

Dear ladies and gentlemen,

As the [REDACTED] Division of Clinical and Translational Genetics at the University of [REDACTED] I am at the forefront of treating patients with different inborn errors of metabolism including Phenylketonuria (PKU). Children with PKU require life-long dietary therapy with costly medical foods to sustain normal development and a healthy, productive life. Without the foods children will suffer from intellectual disability which devastates their lives. There is a gap in coverage for medical foods that are needed to treat PKU.

I therefore urge you to support H.R. 3665, the Medical Foods Equity Act, to ensure that the federal health programs provide coverage for medical foods for the treatment of PKU. Coverage of medical foods at the federal level would greatly influence coverage in the private insurance market.

Sincerely

Olaf [REDACTED]  
[REDACTED]  
[REDACTED]

My husband health insurance thru his job does not pay for the first \$1,500.00 towards any prescriptions so I have to pay out of pocket and can not afford to do so. And therefore I take an OTC such as Alleve, Tylenol or aspirin. When my knees are very painful, I then make an appointment with my rheumatologist for shots in the knees, But for how long will this go on.

Please give me an answer on how I am to continue living with so much pain in my joints. I want to continue working (part - time) and not stay home (I will be turning 62 next month).

Thank you.

Hi,

My name is Paige [REDACTED] I'm 19 years old and I have Friedreich's Ataxia, which is chronic, neurodegenerative, and currently incurable. I wasn't able to answer every question in the article, but here are my answers to a few of them. I hope this is helpful! I know the FA community appreciates your efforts. Thank you,

Paige Myers

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

Yes, the FA community is fairly confident that the disease will be cured within a matter of years. But, as of right now, FA has neither been cured or treated. Since FA is neurodegenerative and significantly life-shortening, time is really of the essence. All of us would love a drug to stop our progression before we get worse than we currently are. There are promising drugs in trials right now, but I do get really frustrated with how long it takes.

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

I think things need to move faster than they do. I know with FA, and I assume most genetic diseases, trials really fill up pretty quickly. People are so desperate for a treatment or a cure that safety concerns become secondary. I really think that the trial process can be accelerated because people are normally more than willing to volunteer. In addition to being slow, the FDA tends to reject drugs where the results are inconclusive. That doesn't mean it's useless, it could just mean it's benefits are difficult to document. If a drug passes safety tests and *might* be beneficial, I don't see the harm in making it readily available anyway.

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

The FA community is not a selfish group at all and when someone finds something that helps them, they let everybody else know. Social media (such as Facebook) is an important tool, and there are also online groups, such as FAPG (Friedreich's Ataxia Parent Group). My dad is a part of that, and finds a lot of info that way. Drugs to maybe try, equipment to maybe try, advice on dealing with specific obstacles, etc.

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

YES! The financial burden is big, both on our family and on our insurance. I have a GP and several specialists, all of whom I see at least once a year. There is also lots of medical equipment to buy. For example, last year I got a new chair and a power drive for that chair, and this year we're renovating a bathroom to make it wheelchair accessible.

I have two sons with Friedreich's ataxia (FA). FA is a rare, genetic, progressive neurodegenerative disease for which there is presently no treatment or cure. We have been living with FA for the last 12 years and over that time I have watched my sons go from perfectly normal to being in a wheelchair. There are so many ways FA affects our family and all the families I know living with this disease.

There are about 5000-6000 people with FA in the United States and around 15,000 world-wide. However, we are a very tight-knit community. We have a parent email support group and a wonderful non-profit organization called the Friedreich's Ataxia Research Alliance (FARA) (<http://www.curefa.org/index.html>) which was started by parents of kids with FA and has made great strides in moving research forward since it started in 1998. We have a patient registry, a robust pipeline, research scientists around the world, outstanding doctors and many dedicated people working to find a treatment and a cure. We have many patient families doing fundraisers to raise awareness and funds. However, things do not move fast enough!!! I know many, many parents who have lost children to the disease over the last twelve years. I have seen countless families split apart because of the emotional toll it takes on marriages. I have watched many bright, beautiful, young kids lose their abilities and have to start using a wheelchair full time – many at very young ages.

The financial burden can also be overwhelming. In order to receive SSI, the person with FA cannot have more than \$2000 in assets and only receives around \$700 per month to live on! This is a shame because these people are truly disabled unlike many people on disability that abuse the system. It is hard to get a personal care assistant (PCA) to take care of them who is competent enough to help since their salary is so low. Many of them take advantage of the person with FA and steal money from them. There is also not enough hours of PCA time granted to the person with FA. Many FA patients would like to work at least a few years while they are able to help with their self esteem and allow them to get out in the world but the SS system actually discourages them from doing this by taking away their support if they are working. This whole situation is a problem to our society as a whole.

The bottom line is that research is not moving forward fast enough. I cannot emphasize this enough. **Research is Not Moving Fast Enough!!!** Laws that are put in place to help rare diseases are just taken advantage of by pharmaceutical companies. Most every disease can somehow get rare disease status by breaking it up into sub disease groups which totally negates the whole advantage of the rare disease designation. This needs to stop. Pharma uses tax incentives for rare disease to their advantage without moving drug development forward in a timely manner. There needs to be more NIH funding for basic science research. We are concerned about the safety of people in our country and spend way too much on a defense system when there are thousands of our citizens dying each year from rare diseases that don't get enough funding.

The FDA does not seem to help move things forward fast enough. The whole system slows down drug development. Clinical trials which originally had three phases now actually have 6 phases because each phase is broken down into a Phase A and Phase B. It takes too many years to move a drug from the research stage to the clinic. Meanwhile countless lives are lost. Probably more money for the FDA would help; but there should be a closer look taken of the whole FDA process of moving a drug forward. It should be streamlined to make it as efficient as possible. Each meeting that is requested of the FDA

by a pharmaceutical company takes too long to wait for – if up to 90 days are required before a meeting can take place the full 90 days are taken and nothing gets done earlier than 90 days. This is really unacceptable.

The time it takes for a drug trial to start once it gets FDA approval is also very slow. There was one FA drug trial that was supposed to start in January at three different sites. It started in January at one site and not until March at the second site and not until June at the third site. This means that a trial that is supposed to take 6 months actually takes double that time. This appears to be because the Institutional Review Board (IRB) at each trial site does not approve the trial in a timely manner. Incentives should be given to these IRB's to get them to approve trials more quickly for rare diseases.

The cost to develop drugs is very high. Pharmaceutical companies are reluctant to start developing drugs for rare diseases because the initial non-proven research may not go anywhere. There has to be more NIH funding for early stages of drug development for pharmaceutical companies so they will not have to spend their own money to get the drug to a phase 1 trial. Also once the drug does go through the whole trial process; there should be a reasonable amount that they should be able to charge so that people with rare diseases can actually afford these drugs. Otherwise, insurance companies and government programs are footing the bill for these astronomically expensive drugs. Also because the drug companies have a patent on these drugs for 20 years, the drug companies continue making these huge amounts of money for several years. Something has to change about this whole process because it is something that affects society as a whole.

All of these delays just mean that patient's diseases are progressing faster than drug development and the result is more lost lives and more severely disabled these FA patients are becoming. The government needs to step in and make this whole process go much faster!! You need to talk to organizations such as the Friedreich's Ataxia Research Alliance (FARA) and figure out where the bottlenecks are and what needs to be done.

Thank you for starting this whole process and I hope that some positive things come out of it to help everyone affected by diseases that need a treatment sooner rather than later.

Sincerely,

Pam [REDACTED]

HI. MY NAME IS PATRICIA AND I AM A 59 YEAR OLD MOTHER OF ONE ( 39 ) AND GRANDMOTHER OF TWO PRECIOUS GRANDCHILDREN AGES 12 AND 16.

I AM ALSO A PKD PATIENT AND HAVE BEEN ON DIALYSIS ALMOST 6 YEARS. I AM LOOSING GROUND WITH THIS DISEASE. THE PAST 2 YEARS MY HEALTH HAS DECLINED AND I FEAR I WILL SOON BE ANOTHER PKD PATIENT WHO HAS LOST THE BATTLE AGAINST THIS DISEASE THAT NOT ONLY TAKES OUT INDIVIDUALS BUT FAMILIES.

MY DAUGHTER HAS ALREADY BEEN DIAGNOSED WITH PKD AND THERE IS A LIKELY HOOD THAT MY GRANDCHILDREN MAY HAVE IT TOO.

IT IS MY PRAYER THAT THERE WILL BE A CURE OR AT LEAST A WAY TO SLOW THIS TRAGIC DISEASE DOWN. IT IS TOO LATE FOR ME BUT HOPEFULLY NOT FOR MY DAUGHTER AND SURELY NOT FOR MY GRANDCHILDREN AND SOME DAY FOR MY GREAT GRANDCHILDREN AND THEIR CHILDREN. AGAIN THIS HORRIBLE GENETIC DISEASE DESTROYS FAMILIES NOT JUST INDIVIDUALS.

PLEASE, PLEASE WE NEED YOUR HELP. FUNDING, RESEARCH AND TRIALS ARE SOME OF THE WAYS THAT WILL HELP FIND A BETTER WAY TO LIVE WITH POLYCYSTIC KIDNEY DISEASE. WE NEED MORE OF THESE TO ADVANCE THE KNOWLEDGE IT WILL TAKE TO SUCCEED IN THIS PROCESS.

THANK YOU SO VERY MUCH.

PATRICIA [REDACTED]  
[REDACTED]

Four years ago I was diagnosed with Relapsing Polychondritis...I had no idea what this meant...I had no idea that I was dealing with a diagnosis that would be doubted because of its supposed rarity..the reality is very different...within 1 yr of diagnosis and my understanding of it through the online support groups( no my doctors didn't help me understand it mostly because they have been told it was very rare) I found 8 people within a 1 mi radius of where I live with all the classic symptoms of swollen cartilage in ears...nose over bridge red and swollen joints but yet to be diagnosed. You see this disease is unique in that it attacks cartilage only...other auto immune diseases attack organs as well...and that is an interesting fact that may be useful in that it may be the door to understanding processes underlying auto immunity...but there are no to little \$ being spent on research this diagnosis because of its supposed rarity. This so wrong as it is rare because of being underdiagnosed.

There are some medications they have tried but my immune system rejected all of them often with painful blisters across my body and even in very private places. But everyday I loose a little more mobility and the meds leave my brain foggy and the first drug of choice prednisone has in 3 years caused my body weight to double...but its the only thing known to slow the progression.. I have shortness of breath as a result of the trachea starting to collapse. I am swollen from prednisone and today there is no way I can get a pair of shoes on these feet and tomorrow will probably be hospitalized again as it is affecting my heart...cartilage is interest in a glue that keeps everything together...and my body has a bad feedback loop that disrupts the precursor for making cartilage. I had to go to Mayo Clinic in Rochester to get a confirmation of diagnosis. I found it amazing that every person on staff there knew what this diagnosis was and worked to get tangible data as to where I was in the progression of this disease. I live in a city with 5 medical schools but very poor coordination of care...compared to my treatment at Mayo its a world apart. As an engineer I appreciate their concern and understanding of this disease and instruction as to how I can help myself...

In my life before this I was an engineer who loved her work...I would turn on like a christmas tree when talking about my work. They had to yell at me to go home...

If I where to be so lucky to be cured of this disease because of the respiratory issues I can no longer work in manufacturing... but would go back to my training and using my skills in orthotics and prosthetics ( my senior design was for a mandible for a young woman)....And right now the only cures for this have come about with stem cell transplantation(9 members of the support group no longer have any symptoms of this disease.after anagolous proceedures) but its no walk in the park but cure IS always better than treatment..I would like to see a lot more done in this area. But because it is still considered experimental and dangerous its not readily available. And the cost is over the moon...to prevent this disease in the progression of e and other autoimmunity diagnoses we have to take it out of the black box realm into a place of understanding. I ask your help in making it possible for me and others to BE cured because right now its not likely as there are no clinical trials of meds for this in my area although there are still the myth that this is a rare disease....we who have it are tossed to winds as orphans.



Patricia [REDACTED]

To whom it may concern. I have Relapsing Polychondritis. It is a rare disease. It is very difficult for the many of us with Relapsing Polychondritis to get the drugs we need because they are not approved by the FDA for this disease, but are very effective in treating This disease. We need help in getting these drugs approved.

Thank you,

Patricia [REDACTED]

[REDACTED]

Good afternoon,

As a dedicated volunteer and advocate of the PKD Foundation, thank you for the opportunity to write on behalf of all individuals who have been impacted by Polycystic Kidney Disease (both ARPKD and ADPKD).

PKD (autosomal dominant) has been present in the paternal side of my family. I have lost five family members before the age of 55 (a grandmother, father, two uncles, and a sister). My brother received a kidney transplant in 2003.

My “brief” history with PKD is as follows:

- Diagnosed at age 16
- Multiple kidney infections that did not respond to antibiotics during the 1970s and 1980s
- Hypertension, again with difficult medications to control easily at a young age
- A liver cyst requiring sclerotherapy four times
- Pain from large kidneys, liver, and pancreatitis
- Hemodialysis began at age 49 for two years
- Kidney transplant at age 51 with bilateral nephrectomy (kidneys donated to PKD Foundation), and left lobe resection of liver due to pain (kidney was received from generous coworker)
- Participated in University of Colorado Health Science Center hypertension study from 1993-2000

Having worked within the medical university setting (closely with nephrologists and transplant surgeons) has given me an edge to learn more about PKD. Through research and the great efforts of the Polycystic Kidney Disease Foundation, medications and better dialysis and transplant management have helped patients living with PKD. 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 our kidneys fail are dialysis and transplantation. While these options are life-saving, having a treatment that preserves healthy kidney function is the best option!

With the attention of Congress to hear our personal impact, it is my desire that my sister, age 48, as well as my niece (age 13), and nephew (age 15) living with PKD will have a better future and hopefully a chance to avoid end-stage renal disease treatment for a longer time.

Thank you for this great opportunity to tell my story. My story is the same story as many of my colleagues who have lost loved ones to PKD at too young of an age. We volunteer strongly to ensure a better future for all patients living with PKD – autosomal dominant and recessive, for.....

***Together we can make a difference in the future of patients with PKD, particularly in the San Antonio community. We “Unite to Fight PKD”***

Kindest regards,

Patti [REDACTED]

[REDACTED]

[REDACTED]

[REDACTED]

[REDACTED]

[REDACTED]

[REDACTED]

[REDACTED]

[REDACTED]

[REDACTED]

[REDACTED]

[REDACTED]

Dear Representative Fred Upton and Representative Diana DeGette,

Thank you for this opportunity to help speed the approval of critically needed drugs for suffering children and adults everywhere.

My daughter [REDACTED] was diagnosed at 9 in 1986 with a progressive disorder called Friedreich's Ataxia. Perhaps the worst part of this disorder is that it typically shows itself between 5 and 15 just as these developing youngsters are starting to dream of their futures and then it trashes those dreams leaving the victims cognitively intact but increasingly physically inoperative. Learn more about Friedreich's Ataxia here.

<http://www.curefa.org/whatis.html>

I have answered your questions below as fully as I can but for the best quality input I recommend you arrange a meeting with the leaders of FARA, Friedreich's Ataxia Research Alliance, for the best comprehensive and focused inputs about the FDA clinical trial processes and how they might be re-balanced and streamlined.

<http://www.curefa.org/contact.html>

Sincerely,

Paul [REDACTED]  
Dad to 6 including [REDACTED], FA, 36  
Not part of FARA

[REDACTED]

[REDACTED]

=====

\* 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 no FDA (nor other country drug safety organization) approved treatments for Friedreich's Ataxia. Thanks in large part to our FA-family-created FARA we are in the blessed position (compared to most other rare/orphaned disorders) of having 8 drugs in clinical trial right now. None of them are approved though, time is passing and our children are dying, so your interest in speeding the approval process is of the utmost interest to us. See the status of FA research here.

<http://www.curefa.org/pipeline.html>

\* What programs or policies have you utilized to support and foster research, such as patient registries, public-private partnerships, and venture philanthropy?  
+ In 1998 some parents of FA'ers saw the need for a FA organization to focus resources and increase awareness and so FARA, FA Research Alliance, was born in that September. Now 15+ years later it is the model organization for other rare disorders. Funding comes from many quarters; grassroots fundraising, a couple of FARA-run fundraisers, many FARA sponsored fundraisers, direct donations, several well-

funded individuals (FA cares not about social standing nor financial success), etc. See <http://www.curefa.org/mission.html> on the right side.

- + FARA recognized early-on the importance of having an effective research and approval process infrastructure. FA-interested drug companies and researchers now come to FARA for
  - o The FA registry ( <http://www.curefa.org/registry.html> ),
  - o FA clinics trained and ready as trial sites ( <http://www.curefa.org/network.html> ),
  - o The FA Natural History Study ongoing results
  - o Grants ( <http://www.curefa.org/grant.html> ) to help further their basic and advanced research work.

- + FARA also recognized the value of collaboration and teamwork among researchers, government agencies and drug companies. Much of our progress toward a treatment is due to this.

- o FARA puts on FA research symposiums (the first one was possible with a grant from the NIH) to pull researchers together for the synergy of sharing information and to create more networking amongst them.

<http://www.curefa.org/conference.html>

- + By browsing the FARA website ( <http://www.curefa.org/index.html> ) you will have a much better understanding of FARA's "value-added" to FA research and the FA community.

- + Also in 1998 Sue [REDACTED] under the urgings of other FA families, started FAPG, FA Parents Group, an emotional, problem solving, and research communication support group whose 600+ members support FARA in various ways.

<http://www.faparents.org/fapg/> More recently Facebook has been added as a significant research communication medium.

