please consider passing The Medical Foods Equity Act HR 3665.

Sincerely,

Kelly [REDACTED]

Hello,

Polycystic kidney disease runs in my family. My mother died of it, after years of dialysis, as did her mother before her. I am the only one of my mother's five children who does not have this disease. On April 20, 2014, my youngest sister and I celebrated the ninth anniversary of donating one of my kidneys to her. She had developed complications with both hemodialysis and peritoneal dialysis and would have died without this transplant.

I am grateful to be free from this disease that impairs and shortens lives, and to have been able to help my sister. How wonderful it would be if a treatment were available that could slow or stop the growth of cysts in people with PKD!

My understanding is that PKD has long been "underrepresented" in regard to research funding, viewed in terms of government spending per person afflicted with PKD, compared to other diseases that might be better known. I hope as part of the 21st Century Cures initiative, this inequity will be corrected. I also hope medical research in general will be given a higher priority in the allocation of limited revenue.

If you have any questions, please feel free to call me at [REDACTED]

Sincerely,

Kelton [REDACTED]

Hello,

My name is Krista [REDACTED] and I have Polycystic Kidney Disease (PKD). PKD is a hereditary disease that is life threatening and incurable. It has claimed the life of my father and the lives of several members of his family. When I was diagnosed 8 years ago at the age of 23, I was most disheartened and frustrated to learn that there was nothing I could do to prevent the progression of the disease, which can ultimately lead to kidney failure and a host of other serious complications. There was no approved medicine to stop or slow the progression of kidney cysts, and that remains true today.

Living with a progressively degenerative disease, especially one in which the full negative effects were played out by close family members around me, is an enduring struggle of mind and body. After learning that Tolvaptan, a fast-tracked drug in clinical trials shown to slow the progression of PKD, was rejected by the FDA not because it wasn't effective, but because one of the largest outcomes of the study--the slow of total kidney volume growth--was not a recognized surrogate endpoint, I was devastated.

There seems to be a hiccup in the way that science and law communicate with one other, and speed is essential when hundreds of thousands of lives are at the whim of government laws that are not as up to date as science and technology. Last year I participated in my first clinical trial and received a different kind of drug than Tolvaptan, which also has the promise to slow kidney cyst growth, and I can only hope that if and by the time it's ready to be approved, the law will be reworked and ready to meet it head on.

Thank you for all the work, research, and resources the committee is dedicating to this initiative, and thank you for taking the time to listen to my personal story.

- Krista [REDACTED]

Hello,

We have a child who lives everyday with arthritis. We have had a hip injection. Our daughter takes weekly medicines to control her arthritis which we know will become ineffective at some point. There are not a lot of treatments for children with Arthritis because eventually they all don't work. Where do the kids go then? What medicines do the kids take then? There isn't anywhere for them to go. We need to find a cure so this is a problem for future children.

Thank you,

LaRae [REDACTED]

I have been diagnosed with multi joint osteoarthritis. It is advanced for my age. (63) Whereas I am otherwise very happy with my health coverage with Kaiser Permanente, I find it incredulous that an organization dedicated to prevention will not afford me the alternative therapies that have been proven effective in alleviating the discomfort from this disease. I have already had a hip replacement. I would think a business would find it prudent to provide a more inexpensive alternative to surgery. No to mention the fact that these people are in the health/helping profession. Osteoarthritis is an aging disease. The longer we live, the more of us will have it.

Sincerely,

Laura [REDACTED]

[REDACTED]

Thank you 21st century cures committee for your consideration of Polycystic kidney disease. There is currently no treatment for PKD. Your kidneys fail and you go on dialysis or if your blessed you receive a kidney. That is a mixed blessing the cost of transplant and the many drugs which have side effects like tremors, hair loss, hair growth on face, bone loss, burst capillaries suppressed immune system etc. In 2014 I was hospitalized twice for ecoli which was very serious. Then in October I had my old diseased kidneys removed which are pictured below. The largest was the size of a football. That surgery was worse than the transplant which was not exactly a piece of cake. The removal left an incision from my breast bone down around and past the belly button. I am fortunate to be alive since my dad and grandfather both died at early age from PKD. My daughter has also been diagnosed with this in curable disease. She has had cysts rupture twice which is so painful her DR prescribed vicadin. It is too late for me, but my dream is my descendants will have a treatment that keeps the cysts from enlarging. Then they could avoid transplant and all it's side effects.

Thanks for your consideration.

Laura 

PKD Facts

Polycystic kidney disease (PKD) is one of the most common, life-threatening genetic diseases affecting thousands in America and millions worldwide.

In autosomal dominant PKD (ADPKD), fluid-filled cysts develop and enlarge in both kidneys, eventually leading to kidney failure.

PKD is the fourth leading cause of kidney failure.

More than 50 percent of people with PKD will develop kidney failure by age 50.

Once a person has kidney failure, dialysis or a transplant are the only options to treat the damage the disease has caused.

It is a painful disease that impacts quality of life.

The average size of a normal kidney is a human fist. Polycystic kidneys can get much larger, some ***** This is me! I'm 52, but I look 6 months pregnant. My kidneys are well over the size of footballs. I am forced to wear maternity clothes. These are real life effects - not marketing speak. getting as large as a football, and weighing up to 30 pounds each.

Parents have a 50 percent chance of passing the disease to each of their children. Unlike some genetic diseases, it does not skip a generation. Because it is passed from generation to generation, ***** 8 known cases in my family of 16 over 3 generations PKD often affects many people in one family.

Approximately 10 percent of the people diagnosed with PKD have no family history of the disease, with PKD developing as a spontaneous (new) mutation. Once they have it, they have a 50 percent chance of passing it on to each of their children. PKD equally affects people of all races, genders, nationalities, geographic locations and income levels.

There is no treatment or cure for PKD.

My Mom was the first known generation of PKD in our family. My child hood was spent going with her to Dr's and hospitals. There, the only " treatment" was to drain the cyst in hopes of alleviating her pain. It worked for 1 year, if she was lucky. The doctors knew next to nothing about her disease and simply treated it with a multitude of pain medication. 35+ years later - not

much has changed. I see every day people who have to teach the General Physicians about PKD to prevent the dr from doing harm.

As a second generation patient with PKD, I have suffered kidney failure, dialysis and transplant. I saw my salary and job duties cut in half due to my illness. Still, I had to consider myself lucky, as most patients are unable to work at all. Nothing short of living through it will convey to you the traumatic experience of being chronically ill, seeing it impact your entire family. Worse yet, knowing that there is a 50 / 50 chance that it impacts each and every child in our third generation and knowing that the only real advances in the disease have come through private efforts. Think about that - 1 person in the first generation was diagnosed, 4 /5 in the second generation were diagnosed and to date 3 have had transplants, now 10 in the 3rd generation - many of whom are afraid to be tested due to insurance rate hikes. Remember - this is just ONE family.

Below is an explanation of the cost of kidney failure via the testimony of Dr. Larry Melton:

Examining Reforms to Improve the Medicare Part B Drug Program for Seniors
Committee on Energy & Commerce
U.S. House of Representatives
Friday, June 28, 2013

When kidney's fail, patients only have two treatment options: dialysis or transplantation. Since 1972, Medicare has covered people with End Stage Renal Disease (ESRD) – permanent kidney failure requiring dialysis or a kidney transplant – without regard to age or SSDI status. There is no Medicare coverage limit for a dialysis patient. By contrast, kidney transplant recipients lose Medicare coverage at an arbitrary 36 months after transplant. In 1972, it was estimated that the ESRD program would cost \$250 million. Today, the program costs in excess of \$250 billion. These figures are staggering and there is no question that a functioning transplant with immunosuppressive drug coverage is vastly less expensive than the cost of dialysis. When renal allografts fail, patients again require dialysis and may even be candidates for re-transplantation, both of which would be covered by Medicare. Extending immunosuppressive coverage beyond the 36 month limit would decrease the risk of allograft failure due to patients not taking their immunosuppression.

A conservative estimate is that 20 individuals will die today awaiting a life-saving donor organ. Donor organs are a precious resource that fall far short of meeting the actual demand. As we have seen again from recent high profile media coverage organ demand far exceeds supply, as a result, the transplant community works diligently to ensure that every donor organ is given the best opportunity to save and extend life for as long as possible. Current Medicare policy inhibits our ability to that.

A variety of national and international medical journals have focused attention on the U.S. policy of limited immunosuppressive drug coverage and the kidney failure that follows. In particular the New England Journal of Medicine (NEJM) highlighted a survey conducted by the American Society of Transplantation (AST) that found that 70 percent

of U.S. kidney-transplantation programs reported that their patients had an “extremely serious” or “very serious” problem paying for immunosuppressive medications, and 68 percent reported deaths and graft losses attributable to cost related non-adherence.” The study further found, “Since patients with kidney failure need either long-term dialysis or a functioning renal allograft to survive, failing to pay for ongoing immunosuppression ensures that Medicare’s initial investment in kidney transplantation is squandered, that patients die prematurely, and the U.S. taxpayers pay for a more expensive but inferior therapy after some transplants fail unnecessarily.” At present, Medicare spends approximately \$70,000-80,000 per year on a dialysis patient, which Medicare covers indefinitely. However, Medicare on average spends less than a quarter of that cost for a kidney transplant recipient after a year of the transplant.