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

- + Have the NIH pay for it since many drug companies won't be interested until translational research shows promise. For Rare disorders the subject of who to fund can be difficult. How much money do you invest for a disorder that only 10 people in the world has? I do not have that answer. FA has about 20,000 patients world-wide and is believed to be a stepping stone to other mitochondrial disorders. Factors like these should guide decision-making.

- + Congress could stop cutting the budget of the FDA and the NIH!! You do not "incentivize" nor "accelerate" by taking away their money. The FDA is being mandated to expand their various scopes of responsibility on a number of fronts and at the same time their budget is constantly at risk and does not increase in proportion to the new work they are instructed to take on.

- + You've already done the perks for orphan designation and fast track. I'm not qualified to suggest other programs.

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

- + Join the collaboration between patient organizations, drug companies and researchers to identify the technologies and how to integrate them into the testing and review processes.

- + This has to be funded. And you cannot reduce the budgets of the FDA and NIH while expecting them to take this on. Won't happen. You ask for more you give them more.

- + Collaborate with and do a next-bench examination of the European Medicines Agency (EMA) way of doing business to see where FDA advances in speed and efficiency might be made.

o Repligen came to the FDA with a FA HDAC inhibitor Phase I trial request and was sent back to the lab. Repligen re-grouped and went to the EMA and got approval for a Phase I trial in Italy. How and why was that possible? Case study time.

\* How do you coordinate your research and outreach with other (FA) patients?

+ Through communication in the FAPG email group, FA Facebook groups, FARA FA Registry notifications and the FARA news distribution list.

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

+ Through communication in the FAPG email group, FA Facebook groups and the FARA news distribution list.

+ When a new trial is starting FARA Emails potential qualified FARA Registry FA'ers with contact information to the trial coordinators.

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

+ That collaboration and teamwork do work. Adversarial relationships do not work as well or as fast.

+ Unfortunately the FDA and government is SO bound up with fears of "conflict of interest" even to the mere possibility of an appearance of same that I fear any significant collaboration is doomed.

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

+ We look to the NIH to help on basic research and collaboration efforts.

+ We look to the FDA to protect our children from "bad" drug treatment alternatives and to move all drug treatments forward posthaste to separate the "wheat" from the "chaff". We can accept risk if it is clearly explained.

+ The FDA is underfunded and understaffed. They also work in an atmosphere of fear-of-retaliation. You cannot work in this field of creation, exploration and marketing without some risk of a "bad" drug slipping through the best of processes. When this has happened "government" did not "have their back" and instead hung them out to dry. Should we wonder why they are risk-averse?? The role of government should be to confirm the "standard of the day" and "process adherence" and then back them up!

Unfortunately that is not the governmental "rule of the day". "Backing one up" is a matter of political expediency and voter damage control. I have no idea how can "incentivize" any organization in this climate of "cover your buns". Good luck.

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

+ Tough question. The FDA has struggled with this for many years. There is the medical-professional benefit-risk statistical perspective and then there is the personal benefit-risk. For the professional choose a historical-precedent model and run the numbers. Pass/Fail? Relatively simple. Look at the exceptions, intervention, side "events". Make a judgement call (erring on the safe side; ie, "Do no harm"). For the FA parent or patient it is much more visceral and emotional. Watch your child or yourself decline knowing there is no "cure" nor progression stoppin/slowing treatment available. Other patients die around you.

Desperation mounts; "We need it now!!" is the cry. "But at what risk?" is usually not heard and if it is heard it probably is not integrated as a real possibility/probability; "Not us/me" is the thought. How do you "work with"

that dichotomy of views?

- + And does the FDA really want to absorb individually the voices of so many emotional people, and should they?

- o Focused surveys within a specific disorder and even a specific drug with knowledge of side effects might be a good approach. A survey administered by the FDA to the 650+ world-wide membership of FAPG for instance might make knowledgeable and evaluable information from the din of individual input.

- o Or working with a FARA assigned group in several meetings focused on a specific drug might be useful.

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

- + Another big question books might be written on.

- + For me public funding should be used when private funding is not forthcoming or inadequate. Again the tough question is that of volume. How much public funding should be expended on disorders that affect only "5 or 10" people in the whole world? I don't have that answer.

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

- + FARA is a success story of the highest order and should be looked at as a model for other disorders, diseases and other areas where research collaboration is needed.

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

- + The Email group FAPG and several Facebook FA groups tie us together daily with opportunities to network and hug at annual FA and Ataxia conferences. Regionally the various fundraising events draw us to one another occasionally.

\* What is the financial burden of your disease?

- + The financial and mental health burden varies state to state, how FA presents itself and the individual phase of progression.

- o Many/most FA'ers never work so they are on SSI and/or on SSDI (retired parent) getting \$600-ish to \$1,000-ish a month to live on. If they live with someone SSI removes \$300-ish for room and board. If they live independently it is a big financial struggle just to live. Parents help to the limit of their own budgets and the limits set down by SSI. Copays, PT/dental/accupuncture not covered, supplements thought to perhaps help, exercise equipment, ramps, bathroom adaptations, wheelchair maintenance, etc are areas of extended cost.

- + Caregiving is needed for many adult FA'ers but even the hours that are given (often none) are not adequate. Parents wear out, get old and get injured/sick. Many FA'ers desiring to live independently cannot because they cannot get/afford caregivers.

\* How would better treatments and cures help save money for your family and the federal government?

- + You know the answer to this as well as I. Less to no caregiving, less to no Medicaid medical costs, some FA'ers might even be employable and off SSI.

\* How can Congress help?

- + Fund the NIH and the FDA at levels appropriate with the responsibilities you mandate them with.

- + Back them up instead of throwing them to the sharks when a "bad" drug is discovered.

Paul



Dad to 6 including [REDACTED] FA

I wanted to pass along information on a new innovative company CarePayment, which helps assist patients with the skyrocketing high deductibles they are incurring. CarePayment is a co-branded effort between our company and the hospital. This program is a new innovative way to help families cope with these high deductibles and not have to bankrupt them or force them to deny procedures because they cannot afford to pay for them. I would love to share with you more about our program and how it helps consumers and fits into this particular roundtable event.

Paul [REDACTED]  
[REDACTED]  
[REDACTED]  
[REDACTED]  
[REDACTED]  
  
[REDACTED]  
[REDACTED]  
[REDACTED]  
[REDACTED]  
[REDACTED]

# An Ethical Solution to Address Consumer Medical Debt



# About Us

## **Aequitas Capital Management**

*A Credit Focused Investment Management Firm*

- Nearly \$500 million in assets under management
- Provides income-producing alternative investment solutions to institutional and high net worth clients
- Lending and fund management platform, with focus on high yielding strategies within education, healthcare and private credit
- Over 100 employees with cross-functional experience
- Headquarters in Portland, OR, and offices in San Diego, Philadelphia, and New York City

## **CarePayment**

*Innovative Healthcare Finance Solutions*

- \$780 million life-to-date balances managed by CarePayment
- 1.25 million patient accounts and growing
- Founded in 2004, with headquarters in Portland, OR
- Owned by Aequitas Capital Management offering tier one, highly stable credit source with significant investor demand to fund CarePayment patient receivables
- Strong team combining deep provider revenue cycle and payer/employer experience

# The Current Market

## For Providers

ACA is driving enrollment in high deductible plans. 80% have chosen silver or bronze plans<sup>1</sup> with family deductibles averaging \$6,078 and \$10,386 respectively<sup>2</sup>.

85.2% of hospitals either have no monthly cash collection goal or consistently fall short of their goal<sup>3</sup>.

In 2012, US hospitals provided \$45.9 billion in uncompensated care, 6.1% of their annual expenses<sup>4</sup>.

Deferring treatment due to cost undermines the provider's ability to improve patient outcomes and drives up care costs

## For Patients

Employees' share of medical costs increase 150% from 2004 to 2014<sup>5</sup>.

40% of consumers have medical debt<sup>6</sup>, with a majority owing \$2,000 or more<sup>7</sup>. The leading cause of personal bankruptcy is medical debt<sup>8</sup>.

71% patients prefer monthly payment plan<sup>9</sup>.

30% of U.S. adults say they, or a family member, have put off medical treatment in the past year because of the cost<sup>10</sup>.

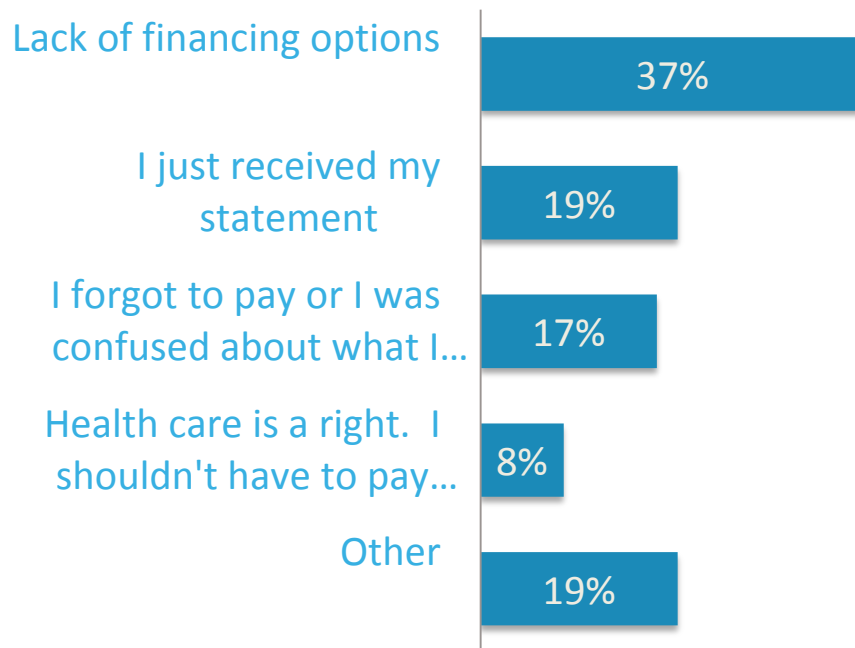
### Sources:

1. Health and Human Services Department, January 13, 2014
2. HealthPocket
3. "Funding Your Mission," RelayHealth and Healthcare Finance News 2013
4. American Hospital Association, "Uncompensated Hospital Care Cost Fact Sheet," January 2014

5. Aon Hewitt analysis, October 2013
6. The Commonwealth Fund Biennial Health Insurance Survey 2012
7. Centers for Studying Health System Change, December 2011
8. Nerdwallet Health study, June 2013
9. InstaMed "Trends in Healthcare Annual Report: 2012"
10. Gallup Poll, November 2012

# Industry Trends

## Why Don't They Pay?



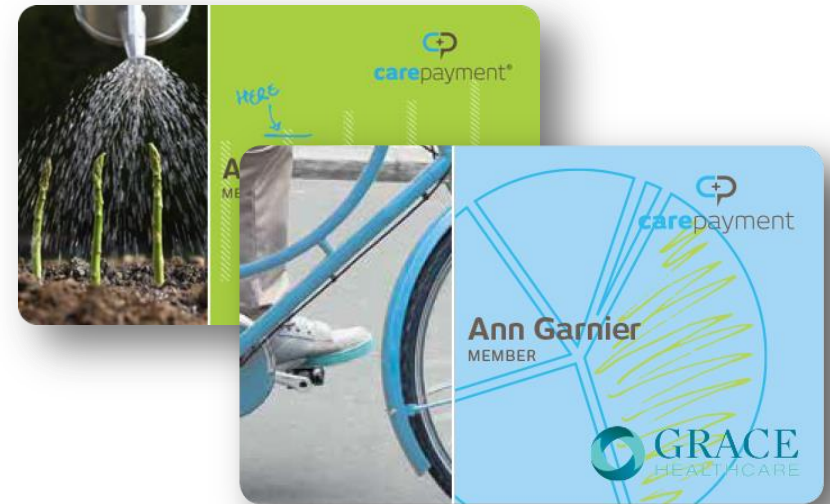
## Increase in Bad Debt

- Consumer bad debt continues to rise, resulting in more than \$65 billion in uncollected revenues in 2010 – McKinsey Quarterly, May 2010
- The majority of households rank medical expenses 7th in importance (after cell phone and internet bills), with only 7% of households rating medical expenses a top priority – McKinsey Quarterly, May 2010

# The Solution

CarePayment® offers an innovative and ethical solution that addresses consumer medical debt while improving hospital financial performance.

As a patient-friendly alternative to collection agencies, we offer a combination of loan funding and account servicing solutions that reduce barriers to obtaining healthcare.



## CarePayment® Program

- 0% APR financing
- Payment plans from 12 to 72 months
- No application required
- Revolving credit line at provider site
- Third party co-branding
- Consumer lending compliance oversight

# Advantages of CarePayment

## Consumer Advantages

- Flexible payment plans enable patients to pay over time with terms ranging from 12-72 months
- At 0% APR, there is no interest expense
- No impact to credit score
- No application required
- Additional medical expenses may be added to the account
- Friendly US-based customer service staff ensures clear patient communication

## Care Provider Advantages

- Guaranteed net financial improvement
- Up-front payment for patient receivables
- Co-branded program extends providers' patient services and increases satisfaction and brand loyalty
- In-house expertise and third party audits ensure compliance with all applicable state and federal consumer credit laws
- Stable funding sources regardless of market fluctuation
- System-wide capabilities
  - Hospitals, health systems, physician groups, and elective care



# Summary

## CarePayment:

- Empowers consumers to bridge the financial gap created by high deductible health plans and/or lack of insurance coverage
- Improves financial performance for health care providers facing an increase in demand for services and self-pay accounts
- Is eager to participate in pilots or other legislative opportunities where our services would provide value



*“Medical bills had led me to the verge of bankruptcy. Keeping my medical bills low and not having interest got me back on solid ground. Thank you CarePayment!”*

*- DENNIS*



# An Ethical Solution to Address Consumer Medical Debt

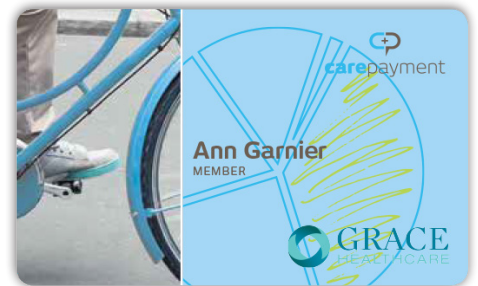
## CAREPAYMENT PROGRAM

- 0% APR Financing
- Flexible payment plans from 12 to 72 months
- No application required
- Multiple payment options
- Consumer lending compliance

It's no secret that out-of-pocket healthcare expenses are on the rise. In fact, they are expected to top \$400 billion by 2016<sup>1</sup>. More than 40% of adults report some type of medical bill problem or debt<sup>2</sup>, while 60% of those with medical debt owe \$2,000 or more<sup>3</sup>. Medical debt is responsible for 57% of bankruptcies in the US<sup>4</sup>.

Meanwhile, hospitals are picking up more of the tab than ever. In 2012, uncompensated care increased to its highest level, totaling \$45.9 billion<sup>5</sup>. As patient financial responsibility grows, there will be a new urgency to collect. The average collection rate for pure self-pay balances ranges between 2-3%, with a 35% collection rate for balances after insurance<sup>6</sup>.

CarePayment® offers an innovative and ethical solution that addresses consumer medical debt while improving [hospital financial performance](#). As a patient-friendly alternative to collection agencies, we offer a combination of loan funding and account servicing solutions that reduce barriers to obtaining healthcare.



## CAREPAYMENT'S CARE PROVIDER ADVANTAGES

- Up-front payment for patient receivables
- Co-branded program extends providers' patient services and increases satisfaction and brand loyalty
- In-house expertise and third party audits ensure compliance with all applicable consumer credit laws

## CAREPAYMENT'S CONSUMER ADVANTAGES

- Flexible payment plans enable patients to pay over time with terms ranging from 12 - 72 months
- At 0% APR, there is no interest expense
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1. National Health Expenditures, Centers for Medicare and Medicaid Services Office of the Actuary  
 2. The Commonwealth Fund Biennial Health Insurance Survey 2011  
 3. Centers for Studying Health System Change, December 2011

4. Nerdwallet Health Study, June 2013  
 5. American Hospital Association, January 2014  
 6. Boehler, A, & Hansel, J. (2006, January 1). Innovative strategies for self-pay segmentation

*Medical bills had led me to the verge of bankruptcy. Keeping my medical bills low and not having interest got me back on solid ground. Thank you, CarePayment!*

- DENNIS

## Polycystic Kidney Disease!

Even though thousands suffer from the disease, most people have never heard of PKD. I had certainly never heard of the disease when I was diagnosed at the age of thirty. The diagnosis changed my entire life. I never had a second child. It was not safe for me to become pregnant. My daughter had nightmares that I would die before she graduated from high school.

I got lucky. When my kidneys failed, my brother was able to donate one of his to keep me alive. But not everyone is that lucky! Dialysis is not a cure and is only a relatively short extension of life. PKD is not all over the news like other diseases so for funding trials and studies is limited.

It is important to encourage American companies to perform research and studies. It is important to find swift ways to bring new treatments to patients quickly. It is important to look beyond drugs to biological treatments that help genes fight and stave off disease. The U.S. government can encourage creativity and cures by streamlining the FDA processes and adopting policies that free companies to develop new treatments.

Thank you

K. L. [REDACTED]

> I have had RA since the age of 11 months. I was diagnosed in July of 1963. My initial treatments consisted of aspirin and indocin. It then progressed to "gold" treatments (for over 25 years) by the time I was 26 I had 2 knee replacements. Within the years after I had 2 hip replacements. In 2003 I went on Enbrel, within months having severe side effects affecting my eyes, which were not at the time noted. I was then put on methotrexate to deal with the eye issue. Again within a short period of time I had severe side effects.

Over the course of this time I have been employed as a special education teacher (in Providence schools since 1993).

Medications work differently on different people, but everyone should have the choice of which treatments would help them.

Paula [REDACTED]  
[REDACTED]

Hi

My name is Peggy [REDACTED] and I have Polycystic Kidney Disease. Please keep funding the research so we can find a cure for this hereditary disease. The funding helped me. We still need it coming for those who still need help.

Thank You Peggy [REDACTED]

[REDACTED]

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 Grandma to a wonderful 15 month grandson who has 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.

[Choose 2-3 bullets or create you own]

- 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.
- Treatment for PKU is currently covered in 39 states through a state insurance mandate or state program. However, this coverage only benefits a small percentage of PKU patients.
- Failure to include coverage for medical foods for all patients with PKU in the federal health programs is not in accordance with the accepted standard of medical care.
- 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.
- 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 and Thank you for your time,

Penny [REDACTED]

[REDACTED]

[REDACTED]

[REDACTED]



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

Re: 21st Century Cures. The Gap in Access to treatment for Phenylketenuria (PKU)

Dear Chairman Upton and Rep. DeGette,

I am writing to you with a concern, pointing out a gap in our healthcare system's insurance fair access for treatment of PKU.

Our grandson has PKU.

It is not right, that not all people, will have equal access to treatment for PKU, based on insurance availability and coverage, or lack thereof. People born with this genetic defect must have medical food for the rest of their lives, or they will be adversely affected, and loose brain cells. They can NEVER eat protein any time, ever in their lives, lest they lose brain cells or they will require increased treatment , and ultimately hospitalization up to , and including institutionalization.

While some states have addressed this at the state level, in no way is the insurance approval available nation-wide. This ultimately results in babies and adults going untreated, through no fault over their own, and becoming a burden on society, when they could be otherwise productive citizens. Our country cannot afford the additional burden of caring for people affected in this way, when any easy solution is available.

However if you pass H.R. 3665, the Medical Foods Equity Act, it will be a Win/Win situation for the patients, the families, and ultimately the American People, in lowering the costs for those that have this terrible, yet treatable, genetic disorder.

Please pass H.R. 3665 so that every person affected by PKU, in each and every state, can grow up to enjoy the same healthy and productive life you and I enjoy, and contribute, instead of becoming an increasing burden on the system.

Philip and Connie [REDACTED]  
[REDACTED]  
[REDACTED]  
[REDACTED]  
[REDACTED]





As you are well aware, 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.

Peter [REDACTED]

Committee Persons; The cost of a new drug is absurd because of regulations-some legitimate most not. It is the same for hospital and doctors costs. Adding insurance and/or gov't funds compounds the expense. De regulate as much as possible and bust the hospital and doctor monopolies as much as feasible-voila the costs will plummet. Thank you for tackling this issue- please do not just 'throw \$ at it which is ,sadly, the expected result from DC. Respectfully, Dr Peter [REDACTED]

From the time I was a teenager until the time I got my transplanted kidney, I was cold. I was always cold. I wore sweaters when others around me were in tee shirts. I would sneak up to the thermostat and turn it up to above 80 degrees, and when my father found it, he would call out “who turned up the thermostat?” But he always knew I was the culprit, without really having to ask.

In 1972, my then-husband was stationed in Baumholder, Germany, and my sixteen-month-old daughter and I went to live in Germany with him. Shortly before it was time to return to the States, I got a letter from my Father. He told me that he had been sick; that he had been diagnosed with Polycystic Kidney Disease. I had never heard of the disease. He told me that it was hereditary.

My daughter and I flew back from Germany when my husband was discharged from the Army in 1973. My parents and two brothers met us at the Atlanta Airport. Well, my mother and two brothers met us at the gate – they told me that my father was too weak to walk the distance from the terminal to the gate, and was waiting back at the terminal. It was a terrible shock to see him when we got back to the terminal where he was waiting. The robust, still-young man that my father had been a mere fifteen months earlier when I left for Germany had been replaced by an emaciated, weak and aged version of himself.

Dad had been waiting to start dialysis. For some reason, the doctors would not or could not put him on dialysis until he had deteriorated to a point where he was barely still alive, and failing rapidly. He and my mother began training to learn how to perform home dialysis shortly after I arrived back in the States, and then a huge machine was delivered to their home. My mother began the nerve-wracking process of attaching the artificial kidney to the dialysis machine, attaching the tubes that took the blood from his vein and cycled it through the artificial kidney and back into his vein, and of hooking him up to the machine, and then monitoring him while the artificial kidney cleansed his blood and removed excess fluid from his system. The excess fluid had to be removed because his kidneys no longer could make urine, and so, he did not urinate any more. Even though he severely restricted his fluid intake, the little bit that he did drink would build up in his system until it was removed during dialysis. They did this three times a week, for four or five hours per session. Mom was terrified each time that something would go wrong, that she would make a mistake that would hurt or harm my father. The stress of performing her role in that process took a large toll on her each time. Every time a line kinked, or anything needed attention, the machine made a loud beeping sound, and my mother would jump and run to see what had gone wrong, terrified each time that something horrible had happened to my father. My father suffered with each treatment, and in between treatments. He had become so weak and debilitated by the disease that he was unable to recover even a semblance of health even though the dialysis treatments were finally cleansing his body of the toxins that his kidneys used to remove before they failed.

He reached a point where he was praying to die. But he would not refuse treatment, even though continuing to live kept him in agony. Finally, mercifully, his heart stopped beating.

For me, I was diagnosed with polycystic kidney disease in the mid-eighties. I was in my early forties. As the cysts grew in number and in size, my kidneys ability to function, to cleanse my

blood, deceased bit by bit. I was constantly fatigued. I remember one Saturday I decided to mop my kitchen floor. I quickly discovered that I could only mop a small section at a time, because once I mopped an area that was approximately four feet by four feet, I was so exhausted that I had to sit and recover. It took me several hours to get that floor mopped.

Finally, in 1994 my doctor told me it was time to get a fistula made, so it would be ready to use when it was time for me to begin dialysis. A fistula is an artificial connection between an artery and a vein. It causes back-pressure on the vein, and that makes the vein expand in size and toughen. It becomes a sturdy location to insert the large cannulae that are used to take the blood from the body and return it to the body. In 1996 my doctor told me that it was time to begin dialysis. I had a full time job, and I was a single mother with a twelve-year-old son to raise.

Most dialysis patients are so tired out by the process that they go home and crawl into bed because they are exhausted by the process. But I wanted to keep my job, because I hoped to get a transplant and resume a normal life. And so, three mornings every week I missed work so I could dialyze, and once I was unhooked from the machine I drove to work and finished out the day. I offered to make up the missed time by working in the evenings, but my boss refused to let me do that. Years later he told me that he used to see me walking down the hall leaning on the wall for support, and he was impressed that I kept on keeping on (not his exact words but I don't remember his exact words).

You know that process of removing fluid during dialysis that I mentioned? If the nurses set the machine to remove just the tiniest bit too much fluid the muscles of the body begin to cramp horribly. The pain of those cramps is something that I will always remember. They do have something that they can inject into the line to relieve the cramps, but you can never get their attention quickly enough once the cramps start, and they can never get the injection accomplished quickly enough, either.

I was very lucky. Eleven months after I started dialysis I received a cadaveric kidney transplant. Most people stay on the list awaiting a kidney for several years. The kidney I received was a perfect match, something that rarely happens. And my transplanted kidney is still functioning wonderfully well. It is my guess that the longevity of my new kidney is due at least in part to the fact that it was a perfect match, because from what I have heard, most transplanted organs do not continue to function as long as mine has. I live an almost-normal life now. I do have to take lots of expensive prescription drugs and I get so tired of taking so many pills. But I'll gladly swallow them each day, because they keep me alive.

My father had six grandchildren. Three of them have PKD. I hope and pray that a new treatment will make it unnecessary for them to go through dialysis or transplantation. I hope that current research and drug trials will yield a better treatment for them - or, even better, a cure.

[REDACTED]

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

Re: 21st Century Cures: The Gap in Access to Treatment for Phenylketonuria / HR 3665

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. 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.

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.

- Treatment for PKU is currently covered in 39 states through a state insurance mandate or state program. However, this coverage only benefits a small percentage of PKU patients.
- 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.
- 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.

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

Preston [REDACTED]  
[REDACTED]

House Energy and Commerce Committee:

Thank you for the privilege of allowing me to respond to this Initiative. The PKD disease has had a glaring gap between the disease and it's treatment for hundreds of years. PKD patients deserve the right to HOPE that this gap can be closed and to REALIZE the actual results.

With genuine concern for all PKD patients,

Reta [REDACTED]

June 12, 2014

Attention: House Energy and Commerce Committee

I am a 77 years old lady living with Autosomal Dominant Polycystic Kidney Disease. My mother, her father, and her grandfather died of this disease. Both my brother and I inherited ADPKD, as well as one of my brother's sons. Two of my children died at birth because of Autosomal Recessive Polycystic Kidney Disease.

There is no cure or treatment to slow or stop the growth of kidney cysts. Each generation of people has told the next generation that science would find a treatment or a cure and life would be better, but for endless decades, NOTHING has happened.

The kidney has a remarkable ability to compensate for damaged cells and keeps working to remove the waste product of the human body, in my case until age 59. But eventually, the kidney is unable to respond appropriately, and the only recourse is to go on dialysis which I did at the age of 60. I choose peritoneal dialysis and later had to go on hemo-dialysis because of a torn lining in the peritoneal cavity. Dialysis is a life-saving procedure but the quality of life for me was highly questionable because of severe fatigue. Dialysis is only treating the symptoms.

I immediately started asking questions about getting on the kidney transplant list. I had suffered a heart attack at the age of 56 which did some damage. (Now we know that a heart attack is also connected to PKD.) I was told by the head of the Nephrology Department at [REDACTED] that "I would never qualify for the kidney transplant list because of my heart problems and there was no use in doing any tests". Thank goodness I got a second opinion from a Phoenix doctor and proceeded to do the testing. The final decision was made by the head of Cardiology at [REDACTED] stating that "my name should be placed on the kidney transplant list". That was a GREAT DAY! I received a cadaver kidney on October 2, 2000, at the age of 63 and have been very thankful for a much better quality of life for almost 14 years.



During the time I was on dialysis, I served on a committee to help improve patient communication with dialysis staff and make some acceptable changes in procedures when possible. Some friends and I connected to form our own support group. The PKD Foundation had a chapter in Tucson for a short time, but leadership was poor. Lack of communication and up to date education concerning PKD for patients in this area and many other areas across the US and the World are two of my major concerns.

I was placed on Medicare when I began dialysis and I also was very lucky to be able to afford a secondary insurance. The government's role in all these health issues was to pay for the remaining cost. Information from the web at [priceconomics.com](http://priceconomics.com) gives statistics for chronic kidney disease (not just PKD) stating that roughly 398,000 patients are on dialysis each year costing approx. \$75K per patient which equals \$29 Billion. The annual cost of failure in the U.S. is approx. \$42.5 Billion as of 2009. Typically Medicare covers costs for seniors 65 and over. However, Medicare has a special exemption with people with End Stage Renal Failure. The result is that the U.S. government pays \$29 Billion towards covering the cost of dialysis and kidney transplantation which is 68% of the total kidney spending. According to the National Kidney Foundation, 600,000 people in the U.S. have PKD. It is the fourth leading cause of kidney failure. It is found in all races and equally found in both sexes. It causes about 5% of all kidney failure. Think of the possible savings for the U.S. government if PKD patients no longer had these expenses.

The good news is that I have been working locally with Critical Path Institute (C-Path). In the last four years, they have been able to put existing data from four main organizations together to form a "yardstick of information" which is what was needed in order for any progress to be made regarding a treatment or a cure for this disease. C-Path is working very closely with the FDA and the FDA is extremely pleased with their results. This is a tremendous milestone because without this new YARDSTICK, no drug company was ever going to spend money to develop any drug because there were no guidelines for measuring success. This organization is finally proving that SOMETHING very significant can be done!!

We desperately need to develop a drug that will slow or stop the growth of PKD cysts. Then, the goal will be to identify the young PKD patient with beginning signs of enlarged kidney cysts that can be treated in the early stages, long before the need to go on dialysis, and prevent the patient from even needing dialysis or a transplant or suffering the long list of side effects that accompany this disease.

Please consider the PKD disease number ONE in your efforts to close the gap between a disease and it's possible treatment.

Sincerely,

Reta [REDACTED]

Good Afternoon to All In Congressional Representatives,

First of All, I want to thank, House Energy and Commerce Committee Chairman Fred Upton (R-MI) and Rep. Diana DeGette (D-CO) recently who announced a bi-partisan initiative, called the "21st Century Cures: A Call to Action". This will certainly help to shed some new light on chronic illnesses, and especially those new diseases and illnesses, that at this time, like many types of Arthritis, (Rheumatoid, Osteoarthritis & Juvenile to name a few), that have NO cure at the time, little known about "why" those who all of a sudden get the diseases, why others may not, what medications we need in the future to either put these illnesses into remission, OR CURE them.... even STOP them before they ever do the severe damage they do to all of us suffering daily dealing with ALL of this issues these horrid illnesses bring into our lives. I hope many of our other Congressional members will stand beside HECC Chairman Mr. Fred Upton and Representative Diana DeGette and help to make this "Call to Action" a huge success for the thousands and hundreds of thousands suffering from various forms of Arthritis and Autoimmune Illnesses.

I am an "Arthritis Patient", that suffers from several various forms of autoimmune arthritis, as well as osteoarthritis. My list of diseases, illnesses and syndromes; Lupus, Rheumatoid Arthritis (RA), Sjögren's, Pernicious Anemia, along with many issues brought on by thee illnesses, such as a heart attack at 40, severe pain, swelling stiffness that all have consumed my quality of life at times so much, that I feel the diseases rule my daily living. At the age of 15 I had my 1st knee surgery. At that time, they knew little about Juvenile Arthritis (JA), and the experts really did not know much about RA, and all of the other arthritic and autoimmune illnesses here in our nation. So, I went for years from one physician to another, trying to find out why all of my joints were "falling apart"... by the time I was 40, I had already had a left elbow repaired, a left shoulder repaired both knees had surgery at least twice back them, plus all of the other "symptoms" that most doctors at the time "blew off" especially when it came to women. They for the most part thought if I woman complained about aches and pains, she was either nuts, a hypochondriac, was having "female" issues, or was depressed. That was the huge one. The answer from most doctors to a woman with "mysterious" health issues was to determine they are depressed and fill you full of anti-depressants that did nothing for the "physical issues" that overwhelmed you. Still I would hear, even with the severe migraine headaches I had, the weakness, & still other signs and symptoms that should have never been looked over, but taken seriously.

In 2009 I FINALLY began to get some "answers" that made sense. I was not nuts, depressed, making it up, had female issues etc.... I had an "autoimmune issues or issues" that were ruining not only my joints, but also causing he severe fatigue, all of the joints and surgeries I was having... including both knees completely replaced and my right shoulder had to be replaced, along with a 4 level cervical surgery, for degenerated disc disease, and now my thumbs, fingers, toes, and ankles are "eaten up" by RA.

I've jumped through hoops and hoops for a very long while over my medications for these diseases. When you have a serious illness, and medications come out that may "halt" or at least give you a huge percentage of reprieve, so your quality of life comes back & your doctor feels the medication is necessary, there should be NO ONE at an insurance company, or throughout Medicare to tell your doctor differently. These ridiculous amounts of forms, paperwork, having to be put through 4 or more medications that DO NOT work before the ONE that does can even be considered, makes for a patient becoming worse by the day, paperwork and red tape costing precious doctors time, insurance people who have no clue what the entire thing is all about, and it is a vicious cycle of "stuff" in order to finally either get the medication you NEED OR be turned down, which really is asinine. I have been through I cannot tell you how many "pre-authorizations", sending medical records, my doctor having to fill out forms which they should not have to, talking for literally hours on the phone trying to get to ONE PERSON who can "fix" the problem... and on the computer sending emails, sending messages, and as I said, all the while suffering from these horrid diseases that each day wreck my joints and my body a bit more each day. Any type of "autoimmune arthritis" such as RA, effects NOT JUST the joints. They are "systemic" in nature & can effect the heart, lungs, kidneys, your hearing, your eyesight, your memory, your blood, your stomach, intestines, and so on. Just about every part of the body can be effected by any type of autoimmune illness, and can reek havoc in just a short time without proper care.

One of my horrors with one of these “Autoimmune Arthritic” diseases is Sjögrens. This is just one that they do not know nearly enough about. The doctors know it effects the mucous membranes, which we have all over our bodies. Our mouth, which is how saliva is formed. It keeps our eyes moist, our intestines, and many “linings” in our organs have these types of member in them. Without them, organs like the inside of the mouth, dries out so badly, you cannot even speak, swallow, and the worse cause horrendous “dental caries” and other issues! Yet, I was not informed of just how quickly and how badly my teeth might be effected. I brushed daily, tried my best to chew sugarless gum, sipped on green tea all day long, and used the special toothpastes and all of the “remedies” for dry mouth. Yet, about a year ago, all of a sudden one evening we were eating dinner, and an entire back of a molar just fell off in my mouth! Within the course of 3 weeks I had 3 more teeth with break off at the gum line, or a half of a tooth break off. Then suddenly almost every tooth had either a large cavity in it, OR would crumble off on the edges, and I knew I had to get assistance quickly because I was losing all of my teeth, and in a time frame that was not going to allow me to wait even a month or two.