The link for the full testimony:

<http://democrats.energycommerce.house.gov/sites/default/files/documents/Testimony-Melton-Health-Medicare-Part-B-Reform-2013-6-27.pdf>

You can see it only makes financial sense to pay for transplantation rather dialysis. Common sense says to pay for the transplant drugs rather than forcing another kidney failure and dialysis on a patient. But why not find treatment and cures to postpone or even prevent the insane costs of this disease?

Autosomal dominant PKD is one of the most common inherited disorders. (Source: excerpt from [Polycystic Kidney Disease: NIDDK](#))

Prevalance Rate: approx 1 in 503 or 0.20% or 540,000 people in USA (source http://www.rightdiagnosis.com/a/autosomal_dominant_polycystic_kidney_disease/stats.htm)

Funding spent by NIH on PKD \$40 Million in 2013 source: http://report.nih.gov/categorical_spending.aspx

This is what I find from a few simple searches:

Number of deaths for leading causes of death:	# of deaths	# rank in deaths	# - rank in spending	Spending FY 2010 non ARRA
Heart disease:	597,689	1	2	1,329
Cancer:	574,743	2	1	5,823
Chronic lower respiratory diseases:	138,080	3		*
Stroke (cerebrovascular diseases):	129,476	4	7	337
Accidents (unintentional injuries)	120,859	5		*
Alzheimer's disease:	83,494	6	5	450
Diabetes: 69,071	69,071	7	3	1,046
Nephritis, nephrotic syndrome, and nephrosis:	50,476	8	4	552
Influenza and Pneumonia:	50,097	9	6	308+93
Intentional self-harm (suicide):	38,364	10	8	36

polycystic kidney disease

**

35

* not included in disease spending

link for data column B - FY 2010

<http://www.cdc.gov/nchs/fastats/deaths.htm>

link for data column C -

2010http://report.nih.gov/categorical_spending.aspx#legend5

** PKD is not listed as a primary cause of death. Kidney failure, heart disease, stroke, diabetes etc are listed, yet the cause is PKD

PKD is not listed as a primary cause of death, thus there is no accurate account of the impact of this disease. Kidney failure, heart disease, stroke, diabetes, etc., are listed as primary causes. Simply put, funding for PKD research, treatment and prevention will vastly impact many other mortality rates as well as health care costs.

Please,

- Increase federal funding for PKD research
- Change FDA policy for approving promising treatments and therapies
- Tackle the shortage of kidney donors
- Address inadequate Medicare coverage of immunosuppressive drugs

Laurell 

Members of the House Energy and Commerce Committee:

I am writing to you today to share my experience as a family member of [REDACTED] [REDACTED] who died of Polycystic Kidney disease in 2012, and his daughter, [REDACTED] [REDACTED] my beloved daughter-in-law, who suffers from PKD.

I met [REDACTED] in 1998 when our children became engaged. He was an accomplished university pharmacologist and later a high school science department head and admired teacher of AP science courses. Given his notable talent in the sciences, it is ironic that he suffered from a disease that science has in no way conquered. Today, as in 1998, there is an absence of real treatments to address PKD once kidneys have failed. The options are dialysis and transplantation. [REDACTED] was on dialysis in 1998. When he came to visit, he had to make advance plans for his treatments at a local university hospital, treatments that were a difficult and regular part of his life at home as he continued his full time work and professional responsibilities. Over the years we watched his energy wane and complications of PKD develop, including heart problems that necessitated complicated by-pass surgery. Ultimately one of his three children, the only one unaffected by PKD, donated a kidney to his father and [REDACTED] underwent a kidney transplant. The transplant liberated him from regular and grueling dialysis treatments, but it had massive complications of its own, many related to his regimen of antirejection medications as well as others. [REDACTED] maintained his spirit and his work until he no longer could. In failing health, he and his wife arranged for the sale of the house that he loved and a move to a community that would be ultimately more appropriate for her as a single person living alone. He died shortly after their move. He was not quite 65 years old. Had effective treatments been available to [REDACTED] early on in the manifestation of the disease, he would have had many more years to enjoy and to contribute to the well being of his community and family~his wife, children, and beautiful grandchildren.

My son's wife, [REDACTED], almost 40 years old, is a superb human being. She is an award winning contributor to her community, to myriad charitable and educational organizations, devoting much personal time, energy, creativity, warmth, and skilled leadership. She was an outstanding and admired high school teacher and is a terrific mother, wife and role model. From the time she was a young child, [REDACTED] was an athlete who became an accomplished swimmer and ultimately the captain of the Duke University Women's Swim Team. [REDACTED] maintains an active fitness regimen and a careful diet devoid of salt to do all she can to maintain her health while she can. When her kidney function deteriorates, we are hoping that there will be drugs available to her to extend her quality of life for many years, without the need for grueling dialysis and/or transplantation.

I'm writing to you to urge you in the strongest terms to do all you can to draw attention to the lack of effective PKD treatments and to work to allocate sufficient funds to foster more research and development. My husband, [REDACTED] and I are earnest and ardent supporters of the PKD Foundation, but personal contributions alone are insufficient to the task. We need your help. Thank you for your consideration.

Sincerely,

Lauren [REDACTED]

[REDACTED]

[REDACTED]

To The House Energy and Commerce Committee:

I am writing in regards to Polycystic Kidney Disease. PKD is a life threatening disease that affects from the very young to the old.

When I was 30 years old I was diagnosed with the disease, that was almost 30 years ago. I was shocked because I had not only never heard of the disease, I didn't realize it was heredity. I called my parents immediately and they had to do some asking of many family members to see if anyone in the family had ever been diagnosed with PKD. After much asking and researching they "think" my Aunt died from the disease at the age of 49, this was over 40 years ago. She had kidney failure but that was all that was said. So we came to the conclusion that it was my fathers side of the family. I insisted that all my family members get tested and well, my father had it and 5 of his 8 children had the disease as well. Four of my five brothers and myself were diagnosed. As you can imagine we were in shock, that 5 of the 8 of us would have this life threatening disease.

Fortunately when I was in for a physical I had a very astute woman doctor who told me my blood pressure was too high. She had me go to the clinic for 3 weeks, different times every day. She wanted to see if there was any changes and there sure was, it was so erratic all the readings were different and didn't matter, day or night. She then had me go for further testing on my kidneys. She explained to me that I was too young to have this erratic high blood pressure. So the results were I had Polycystic Kidney Disease. I thank God that she was my Doctor and didn't just "chock" it up to high blood pressure and put me on pills. I then went to a Kidney specialist to see how "bad" it was. I had them in both kidneys but not as bad as it could have been.

I went on blood pressure medication and immediately got off salt and changed my diet. Now I do "cheat" on my diet, but when I cook at home I do not use any table salt and I cook fresh meals, usually nothing processed and of course exercise is important.

I see my Doctor annually and have my blood pressure checked every 6 months by her, as she likes to monitor it regularly. I see my kidney doctor as well every several years now because my kidneys have stayed the same. I believe with constant monitoring of my blood pressure, my diet and exercise it has helped me and the fact that I found out so young. But I think I am one of the lucky ones, that they have not grow at an alarming rate, as some with the disease.

Unfortunately I have two brothers that have not been so lucky. They are both on dialysis and they need help. We need a treatment NOW to stop the cysts from growing, which in some people they seem to grow at a much faster pace. There are very young children with this horrible disease and I believe when they start out at such a young age they need options now!

This disease is more widespread then you can ever imagine, many people may never know they have the disease until it is way too late.

Dialysis and a Kidney Transplant (very long waiting list and finding a match) should not be their only option, because by then it is usually too late!

Thank you,

Linda [REDACTED]
[REDACTED]

I'm trying to find out why the CFC <free> version of Intal asthma inhaler is not approved for use in the United States, even tho the earlier Intal inhalers that <did> use CFC propellant was approved! Makes no sense at all.

Intal is a ridiculously safe medication to control asthma, with virtually no side effects, unlike all the other asthma medications currently approved for use here.

Pfizer was the original source for Intal, which used a CFC propellant. When CFCs were banned, they stopped making it and transferred rights to Sanofi-Aventis.

Sanofi-Aventis now sells a CFC <free> version of Intal (same medication, different propellant) which is for sale in the Commonwealth (UK, Canada, Australia, New Zealand).

I've had several email exchanges with the FDA (very quick & responsive), but they say they are not allowed to so much as tell me whether or not Sanofi-Aventis has applied for approval to sell Intal in this country. And have referred me to Sanofi-Aventis.

I assume the FDA regulations require Sanofi-Aventis to go thru the same protracted trials and crap even tho it's exactly the same medication as was previously approved.

I am so glad to see there is an effort to overhaul the process for FDA approval. I hope I live long enough to see Intal not only get approved for sale in the US, but made available OTC, as are the nasal spray and eye drops with the same active ingredient.

Thank you. Linda [REDACTED]

Dear Committee Members,

I am writing to urge you to pass H.R. 3665, the Medical Foods Equity Act, to ensure that federal health programs provide coverage for medical foods for the treatment of PKU!

My grandson, who is 15 months old, has PKU and our family is now acutely aware of the potentially devastating effect of this condition if this child, and later adult, does not eat the proper foods. He is now a healthy, happy and incredibly smart toddler and we are all very aware of how important it is now, and will be in the future, to strictly control his diet so that he remains as healthy as he is today.

The foods he is allowed to eat are very few and, as he grows, we are finding that most of the allowed foods are very expensive. It is crucial that his parents, who are both hard working adults, be able to properly care for him and not be unreasonably burdened by the very high cost of special food necessary for their child's health and well-being.