So, I began trying to find an “Oral Surgeon” and/or an Oral Surgeon/Maxillary surgeon that would take my Medicare Advantage Plan “Humana Insurance”. Well, I have yet to find ONE dentist, oral surgeon, and so forth that will take my insurance, EVEN THOUGH this problem is caused from a physical ILLNESS, NOT just regular dental caries. This is a serious matter, that I spent weeks and weeks calling dental offices, and researching online, first of all, WHAT I truly needed done, and a Dental Doctor that could do the procedure, and try to help me get some of it paid for by my Medicare Advantage Plan. I literally spent days and days sending emails, making phone calls, sending messages to dentists offices, all to no avail. In other words, (unless you have had to price these types of dentures called “mini implants”) I was going to have to cough up anywhere from \$8,000.00 to some the charge \$16,000.00! Now these are the dentists that have been through enough “training” to do certain types of “oral surgeries”, not the Physician Dentists, the true Oral and Maxillary Surgeons MD. I am sure when you look at the fact they usually put you under complete anesthetic in an operating room, and have several nurses and so on assisting them, they charge I would say \$25,000.00 and UP! By the time you pay his services, a anesthesia doctor, all of the charges for an “outpatient” stay at a day surgery or hospital setting, I know from the extensive surgeries I’ve had due to all of these horrid, life altering chronic illnesses, it certainly would be a great deal more than \$8,000.00. And “they” MAY be able to get my Medicare Advantage Plan to “pay”... with some pushing, pulling, red tape, & lots of time before I could have anything done, BUT the insurance ONLY PAYS 60% of any type of “dental” procedure no matter if the problem has been caused by an illness! So, that means by the time you added up all of those “extra’s” involved where I would have to travel back and forth from Dallas many times to get it all done (so far I have had about 5 appointments to do all of this, and still have another at least 3 to go)... thus you are looking at the cost of gas, parking, and what if something happened after hours or a weekend? Like a dry socket, of which I had one, and also the start of an infection. But, I am within 5 minutes of my dentist/oral surgeon, thus I can be in there and getting care within an hour or two. And I am sure if something happened over the weekend that called for attention, he would go to the office and see me. Now, as it is, after an almost “deadly” car accident that my husband (who more or less has been my “caretaker” now for almost 10 years) last March. An 18-wheel tractor trailer “ran over” him from the back. Aa of now he is barely able to walk, much less drive anywhere. So, I have only myself to depend upon. My 2 children live 8 or more hours away, and my Mom, who I am “watching after” and helping out at times, can barely drive to the grocery store and back home that is about 10 blocks or less, thus there is NO way I could have been going to Dallas, being “put under” and driving myself home.

This part of my story only touches the “iceberg” of what I’ve had to endure. Lupus, RA, Sjögren’s, Raynaud’s, Osteoarthritis, Migraines, two heart attacks, one at the age of 40 (now they think the Lupus may have caused them, but that was years ago before I was diagnosed), plus all of the complications that go along with these illnesses. They can “attack” just about any part of the body, from the brain, to kidneys, to your heart and lungs, blood vessels, skin, and that list just goes on and on. As I have come to find out also, once they “finally diagnose” you, more than likely you have “several AI’s, causing the problems. (AI- Autoimmune diseases). We NEED HELP! WE NEED more RESEARCH, TRAINED PHYSICIANS ON THESE ILLNESSES, including DENTISTS, MORE MEDICATIONS, TO BE

DIAGNOSED EARLY BEFORE all of your body is falling apart from them. We need to find out what causes these chronic illnesses, whether heredity, or caused by something later in life. The number of people (WOMEN rank MUCH HIGHER in getting any of these than men), grows each day. Some of us, like myself, may have been ill for many years, 10 years, or more BEFORE they finally get a doctor who takes the time to sit down, and put together the pieces of the puzzle of someone with one or more of these illnesses. But, doctors are overwhelmed, underpaid, and just do not have the time, even as specialists, like Rheumatologists, to really EXAMINE a patient, and there past medical issues to find out their patient is ill with a disease that could have been there for a decade or more, By the time mine were found out, I had already had both knees replaced (after several arthroscopic surgeries), an elbow surgery, arthroscopic surgery on both shoulders, my right one now replaced completely. I have osteoporosis, and mine is "severe" in range. I have the bones of someone 80 plus years old.

This past 6 weeks have been especially an extremely fought time for me. Due to the Sjogren's literally eating away from the inside out, and no one knowing it until it was too late, I have had to endure having two sets of 5 at a time teeth pulled, much of it difficult due to the teeth being so brittle, thus difficult to excise, then after going through a couple of times trying to make sure the complete regular dentures will fit right for now, I went through a couple of visits for that. I finally got my "new teeth" just this past week on August 12th, 2014. Yet, I still had 11 teeth that had to be pulled all at once, then my gums were sutured shut, after my dentist had to do some "filing" to my bone so the dentures will fit properly, and then my gums were sutured closed in the front, top and bottom. I did then immediately get my dentures put in, but of course with extremely swollen gums, suturing and a great deal of bruising due to all of the local anesthetic being put in the syringes, then the extracting in itself, my gums developed some very sore spots, and even several mouth ulcers. Those I get with Sjogren's to my dismay. I HAVE to be able to wear these teeth, since I have none of my own now. And I cannot have them "modified" to be held in with the "mini implants" until the bone is completely healed from where all of the teeth were extracted first. This process could take 90 to maybe even 120 days. My problem is trying to have my own mouth accept them, due to the issues of ulcers, and other problems I develop in my mouth due to the Sjogren's. Then even at that there is already over \$4,000.00 paid all of my pocket to get this far with the dentures. Then in another 90-120 days, there will be another \$1200.00 (what was "left off accidentally the original treatment plan to pull all of the 11 teeth left) that we were not expecting, then another over \$3,000.00 just to have those four "mini-implant posts" put into place in my gums, then the dentures modified to fit properly on those posts. If I could wear the dentures as they are now, I may not have to incur the last \$3,000.00 plus BUT I can already see since I am suffering from ulcers, (probably a bad flare of the Lupus, Sjogren's, and RA) from all of the trauma to getting this far with the dentures, and all of the other very stressful things going on in my life at the present, it has brought a bad flaring up of the autoimmune illness. When this happens, I can have everything from those types of very painful mouth issues, severe Lupus Headaches, severe fatigue, swelling of my joints, the "mylar rash" on my face, & numerous other symptoms that are from these illnesses. It is shameful the amount of time, the loss of quality of life I have sufferer and continue to suffer, the lack of enough research, medication, doctors, and other medical items that may help to put these illnesses in permanent remission, or not get them at all, then I have to fight "tooth and nail" (no pun intended) to get my Medicare Advantage Plan to pay a dime, especially when it comes to anything to do with the huge burden of expense of all that I have endured with my mouth, and the Sjogren's that causes it. I want to see this change, for myself, and also for many others, some I have met recently that are going through the same situation that I am. Most of them like myself, really taking on a very heavy financial burden because they cannot get Medicare or a Medicare Advantage Plan to cover any of this even though it is a "medical" problem, not dental per se. For some \$9,880.00 plus (because I already had several teeth that rotted down to the gum and had to be pulled before the procedure of getting my mouth ready for the dentures) may not think that is a great sum some of money. BUT, someone living on Social Security Disability, and the spouse (my husband) that had been my "caretaker" for the most part, then he was in a severe, almost life taking vehicle accident on March 26th, 2014 and spent almost 4 months in the hospital, with many complicated injuries including his spinal cord. Thus now he had massive health needs, puts even more of a burden on my life and on his that I an barely begin to tell about. He was hit from behind by an 18 wheel tractor trailer that day, and rushed to Baylor Emergency in Dallas, where he was operated on for basically a "broken back", 7 broken ribs, and the list of injuries goes on and on. So, us trying to take care of one another is a daily struggle. Again we begin all over to try and find proper physicians for him through the system, and much like I have come to find out, many doctors are refusing

to take any Medicaid patient, and now even worse, are refusing to take Medicare patients, due to not getting paid properly, and in a reasonable time length.

I would like to see for one Medicare or these Medicare Advantage plans cover MORE on something such as Sjogren's, that is not "dental" in nature, but caused by a physical illness. I would also ask that rather than put up a road block, where the patient cannot get through to anyone at Medicare to explain the problems, or be able to get our doctors and dentists to be able to help get these types of things paid for just as any other type of chronic illness. I cannot express enough the huge amount of emotional, physical, mental, and financial anguish I have been through, and still have more to go just to be able to eat. But, my teeth are what can sustain my life, and without them, I am at risk for all types of other physical and emotional issues.

I realize we have many people in our nation, and around the globe in need of all types of medical attention. Yet, in a great nation such as the United States, our people should not have to bear such a hardship, and not have anywhere to turn to get any type of financial assistance with something so critical as your teeth. I did NOT cause this, the disease did.

I ask you to see if there is a way to change this system, give help to get things like this paid for or at least a good portion paid for by Medicare and/or a Medicare Advantage Plan. I ask you to get the funds out there to get more specialists, so we have enough doctors to are qualified and trained, that have went through a major study of these life altering illnesses, and can give us back our quality of life, and find these illnesses and have the ability to treat them before 5, 10, of more years go by and the damage has already been done.

I feel I do my part by voting, by participating as a volunteer, activist, and Ambassador for several organizations that are trying to change the face of Arthritis, Autoimmune Diseases, and all of the other medical problems that come with them, I sign petitions, I send letters, make calls, and send emails. I am somewhat limited to do what I can medically and monetarily, but in the ways I can try to stand up and make a difference, I give it my all. Now, I ask you do to the same. Stand up for all of us that want our quality of life back, to be able to do the simple things in life like eat, do a hobby, work in a garden, or whatever you may want to do, by giving the way with funds, research and voting for bills that will reduce the terrible burden off of those like myself.

Thank you for your time and for listening. I hope my "one" voice can help to make a difference.

Best regards, Pamela [REDACTED]

Congressman Upton and Congresswoman DeGette,

Thank you very much for your efforts to accelerate the pace of cure discovery, development, and deployment.

I am currently the President of the [REDACTED]. Also for nearly 15 years I, have been actively involved in academic drug discovery. Prior to coming to [REDACTED] last July, I was director of the [REDACTED]. At [REDACTED] we are establishing a Drug Repurposing facility to complement our professional-level In Vivo Facility that helps both academics and biotechs advance drug programs.

My own lab is actively pursuing a novel cure for Scleroderma and other diseases of fibrosis.

[REDACTED]

[REDACTED]

I am heartened to see the emphasis and excitement about advancing cures. As you point out in the video around the recent hearing, the potential for making substantial advances is huge. I am sure that you are going to get input on many fronts but I would specifically like to comment on the transition from discovery (generally in the academic sector) to the commercialization pipeline.

The cost to move a new chemical entity through clinical trials is very high so companies have become increasingly reluctant to take on a new development project unless it has been substantially de-risked by prior work. Improving the approval process will help that some but, realistically, one will still need to test compounds in significant numbers of individuals to ensure the level of safety expected by the American people. Even with innovations, this clinical testing will remain expensive. So pharma companies, where most of the cost of final clinical development occurs, expect projects to be farther along the development path – often to Phase I or even Phase II level.

This represents the well-known “Valley of Death” of innovation – especially in drug discovery and development. Drug companies are moving out of early stage discovery and even early development. While NIH generally provided most of the initial discoveries that lead to novel drugs in the past, now even early development is moving out of the pharma sector. The NIH has stepped in to provide some resources to help bridge this gap but the number of projects that they can take on is presently very limited. Also, with negative budget increases over the last 10 years (accounting for inflation), taking on such a “new” endeavor risks starving the pipeline that will feed new drug discoveries in 10-15 years time. While there is a critical need now to advance discoveries to the clinic, we shouldn’t have a short-sighted view that compromises the future.

I would encourage you to support a multi-faceted strategy to enhance the transition of therapeutic ideas from the lab to the clinic. This should include federal support – with new \$ for NIH to avoid destroying the basic research efforts that fill the drug pipeline – increased SBIR/STTR funds as well as ways to encourage the private sector to take back early phase drug development, perhaps tax credits or other incentives for early phase (pre-IND) discovery/development projects.

The NIH Molecular Libraries Program has spawned a cadre of academic scientists involved in drug discovery as well as many novel compounds and therapeutic ideas. I am discouraged to see how many of those stall as they begin the march toward early development.

I applaud your efforts to streamline the approval process but we also need to prime the pump. In a recent visit to China (Shanghai and Beijing), I was blown away by their current investments in biomedical research and drug discovery. While we still have the edge on them in creativity and innovation, that won't last very long if our research enterprise is spending the majority of its effort just chasing grant \$. Modest additional investments will have major benefits in advancing novel cures.

Thanks again.

Rick [REDACTED]

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

[REDACTED]  
[REDACTED]  
[REDACTED]



Dear House of Energy, Commerce Committee and 21<sup>st</sup> Century Cures Initiative,

Allow me to introduce myself. My name is Risa [REDACTED] and I am a Polycystic Kidney Disease (PKD) patient who has been impacted by generations suffering the full effects of this insidious disease that has no cure.

My father died from PKD in his early forties. My grandmother died before I was born. My brother has been on dialysis twice, in between two transplants—and I am now a transplant recipient as well. What you may not know about PKD is that it is one of the most common, life-threatening genetic diseases, causes multiple fluid-filled cysts to grow in the kidneys. It is a painful disease that can affect other organs, like the liver causing Polycystic Liver Disease. As you can see, those affected by PKD are dramatically impacted as is their quality of life.

While the average size of a normal kidney is the size of a human fist, the kidneys of a person with Polycystic kidneys can be large as a football—weighing up to 30 pounds each. If a parent has PKD, they have a 50 percent chance of passing it to each child. In my family we had two of two children effected. While PKD is genetic, it can also develop spontaneously. About 10 percent of the people diagnosed have no family history of the disease. Yet, once they have it, each of their children has a 50 percent chance of being born with PKD. Because it's passed from generation to generation, PKD often affects many people in one family.

Another important fact to consider is that PKD is the fourth leading cause of kidney failure with fifty percent of PKD patients' kidneys failing by age 50. Some are affected earlier, even in their teen's—or, like my father or brother who were impacted in their late thirties.

The only remedies for PKD patients once their kidneys fail are dialysis or a kidney transplant. While these options can be life-saving, having a treatment that preserves healthy kidney function—and ultimately a cure would be the best option.

To date, there is still no treatment to slow or stop the growth of these PKD kidney cysts, or cure to end PKD. Research is the only way to find treatments to stop or slow the growth of these cysts and prevent kidneys from failing.

It is my understanding that the 21st Century Cures is all about patients. I appreciate your consideration from this invaluable perspective, by putting the patient-first. It is my hope that you will be charged by including PKD research in the 21<sup>st</sup> Century Cures Initiative. It is also my hope that you will position PKD as a worthy cause, which deserves immediate attention (and action) to fill this unmet need.

The time has come to END PKD.

Respectfully,

Risa [REDACTED]  
[REDACTED]  
[REDACTED]

[REDACTED]

I never had symptoms of RA until a month or two after a car accident. For a long time I have been used as a lab rat for the ever elusive remission. I have had so many side effects from the many medications from sleep walking, to seizures to unbearable pain. It's hard because from month to month your symptoms are never the same. When one thing doesn't work you are immediately given something else. The wear and tear on your body from the medications are just as bad as the symptoms of the disease itself. I believe not only our physical health should be treated but it should also include our mental health. We are not the same people we were before we got sick physically, mentally or spiritually.

To Whom It May Concern,

I am writing to express my frustration with the current state of medical care for polycystic kidney disease, otherwise known as PKD. This disease currently has no cure yet affects more than 600,000 Americans today.

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.

The only remedies for PKD patients exist only once their kidneys fail, which consist of dialysis and transplantation. While these options are life-saving, having a treatment that preserves healthy kidney function is the best option.

I have been part of the clinical trial for Tolvaptan which sought to reduce the rate of growth amongst the cysts. It showed promising results and yet, against all public input and scientific data, the FDA decided it was not suitable for PKD patients; although it is being used for other ailments.

I implore you to put as much funding behind this disease as possible, while it may be too late for me, it may help my children who have a 50% of developing this genetic disease.

Thank you for your time,  
Robert [REDACTED]

Last fall, I sent a saliva test to 23andMe to obtain my genetic profile. Immediately thereafter, the FDA issued an order preventing this laboratory from releasing any medically relevant information except that dealing with my genealogy. The stated rationale for this ruling was that genetic information indicating that I could be at risk for certain diseases is inappropriate for me to know. To say that this ruling infuriates me is an understatement. I am 3 1/2 years post prostatectomy and 3 years post IMRT for Gleason 8 prostate cancer (an aggressive cell type). I have biochemical recurrence, but no evidence of macro-metastatic disease. I have also had my cancer tissue profiled by GenomeDX in California. In this case they were allowed to give me useful prognostic information, but were prohibited from giving me the RNA data of the tumor critical to optimizing my treatment. I am potentially a candidate for targeted therapy which could result in long term remission or possibly even cure. Aside from the cancer, I am in extremely good health and am pro-active in seeking the most appropriate treatment of my cancer. As long as the FDA can make such high-handed decisions about what patients may know about their genetic profile, cancer treatment will continue to be severely compromised, As a retired physician I believe I am capable of knowing such critical prognostic information without being emotionally crippled by the data. The pace of development of new therapeutic agents for cancer is accelerating, but the FDA's bureaucratic approval process significantly delays medical progress in cancer therapy.

Robert [REDACTED]

\*Those Concerned:

Viewed your round table discussion on May 14th, 2014 last night on C-SPAN 3.