PLEASE CORRECT THE CURRENT GAP IN COVERAGE FOR MEDICAL FOODS SO THAT MY VERY SPECIAL GRANDSON -- AND ALL THE VERY SPECIAL PEOPLE IN OUR COUNTRY WITH PKU -- CAN HAVE REASONABLY AFFORDABLE ACCESS TO THE FOOD THEY NEED TO KEEP THEM HEALTHY!

Thank you so much for your consideration of this matter. We are counting on you to do the right thing.

Lisa [REDACTED]
[REDACTED]
[REDACTED]

Questions For Patients

- What is the state of discovery of cures and treatments for your disease? Are there cures and treatments now or on the horizon?

My son has Noonan Syndrome, a RASopathy. A RASopathy is a genetic disorder that affects cell growth and cell regulation on the RAS-Mapk pathway. A protein defect. Our goal is to find ways to regulate the pathway to normalcy to help cure several different diseases including Noonan Syndrome. Currently there is only one treatment for NS, Growth Hormone that can only be used in part of our population. Many in our population are restricted from taking the drug because of their history with cancer or hypertrophic cardiomyopathy.

- What programs or policies have you utilized to support and foster research, such as patient registries, public-private partnerships, and venture philanthropy?

We recently partnered with the University of Pennsylvania for the Million Dollar Bike Ride to raise funds for a RASopathies grant that any scientists in the world will be able to apply for. (Fundraising for research is hard when no one knows anyone with the disease and those affected have so many medical bills.) We have also held a scientific symposium every other year bringing together the top RASopathies researchers so they can learn and share with each other to expedite research.

- How can Congress incentivize, coordinate, and accelerate basic research for diseases we know relatively little about?

We need more funding for rare diseases. We need incentives for research institutions to share their knowledge with each other. We need incentives for drug companies to be interested in rare diseases. FDA guidelines need to be different for the rare diseases and doctors who review potential drugs should be knowledgeable in that disease state.

- How can we work together to better translate advances in science into safe and effective new therapies for patients?

We all need to communicate with each other. Find out what everyone needs and work together. We need to be able to have an open dialogue with drug companies and current rules and regulations of big pharma prevent that.

- How do you coordinate your research and outreach with other patients?

Facebook, Websites, Family Meetings that run with the Scientific Symposium

- How do you learn about new treatments and cures? How do you communicate with other patients regarding treatments and cures?

There haven't been any I've come across, so none to pass along!!

- What can we learn from your experiences with clinical trials and the drug development process?

I have no first hand experience. I'm just trying to learn all the pieces of the puzzle. The bench side to bed maze is not an easy map to follow. I have been trying to work with Global Genes on creating a roadmap for patient organizations with this exact information. It is so hard to learn the rare disease world. It is like when you get the diagnosis. You don't get 20 pamphlets, books, support groups, free yoga classes and a specialty center. You get a name of a disease and you are left by yourself to figure it out with absolutely no guidance.

- What is the role of government in your work, including any carriers to achieving your goals and advancing breakthroughs?

I live in [REDACTED] so I am a part of [REDACTED] a working group of patient advocates in [REDACTED] to raise awareness of rare disease and help support bills that will help advance research. Government is extremely important. Without the Orphan Drug Act companies would have no interest in getting involved in the rare disease space. We need the government to help encourage companies to be involved in rare diseases, to keep clinical trials in the U.S., to help fund research that will help create jobs, to change the FDA's rules to understand that we might not have good natural history studies, that we are going to have small number of patients in our studies and clinical trials, and that our endpoints might be different than studying a "normal" population.

- How should regulators evaluate benefit-risk? How do you work with regulators regarding benefit-risk? Can this process be improved?

For those patients who are dying, benefit-risk is going to be different than those diseases who are not in that circumstance. You need to understand that.

- What is the role of public and private funding in the research and development of cures and treatments?

I think the government needs to devote more money to funding research. I truly believe that the government will get their money back two fold when treatments are created.

- Are there success stories the committee can highlight and best practices we can leverage in other areas?

- How have you worked with other patients to support one another?

Yes. Always. We are better in numbers. I work with [REDACTED] [REDACTED]. We might have different diseases but we all have the same common goal.; to eliminate the challenges of rare disease. I am so glad the government is also

starting to get involved. The Rare Disease Caucus, this survey is making me hopeful that positive changes in our community are on the way!!

- What is the financial burden of your disease? How would better treatments and cures help save money for your family and the federal government?

We would be able to see less doctors. Less time away from our other family members, less time away from work. If we spent less on healthcare, we would be able to spend money on goods to increase economy.

- How can Congress help?

Fund more research!!! Create bills to incentivize the drug companies to stay interested in rare diseases. Create ways we can rare purpose drugs. Change FDA regulations to make it easier to get rare disease drugs from bench to bedside in a safe and effective manor. The rules currently in place were written for the norm, not those with rare disease.

I really appreciate you taking the time to hear what we have to say. I hope in the future we will have more cures for the 7,000 rare disease that are out there. This is a good step in the right direction. Thank you!!!

Warm Regards,

Lisa [REDACTED]
[REDACTED]
[REDACTED]
[REDACTED]
[REDACTED]

I am a 50 year old female with polycystic kidney disease. I am in the 4th stage of failure. 16 to 16% function of my kidneys. This disease has affected my whole family. My Father, Brother, Sister, Daughter, and I have Polycystic kidney disease. I am writing to you to let you know there is no cure for this disease. We need to work on a cure fast. The only option I have is dialysis or transplant. Parents have a 50 percent chance of passing the disease to each of their children. It is a painful disease that impacts quality of life. Polycystic Kidneys can get as big as a football and weighing up to 30 pounds each.

There is no treatment or cure.

Please help

Lori [REDACTED]
[REDACTED]
[REDACTED]

Thank You

[REDACTED]

Hello my name is Lori [REDACTED] on February 7 2011 at 1:54 p.m. my life changed i gave birth to my beautiful baby girl she weighed 6 pounds 10oz her name [REDACTED] her heel prick tests came out normal it wasn't until she was a month that we started noticing she was having milk intolerance we were referred to a gastroenterologist by the time she was 4 months she first contracted RSV at 10 weeks old she had pneumonia by the time she was 5 months the problems continued she has had numerous hospitalization starting from around three months. Failure to thrive was her first diagnosis she had fallen off the growth chart, then Feb 15th 2012 she was diagnosed with Cystic Fibrosis by Dr Macias. May 30th 2012 I was told by infectious disease dr to make arrangements having contracted Pseudomonas, Pneumonia, Mycoplasmosis, and the flu all at once she had been in the ICU for quite a while had a port placed to receive antibiotics IV at home. There isnt a worse feeling then watching helplessly your child that you love more than anything in the world suffer fight for every breath. By the grace of God she pulled through my daughter sees many specialists 9 different doctors to be exact she has been called a puzzle that could not be put together she has been diagnosed with Autism, Chiari Malformation, Tracheomalacia, Gastric Reflux, Ezcema, low B cells, Aspiration, and most recently diagnosis is Syngap1 a rare chromosome disorder that only affects around 41 children [REDACTED] strand is the first of its kind, mental retardation. She has been evaluated by The Meyer Center Texas Childrens Hospital social and speech at age of 7 months, fine motor skills 11 months old, and gross motor at 15 months old she they contribute her Dysmorphic features to genetics also. The WES was done ordered by Dr. [REDACTED] who discovered the synap. My daughter has many milestones she is almost always on antibiotics and sick yet she is able to smile she is able to laugh she is able to love she has the most beautiful spirit. I am torn to see her struggle i wish i could take her place but i cannot I can only speak for her because she cannot. She deserves so much better she is terrified when she goes to any doctor because of all the tests treatment and bloodwork she is currently receiving therapy and will soon start ABA therapy which her insurance does not cover and we have to pay 100% out of pocket. But we will do anything to help her. Most parents have hopes and dreams for their precious children my hopes and dreams went from all the normal to finding a cure and to create a better life for her She has been through more then anyone should ever have to suffer through and can still smile even when she is weak that is worth fighting for. I would like to add that vaccinations to not help her she still gets the flu and rsv even after vaccines she cannot go very many places because she picks up llnesses so easily. Thank you for your time if you have any questions please feel free to contact me please help my child and all the children who were born innocent and inflicted with such problems.

- Lori [REDACTED]

To whom it may concern,

I am writing to raise government awareness of the lack of funding and research to treat and/or cure kidney disease in our country. I would like to share with you our family's story about living with Polycystic Kidney Disease (PKD). PKD is an inherited genetic disease. There is currently no treatment to slow or stop the growth of the kidney cysts that plague generations of families suffering from polycystic kidney disease (PKD). The only remedies for PKD patients once their kidneys fail are dialysis and transplantation. While these options are life-saving, having a treatment that preserves healthy kidney function is the best option. Some information about PKD:

- PKD affects both children and adults, with a prevalence of AD(Autosomal Dominant)PKD of 1 in 1,000 persons among children and adults in the U.S. and a prevalence of AR(Autosomal Recessive)PKD of 1 in 10,000 neonates and children
- ADPKD is the fourth leading cause of renal failure, accounting for 3% to 4% of all end stage renal disease, and is associated with a perinatal mortality rate of 43%
- From 30% to 50% of neonates with ARPKD die shortly after birth due to sepsis and respiratory failure

My mother suffered renal failure in her early 50s due to Polycystic Kidney Disease. She was fortunate enough to have a cadaver donor save her life. She is dependent on multiple medicines daily to discourage rejection of her transplant. She had to give up her job and now is on disability. She loved being out and active in her community and now has developed other health issues due to her compromised immune system; including breast and skin cancer.