It was refreshing to see an almost intelligent discussion on Big Pharma in this country. There is little doubt that when the Federal Government offers up \$30 Billion dollars a year and the Private Sector offers up \$80 Billion dollars a year for R & D Research either in Grants or Direct Investment through HHS, NIH, FDA and CDC.....the limits of Government oversight will be stretched. At the influence of AMA and the Drug Manufacturers and their influence on Research Grants and Direct Investment to identified Universities and Colleges and pretty soon you have a leviathan which can grow out of control far too easily.

Our position has simply been that the entire Industry is driven by Corporate Profits which create NO Cures, but many Daily Therapies which require drugs that are designed to NEVER CURE the disease being treated. This is purely a total lack of creativity and independent thought. The Organizations and their fiefdoms have become self perpetuating. The hand to mouth existence driven by the resources of Big Pharma create an untenable situation. This allows unethical television advertising of meaningless or harmful drugs with a high ticket price. This allows the AMA to turn a blind eye to the direct referral payments of the drug companies to both Doctors and Hospitals. This is certainly a conflict to public interest. The system has become self sustaining and influenced by the direct or indirect payment of re-election campaign funds to various members of the Congress and the various agencies involved in the Administrative process.

We will give you one example of this conflict of interest: Tamoxifen is a 40 year old drug given to most Breast Cancer patients at whatever level 1 through 4 in order to block estrogen. The drug is suggested for all patients that have not gone through the change of life and then that drug is replaced with an Aromatase inhibitor which has different side effects...many worse. This drug has gone from prescribing for five years to now ten years. The side effects are horrendous. In 40 years there has been no effort to mitigate these awful side effects. Why? The drug is now made in India with Quality Control issues as are many of the drugs being manufactured and passed around as Brand name drugs. Recently, Tamoxifen received such bad publicity that the drug is now being offered FOR FREE! Why would the drug companies ever give anything away for free? Because five other drugs are required to deal with the SIDE EFFECTS of Tamoxifen. This is just the tip of the iceberg and side effects need a strict oversight. If any drug offers life threatening side effects....it should never be allowed to be advertised on television EVER! We suppose the concept is to make the Doctors except for prescriptions that kill.

There is something very important which should come out of your proceedings: (1) Fix the Anti-biotic Crisis immediately. Infections are killing people - just coming out of surgery in out patient hospital procedures. Chronic Staff Infections and the like. We will not bother to list all the Middle East Bronchitis - MERS issues. (2) The Congress needs the HHS, NIH and FDA to determine a priority list of drugs which CURE the most important diseases in our society. Pancreatic Cancer, Alzheimer's and the list goes on. We need approvals for nano and vaccines which destroy cancer tumors anywhere in the body. These CURES, not therapies need to become ubiquitous and not in 4, 6, 8 or 10 years. We need an original space program effort to implement these cures in one to two years. The Cancer Treatment Center is doing this work today. Why is not being utilized, everywhere?

We applaud the efforts of Chairman Fred Upton from Michigan, Leonard Lance of New Jersey and Diane DeGette from Colorado and Director Francis Collins from the NIH by bringing these issues in front of the citizens of the United States. As they pointed out this effort is and should be Global in nature. What cannot continue to occur is to reward Drug Companies for adding therapies, when CURES are not only available, but are being mitigated by engineering the cure out - in order to create a therapy drug which needs to be taken and purchased. The funding mechanisms need a complete re-work project. We suggest a Special Committee on Funding created especially for this purpose. One HHS, One NIH, One FDA, One CDC, One AMA members and seven Ad Hoc members selected by the President and the Administration.



To whom it may concern:

I'm presently taking Celebrex for my arthritis. I had a full knee replacement because of arthritis.

I also have arthritis in my lower back.

I wish the drug companies could develop a safe drug to ease the pain, and better yet, something that will cure it.

Roz [REDACTED]

[REDACTED]

[REDACTED]



As you know there is No treatment for Polycystic Kidney Disease. My mother,three of her brothers and her father died from PKD. For an infinite number of generations my family has been plagued with this evil disease. My two cousins and myself, have Pkd and have been fortunate enough to have received kidney transplants.

Research must continue to find a treatment that can delay or stop the continual growth of cysts.

Sincerely

Ruth [REDACTED]

To Whom it May Concern,

On this day, June 5th, in the year 2008 my 22 y/o daughter [REDACTED] was diagnosed with a rare form of liver cancer called Fibrolamellar Hepatocellular Carcinoma. Of course we had never heard of it and the doctors didn't really seem to know much about it. Searches on the internet found little to no information except to say she probably would not survive past five years. It was difficult to find other patients because this disease affects only about 1 in every 5,000,000 people or 3 in the United States each year.

[REDACTED] tumor was very large, 14X10X8 cm and the location was concerning to the doctors because it was pressing on many of her primary organs within her abdomen. Surgery was a risk but was the only option as she was continuing to lose weight and was not healthy enough to withstand Chemo. She could barely keep any food in her. So the surgeons at University of Maryland Medical Center in Baltimore did her surgery to remove the primary tumor from her liver and some affected lymph nodes, but could not get all of it.

While [REDACTED] recovered from surgery, my sister dug into the internet and was able to find a group and by August had located a young man in Greenwich, CT who had also been diagnosed. In 2009, this young man started the Fibrolamellar Cancer Foundation ([www.fibrofoundation.org](http://www.fibrofoundation.org)) which raises money and uses 100% of donations for research. This foundation has assisted the Tucker Davis Research Lab (named in honor of the founder of the foundation) at Rockefeller University to begin research on this cancer.

The director of this lab has created a tissue bank so patients can donate their tumor tissue to be used to research this cancer. The tissue bank has little to no funding. The Director of the lab is [REDACTED] and his daughter [REDACTED] who had the cancer when she was 12, recently completed some research that identified a break in DNA in 15 of 15 tissue samples tested. [REDACTED] was at the White House Science fair just last week presenting her research to the President.

I say all of this to say that this cancer affects many and although it remains rare, it is killing our young. My daughters battle was a short

3 1/2 months from diagnosis on June 5, 2008 to death on September 18, 2008. More funding is needed to continue the research and develop a blood test to identify early on if a child had the genetic makeup that could cause them to have this cancer.

Currently there is no "cure" for this cancer. Surgery remains the best option to remove tumor's however; often the tumors are not discovered until they are too larger for surgery, they invade the entire liver, or it has metastasized and doctors are unwilling to do surgery. There is no known chemo that definitely works on this cancer, some chemo's have held tumors stable for a period of time in patients while others it does not help at all.

There is currently a clinical trial being conducted specific to this cancer and sponsored by Memorial Sloan Kettering, NYC. More information can be found at this link <http://www.clinicaltrials.gov/ct2/show/NCT01642186?term=fibrolamellar&rank=1>.

I would be happy to discuss any further information with your committee at your request.

Thank you for your time

V/R

Sandra [REDACTED]

Listed below is a section from a recent National Kidney Foundation letter to Congress I wanted to pass on again as I felt I couldn't say what needs to be said any better.

"How can Congress help?

Even in the immediate absence of breakthrough cures and treatments, Congress can take steps to reduce CKD progression and improve mortality and morbidity. To improve outcomes for patients with CKD and spur innovation Congress should:

1. Protect Medicare's investment in kidney transplants, prevent recipients from needing to return to dialysis, and potentially increase the number of kidneys available for transplant, by passing Comprehensive Immunosuppressive Drug Coverage for Kidney Transplant Patients Act of 2013 (H.R. 1428 and S.323).
2. Pass legislation that seeks to identify research gaps CKD and strengthen kidney disease research coordination across agencies (H.R. 4814).
3. Invest in research specific to kidney disease to address known gaps.
4. Encourage the FDA to use a surrogate endpoint of eGFR less than 30 in clinical trials for

drugs seeking to prevent or delay progression to ESRD, consider patient reported outcomes for approval of new CKD therapies, and include CKD in the FDA patient focused drug development initiative.

5. Explore solutions to encourage early detection and diagnosis of kidney disease in the Medicare program.

6. Provide funding for a national patient registry to allow evaluation of the course of CKD, the compliance with evidence based treatment guidelines and how clinical practice affects outcomes."

Thank-you for your time.

Sandy [REDACTED]  
[REDACTED]  
[REDACTED]

**Dear House Committee on Energy and Commerce,**

**Our beautiful 12 year old daughter [REDACTED] has Friedreich's Ataxia which is a debilitating, life-shortening, degenerative neuromuscular disorder. Thank GOD it does not impact her beautiful brain and brilliance! Presently there is no treatment or cure. It is so hard as a parent watching this awful disease take hold of our baby in a way that makes day to day tasks you and I do, a challenge for her. Please provide research funds and expedite the whole process of approving treatments and even better cures to stop this progressive disease quickly! I have done my best to complete questions below and am so grateful to be able to voice my opinions. From the bottom of my heart, thank you so much!**

**I don't know what I don't know but for me it looks as if the present FDA process was designed some time ago when test models, technology, medicine and understanding of these diseases was not understood! Today, for example for Friedreich's Ataxia the test models are superior, as the medicine and understanding of this disease is too! I would have no problem with some fixable side effects vs. debilitating life threatening disease as Friedreich's Ataxia!**

**Whatever I can do to help this cause, please let me know! Call me any time with any questions and if face to face opportunities to voice opinions would be beneficial, I will be there! I love my baby so much and know she can be treated and/or cured by something in one of these present more than 20 trials which makes it so hard not being able to help her now! Please help her and the many other families impacted by Friedreich's Ataxia!**

**Much Gratitude,**

**Sarah [REDACTED]**

**[REDACTED]**

**Answers to your questions are shown below under questions. Again thank you so much for this opportunity to express my opinions on this very important, most needed 21<sup>st</sup> Cures Bill!**

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

**It is so hard to wait as our brilliant and beautiful 12 year old daughter's Friedreich's Ataxia progresses and there is no cure or treatment. There is however a lot to be hopeful for with over 20 trials in progress and 8 of them are in clinical trial for this awful debilitating life shortening neuromuscular disorder. Friedreich's Ataxia is the most common hereditary ataxia with an estimated 5,000 to 10,000 patients in the United States. As my daughter was easily misdiagnosed for over six years, I can only imagine how many others are living with it but not properly diagnosed. My worries are that my daughter and others with this progressive disorder maybe able right now to have a treatment that can HELP her live a normal healthy life but can't get it yet but when? It would not be ok for it to be in a couple years when it is too late and she is in a wheelchair or potential worse scenarios. The big present challenge is getting something worthy approved quickly.**

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

**I now go to the National Ataxia Conference yearly and donate and volunteer for FARA.**

**Friedreich's Ataxia Research Alliance (FARA) is a foundation whose mission is to marshal and focus the resources and relationships needed to cure this awful disease by raising funds for research, provide AWESOME dedicated Doctors to patients, align government agencies, scientists and pharmaceutical companies dedicated to curing FA! There are over 20 trials going on right now and all our support to help FARA expedite and get the right tools to the right resources to find the cure does make a BIG IMPACT!!!!**

**\* How can Congress incentivize, coordinate, and accelerate basic research for diseases we know relatively little about? We know a lot about Friedreich's Ataxia, I worry it is more that there is not enough funding and research that has been done due to it being a rare disease. The ideal end results for this bill, in my opinion are:**

**Each trial should be looked at individually on its own merit to determine best process to prove effective and not harmful in an expedited fashion for Adults**

**and Kids alike. A treatment and/or cure that may benefit a child more than an adult hence to delay giving to a child too would be wrong on many levels!**

**FDA should have adequate resources to help review and expedite these life changing/saving trials effectively.**

**Someway, somehow it should be made possible so that everyone that needs these new treatments and cures can get them. These treatment costs should not be gauged to where the prices are unethical just to make a buck off of such a vulnerable community...that is not okay.**

**As FARA is our hope for a cure and have been a role model on how to help find other cures too, it would be great if they could get research, funding and FDA support to help find and expedite getting treatments to patients once known of healing properties.**

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

**Support the NIH to pay for research since many drug companies won't be interested until translational research shows promise.**

**Please enhance, not take away, monetary and resource support to FDA and NIH so they are best equipped to save lives by expediting trials, hence treatments and cures. This is a win/win as it will cost the government a lot less to cure then support for patients for a lifetime!**

**\* How do you coordinate your research and outreach with other patients?  
FARA, FAPG Group, FA Facebook groups**

**\* How do you learn about new treatments and cures? How do you communicate  
with other patients regarding treatments and cures? The internet, FARA, FAPG group, FA Facebook groups and friends**

**\* What can we learn from your experiences with clinical trials and the drug development process? Collaboration and teamwork from NIH and FDA that is well funded and has the resources to respond quickly and help to get cures to those in need as fast as possible. Specific to Friedreich's Ataxia work closely with FARA to help too.**



**\* What is the role of government in your work, including any barriers to achieving your goals and advancing breakthroughs? Support FDA and NIH to do the best job they can to research, provide safe treatments and cures to ALL those that need it as quickly and safely as possible! This will require more funding and resources within an environment that is supportive, positive and rewarding .....we all make mistakes, FDA should not be punished for this.**

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

**1.You should know for diseases like Friedreich's Ataxia to slow progression but have a side effect or two would be tremendously appreciated and is needed NOW even if not perfect, please help!**

**For example: In Europe Friedreich's Ataxia patients take Idebenone and the many of these countries governments pay for it and these patients benefit with less heart problems, more energy and better health, potentially even more years of life. This trial in the USA measured the wrong benefits hence a supplement that could help was not approved in the USA. This shouldn't of happened, there is little to no side effects and it has been proven to help many but yet it is not approved? Again as a parent to an American child that is not ok and is perfect example of what should not happen! I don't know what I don't know but I would think there are many lessons learned here and hope that they are being captured! The biggest lesson being it should be approved and why isn't it?**

**2.Focused surveys within a specific disorder and even a specific drug with knowledge of side effects might be a good approach.**

**3.Working with FARA and other groups similar for others focused on a specific drug may likely be very helpful!**

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

**The challenge is with rare diseases it is hard to get the momentum and funding needed to provide treatments/cures but yet there is still many afflicted and many yet to be born with these genetic disorders and by treating much**

**government money will actually be saved with the obvious lives saved and/or significantly enhanced to where they can now become contributing members in society. The public should help with these causes because it would be inhuman not to!**

**\* Are there success stories the committee can highlight and best practices we can leverage in other areas? What works to help one disease usually can be used to help another, it is typical to see a domino effect. FARA has many success stories and are worthy working with to learn what has worked and what doesn't.**

**\* How have you worked with other patients to support one another? We are in a Friedreich Ataxia parent group sponsored by FARA and it has been very helpful and informative in helping our daughter. I don't know what I would do without this group, so grateful!**

**\* 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 huge between modifications to house, caregivers, wheelchairs, physical therapy, in ability to work but these kids are so smart! If Friedreich Ataxia is cured they could be working and supporting community! Please, Please help make this happen!**

**\* How can Congress help?**

**1.Each trial should be looked at individually on its own merit to determine best process to prove effective and not harmful in an expedited fashion for Adults and Kids alike. A treatment and/or cure that may benefit a child more than an adult hence to delay would be wrong on many levels!**

**2.FDA should have adequate resources and funding to help expedite these life changing/saving trials effectively.**

**3.Someway, somehow it should be made possible so that everyone that needs these new treatments and cures can get them. These treatment costs should not be gauged to where the prices are unethical just to make a buck off of such a vulnerable community...that is not okay.**

**4.As FARA is our hope for a cure and have been a rule model to help find other cures too, it would be great they could get research, funding and FDA**

**support to help find and expedite getting treatments to patients once known of healing properties.**

**Again, Thank you so much for giving me a voice, this means the world to me, I love my baby girl so much and she has such great potential if only she is cured! Much GRATITUDE!!!!!!!!!!**

COMMITTEE MEMBERS:

I am a PKD patient .MY kidneys are still functioning.

The thought of dialysis or transplantation fills me with fear , and sorrow .My wife also worries silently. At this time there is no cure or treatment that would stop this disease from damaging me, and others like me.It is inevitable.

Please help those that are working to solve the problem of PKD.Only adequate funding can help.

Stopping PKD will save the enormous costs of dialysis and transplantation, and allow PKD patients to live a more normal life.

Sincerely,

Saul 

Honorable Fred Upton:

I am afflicted with 4th stage Polycystic Kidney disease. I am too old to qualify for a kidney transplant, but I still work and consider myself productive and a contributor to society. Nonetheless, I am living a slow, irreversible death sentence.

PKD is a disease for which no medication or preventive protocol has been discovered. Other than total surgical replacement, no recourse exists to slow or reverse the progress of this lethal disease.

I am the equivalent of a criminal on death row, waiting for my life to be taken by those who impede or prevent progress that would save my life. At this time, only the US Congress can provide the magnitude of resources necessary to make progress toward finding a cure.

You are my only hope. Please do not disappoint me and the myriad others here in the United States and worldwide who are so dependent upon your help to eradicate Polycystic Kidney Disease from the list of progressive, murdering illnesses.

Please remember us.

Seymour

[REDACTED]

[REDACTED]

I am a 56 years old female PKD patient. I would like to see some more research in finding a cure for this disease because it would help patient like me have a normal healthy life. I have already had a transplant with the polycystic liver disease nine years ago and I pray every day that a cure is find and I would not have to go on dialysis and/or transplantation. This does not just hurt the patient but the family too.

Please give to help in research to find a cure for this disease.

Thank you very much.

I am a PKD patient (inherited the disease from my father) and have been under treatment for the past decade; however, I use the term "treatment" rather loosely.

I have ben on a strict low potassium-low sodium-low magnesium diet , but there are no renal dieticians who understand my food. Ergo: I have essentially been hungry in a first world country. This, of course, comes with a variety of other health problems.

As if that were not enough, things are getting worse.

I have had Aetna as my health care provider from my employer, and I have bought the most expensive plan available to me. It is finally time to consider dialysis and transplant, so I began to the process for an initial diagnosis.

Guess what? The nearest "in network" facility is 250 miles from my house! So now, in addition to being seriously compromised in life style and physical ability, I have to consider whether I want to spend more money for an "out of network" facility or spend even more money on travel, accommodations for an "in network facility.

I have opted for the out of network facility because in the final analysis, it would be cheaper.

Please, can we not have enough support and help for people like me? Can we have affordable dieticians or advisors? Can we have more affordable care for PKD patients?

We are also dreading the future, since my 20 year daughter was also diagnosed with PKD last year.

It seems that there is no help, no real support for us.

Sincerely,

Shefali [REDACTED]  
[REDACTED]

To Whom It May Concern:

My one year old son has phenylketonuria (PKU) which means his body cannot break down the amino acid phenylalanine found in protein. He has a restricted diet in which he can only have a limited amount of protein per day, currently 5-6g total. This is less than the amount of protein found in an 8 oz glass of milk. It is very challenging to find low protein foods that he can eat, let alone getting enough of the food to fill him up. The low protein food offered by companies that specialize in this market are very expensive and not practical for everyday consumption. Since this food is medically necessary to ensure proper mental development, I feel this food should be covered by insurance. If we do not monitor my son's phenylalanine level and disregard the amount of protein he consumes, this will lead to mental delays and in severe cases, mental retardation.

Currently there is a gap in coverage for medical foods that are needed to treat PKU. This can be solved by passing H.R. 3665, the Medical Foods Equity Act, to ensure that the federal health programs provide coverage for medical foods for the treatment of PKU. Coverage of medical foods at the federal level would greatly influence coverage in the private insurance market. We need all the help we can get because low protein food is very costly!

Thank you for your consideration,  
Sheryl [REDACTED]

[REDACTED]  
[REDACTED]



To Whom It May Concern;

When I found out that I had PKD in 1981, my mother, who had passed the disease on to me, promised me that there will be a cure in the next 20 years and that I will not have to suffer the way she had. Well, it's been over 20 years and there's still no cure. I've watched my mother and my brother experience dialysis and all the problems related to it. There's never a day when I'm not remind that I have a limited time until it's my turn to get hooked up to a machine. I call upon you, for myself, my children and the thousands of others afflicted with this disease, to help find a cure that could preserve healthy kidney function.

Thank you.

Stephanie [REDACTED]

\* What is the state of discovery of cures and treatments for your disease?  
Are there cures

and treatments now or on the horizon?

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

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

\* How can Congress incentivize, coordinate, and accelerate basic research  
for diseases we

know relatively little about?

\* How can we work together to better translate advances in science into safe  
and effective

new therapies for patients?

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

Good Morning,

I appreciate your time as I know the hustle and bustle of your everyday lives. Waking at 5:00am, showering, getting the kids dressed and off to school, grabbing your cup of coffee, fighting the rush hour traffic to work. I remember those days as well.

Until January of 2009, our lives changed. I will never forget the day our daughters neurologist called with her genetic test results. Our precious 13 year old daughter was diagnosed with a rare genetic disease called Friedreich Ataxia.

I had done research myself prior to the dreaded phone call, I had her symptoms narrowed down to MS or a disease I had never heard of, Friedreich Ataxia. During my research I knew there was not a treatment or a cure but several possibilities on the horizon. The following month, February 2009 we went for her first visit to a wonderful, caring doctor who specializes in Friedreich Ataxia. We were told of several possibilities in clinical trials but the drugs were still in clinical trials. As a mother I stayed positive and more praying than I had ever done.

During the year our daughter took a health class and got interested in her nutrition and constant Physical Therapy.

February 2010 roles around and it was time for her 2nd annual appointment with the same wonderful caring doctor.

Much to his surprise, our daughter had improved from the following year. He talked about the research, he was very positive the treatment was getting closer. Once again, as a mother I stayed positive and more praying. Our daughter continued her nutrition and physical therapy.

Oh did I mention, our daughter not only went to her therapist weekly for physical therapy but also did her physical therapy every day on her own.

February 2011 roles around and it is time for her 3rd annual doctor appointment with the same wonderful caring doctor. Our daughter improved or maintained her stability from the previous year. The doctor was very positive about all the clinical trials in progress. Once again, as a mother I stayed positive and more praying. We return home and continue our routine, healthy diet and nutrition along with physical therapy everyday.

Then in January 2012 our daughter got the flu. Her daddy and I also got the flu which lasted about a week and we where back to normal. But our daughter took longer to recover. It took her approximately 2 weeks before she had the energy to get out of bed. Slowly she got her strength back enough to go to the bathroom and shower. Eventually she started her physical therapy exercises, but did not regain the stamina to do all the exercises she did prior to the flu.

She never gave up and continued increasing her exercises everyday. You see she knew she had her annual doctor appointment coming up. She was determined to be able to walk the 25' alone, this is just one of the test the doctor does annually. Please take just a moment, look across the room, estimate 25 feet. Yep, it doesn't seem very far to you and I, does it? But to our daughter that 25 feet seems like a mile.

We scheduled her 4th annual appointment a little later in May 2012 with the same wonderful caring doctor. Thru determination and the grace of God, our daughter walked the 25 feet that year. The doctor talked about all the clinical trials and research going on and getting closer. Once again, we returned home, as a mother I stayed positive and more praying.

Our daughter continued her nutrition and physical therapy. She still isn't able to do all the exercises she could do before the flu. But her determination kept her going and doing all she could.

May 2013 we scheduled her 5th annual appointment with the same wonderful caring doctor. Our daughter had built back about 90 percent of her strength from the previous year after having the flu. She was able to take that 25 foot walk alone, just as she did in the previous years. We just knew the clinical trials where progressing. Positive signs with certain drugs.

Once again we head home with high hopes that in 2014 we will see a treatment. As a mother I stayed positive and more praying. We return home and our daughter continues her nutrition, healthy eating habits and physical therapy everyday. Pushing herself far beyond what most people do but we start noticing less stability. As the year continues, her daddy and I are assisting her across the living room to the bathroom. She is not able to take that 25 foot walk across the living room to the bathroom without our arm to stable herself. But we still stay positive that the treatment will be here soon. How could it not, we have the best doctors, pharmaceutical companies and families raising money for the trials.

May 2014 we scheduled her 6th annual appointment with the same wonderful caring doctor. I almost cancelled her annual appointment, but I thought, no this is the year. He will have the news we have been waiting for the past 5 years. We have the

drug that will improve your way of life. But much to our dismay, we didn't hear the wonderful news we had been praying for, the news that our daughter has been working so hard for everyday of her life. But we still are staying positive and praying for the Miracle!

I do not have the answers to the list of questions which follow. I only have our families everyday hustle and bustle of life story. Yes, it differs from most of yours but at the end of our day your decision makes a difference in our lives.

Thank you for your time.

As a Mother, I will stay positive and continuing praying for the treatment and the ultimate cure!

Believe in Miracles!!!

\* How do you learn about new treatments and cures? How do you communicate with other

patients regarding treatments and cures?

\* What can we learn from your experiences with clinical trials and the drug development

process?

\* What is the role of government in your work, including any barriers to achieving your

goals and advancing breakthroughs?

\* How should regulators evaluate benefit-risk? How do you work with regulators regarding

benefit-risk? Can this process be improved?

\* What is the role of public and private funding in the research and development of cures

and treatments?

\* 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?

\* What is the financial burden of your disease? How would better treatments and cures help

save money for your family and the federal government?

\* How can Congress help?

<http://energycommerce.house.gov/press-release/latest-white-paper-seeks-input-patients-and-advocates-regarding-gap-between-known>

All comments can be sent to [cures@mail.house.gov](mailto:cures@mail.house.gov) by June 13, 2014.

Response from Steve [REDACTED] Patient Advocate. Father of Patient with Rare Disease. Actively communicated with over 160 Patient Advocacy Groups. Advocate since 1999.

My son has a rare disease for which there has been no treatment until the first drug was approved on February 14, 2014. This is an “enzyme replacement therapy” and should be helpful to patients, but not a cure. My related observations over some years brings issues to my attention that can be addressed in part by legislative change, and by congress exerting influence on the FDA to use its existing authority to drive more use of the accelerated approval pathway, and to be more flexible about approving drugs based on surrogate and intermediate clinical endpoints.

For my son this newly approved drug (Feb 2014) comes too late since the damage that has been done by the disease is irreversible. He is 27 years old and is three feet and one inch tall. Joint and tissue abnormalities make him unable to walk and he is dependent on an electric wheel chair. He has lost much of his ability to hear, is classified as “profoundly deaf” and uses a cochlear implant and hearing aid. He is not mentally affected and works and lives independently. But the disease is always in his way.

This treatment “should” have been approved in the year 2000 rather than 2014. Arguably, there were numerous issues of evidence and the regulatory process that we might never untangle in a debate about the past. But given the advances in computerized clinical data collection systems (e.g., used for clinical trials), and changes in science, we are much clearer today that the regulatory process needs to to “catch up.”

The irreversible damage that the untreated disease has caused all these years was, in retrospect, avoidable. In today's terms, the clinical trials process, designed many years ago when science was different, was largely the problem. This is true for many diseases and is one reason why of the 7000 rare diseases, we only have treatments (not cures) for about 500, or fewer, of them.

In this particular case, the clinical trial confirmed, in 2013, that the predictions regarding patient response and side effects were true. These predictions were made in 1999, when I first toured a lab and saw a vat holding the "enzyme" that was made to treat people with my son's disorder. The predictions were that side effects would be mostly mild, and patients would not be cured, but would improve, raising quality of life. Yet it would take 14 years to approval. Note that by 1999, there had already been ten years of "enzyme replacement therapy" for people with a biochemically similar disorder, Gaucher Syndrome (Genzyme made Cerezyme). By that time, 1999, people with Gaucher Syndrome had seen their quality of life rise. Side effects of the drug were primarily an allergic reaction to the substance, that was correctable by careful monitoring and dose regulation. Very similar "enzyme replacement" therapies then came out for diseases that were also biochemically similar to my son's, including for the diseases: MPS I, MPS II, MPS VI, Fabry Syndrome, and Pompe Disease. In all these cases, the main side effect was a manageable and temporary allergic reaction, handled by dose regulation.

So why, given the above, did we have to wait for each disease to get a treatment one after the other, as if waiting in line? Why can't the disorders be grouped and researched together? The answers given relate to science (the diseases in question are different enzyme deficiencies and require different trials to test, goes the argument). Also the cost is very high.

Investors cautiously watch progress of what goes before. The hurdles imposed by the FDA's required process and data collection methods 15 years ago FDA process were well known to be an issue.

So projects were slow to be started, slow to be approved, and took many years to reach patients. In retrospect, we could have approved all those drugs in the year 1999 or sooner. The hypothesis of 1999 were correct: minimal side effects, substantial benefit. In the meantime, many have died and had their lives irreversibly harmed. A related issue, the imposition of the double-blind placebo trial on these trials, in the name of safety, is inadvertently cruel.

We can find within the clinical trials *process*, of then and now, avoidable obstacles, and there are signs of improvement. Some obstacles have been targeted by the language of FDASIA, but not all of them. Even so, PDUFA V is not correct in its implementation since "Orphan Diseases" are still not being addressed as the language of PDUFA V states. We await specific mention and guidance from the FDA on a number of issues as previously documented by a letter to the FDA signed by many members of Congress in the Fall of 2013. We await helpful FDA "guidance" when a rare disease trial is proposed (IND) regarding surrogate markers, but are distressed to see that this has still required congressional pressure to come out right.

Regarding the surrogate marker issue, in 2003 I testified as a patient advocate asking the FDA to reduce the hurdles regarding validation of "surrogate biomarkers" that should be allowed instead of traditional clinical markers. Many rare diseases, but also other serious and life threatening diseases, can not be measured using clinical markers in a reasonable timeframe. That FDA Advisory Committee in 2003 (for the approval of Aldurazyme by BioMarin Pharmaceuticals) agreed with me, as each member gave comments just after I spoke. The consensus was that the FDA puts



far too many proof hurdles up regarding the validation of data. In effect, under those conditions, many trials' results could never be approved. This obstacle continues today, over 10 years later, to prevent much promising science to ever get out of the lab and into a trial since the trial is considered unapprovable, too risky, under the current FDA regulations.

Sound analysis has been made to indicate that the FDA already has the authority it needs to be more flexible and more actively promote the use of the existing accelerated approval pathways. Pressure from congress is needed to make this happen apparently. More use of ICE (intermediate clinical endpoints) has been proposed as one way, for example, for the FDA to lighten its load when reviewing a drug, rather than having to assess a trial based on unvalidated surrogate markers.

So it is remarkable, that today we are still waiting for such issues of rare disease research to be resolved by the FDA: these include:

- The FDA should use its existing authority and routinely consider the accelerated approval pathway for each new IND. (See Frank Sasinowski Hyman Phelps & McNamara research and testimony to PCAST.)
- small patient populations and the heterogeneity of response. Allow a diverse set of improvements to “count” even when the trial populations response is divided into numerous varied responses. This is the nature of these disorders. This means a wider range of endpoints should “count” in a single trial including when trial populations are very small. (Yes, that means that statistically there will be little “power” in the data.)
- use much more Risk vs. Benefit decision making. When patients will be irreversibly harmed, or die, from their serious disorder, they are willing to undergo more risk. Yet trial rules only give minor nods in that direction. Very few have ever actually benefitted from the existing

“compassionate use” rules. Many die waiting. Yet compassionate use is sometimes cited as a reason why “we don’t need” improvements to the trials process.

- The requirement of the so-called “Gold Standard” of the double-blind, placebo trial is obsolete. In cases of serious and progressive disorders, this should be eliminated. Market release of a drug after safety and minimal efficacy should be allowed, with more reliance on post-approval surveillance trials that continue to measure how patients are responding. So early in Phase II trials, using today’s terminology, many of these drugs should go to market. In some cases, earlier than that.

What has changed that should allow reduced use of placebo trials? Science has changed since the double-blind placebo standard was created. So has the ability to use computers to improve the design, planning, data collection, data analysis, safety reporting, and management of a clinical trial. These changes should allow us to look at patient and clinical trial data more effectively across diseases and patient groups.

In recent public meetings facilitated by the FDA under the Patient-Centric Drug Development, I see progress since first focusing on this area in 1999. But many of the very same questions are still being asked (e.g., would you consider a different Risk vs Benefit paradigm if you have a serious disorder?). The progress I see is the result of Congressional mandate (PDUFA V and FDASIA, Breakthrough Therapy Designation and Priority Review). The slowness however is marked. One gets the sense that there is simply a newly minted group of FDA officials, but with the same constraints, or perceived constraints, and that the agency is not a “learning organization.” That is why more congressional mandate change for the FDA is needed.

To support congressional mandated changes, both FDA and NIH need steady, reliable funding that is not interrupted by sequestration and political wind shifts

and 6 we want you to do. They agreed, and fulfilled my requests. But many of the issues persist. In 2010/2011 I contacted over 160 Patient Advocacy Groups to ask them to endorse language that to some significant extent found its way into PDUFA V to improve the trials process. From this basis of advocacy community experience I believe my comments mirror the issues of many others across many serious disease groups.

Hon. Rep Upton

If you are truly looking for cures, it's time you start thinking and looking outside the box. Md doctors are by law the only ones allowed to use the cure word, yet they cure very little. Because drugs and surgery are their only tools. The fact is all of us are nutritionally deficient. This and only this is the cause of most of our diseases. This is where you must think differently about nutrition. Nutrition isn't just carbohydrates fat and protein, 60% of the nutrition you need every day is in the form of minerals. It is impossible to get the nutrition you need from the food supply, I can't emphasize this enough, it is impossible. If you are not aggressively supplementing the 90 essential nutrients, it's only a matter of time before you get a disease. Calcium deficiency is connected to over 270 diseases.'

Here is the biggest problem with health care in this country. MD doctors are **NOT TRAINED IN NUTRITION, yet they routinely hand out nutritional advice.** Not only are they not trained in nutrition, when you suggest a vitamin or mineral can reverse or heaven forbid "CURE" something, you will be ridiculed or referred to as a quack. MD doctors practice disease management. Wholeistic and Naturopath's are the only doctors trained in medical nutrition and yet they are referred to as quacks.

MD doctors have been telling us to eat a low fat, low cholesterol diet since the early 1960's. Your hormones are 95% by weight cholesterol, your nervous system is 75% cholesterol, your brain is about 75% fat and most of that is cholesterol. If you eat a low fat diet, exercise regularly or take cholesterol medicine, you are almost 100 assured you'll get Alzheimer's.

Take type II diabetes, if you go to an MD doctor you'll get a blood glucose monitor and insulin or metformin. I could get into the technical parts of how diabetes works, but suffice to say once you become a diabetic, and you continue to see an MD doctor, you will be diabetic the rest of you life. If you see a Naturopathic doctor, you will be an ex-diabetic in two weeks and off your medication in 2 to 6 months. I said ex-diabetic because Naturopaths aren't allowed to use the "CURE" word, that's reserved for MD's even though they do not know how to cure type II diabetes. Yet 100% of the time naturopaths fix diabetes, the only caveat is if your 99 years old and have been diabetic for 60 years, your screwed, because you can only abuse the body for so long before it's irreversible.