I also suffer from PKD. My cysts continue to grow and I am exposed daily to the risk of having cysts burst during my very active job of working with children on the autism spectrum and other developmental disabilities. I love my job and hate that I must be on guard at all times. I also take several medications to treat high blood pressure, painful swelling in my ankles, prevent repeated kidney stones and attempt to make my kidneys function as long as possible so that I can continue to live a full life.

My 17 year old son suffers from PKD as well. It was discovered when he was elbowed in the back while playing basketball; resulting in severe flank pain and blood in his urine. He has already developed high blood pressure due to this disease and is prevented from playing any contact sports due to previous encounters during play with peers that have cause his cysts to burst. He was passionate about football, soccer, and basketball. He now suffers from depression at not being able to engage with peers in these activities and watch from the sidelines.

Taking into account our personal story; which not only effects our lives on a very personal level, but countless other generations of families...the money our country is spending on medications, treatments, transplants, associated medical complications and disability checks is astronomical.

An effective treatment to halt the growth of the cysts or cure of this deadly disease is in everybody's best interest for the health and financial destruction it has on families and our healthcare system.

Please advocate for research to help our families destroy this deadly disease,

With appreciation and hope,

Loraine [REDACTED]

[REDACTED]

[REDACTED]

[REDACTED]

Dear Chairman Upton and Committee Members,

Thank you very much for looking into ways to speed the process of drug development. We have a debilitating genetic disease in our family, Friedreich's Ataxia, that affects our son and greatly reduces his prospects for the future. Hope for a cure is what keeps him going.

We have seen many exciting developments in the 4 years since he was first diagnosed, but the length of time from discovery to FDA drug approval - 10 to 15 years - means he would be in a wheelchair, if not dead by the time any of these developments were approved.

In answer to some of the questions on your patient input questionnaire:

*What is the state of discovery of cures and treatments for your disease? Are these cures and treatments now or on the horizon?

Very promising genetic experiments have happened on mice in France that have completely reversed the damage of FA and allowed the mice to live a normal (mouse) lifespan. In England there are some promising drug trials using a basic, cheaply and widely available vitamin B3.

In Massachusetts, our geneticist has been waiting for 2 years for approval to begin an experiment on human cells, I don't know why it takes so long or whether it takes this long in other countries too.

*What programs or policies have you utilized to support and foster research, such as patient registries, public-private partnerships, and venture philanthropy?

We are involved with the Friedreich's Ataxia Research Association (FARA) Our son is registered in their patient registry and is involved in 2 studies. We also actively raise funds for FARA.

*How should regulators evaluate benefit-risk? I would greatly appreciate regulators and regulations allowing patients and their families to assume more risk in trials.

*What is the financial burden of your disease? Even though he is very intelligent and we have the resources to send him to college, it is unlikely our son will be able to work for long, if at all. His genetic disorder, FA, robs him of the ability to move, beginning with difficulty walking, progressing to writing and speaking. In his final years, he will need full-time care.

Regards - Lucia [REDACTED]

[REDACTED]
[REDACTED]
[REDACTED]

I am writing because I have **Polycystic Kidney Disease** an incurable genetic disease. As you may be well aware, there is currently no treatment to slow or stop the growth of kidney cysts that plague generations of families suffering from **polycystic kidney disease (PKD)**. The only remedies for PKD patients once their kidneys fail are dialysis and transplantation. While these options are life-saving, having a treatment that preserves healthy kidney function is the best option. Research is an important factor in finding a cure for PKD, and although some work is being done, certainly not enough. Medical technology is making huge advancements and the focus should be on PKD. Even from a financial advantage, if you find a cure for PKD the cost would be less than a transplant and medication or dialysis.

Thank You

Maggie



My daughter was diagnosed with JRA at 18 months and is 5 years old. After tons of tests to figure out what was wrong, they determined she had JRA. They immediately began her on Methotrexate shots once a week as well as Naproxen liquid. She has been on Methotrexate since 20 months of age...either via subcutaneous, pills or liquid. We've upped her dosage recently, since she has been battling uveitis (inflammation of the eyes) for several years as well. She also has been on Prednisolone eye drops for years as well. That's our story in a nutshell.

Mandie [REDACTED]
[REDACTED]

Dear Members of The House Energy & Commerce Committee,

My name is Marc and I was diagnosed with Mucous Membrane Pemphigoid in 2007. Mucous Membrane Pemphigoid is a chronic autoimmune disease characterized by blistering lesions that primarily affect the mucous membranes of the body, but also affects the skin. In my case, the mucous membranes of the mouth and eyes were also affected. My blistering lesions did eventually heal but not without scarring causing me to be completely blind in one eye.

As a representative of the International Pemphigus and Pemphigoid Foundation I am happy to hear that The House Energy & Commerce Committee is seeking feedback on the 21st Century Cures Initiative. Additionally, there are several areas that patients like myself and the patient community that I advocate for feel need to be addressed.

First, FDA staff education is crucial as the science behind rare diseases is constantly evolving, and I am concerned that FDA staff does not have enough exposure to the evolving science when reviewing applications for products to treat rare diseases. I believe that FDA staff should have access to a robust educational program to stay on top of emerging science and trends within our disease community.

Next, the Compassionate Use Programs that are currently in place need to be expanded. There are no incentives for companies to provide drugs in development for compassionate use to patients seeking them. Often, the FDA is unfairly targeted as the reason why compassionate use is so difficult to obtain, however, in reality many companies developing their products have no incentives to provide their drugs to patients. These companies are required to take all the risk (primarily financial) with zero benefit. As a result, this slows the ability for patients to be treated. We would like to see some kind of incentive programs created for companies to participate in more compassionate use programs.

Lastly and perhaps most importantly, drug re-purposing is desperately needed. Many patented drugs are already developed and approved for common conditions which might effectively treat rare diseases like mine. A single targeted drug is likely to have multiple therapeutic uses. However, rare disease indications will not be developed for patented drugs, because the perception of risk to a billion dollar product is too great to allow any rare disease development. There is worry from manufacturers that potential adverse events in clinical trials on very sick patients would risk the product's market. Additionally, there is minimal financial incentive to do so, as adding a hundred or a few thousand rare disease patients may not increase market revenue enough to justify the costs of re-purposing or the potential risk. A new orphan product patent exclusivity extension is needed to incentivize re-purposing for the rare diseases.

The 21st Century Cures Initiative is a bold and brave step in helping the nearly 30 million Americans that affected by Rare Diseases and I thank you for the opportunity to share my story and comments with you.

Respectfully,

Marc [REDACTED]

[REDACTED]

[REDACTED]

[REDACTED]

[REDACTED]

[REDACTED]

[REDACTED]

[REDACTED]

To the committee:

Re: this communique from the American Academy of Ophthalmology:

Lawmakers Seek Input to Speed Up Advances in Health Care

The U.S. House Committee on Energy and Commerce is seeking input on how to advance health care. The [21st Century Cures initiative](#) is the committee's bipartisan effort to ensure that laws and regulations keep pace with rapid advances in biomedical research and innovation. Committee members are exploring how Congress can help accelerate the discovery, development and delivery of promising new treatments to patients. To provide feedback to the committee, send an email to cures@mail.house.gov. The deadline is June 1.

I find it disappointing that US doctors must look to other countries for evidence that evolving treatment modalities are safe and effective since they are not allowed to be used or studied here (prime example: accommodating intraocular lenses). There are very few doctors that do not wish the best for their patients. Placing new therapies/technologies in our hands and letting us decide what works best is a self correcting policy. Modalities that do not work well are quickly abandoned by physicians. Regulatory agencies need to trust the physicians that have jumped through more hoops than any other medical system in the world. Give US doctors more latitude to explore and use tools that can benefit patients. You may become one of these patients yourself who would benefit from the latest treatment under the most skilled hands in the world.

Mark [REDACTED]

[REDACTED]

[REDACTED]

[REDACTED]

[REDACTED]

[REDACTED]

[REDACTED]

[REDACTED]

[REDACTED]

Dear Congressman Upton,

I am writing on behalf of many Americans who suffer from PKD throughout the United States. This is a disease to which there is no cure. It has been around for quite a while and many never realizing what they have because it is often brought on by high blood pressure which a lot of Americans have. For the last 5 years I have come to Washington to be an advocate for all Americans who suffer from this genetic disease. I have met several Congressman including Rep. Rush Holt who have promised to continue to support more money for research in trying to find that cure that hopefully my two girls will never have to worry about renal failure or dialysis in their lifetime or any generations to follow.

I was diagnosed when I was 33 with high blood pressure. Many doctor visits followed. Over time cysts had formed on my kidneys and in 2005 I suffered total renal failure. I was on dialysis for 3 1/2 years 3 nights a week for 3 1/2 hrs a night. It is quite painful. I watched my mom die from this disease at age 57. I was now 58 years old.

Fortunately, in Sept. 2008 I received a kidney transplant and almost 6yrs later I have a new lease on life. I'm married now for 39yrs and my wife and I own our own business. We have been self-employed for 16yrs. I went to work every day and went to dialysis those 3 nights. I did not want to become part of the system. I truly believe none of us do. We just need more support from Congress to reach our goal.

Thank you for your attention in this matter.

Sincerely,

Mark [REDACTED]

To whom it may concern:

I lost my father to complications from PKD. He was a WWII veteran that was awarded three purple hearts and an oak cluster. His tank was destroyed and he was injured by a land mine that exploded at the same time. All of his war related injuries and disabilities he lived with and recovered from but the PKD killed him in 2001 - he was 79.