Dr Joel Wallach has reversed more diseases and fixed more disease problems than MD doctors could hope too. The only problem is there is no money in curing some one if your using nutrition and they god give ability of the body to fix things. Dr Wallach found cystic fibrosis in a rhesus monkey, proved it was a nutritional deficiency and when he went to tell the world, he was fired and black balled by the NIH. The very people who claim to want to find cures for diseases, but only if it drug or surgery related, and that is one of the biggest problems with health care. When I can take a nutritional supplement and lower my blood pressure, loose weight, grow back bone I've lost, and fix my hypothyroidism without the help of an MD doctor, that is amazing. In fact I have done all of this over the last 6 months. Just for the thyroid alone, I have been going to an MD since 2001, and nothing they did cured the problem, they just managed it and didn't do a very good job.

The fact is, we are putting our faith and money into a system that only manages diseases and has no intention, or the knowledge to cure even some of the simplest problems. MD's can't even cure acid reflux, but if you follow their advice they will make things worse. The facts are unmistakable, diseases have sky rocketed since the MD's have been driving the bus.

I'm just a truck driver who follows and listens to a very good Naturopathic doctor and I'm healthier than I have been in decades, I'm 58. Until we start to include Naturopath's in the health care system, we will all suffer. MD doctors are absolutely necessary for trauma, infectious diseases and surgery. But if you have a chronic disease (most chronic diseases are caused by nutritional deficiencies), they are the last people you want to see.

Contact Dr Wallach or Dr Peter Glidden for anything Naturopathic. Dr Wallach is the top Naturopath in the world, and don't believe that is an exaggeration. It's time to pull back the curtain on the MD's because they failed us in so many ways and have needlessly cost the people of this country hundreds of billions of dollars.

Here are some links to Dr Glidden's webinars, they both have many on youtube also. It doesn't matter what diseases you have, it is highly likely it can be reversed or some of it's affects mitigated through nutrition.

Steven [REDACTED]  
[REDACTED]

Cholesterol myth busting [https://connectpro84292131.adobeconnect.com/\\_a993364313/p29x2n0ae6b/](https://connectpro84292131.adobeconnect.com/_a993364313/p29x2n0ae6b/)

Osteoporosis [https://connectpro84292131.adobeconnect.com/\\_a993364313/p12o8ufe3ct/](https://connectpro84292131.adobeconnect.com/_a993364313/p12o8ufe3ct/)

Muscular dystrophy <http://www.youtube.com/watch?v=NqdlwSq7nL8&feature=youtu.be>

Bone and Joint <http://webinarjam.net/webinar/go/replay/1529/e26a2ad5d0/accessok/>

Calcium [https://connectpro84292131.adobeconnect.com/\\_a993364313/p8w4pikor5h/](https://connectpro84292131.adobeconnect.com/_a993364313/p8w4pikor5h/)

Stroke [https://connectpro84292131.adobeconnect.com/\\_a993364313/p1gux3zgzjc/](https://connectpro84292131.adobeconnect.com/_a993364313/p1gux3zgzjc/)

Osteoarthritis/Rheumatoid arthritis

[https://connectpro84292131.adobeconnect.com/\\_a993364313/p341voidro2/?launcher=false&fcsContent=true&pbMode=normal](https://connectpro84292131.adobeconnect.com/_a993364313/p341voidro2/?launcher=false&fcsContent=true&pbMode=normal)

Healthy heart [https://connectpro84292131.adobeconnect.com/\\_a993364313/p9qynlihiy/](https://connectpro84292131.adobeconnect.com/_a993364313/p9qynlihiy/)

Healthy blood sugar [https://connectpro84292131.adobeconnect.com/\\_a993364313/p5bg6sdpn7i/](https://connectpro84292131.adobeconnect.com/_a993364313/p5bg6sdpn7i/)

Thank you for this opportunity to help speed the approval of critically needed drugs for suffering children and adults everywhere. They are everywhere, in every country.

My grandson, [REDACTED] was diagnosed at in 2008 at age 9 with a progressive disorder called Friedreich's Ataxia. He turned 15 just last month and is now in a wheelchair, slowly losing his ability to walk and use his hands and arms. This orphan disease will kill him, slowly..... Perhaps the worst part of this disorder is that it typically shows itself between 5 and 15 just as these developing youngsters are starting to dream of their futures and then it trashes those dreams leaving the victims cognitively intact but increasingly physically inoperative. there is not treatment, no cure. Learn more about Friedreich's Ataxia here.

<http://www.curefa.org/whatis.html>

I have answered your questions below as fully as I can but for the best quality input I recommend you arrange a meeting with the leaders of FARA, Friedreich's Ataxia Research Alliance, for the best comprehensive and focused inputs about the FDA clinical trial processes and how they might be re-balanced and streamlined.

Sincerely,

Susan [REDACTED]  
Granmother to [REDACTED] age 15, Friedreich's Ataxia

\* 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 no FDA (nor other country drug safety organization) approved treatments for Friedreich's Ataxia. Thanks in large part to our FA-family-created FARA we are in the blessed position (compared to most other rare/orphaned disorders) of having 8 drugs in clinical trial right now. None of them are approved though, time is passing and our children are dying, so your interest in speeding the approval process is of the utmost interest to us. See the status of FA research here.

<http://www.curefa.org/pipeline.html>

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

+ In 1998 some parents of FA'ers saw the need for a FA organization to focus resources and increase awareness and so FARA, FA Research Alliance, was born in that September. Now 15+ years later it is the model organization for other rare disorders. Funding comes from many quarters; grassroots fundraising, a couple of FARA-run fundraisers, many FARA sponsored fundraisers, direct donations, several well-funded individuals (FA cares not about social standing nor financial success), etc. See <http://www.curefa.org/mission.html> on the right side.

+ FARA recognized early-on the importance of having an effective research and approval process infrastructure. FA-interested drug companies and researchers now come to FARA for

o The FA registry (<http://www.curefa.org/registry.html>),

o FA clinics trained and ready as trial sites (<http://www.curefa.org/network.html>),

- o The FA Natural History Study
- o Grants (<http://www.curefa.org/grant.html>) to help further their basic and advanced research work.
- + FARA also recognized the value of collaboration and teamwork among researchers, government agencies and drug companies. Much of their progress is due to this.
- + By browsing the FARA website (<http://www.curefa.org/index.html>) you will have a much better understanding of FARA's "value-added" to FA research and the FA community.
- + Also in 1998 Sue Kittel, under the urgings of other FA families, started FAPG, FA Parents Group, an emotional, problem solving, and research communication support group whose 600+ members support FARA in various ways. <http://www.faparents.org/fapg/> More recently Facebook has been added as a research communication media.

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

- + Have the NIH pay for it since many drug companies won't be interested until translational research shows promise. For Rare disorders the subject of who to fund can be difficult. How much money do you invest for a disorder that only 10 people in the world has? I do not have that answer.
- + Congress could stop cutting the budget of the FDA and the NIH!! You do not "incentivize" nor "accelerate" by taking away their money. The FDA is being mandated to expand their various scopes of responsibility and at the same time their budget is constantly at risk and does not increase easily.
- + You've already done the perks for orphan designation and fast track. I'm not qualified to suggest other programs.

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

- + Join the collaboration between patient organizations, drug companies and researchers to identify the technologies and how to integrate them into the testing and review processes.
- + This has to be funded. And you cannot reduce the budgets of the FDA and NIH while expecting them to take this on. Won't happen. You ask for more you give them more.

\* How do you coordinate your research and outreach with other (FA) patients?

- + Through communication in the FAPG email group, FA Facebook groups, FARA FA Registry notifications and the FARA news distribution list.

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

- + Through communication in the FAPG email group, FA Facebook groups and the FARA news distribution list.

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

- + That collaboration and teamwork do work. Adversarial relationships do not work as well or as fast.



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

+ We look to the NIH to help on basic research and collaboration efforts.

+ We look to the FDA to protect our children from "bad" drug treatment alternatives and to move all drug treatments forward posthaste to separate the "wheat" from the "chaff".

+ The FDA is underfunded and understaffed. They also work in an atmosphere of fear-of-retaliation. You cannot work in this field of creation, exploration and marketing without some risk of a "bad" drug

slipping through the best of processes. When this has happened "government" did not "have their back" and instead hung them out to dry. Should we wonder why they are risk-averse?? The role of government should be to confirm the "standard of the day" and "process adherence" and then back them up! Unfortunately that is not the governmental "rule of the day". "Backing one up" is a matter of political expediency and voter damage control. I have no idea how can "incentivize" any organization in this climate of "cover your buns". Good luck.

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

+ Tough question. The FDA has struggled with this for many years. There is the medical-professional benefit-risk statistical perspective and then there is the personal benefit-risk. For the professional choose a

historical-precedent model and run the numbers. Pass/Fail? Relatively simple. Look at the exceptions, intervention, side "events". Make a judgement call (erring on the safe side; ie, "Do no harm"). For the FA

parent or patient it is much more visceral and emotional. Watch your child or yourself decline knowing there is no "cure" nor progression stoppin/slowing treatment available. Other patients die around you.

Desperation mounts; "We need it now!!" is the cry. "But at what risk?" is usually not heard and if it is heard it probably is not integrated as a real possibility/probability; "Not us/me" is the thought. How do you "work with" that dichotomy of views?

+ And does the FDA really want to absorb individually the voices of so many emotional people, and should they?

o Focused surveys within a specific disorder and even a specific drug with knowledge of side effects might be a good approach. A survey administered by the FDA to the 650+ world-wide membership of FAPG for instance might make knowledge and evaluable information from the din of individual input.

o Or working with a FARA assigned group in several meetings focused on a specific drug might be useful.

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

+ Another big question books might be written on.

+ For me public funding should be used when private funding is not forthcoming or inadequate. Again the tough question is that of volume. How much public funding should be expended on disorders that affect only "5 or 10" people in the whole world? I don't have that answer.

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

+ FARA is a success story of the highest order and should be looked at as a model for other disorders, diseases and other areas where research collaboration is needed.

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

+ The Email group FAPG and several Facebook FA groups tie us together daily with opportunities to network and hug at annual FA and Ataxia conferences. Regionally the various fundraising events draw us to one another occasionally.

\* What is the financial burden of your disease?

+ The financial and mental health burden varies state to state, how FA presents itself and the individual phase of progression.

o Many/most FA'ers never work so they are on SSI and/or on SSDI (retired parent) getting \$600-ish to \$1,000-ish a month to live on. If they live with someone SSI removes \$300-ish for room and board. If they live independently it is a big financial struggle just to live. Parents help to the limit of their own budgets and the limits set down by SSI. Copays, PT/dental/accupuncture not covered, supplements thought to perhaps help, exercise equipment, ramps, bathroom adaptations, wheelchair maintenance, etc are areas of extended cost.

+ Caregiving is needed for many adult FA'ers but even the hours that are given (often none) are not adequate. Parents wear out, get old and get injured/sick. Many FA'ers desiring to live independently cannot because they cannot get/afford caregivers.

\* How would better treatments and cures help save money for your family and the federal government?

+ You know the answer to this as well as I. Less to no caregiving, less to no Medicaid medical costs, some FA'ers might even be employable and off SSI.

\* How can Congress help?

+ Fund the NIH and the FDA at levels appropriate with the responsibilities you mandate them with.

+ Back them up instead of throwing them to the sharks when a "bad" drug is discovered.

VIA EMAIL ([cures@mail.house.gov](mailto:cures@mail.house.gov))

June 16, 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 allied disorders: PKU. I am a mother of a 6 year old PKU boy who resides in Sussex County, Delaware. 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.

•

Treatment for PKU is currently covered in 39 states through a state insurance mandate or state program. However, this coverage only benefits a small percentage of PKU patients across the country, based on their insurances or lack thereof.

•

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 (\$15 loaf of bread, \$10 per pd of pasta).

•

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,  
Susan [REDACTED] Parent of 6 year old [REDACTED] with PKU

[REDACTED]

I was diagnosed w/ Severe RA roughly 11 yrs. ago. The doctor's choice of drugs was quite limited due to what was on the market @ the time. Currently I am taking the weekly injections of Orencia.

Catamaran is working w/ our group insurance company, Cigna, in handling prescriptions. Because of my RA resultant side effects, I require meds such as Nexium, etc. The drug manufacturer states one capsule daily as the normal dosage. My Rheumatologist wrote my most recent script for pills twice daily due to GERD from OTC pain meds. To save money, rather than consider my health, the ins. co. denied the prescription co-pay. The insurance company determining my health care regimen has occurred too often. Who gave insurance companies the power to oversee what the best meds should B taken in my case & all other cases? Isn't this why we entrust our care to physicians?

Please take a detailed look @ insurance companies determining which Rx. our doctors should order for their payients. I do not want to pay 100% cost of my meds, but if it becomes the choice of treatment/prevention I have no option. More dollars come from our pockets so the insurance companies can pay higher dividends.

Please help everyone in this same predicament; you'll be surprised how many of us there are.

Susan [REDACTED]

I was diagnosed in 1986 at age 36 with osteoarthritis caused carpal tunnel syndrome. The treatment was Ibuprofen for pain & inflammation plus wear a wrist splint. In 1994 I had a Carpal Tunnel release on the right. Thousands of dollars cost for surgery to relieve the pressure on the median nerve, successful. Surgeries for the right hand in 1996. Then spinal arthritis in 1994 treatments were physical therapy, epidural injections, and cervical disc fusion finally in 1994 and again in 1998. Additional hand surgeries for arthritic thumb joints 1998 and 2003, and finally lumbar laminectomy in 2013 and cervical Discectomy and fusion again for cervical spine arthritis in 2014. Medications Ibuprofen and limited narcotic pain relievers after surgery. That is what my treatments have been for the last 28 years.

My name is Tanya [REDACTED] and I am 42 years old. I am a 5th generation PKD carrier for my family and I passed it to my daughter and possibly on down to my granddaughter which would make 7 generations strong. So far in the my family all of us have went into full renal failure by age 40 if not earlier and have either had transplants, dialysis or passed away. We have not had the luxury of having the disease and it not shutting the kidneys down. The trend we are seeing in our particular family is that it is getting worse with each generation.

I myself have sought out trials like having CT-guided cysts aspirations at Johns Hopkins and Memorial Sloan Kettering to try to save the good kidney and also for pain control. My family has tried to work through the transplants, dialysis and even sickness. My mother still works 45 hrs. a week after her 2nd transplant. while she was on home dialysis she still made it to work on time. My grandfather worked shift work at Tenn. Eastman for 13 years while on dialysis before getting to sick to work. My only Aunt worked for the Veterans Affairs at Mountain home for 20 years while she was sick, then was able to get a transplant.

It seems to be hitting my family so hard and it seems we would be a great candidate for genetic testing considering there are something getting stronger in the gene as it goes done the line with us. My aunt has had both kidney's removed and a transplant. My mother had her 1st transplant in 1999 and worked fulltime and took very good care of it for 13 yrs. She went into renal failure while we were waiting to bury my dad in Arlington National Cemetery in 2012. We Buried my dad with full rites on August 13, 2012 at Arlington National Cemetery and mom went into the hospital in renal failure a week later and started dialysis. She received her second transplant on fathers day 2013. My brother was 39 yrs. old when he went on dialysis in his 20's and was on it for 8 yrs. until he received his kidney in 2011. He has had both kidneys removed with each weighing over 20lbs each. My baby sister was diagnosed at age 23 and as of now age 35 she is still doing ok. My daughter was diagnosed at age 4 and has been on high blood pressure meds since then. She has had cardiac issues several times due to this and also her creatine levels are starting to rise. Now I have a 12 mo old grandbaby that I look at and wonder what the future holds so I write to you in the hopes that we could please get something done to help families like us that right now have no hope other than 3 options 1- dialysis which is no way to live your life this young and it is a hard life 2. you have to wait on someone to pass to donate because my whole family has this so no one can donate to us 3. DIE

Please Help Us  
Thank you

Tanya [REDACTED]  
[REDACTED]  
[REDACTED]  
[REDACTED]

To Whom it May Concern,

I am Teresa [REDACTED] 51 years of age, suffering from polycystic kidney disease. At my age I'm not as much concerned for myself but I am VERY concerned for my two children and grandchildren. As of this time the only options are dialysis, which I've seen the slow decline of health until death with my father and transplant, which is a better option but certainly not a cure. My prayers are that there will be a cure or at least a way to slow progression of this disease by the time my children will suffer from it.

I keep updated on reasearch. I know there are people out there who given the tools could find a cure. Please we need help for research to have a successful end to this disease.

Thanks for your time,  
Teresa [REDACTED]



June 10, 2014

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

Re: 21<sup>st</sup> 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 grandmother of a child with 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.
- Treatment for PKU is currently covered in 39 states through a state insurance mandate or state program. However, this coverage only benefits a small percentage of PKU patients.
- Failure to include coverage for medical foods for all patients with PKU in the federal health programs is not in accordance with the accepted standard of medical care.
- 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.
- 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.

My 2 yrs old grandson has PKU and recently his insurance coverage for his protein formula was dropped because he is no longer considered an infant. This formula is essential for his continuing health and he will have to drink it daily for the rest of his life. The cost for a month's supply of formula is \$800. Needless to say, this is a significant financial burden on his parents. This formula is as necessary for him as insulin is for juvenile diabetics, yet coverage is being denied because the word "infant" is inserted in the insurance policy as a condition for coverage. You do not outgrow PKU! Please do all you can to ensure that federal health programs provide coverage for these medically necessary dietary products.

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,  
Terry [REDACTED]

Dear Chairman Upton and Ranking Member Degette:

On behalf of the biomedical research community let me thank you for proposing the 21st Century Cures Initiative. This investment in research will benefit our entire country and the rest of the world. Let me reinforce one important point.