My family carries the "gene" that delivers or facilitates PKD. I watched my dad slowly die from the complications of PKD and it was a hopeless and devastating experience. I urge the readers of this email to consider PKD and its impact on the families of those who carry the PKD disorder. After seeing it's impact first hand, I believe that it deserves a reasonable amount of attention. I hope that my words will convince you to consider a closer look at the devastation of PKD.

Mark [REDACTED]

Hello,

My name is Mary [REDACTED]. I have answered your questions and hope that they can be of some help to you. Thank you so much for your interest in curing diseases and thank you even more for acting on this interest.

I am so grateful for all your hard work and hope to be able to continue to offer hope to my two daughters who live each day of their lives with a rare progressive disease called Friedreich's Ataxia. One day I would love to say there is some offering of a treatment.

Please do not hesitate to contact me if I can be of any further assistance.

Best Regards,

Mary [REDACTED]
[REDACTED]

What is the state of discovery of cures and treatments for your disease?

My family has been living with Friedreich's ataxia for 20 years. In the beginning the gene was not even known, there was nothing. We have worked hard as a family to raise awareness of the disease, doing fundraisers for research and just concentrating on living lives as an integral member of society. Presently there is so much work being done however, no treatment no cure :(

Are there cures and treatments now or on the horizon?

YES! So close yet so far. Thank you so much for the outreach such as this survey. I feel that if we can find a cure for Friedreich's ataxia it will lead to discoveries in other diseases. The model of The Friedreich's Ataxia Research Alliance can help so many diseases.

What programs or policies have you utilized to support and foster research, such as patient registries, public-private partnerships, and venture philanthropy?

As a founding member of The Friedreich's ataxia Research Alliance formed in 1998, I have seen the difference between times when there was no hope to the most hopeful of times. We have utilized FARA's patient registry, which has turned out to be incredibly valuable to both the patient base and drug companies/researchers doing the work. We have visited the FDA to give them a patient's view as well as worked with NORD on Rare Disease Day at the Connecticut Capital. We are grateful and always happy to help in any capacity. I might add that we are incredibly grateful to Congress for recognizing the need for cures in the upcoming future.

How can Congress incentivize, coordinate, and accelerate basic research for diseases we know relatively little about?

Congress can instill the importance in open communication and continue to back the need for incentives to drug companies as well as university researchers. Funding the NIH and FDA is paramount for success.

How can we work together to better translate advances in science into safe and effective new therapies for patients?

By surveying more patient populations and offering constituents the opportunity to speak to both FDA and Congress

How do you coordinate your research and outreach with other patients?

That is a wonderful and important question. When my older daughter was diagnosed in 1995, I was told I would never meet another family with the same diagnosis and given a very grim look at the future. I believe that by banding together, whatever the outcome is each person is no longer alone. Families must connect. It is paramount to living a happy and productive life.

A small group of people began a parent support group on the Internet years ago (1998), it has grown to over 600 parents. This has been a valuable tool in finding and sharing information with other affected

families. FARA has been instrumental in bringing families together by way of symposiums and newsletters. This has been such a valuable experience for many people in the FA community.

How do you learn about new treatments and cures? How do you communicate with other patients regarding treatments and cures?

FARA newsletters and press releases. Our physician who is part of CHOP's Center of Excellence. He is extremely up to date with the latest research news. Dr. David Lynch is a physician like no other. Recognizing outstanding people in the field would also be a great incentive for Congress. The FA community is a very tight and caring group.

What can we learn from your experiences with clinical trials and the drug development process?

A patient community is the best link into the patient's view of drug trials. It is also the only way to get drugs and "cures" to come about. Without the participation of patients nothing would ever be possible. As a family we have learned so much about the drug development process, something that most would never understand, that is unless they were part of a patient community. If there were a way to do outreach on the cost and importance of drug development I feel it would be embracing to all people.

What is the role of government in your work, including any barriers to achieving your goals and advancing breakthroughs?

I think the government holds the key to the most important factor in advancing breakthroughs through government funding. Especially when talking about a rare disease. There is very little hope in large pharma companies being interested in rare disease unless there were more financing opportunities. Setting more incentives for larger companies would also contribute to the success of research.

How should regulators evaluate benefit-risk? How do you work with regulators regarding benefit-risk? Can this process be improved?

I can certainly understand the risk involved when weighing benefit against risk. I know with my daughters as the disease progresses there is much more at stake for a cure. The possible benefits begin to outweigh any potential risks.

What is the role of public and private funding in the research and development of cures and treatments?

As I have said, with rare diseases I believe Government funding is crucial to the development for a cure or treatment for rare disease.

Are there success stories the committee can highlight and best practices we can leverage in other areas?

Yes, organizations such as The Friedreich's Ataxia Research Alliance can be a model for other rare diseases. The organization has done more to forward research as well as collaborating and networking among private, government, pharma and patients.

There is also the success with The Children's Hospital of Philadelphia who has opened a Center of Excellence. Our physician Dr. [REDACTED] encompasses everything needed in a top notch physician. He is kind, accomplished, humble and hard working. He makes each day livable for our family.

Our family has also taken on the task to reach out to researchers and show our gratitude. We try to visit one company/researcher a year.

I envision a National Researcher's Day! 😊

How have you worked with other patients to support one another?

The FA Community is a tight knit group who works together in both fundraising and awareness. They are amazing people who understand the importance of collaboration.

What is the financial burden of your disease? How would better treatments and cures help save money for your family and the federal government?

The financial burden is enormous. As a single mom with two adult daughters I am constantly balancing the need for living life as normal as possible against maintaining the best possible health for my daughters. Trying to live life as others do means you must be out in society in order to feel a part of life. This means huge expenses for adapted cars, lifts in our home, etc. Both of my daughters would like to work like others do, however, although educated most employers do not want to hire nor do they understand the positive outcome to their workforce to have persons with disabilities in their employ.

Having no cure or treatment means that many of the potentially helpful vitamins/food/drugs/treatments are not covered by insurance.

I truly believe that a halt of the progression means a chance at living life as others do.

How can Congress help?

Congress must continue to fund the NIH and the FDA, they can continue to encourage open dialog between government and patient populations and patient advocacy groups. Striving to create an environment where there is continued collaboration between the public and government will only help to continue positive collaboration. Continued positive collaboration will only help to expedite a cure for Friedreich's ataxia and other rare diseases.



I have polycystic kidney disease and THERE ARE NO REAL TREATMENTS.

I have had to live with that hard reality for over 30 years. I have lost my grandmother, my mother, and my aunt to this progressive nightmare. I watched them battle its complications, suffer its horrific pain, and succumb to its inevitable outcome. My brother has already had a kidney transplant; he was one of the lucky ones. But there are trade-offs, side effects; not even that is a solution. Both of his children have the disease. So far, one of my three children has discovered he has the disease; the other two aren't sure they want to live with the cloud over their heads.

And that's what it would be—a very dark ominous cloud. There are only two temporary solutions to this dreaded disease—dialysis or transplant. Both are grueling on the body; neither one is permanent. Not everyone can do dialysis, and even if they can, the side effects can be debilitating. And not everyone is lucky to secure a donated kidney. Even then, the transplanted kidney doesn't last forever. If you're young when you receive the organ, chances are you may have to go through it again—that's if your body and age will allow it. THERE ARE NO REAL TREATMENTS.

One doctor told me that of all the kidney diseases out there, PKD is the one to have because it progresses slowly. Really? It's like having a date with death slowly creep up on you as you watch your blood chemistry get worse and worse over time, knowing there is no cure out there and being helpless to do anything about it. Yes, I keep my weight down. Yes, I'm on a blood pressure medicine to keep my kidneys as healthy as possible. Yes, I drink water. Yes, I watch my potassium intake, sodium intake, caffeine, etc., etc., etc. But after all is said and done, I have no control over this disease. My kidneys are huge, my numbers continue to get worse, the doctors will try different pills, they'll put me on a transplant list. I dread that day and it colors my world. My heart breaks for my children. And I can do NOTHING because THERE ARE NO REAL TREATMENTS.

There are not enough organs to go around. This disease robs you physically, emotionally, and financially. It makes sense physically to treat and preserve the native kidneys; it makes sense economically to unburden an already strained medical community. We need real solutions, REAL TREATMENTS, for polycystic kidney disease. And we need them now.

Sincerely,
Mary [REDACTED]

[REDACTED]

Energy and Commerce Committee:

I am sending this letter to request much needed help with insurance gaps concerning medical food coverage for metabolic disorders such as PKU. My son has PKU and requires a medical formula as well as specific low protein medical foods. Currently, insurance helps with formula but not with any kind of food. Insurance coverage varies widely and we are always at risk of his formula not being covered through our insurance plan. The passage of H.R. 3665, the Medical Foods Equity Act, would ensure that federal health programs provide coverage for medical foods for the treatment of metabolic disorders. This would be a huge step in helping ensure coverage in the private insurance market! Please take some time to get acquainted with this issue and do your part to get H.R. 3665 passed so families can be assured of coverage for their medical needs. THIS COVERAGE IS ESSENTIAL FOR PROPER GROWTH AND DEVELOPMENT IN PATIENTS WITH METABOLIC DISORDERS. PLEASE HELP US GET THIS BILL PASSED QUICKLY!!!

If you have further questions or would like to speak with me personally about these concerns, you may reach me at [REDACTED]

Thank You,
Mary [REDACTED]

[REDACTED]

1 June 2014

Energy and Commerce Committee
United States House of Representatives
Chairman Fred Upton

Re: 21st Century Cures: The Gap in Access to Treatment for Phenylketonuria

Dear Chairman Upton and Rep. DeGette:

I am writing to express my concern about the current gap in our health care system to access treatment for PKU. I am the mother of a 7 year old boy who has Classical PKU. PKU has been successfully treated in the United States for more than 50 years, yet many children and adults cannot access the treatment needed to manage the disorder. We must ensure that everyone with PKU has access to the treatment they need for this rare genetic disorder.

Every baby born in the United States is screened for the early identification of PKU as a public health activity to prevent severe disability. The treatment for PKU includes the daily use of medical foods and foods modified to be low in protein that must be continued for life. However, this treatment is out-of-reach for most patients with PKU because of a lack of insurance coverage. Providing coverage for medical foods for the treatment of PKU is medically supported, cost-effective, and the right thing to do. I am writing to ask you to pass H.R. 3665, the Medical Foods Equity Act, so that federal health programs provide medical foods coverage for the treatment of Phenylketonuria (PKU). This will be a significant step forward in closing the gap in coverage.

- Medical evidence has demonstrated the safety and efficacy of medical foods as treatment for PKU for more than 50 years. Just recently, the American College of Medical Genetics and Genomics issued the first-ever treatment guidelines for PKU that confirms the necessity of medical foods treatment for PKU for life.
- The impact of the lack of coverage on patients with PKU is disastrous and expensive. The average family cannot afford to pay for medical foods without insurance coverage.
- The long-term costs to the government for the care of untreated children and adults with PKU far exceed the cost of providing this essential treatment.

Decades ago, before the implementation of newborn screening and treatment with medical foods, children with PKU were doomed to a life of intellectual disability and costly institutionalization. Now, because of mandatory newborn screening and the proven treatment with medical foods, children and adults with PKU can lead normal and healthy lives. Don't put these lives at risk. Please ensure that medical foods for the treatment of PKU are provided by the federal health programs and pass H.R. 3665, the Medical Foods Equity Act, so that everyone with PKU can grow up and become healthy and productive citizens of this country.

Sincerely,
Mila [REDACTED]

Members of the House Energy and Commerce Committee:

There is no treatment available to stop or slow the growth of kidney cysts due to polycystic kidney disease. My son was diagnosed at age 24.

My son applied for the TEMPO ¾ clinical trial in 2008. He was not accepted. Tolvaptan was studied to determine if it could block the cysts. It has not been accepted as an effective drug.

April 10, 2014 at the age of 39, my son was at end renal stage and had a kidney transplant with his brother as the donor.

Sincerely,

Mildred [REDACTED]

Dear House Members,

I am writing to tell you my story with Lyme disease (with coinfections). I was infected in CA nearly 7 years ago. I was an avid hiker and yet had never really heard of it. I became infected and within two months I had seizures, a transient stroke, respiratory distress (Babesia) severe memory loss, intestinal bleeding, heart problems, passing out, depression, anxiety, pain everywhere and many other symptoms. I was an extremely healthy, happy 30 year old just before with no significant health issues. I went to 14 doctors and several ER visits in those 2 months who all thought I was crazy and finally figured out on my own that I had Lyme, went to a very expensive Lyme doctor, got tested, and came up unequivocally positive over and over again according to CDC standards. Yet new doctors still thought I was crazy. My brain spect scan showed significant reduction in activity. By that time it was too late, it had spread to my brain and all my other organs. Within two years, I was bankrupt, homeless and still in life threatening condition on a regular basis, in an out of hospitals (several times ICU). I had insurance, but it covered pretty much nothing. I was regularly emotionally abused my medical care providers who said I was just making it up, despite my vital signs and lab tests being way off. Fortunately I had a small savings and credit cards as well as a top doctor that helped me through part of it. I was on a PIC line for 7 months and almost died from sepsis because the insurance wouldn't cover the ongoing PIC maintenance. I was disabled for 4.5 years. And miraculously as my disability and unemployment ran out, I started to get better. This is because I met my partner, who belonged to an international engineering firm that had excellent insurance that didn't ask questions or require prior authorization for standard antibiotics. I only felt good on antibiotics, I relapsed for years off it with life threatening symptoms (heart issues). In the year before I met him, I contemplated suicide daily, because there was no hope. Today, I am better and I am the director of the international [REDACTED] I am engaged and hoping to have a family. We are not crazy. We need medical help and antibiotics do work. I didn't have to go through all this. What probably cost over \$300,000 in medical bills, not to mention lost income, could have been a \$150 misadventure.

Sincerely,

Mira [REDACTED]

Good Afternoon,

I am a mother and a wife of someone who suffers from PKD (Polycystic Kidney Disease). There is currently no treatment to slow or stop the growth of the kidney cysts that plague generations of families suffering from polycystic kidney disease (PKD). For PKD patients, the only remedies are dialysis and transplantation once their kidneys fail. While life-saving, having a treatment that preserves healthy kidney function is the best option.

PKD has affected my family, my husband has been forced to retire from his job at the age of 50 and I have watched his health deteriorate rapidly over the last 5 years. He was a successful business owner, he was always active, coaching little league and basketball for over 15 years, camping, riding ATV's and participating in many outdoor activities to someone who has to conserve his energy to do a few daily tasks and needing a nap every day. His mom and grandmother passed away through complications of the disease. His sister is battling it and my daughter has been diagnosed with the disease. I have 2 more children who have not been tested as of yet since there is no treatment or cure, who wants that on their medical records. To complicate issues, my husband had prostate cancer 1 year ago and has been told he is not eligible for transplant for a minimum of 5 years after being rid of the cancer. With kidney function at 16%, how long do you think he will last???

If there was something that could have slowed the progression of the disease he would have many happy years ahead of him instead of wasting away with depression and not feeling well.

My hope is that there is a treatment or better yet a cure by the time my daughter starts having complications from the disease. Without the funding they need, this will never become a reality. I will personally never stop fighting for this cause!!!

Monica [REDACTED]

To Whom it may concern,

Please find the attached answers to the questions submitted at the request of the Energy and Commerce Committee for your consideration. Thank you for your attention to this very important issue that effects our family and most importantly, my son [REDACTED] My family and I thank you for giving us this opportunity to tell our son's story to you.

Best Regards,

Monica [REDACTED]

June 10, 2014

To the Honorable Members of the House of Representatives:

I would like to share a brief story about my son [REDACTED] who has a rare Chromosome 6 disorder called SYNGAP.

In November of 2012 we found out that my son [REDACTED] has been diagnosed with a very rare Autosomal dominant disorder called SYNGAP-1 gene (6p21.3). I didn't know whether to cry or breathe a sigh of relief. He was the first to be diagnosed at [REDACTED]
[REDACTED]

Some of the symptoms of this disorder are seizures, schizophrenia, and moderate to severe mental retardation, speech delay, and autism type behaviors. [REDACTED] has been in multiple therapies that include OT, PT, Speech therapy, play therapy and music therapy.

I knew that something was not right when [REDACTED] was 4 months old. He was not sitting up or meeting the same milestones as his twin sister. I began my search for an answer beginning with my general practitioner, then adding 19 more specialists to the list since his birth. After many tests such as an MRI, EEG, Cat-scan, Microarray and metabolic tests at 2 years old we found out everything was "normal." But we knew that it was everything but "normal."

He was unable to walk, feed himself, babble or talk. We waited 14 months to get an appointment to have an evaluation at Texas Children's Meyer Center for Autism. They concluded that our son was going to have intellectual disability and a severe speech delay, but still no real answers. The Meyer Center then referred us to Texas Children's Genetics Clinic for Whole Exome DNA testing. We were very blessed to have our insurance cover the cost of the test.

After the diagnosis I decided to help our son and myself and began to blog about his progress and try and raise awareness of SYNGAP and find others like him. It took almost 4 years to get a diagnosis for our son.

When I posted our diagnosis on my son's blog I began to reach people from all over the world who were like me. I have created an information page through Facebook that is strictly about research on SYNGAP and brain based research that is related to SYNGAP. Another parent and I have set up a closed Facebook group for parents looking for support and a place to talk about our children's medical progress and challenges.

As the Whole Exome DNA test becomes more common we see our group growing worldwide. The network we have created in this group has brought several international doctors together to work on research to define our rare disease and continue research to help find a treatment that will help our children. A group of parents from the Facebook group are now working to establish a nonprofit for SYNGAP and raise awareness and fund research. I have to conclude that the journey I am on I wouldn't change for the world. I have learned more about myself and the love I have for people that I never thought I would have if I had not been placed in this situation. I am very grateful and appreciate life more than I ever have.

Please see the answers I have provided for your questions in the following pages.

Thank you,

Monica [REDACTED] – Mother to [REDACTED]

21st Century Cures – Patients with the SYNGAP Mutation

1. What is the state of discovery of cures and treatments for your disease? Are there cures and treatments now or on the horizon?
Currently there are two doctors working on research for both treatments and cures. Dr. Gavin Rumbaugh working at Scripps Labs, located in Florida, USA is working on finding the window of time for treatment and a cure. Dr. and Dr. Jacques Michaud from Montreal, Canada; in partnership with Dr. Michael J. Parker from the United Kingdom are defining the disorder and publishing research on EEG patterns and seizures caused by the mutation on Chromosome 6p21.3 (SYNGAP). Research that has been done by Dr. Gavin Rumbaugh and has mouse model data showing that if the gene is treated in early onset of development that the symptoms that are caused by the mutation on the SYNGAP gene is successfully reversed. Dr. Rumbaugh now must research the human window of time to treat the mutation that causes the protein needed to control the growth of the neurons (nerve cell) in the brain.

Research published by Dr. Jacques Michaud:

<http://www.ncbi.nlm.nih.gov/pmc/articles/PMC2925262/>

Please see the following article published by Dr Gavin Rumbaugh:

http://www.zoominfo.com/CachedPage/?archive_id=0&page_id=6455442106&page_url=//www.eurekalert.org/pub_releases/2012-11/sri-sus110512.php&page_last_updated=2012-11-08T21:34:15&firstName=Gavin&lastName=Rumbaugh

2. What programs or policies have you utilized to support and foster research, such as patient registries, public-private partnerships, and venture philanthropy?
The parents of the patients that have the SYNGAP mutation have come together through a Facebook group and gathered information from doctors internationally and encouraged our own Parent Driven study to gather information to continue the research process. We have brought doctors together to partner up and define our specific condition and to be a specific diagnosed disorder. The parents are in the process of creating a 501c non-profit to raise funds for research on SYNGAP.
3. How can Congress incentivize, coordinate, and accelerate basic research for diseases we know relatively little about?

Congress can help accelerate the process by passing H.R. 460, the Patient's Access to Treatment Act (PATA), and H.R. 1591 – The Charles August Long Undiagnosed Diseases Research and Collaboration Network Act of 2013 into law which allows doctors to collaborate together and combine efforts that will help expedite research efforts. Members of Congress can also help by joining the Rare Disease Caucus. This would create an opportunity for law-makers to be aware of the process that occurs when a rare disease has been discovered and what is required for research to begin.

<http://rareadvocates.org/category/caucus/>

4. How can we work together to better translate advances in science into safe and effective new therapies for patients?

Creating an incentive program that will allow the drug companies to be able to use new drugs and repurpose drugs that have already been developed for compassionate use for patients with rare diseases and genetic disorders. It would be beneficial for FDA leaders and employees to be educated on the current and on going science behind the research on rare diseases.

I believe that FDA staff should have access to a strong educational program to stay on top of emerging science and trends within our community.

Research will help develop newer tests, cost effective tests, and quicker diagnoses for patients. The sharing of information and creating a data base for doctors to access will help with a quicker diagnosis. Creating an agency that will help coordinate both research and a networking agent for doctors to create new therapies faster.

5. How do you coordinate your research and outreach with other patients?

Currently, as a parent advocate for SYNGAP (6p21.3) I blog about my son's progress on Google's Blog Spot. I am on a mission to increase awareness of SYNGAP mutation and through my blog. I have reached people all over the world who are looking for answers. Since my son's diagnosis in November of 2012, I have made contact with other parents who have reached out to me. The other parents and I have created a closed Facebook group to include any parent or caregiver of a person with the SYNGAP mutation. We communicate the majority of the time is through the social network we have created and through email. I also communicate with other organizations such as Global Genes and other Facebook groups of families and parents that have not been diagnosed. We keep each other updated on current news about new symptoms and treatment or the non-treatment of those symptoms. We cry, laugh, console each other, vent our frustrations and celebrate the milestones our loved ones with SYNGAP accomplish. We have used each others connections to connect doctors who otherwise would have never collaborated if our group had not been organized. This has put in motion a hope that we are not lost or forgotten. Our efforts will continue, but the help of our United States Government will be crucial to continue the efforts of others and myself that are trying to get diagnosed and research continued for treatments or cures.

6. How do you learn about new treatments and cures? How do you communicate with other patients regarding treatments and cures?

I find out of new research through reading recently published articles on SYNGAP, communicating with our genetics doctor, and making contact with the doctors who are actively doing research on our disorder. I communicate with other patients as stated above, through our social networking site. I also have a SYNGAP information page on Facebook that specifically focuses on SYNGAP and post articles and other information on research being done.

7. What can we learn from your experiences with clinical trials and the drug development process?

Due to the rareness of the SYNGAP mutation, clinical trials are not available and drug development is not available.

8. What is the role of government in your work, including any barriers to achieving your goals and advancing breakthroughs?

The biggest barrier that we have experienced is that our disorder is not an official disease. Due to the rarity and lack of availability for the genetic test needed to diagnose SYNGAP, there have been less than 100 world wide identified. It is believed that this disorder is at least 100,000 to 1,000,000 individuals world wide. The cost of the Whole Exome DNA test averages \$15,000. The average family can not afford this test and often insurance does not cover the cost.

9. How should regulators evaluate benefit-risk? How do you work with regulators regarding benefit-risk? Can this process be improved?

At the current time I do not work with regulators

10. What is the role of public and private funding in the research and development of cures and treatments?

The most critical role for research and development is funding. At the present time there is not an organization that specifically raises money for research on SYNGAP. Corporations and scientists must write grants to fund research. The non-profit that our SYNGAP parents intend to create will be the only non-profit world wide that will fund research on SYNGAP.

11. Are there success stories the committee can highlight and best practices we can leverage in other areas?

The ability for doctors to access information quickly and network as a team to diagnose an individual with the SYNGAP mutation has been effective in treating patients with their known symptoms early on. It is crucial for these individuals to be diagnosed early, due to the fact that some of the symptoms are on set later in childhood. Knowing the diagnosis early can help parents and caregivers know what to expect and how to treat early to reduce the effects of the symptoms, such as the onset of seizures. The only known clinic that has been working with the research and diagnosis of patients is with the SYNGAP mutation is Dr. Jacques Michaud. He is the Head Division of Medical

Genetics, CHU Sainte-Justine; Professor of Pediatrics and Biochemistry, Université de Montréal, CHU Sainte-Justine Research Center, 3175 Côte Sainte-Catherine, Montréal (Québec)Canada. We have successfully partnered Dr. Michael Parker from the United Kingdom with Dr. Michaud and are in the process of building partnerships with doctors from Texas Children's Hospital to come aboard and join forces to continue research on the SYNGAP mutation.

12. How have you worked with other patients to support one another?

We created a Facebook Group where parents and caregivers can share experiences and coordinate efforts to raise awareness.

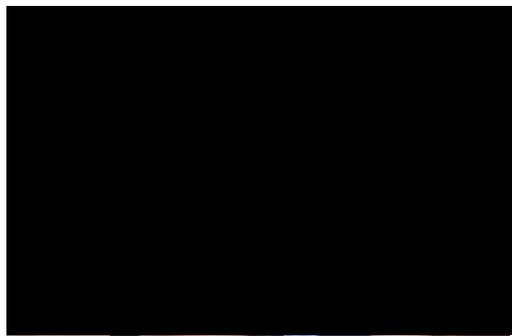
13. What is the financial burden of your disease? How would better treatments and cures help save money for your family and the federal government?

our child that has the SYNGAP mutation has many therapies and medications that he has to have on a daily basis. He also has follow up EEG's and blood work done on a regular basis. On the average our family spends out of pocket any where from \$10,000 to \$19,000 per year in medical costs. He sees on average about 9 -10 doctors every year. He has seen approximately 19 doctors total and approximately 342 doctor's visits and 2 major surgeries in his life time. He is only 6 years old. He has atypical absent seizures, intellectual disability, expressive/receptive speech language disorder, ODD, SPD, ADD, pronated feet, hypotonia, severe constipation, acid reflux, Amblyopia (lazy eyes), allergies to many antibiotics and seasonal asthma. A cure to replace the protein that his brain doesn't make, if found in time, I believe would relieve some of the therapies he needs to function. If it is too late for him, it would not be for the other children with early diagnoses. I know that if funding went to research, in time the need for reimbursements on health tax deduction costs would be reduced for the government.

14. How can Congress help?

I am restating the same answer as #3 of how Congress can help.

Congress can help accelerate the process by passing H.R. 460, the Patient's Access to Treatment Act (PATA), and H.R. 1591 – The Charles August Long Undiagnosed Diseases Research and Collaboration Network Act of 2013 into law which allows doctors to collaborate together and combine efforts that will help expedite research efforts. Members of Congress can also help by joining the Rare Disease Caucus. This would create an opportunity for law-makers to be aware of the process that occurs when a rare disease has been discovered and what is required for research to begin. Vote these pieces of legislation into law to help our future generation.



To whom it may concern,

Please consider granting funds toward research into finding a cure for Polycystic Kidney Disease. There is currently no treatment to slow or stop the growth of the kidney cysts that plague generations of families suffering from polycystic kidney disease (PKD). The only remedies for PKD patients once their kidneys fail are dialysis and transplantation. While these options are life-saving, having a treatment that preserves healthy kidney function is the best option. My husband is in stage 4 of PKD and we just found out that our 18 year old son also has PKD.

Sincerely,

Nanci [REDACTED]

What is the state of discovery of cures and treatments for your disease?

Are there cures

There are several trials underway in the US and Europe for treatments. The ones that have made it the farthest through the phases of research are treatments. There is one in the animal stage that has reversed the disease in the hearts of mice, but it hasn't been applied to the nervous system which is also affected

Are there cures and treatments now or on the horizon? No

* What programs or policies have you utilized to support and foster research, such as

FARA and its research affiliates often petition for orphan drug status, fast track status and abbreviated FDA requirements for drug approval.

patient registries, public-private partnerships, and venture philanthropy?

Our family has registered our two daughters in patient registries, but they are advanced and less likely to be used in a study. We have supported the Friedreich's Ataxia Research Alliance personally through donations and have helped raise over one million dollars over 10 years of fundraising.

* How can Congress incentivize, coordinate, and accelerate basic research for diseases we know relatively little about? Congress has not played a major role to advance research for diseases such as Friedreich's Ataxia. Almost all progress is linked directly to FARA.

* How can we work together to better translate advances in science into safe and effective new therapies for patients? FARA shares all of its research conclusions with several other diseases that impact a small percent of the population. Some of these are Fragile X, Parkinson's,

* How do you coordinate your research and outreach with other patients?

Through the registries, our Ataxia Clinics across the country and through the web presences of FARA

* How do you learn about new treatments and cures? From other parents on a support group, but mainly on communication from the Friedreich's Ataxia Research Alliance.

How do you communicate with other patients regarding treatments and cures? Online support groups, National meetings of the National Ataxia Foundation and FARA.

* What can we learn from your experiences with clinical trials and the drug development process? No major drug company will devote significant research funding to minor diseases. The risk of drug failure in FDA studies combined with limited opportunity to recover the investment will always favor doing very little. Congress has funded millions of dollars for AIDS research - a disease that is totally preventable in many cases while doing little for genetically linked diseases that have no cure. Abbreviated FDA trails should be designed to ensure safety with less emphasis on complete efficacy. A partial solution is better than no solution.

* What is the role of government in your work, including any barriers to achieving your goals and advancing breakthroughs? FARA needs more funding for research. Congress has the ability to address that issue without changing any laws or procedures.

* How should regulators evaluate benefit-risk? How do you work with regulators regarding benefit-risk? Can this process be improved?

As previously stated, regulators should focus more ensuring safety and placing less emphasis on achieving efficacy > 80%.

* What is the role of public and private funding in the research and development of cures and treatments? Both are critical and should be encouraged. I would suggest more tax benefits for major pharmaceutical companies that invest research efforts toward minor diseases.

* Are there success stories the committee can highlight and best practices we can leverage in other areas? I would highlight FARA and the coordination of research exports worldwide to hold annual meetings, share research findings and collaborate on future studies. This effort by FARA has accelerated the research effort in searching for a cure for FA.

* How have you worked with other patients to support one another? In grassroots fundraising, and one of my daughters is a FARA ambassador

* What is the financial burden of your disease? The financial burden is great. We have kept our daughters on our private insurance and they are now 31 and 34. We have to have accessible vehicles, an accessible home. lots of money for supplements which are not covered by insurance, They require 24 hour care, and as their mom I have been unable to work full-time, or even part-time for the past 10 years. Our income has suffered because of that. They have also had several surgeries which has taken much time from work.

How would better treatments and cures help save money for your family and the federal government? Drugs approved by the FDA would be covered by insurance

* How can Congress help?

Support FDA with earmarked funding for research devoted to minor diseases.

Nelda [REDACTED]

[REDACTED]

[REDACTED]

[REDACTED]

My life changed for the second time due to Rheumatoid Arthritis. I was first diagnosed when I was in the fourth grade with juvenile arthritis. I was told it would go away and I would grow out of it. By the time I left junior high school there was little sign of it. Sometimes my knees or hands would hurt. Then suddenly it returned three years ago. The first year doctors couldn't give me a proper diagnosis. Then they finally figured out it was RA.

I barely work part time. I am holding on to that job and it is a very stressful job which only created more pain. I found out this year that I also have fibromyalgia. I am a full time student online and it is also very stressful. I now am not sure if going to school to become a teacher is such a good idea because I my days are treacherous often and very little days of relief. When I do have good days I take advantage of them by doing all that I can then the next day I am down for the count for a day or sometimes days experiencing flares.

I cannot file for disability because you can't work so if I stop working I will have no income and will not be able to provide for myself and my daughter. I am barely making ends meet. I hate RA and this fibromyalgia and what it does to my life and my body and my mind. I am often down because of it but I have to put on a smile for my daughter and others so they don't worry.

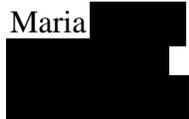
Nicole [REDACTED]
[REDACTED]

Hello,

I have RA, PsA, Hashimotos thyroiditis, and Sjogrens. Once you get a diagnosis of an autoimmune disease other diagnoses seem to follow. The medication are quite pricy and totally too expensive for those without insurance or with medicade or medicare as only infusions are covered and not totally. We need access to more affordable prescription medications for those stricken with autoimmune issues.

Thank you,

Maria



To Members of the 21st Century Cures,

On behalf of the more than 1 million Americans suffering and dying from Myalgic Encephalomyelitis (sometimes referred to as Chronic Fatigue Syndrome but preferably as ME/CFS by government agencies and patient groups) we ask that the following be included in your 21st Century Cures Legislation.

1) The 2015 NIH budget should be commensurate with the size of the disease and/or the cost to the nation. For example, Multiple Sclerosis (MS), a disease the FDA states is similar in seriousness to Chronic Fatigue Syndrome but with only 400,000 patients - receives a \$115 million per year in NIH funding - and there are already nine existing drugs for MS. People with ME/CFS have NO approved treatments yet more than 1 million people are sick (versus 400,000 for MS), and it receives only \$5 million from the NIH. That is only \$5.00 per patient! This is a ridiculously small amount. Yet CDC studies estimate it costs the nation \$22 billion per year. Other funding levels for 2014:

~ Anthrax \$72 million (who gets anthrax?)

~ Attention Deficit Disorder \$50 million (not life threatening)

~ Smallpox \$31 million (no one gets smallpox anymore)

2) One drug – Ampligen - has shown promising results against ME/CFS. There were significant improvements in approximately 40% of the patients on Ampligen! Many patients in the trials have been safely on this drug for years. Yet the FDA continues to drag its heels in approving it. We request that the legislation reflect that if a drug shows promise and the top experts in the disease believe the drug should be on the market, then FDA should use its existing regulatory authority to approve the drug, and in collaboration with the experts and the sponsors develop the appropriate follow-up studies and REMS program. The expert physicians that work with the patients and the drug have the expertise and knowledge needed to provide sound judgment. This will allow for a large proportion of new ME/CFS patients' lives (and those with other similarly ignored diseases) to improve. Leaving patients without treatment is NOT in the best interests of public health; ME/CFS patients die early deaths from cancers, heart disease, suicides (because there are no drugs and no hope).

3) FDA should be required to provide the same type of flexible approach to chronic illnesses with few or no FDA-approved treatments that it does to those considered fatal (able to cause "imminent death"), to rare disorders, or to disorders that produce epidemics. ME/CFS falls between the two as do many chronic illness, yet cost families and the nation greatly. The fact that 1,000,000 people with ME/CFS have suffered for decades without a single FDA-approved drug requires the FDA recognize that a new approach with more flexibility is called for in this disease and other diseases which the FDA has failed to support. After DECADES of inaction, IT IS TIME.

Thank you for your attention and help on this vital issue.

Mary



To Members of the 21st Century Cures,

On behalf of the more than 1 million Americans suffering and dying from Myalgic Encephalomyelitis (sometimes referred to as Chronic Fatigue Syndrome but preferably as ME/CFS by government agencies and patient groups) we ask that the following be included in your 21st Century Cures Legislation.

1) The 2015 NIH budget should be commensurate with the size of the disease and/or the cost to the nation. For example, Multiple Sclerosis (MS), a disease the FDA states is similar in seriousness to Chronic Fatigue Syndrome but with only 400,000 patients - receives a \$115 million per year in NIH funding - and there are already nine existing drugs for MS. People with ME/CFS have NO approved treatments yet more than 1 million people are sick (versus 400,000 for MS), and it receives only \$5 million from the NIH. That is only \$5.00 per patient! This is a ridiculously small amount. Yet CDC studies estimate it costs the nation \$22 billion per year. Other funding levels for 2014:

~ Anthrax \$72 million (who gets anthrax?)

~ Attention Deficit Disorder \$50 million (not life threatening)

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Thank you for your attention.

Nancy



I am very eager to hear what you are considering for all of the pathogens carried by ticks? For example, Babesia is found in the blood supply. This pathogen is a relative of malaria. So, in essence, a malaria like organism is found right here in the United States. Please consult the Red Cross about this. How about Bartonella? This is another tick borne pathogen. What are you doing to consult the Tick Borne Disease community and their advocacy groups? I don't see any consultations with the Tick Borne Disease Community Listed. Thank you, Karen [REDACTED]

To Whom It May Concern,

I wholeheartedly agree with your initiative. It think the first thing you should do is FUND scientific research. I am on a 2nd post doc - in [REDACTED] - and have an unfunded position at [REDACTED] University that I am trying to fund with grants. The last four positions I applied for had more than 120 applicants. The funding rate for new PIs is 5%. We cannot innovate if we cannot test our ideas. I've written at least 10 major grant proposals. They get good reviews. There is just no money.

I'm not quite sure how you do that given the state of our Congress and the inability for people to have a normal dialogue about anything let alone science.

But you have my support.

Best,

Michael [REDACTED]
[REDACTED]

Please reach across the table and try to come to an agreement with the Democratic congressman in establishing something for the veterans who are suffering into HR bill 5680 if you could construct something equivalent or better in this proposal that was turned down last year we would appreciate it
Ken [REDACTED] Sent from my iPad

Thank you for all the work you do Fred! I love getting the current updates. Everything takes time, so hang in there - we're behind you!

Linda [REDACTED]

Sent from my iPhone

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Thank you for your attention.
Kristy [REDACTED]