When I started my career 42 years ago, we had lots of questions and few answers about fundamental processes in biology, so most of medical practice was empirical rather than based on true understanding about how things worked. Since then the advances have been breathtaking. Furthermore, over this time biologists have developed tools to answer essentially any question about mechanisms in normal and diseased cells. This means that we will eventually know enough to deal rationally with the prevention, diagnosis and treatment of most diseases.

Much work is yet to be done, so the question is how long will we have to wait? Progress in biomedical research is now limited by the funds available to do the work, not by a shortage of ideas or methods. To make matters worse, the purchasing power of the appropriation for the National Institutes of Health has declined by 25% over the past decade. Just when the opportunity is the greatest, the understanding of disease is now limited by funds to do the work.

I hope that the 21st Century Cures Initiative will reverse this downward trend in the support of this vital work.

Tom [REDACTED]

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Thomas [REDACTED]

[REDACTED]

[REDACTED]

[REDACTED]

As a parent with a child living with arthritis it is very hard to find a qualified doctor to treat and diagnose a child with Arthritis. The treatments although very promising are incredibly expensive. Living with a child that wakes up out of their sleep screaming because they cannot move and finding them crawling to the bathroom is not something any parent should live through. Please consider funding of arthritis research for a cure.

Tim [REDACTED]  
[REDACTED]  
[REDACTED]  
[REDACTED]  
[REDACTED]

Please fund research for pkd therapy

Thank You

21 st Century Cures,

My son has Friedreich's Ataxia. This is a genetic disorder which is progressive with no cure or treatment. It is commonly diagnosed from age 5 to 20. About 1 in 50 thousand people have this disorder. My son uses a wheelchair full time, lives and works on his own, and enjoys life very much. He is hoping for a treatment for FA while it will benefit him, but understands that a treatment later that benefits the next generation of kids with FA will still be a victory.

It causes loss of coordination and muscle weakness from the fingers to the toes, is life shortening, scoliosis requiring spine surgery, heart disease which can be fatal, diabetes. It does not affect mental acuity.

FARA, Friedreich's Ataxia Research Alliance, was started by and is supported by parents of children with this disorder and includes many parents who have lost their children to FA.

FARA supports research and research collaboration combining public and private research funds.

There are multiple drugs in our research pipeline which show great promise for treating FA.

FARA uses patient registries, public private partnerships and venture philanthropy.

Congress can expand orphan disease status, Fund NIH and Fund FDA.

FARA has created a great model. Ask FARA how to help.

Our son has been on SSI and Medicaid. He has now graduated college, works full time and lives in an apartment with a helpful roommate, his brother. Without family and parental help, he never could have done this.

SSI is not enough. Government should allow parents and family to help their disabled adult children without decreasing SSI benefits. Home PCA hours should be generous, as living in a home with help is much less expensive than living in a care facility.

Congress should adequately fund NIH and FDA.

Thank you,

Thomas [REDACTED]  
FA parent

Hello E&C,

I am happy to see that you have decided to work on decreasing the time it takes to get an idea from discovery into development. I have been in R&D for over ten years, and currently work in the (laboratory) animal welfare compliance group at my company. One of the most costly parts of drug development is the procurement and care of the animals that are used in research and testing. The cost isn't just monetary either; there is an emotional cost for those of us who work with research animals, and for society as a whole. Animal advocates often ask how it can be "justifiable" to take so many animal lives to *potentially* save human lives.

I feel that most pharmaceutical companies would use alternatives to animals (3Rs = replace, reduce, refine) in research and testing if there were more opportunities to do so. Companies are not going to spend millions of dollars and waste precious time-to-market resources, if it is unclear that the FDA or USDA will accept the data generated by the alternative approaches.

If you want to shorten time-lines, spur innovation and reduce social pressures at the same time - start a task-force to replace animals (*in vivo*) with *in vitro* alternatives, then communicate to the R&D world what alternative methods can/should be used in new drug submission applications. It's unfortunate that animal models are the "gold standard." We can and should do better.

Thank you for your time and commitment to science. If I could be of future service, please let me know. I've always got an eye out for adventure, and have a passion for the 3Rs and alternatives.

Kind Regards,

Torrie [REDACTED]  
[REDACTED]

## 21<sup>st</sup> Century Cures –Patients

### What is Batten Disease?

Batten disease is named after the British pediatrician who first described it in 1903. Also known as Spielmeyer-Vogt-Sjogren-Batten disease, it is one of over a dozen forms of a group of disorders called Neuronal Ceroid Lipofuscinoses (or NCL).

Although Batten disease is usually regarded as the Juvenile form of NCL, the term “Batten Disease” has now become the term to encompass all forms of NCL. The forms of NCL are classified by age of onset and have the same basic cause, progression and outcome but are all genetically different, meaning each is the result of a different gene. Over time, affected children suffer mental impairment, worsening seizures, and progressive loss of sight and motor skills. Eventually, children with Batten disease/NCL become blind, bedridden and unable to communicate, and, presently, it is always fatal.

1. **What is the state of discovery of cures and treatments for your disease? Are there cures and treatments now or on the horizon?**
  - A. Genes have been discovered in 12 known forms of Batten Disease (Neuronal Ceroid Lipofuscinosis-NCL)
  - B. In 3-4 of these we have identified the function of the gene, so we know what the problem is. In these known forms, an enzyme is not produced that would usually clear waste from the cells. This failure to clear waste manifests in the brain first causing progressive deterioration and atrophy of the brain ultimately leading to death. Essentially this disease causes a once healthy brain to dissolve from the inside-out. A once healthy and happy child begins to regress, losing all skills and function until their early death.
  - C. In Batten disease we are broadly focusing on 4 therapeutic strategies.
    - 1) Enzyme Replacement Therapy. European clinical trial underway...Awaiting approval here in the USA.
    - 2) Gene Therapy. 1 clinical trial completed 1 clinical trial underway. 2 other groups in pre-clinical development.
    - 3) Stem Cell Therapy – 1 clinical trial completed.
    - 4) Small molecules – 1 clinical trial underway. Several groups in pre-clinical development
  
2. **What programs or policies have you utilized to support and foster research, such as patient registries, public-private partnerships, and venture philanthropy?**
  - A. Registries- Multiple registries of various quality. More standardization required.
  - B. Clinical Database – New worldwide database created in 2013
  - C. Validated Rating Scales – Multiple rating scales for different forms of Batten disease
  - D. Animal Models- several colonies currently maintained (dog, sheep, mouse, fruit fly, zebra fish)
  - E. Bio-Markers – Working with multiple partners to develop and validate biomarkers in the following areas: Metabolomics (plasma & CSF), Proteomics (plasma and CSF), MRI imaging.

- 3. How can Congress incentivize, coordinate, and accelerate basic research for diseases we know relatively little about?**
- A. Maintain the Orphan Drug Tax Credit.
  - B. More resources allocated to NIH research into rare diseases. Expansion of NCATS.
  - C. More resources allocated to FDA Office of Orphan Products.
  - D. A clear regulatory path is necessary at FDA. By de-risking this path, we could open the market to new investments from biotech / pharma.
  - E. Implement sec 901 of FDASIA in the manner in which its writing was intended and written into the [LAW](#). If FDA's current proposed guidance is implemented we are basically no better off than we were 3 years ago. There is an alternate set of guidelines developed in collaboration with rare disease experts and industry that does fulfill the requirement of the law...I hope you take a serious look at this set of alternate guidelines. [HERE](#) and [HERE](#). Current FDA draft guidance falls woefully short.
  - F. FDA Staff Education: The science behind rare diseases is constantly evolving, and we are concerned that FDA staff does not have enough exposure to the evolving science when reviewing applications for products to treat rare diseases. We believe that FDA staff should have access to a robust educational program to stay on top of emerging science and trends within our community.
  - G. Compassionate Use: Currently there are no incentives for companies to provide drugs in development for compassionate use to patients seeking them. Often, FDA is unfairly targeted as the reason why compassionate use is so difficult to obtain. In reality, companies developing the products have no incentives to provide their drugs to patients, as the companies are required to take all the risk (primarily financial) with zero benefit. This slows the ability for patients to be treated, and we would like to see some kind of incentive program created for companies to participate in more compassionate use programs.
  - H. Drug Repurposing: Many patented drugs are already developed and approved for common conditions which might effectively treat rare diseases. 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 patient would risk the product's market. Additionally, there is not financial incentive to do so, as adding a hundred or a few thousand rare disease patients does not increase market revenue enough to justify the costs of repurposing or the potential risk. A new orphan product market exclusivity extension is needed to incentivize repurposing for rare and pediatric diseases and cancer.
  - I. Undiagnosed Disease Network. The CAL Network (H.R. 1591), introduced by Rep. John Carter (TX), would provide a registry for primary care physicians to collaborate and find answers for the many men, women, children, and military Service Members and Veterans who have unexplained symptoms and medical problems. It would help physicians and researchers better outline demographic factors, and essentially provide physicians who are handling undiagnosed cases to search for similar cases and to network with other physicians handling similar cases in order to find a diagnosis.



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

- A. Patient advocacy organizations often seem to be left out of the equation when it comes to funding at the federal level. I would suggest a non-profit/NIH grant program that would match non-profit funding for specific disease groups on a 2 to 1 ratio with some maximum amount to be determined.

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

- A. Our national organization [REDACTED] runs a competitive RFP process modeled after the NIH review process. Proposals are scored by a multidisciplinary group of peer scientists; proposals are ranked then presented to [REDACTED] Board of Directors for award of research dollars. [REDACTED] often reaches out to smaller, partner non-profits or individuals to partner with them in funding proposals that scored well.

**6. How do you learn about new treatments and cures?**

- A. Attending scientific meetings and seminars focusing on rare disease. One of the most helpful has been the Lysosomal Disease Network meeting held every year. This meeting brings together scientists, industry and advocates and is part of the Rare Disease Clinical Research Network (RDCRN) funded by the NIH.
- B. Reading medical journals although this is cost prohibitive as I self-pay for access to the articles. I would recommend some reform of the medical journal system for publication of new findings to allow more open access.
- C. Multiple “google alerts” on subjects relating to batten disease and companies working in the rare disease space.

**How do you communicate with other patients regarding treatments and cures?**

- A. Our international patient organization [REDACTED] organizes an annual family conference each year with about 350-400 attendees each year.
- B. Many parents stay in touch and share experiences via social media – primarily Facebook-.

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

About 6 weeks after my son [REDACTED] diagnosis, we formed our organization, [REDACTED] and for the first three years, we funded a lab at Rutgers working to synthetically make the enzyme our children were missing. Eventually this synthetic enzyme caught the interest of a biotech firm who had experience in developing these enzyme replacement therapies for other similar diseases. I can't really explain the feeling of joy I had almost 3 years ago as I watched via webcast as the CEO of the company officially announced they would be developing a therapy for Batten disease ....a disease that was stealing my 2 children.

Recently however, I have learned that this trial will actually be taking place in Europe with the **possibility** of it starting here “at some time in the future”. We did have hopes that my daughter Laine would be able to participate. With this trial having been delayed here in the US, my daughter will not be eligible

due to disease progression. So my question is: Why...with the same data package .....the same trial design,..... the same therapy....is this clinical trial given the green light in Europe and not here in the United States? And this is certainly not the first time this has happened. I have heard this story over, and over again from other rare disease groups....promising therapies exported to foreign soil. This theme came up several (5-6) times during the June 10, 2014 public FDA meeting on Inborn Errors of Metabolism. However, I very much doubt this will be reflected in the final FDA report on this meeting. Our various rare disease groups have painstakingly raised the dollars to invest in preclinical research here in the United States. We have recruited academic researchers to invest their time and resources -- including NIH funding, here in the United States. We have developed registries and natural histories, here in the United States. We are working on discovering and validating biomarkers for these diseases, here in the United States. We have created and maintained animal models here in the United States. And we finally garnered the interest of an American biotech to take a chance on developing a therapy for a rare and universally fatal disease that affects innocent children. This company, and others like it are not investing small amounts in these rare diseases....billions and billions of dollars are being spent collectively, creating high-paying biotech jobs, expanding tax bases and furthering innovation here in the United States.

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

- A. NIH funding of basic research has been critical to previous work done in Batten disease. Our researchers on the cusp of several important breakthroughs and we are seeing clinical trials happen or at least be in the planning stages. However recent cuts to NIH funding are having major deleterious effects. Labs around the country are being forced to cut back or abandon promising work. Postdocs are leaving research altogether as they see no viable career path for them and their young families. Additionally animal colonies that are critical to moving forward are in danger of being lost due to these cuts.

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

In the case of rare and terminal diseases, I think patient advocates have made it quite clear to the FDA that we are willing to take more risk—perhaps not even for improvement of symptoms, but to have a chance at life. In a recent hearing at the FDA which I attended, a panelist from the FDA seemed to think that a therapy needed to be perfected for life-long therapy for a universally fatal disease. I made the point that our researchers are working on 2<sup>nd</sup> and 3<sup>rd</sup> generation treatment options that we expect will be an improvement over current treatment options being reviewed by FDA....so we do not need this therapy to be perfect...we just need it to bridge the gap until the next treatment is available.

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

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

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

**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?** Initial diagnosis took 18 months and over \$90,000. Our family’s out of pocket for out 2 affected children for medical related expenses during the past 5 years has been relatively high. I would estimate to be greater than \$100,000. Insurance billed is difficult to estimate but would expect this to be 5-7 x our out of pocket at least. These expenses do not capture home modifications we have made (50K). My wife has also had to quit her job to be able to care for our 2 affected children.

**14. How can Congress help?**

The way FDA is looks at rare diseases is broken...it’s not fatally flawed, and some very small changes are happening, but it’s not enough....some bigger changes need to be made....and made quickly.

1. Let’s have a more flexible application [of International Conference on Harmonization guidelines](#) here in the US – especially for well understood rare diseases, and stop exporting therapies denying opportunities to thousands of vulnerable Americans
2. Let’s make sure that sec 901 of FDASIA is implemented in the manner in which its writing was intended and written into the [LAW](#). If FDA’s current proposed guidance is implemented we are basically no better off than we were 3 years ago. There is an alternate set of guidelines developed in collaboration with [rare disease experts and industry](#) that does fulfill the requirement of the law...I hope you take a serious look at this set of alternate guidelines. [HERE](#) and [HERE](#)
3. Let’s keep the door open to innovation and jobs here in the US via the [Orphan Drug Tax Credit](#).
4. And let’s get the FDA & NIH the resources they need to make these changes happen.

We are not talking about lowering safety standards, but rather taking a common sense approach to the unique challenges faced by rare diseases. And once again giving hope to many who have none. Thank you for taking the time to consider my comments. I would be willing to provide further input upon request.

Kind regards,

Mr. Tracy [REDACTED]

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

Dear congress ,

As a 2nd generation pkd patient. My father being the first and a transplant patient because of it

I do as many studies about this but not to help myself, but to help future generations. Seems to be we are no further to slowing or stopping the growth of pkd, we have ways that we think slow the growth down but nothing proven. I am looking at a transplant in the next 5 years if the trends we continue to see at my annual visits with my doctor.

Thank you

Tucker [REDACTED]  
[REDACTED]

Dear House Energy and Commerce Committee;

As a Polycystic Kidney Disease (PKD) patient, I applaud your 21st Century Cures initiative. While PKD is one of the most prevalent life-threatening genetic diseases in the world, it largely remains under the radar of public awareness. The cost of PKD to the United States in terms of dollars and lost productivity is enormous. Currently the only treatment for PKD is kidney dialysis or kidney transplantation, both very expensive remedies.

If one of the goals of the 21st Century Cures initiative is to facilitate medical research to help the largest number of Americans, then finding better treatments and an eventual cure for PKD should be a top priority.

Sincerely,

Vicki [REDACTED]

[REDACTED]  
[REDACTED]  
[REDACTED]

Dear Members of Congress:

Thank you for offering rare disease patients and their families a chance to offer information on our struggles with rare diseases. I'll start by telling you our story, then try to address the questions you put forth in your request for input.

My mother, age 60, was diagnosed with Multiple System Atrophy (MSA) the day before Mother's Day weekend this past May. MSA is a very rare disease affecting perhaps 4 in 100,000 people. It is sporadic; in a few rare cases it has been shown to be hereditary. In my mother's case - MSA-C, which begins by affecting the part of the brain called the cerebellum - over the course of years, she will slowly lose her ability to walk, balance, and speak. MSA also messes up our body's autonomic functions; my mother no longer sweats and, thus, cannot regulate her own body temperature. At the end of life, MSA patients are bedridden without the ability to move on their own or communicate, but their cognition remains intact. It is similar to ALS in that way. It is a cruel disease, one I have no idea how to explain to my 4-year-old son, my mother's grandson. He wants to know why the doctors can't help her.

To your questions:

- The state of treatments or cures for MSA? There are none. There is one clinical trial taking place by the Mayo Clinic regarding stem cell treatment. It is the only one currently that shows any true promise for treatment; however, the results will not be known until at least a year from now.

- What programs have I used to foster research? There are online Facebook groups and the MSA Coalition, which offer support. I'm trying to do some personal fundraising for research. There is no patient registry for MSA that I know of. Our local office of the aging only offers caregiver support services.

- How can Congress incentivize, coordinate, and accelerate basic research? Fully fund the NIH. Reward researchers for attention to rare diseases. Create a national registry for rare disease sufferers, so that researchers can locate patients. Reward pharmaceutical companies for research into rare diseases (I know some work has been done in this area; do more).

- How can we work together to better translate advances in science...? A national registry for rare disease sufferers could help this.

- How do you coordinate your research and outreach with other patients? A facebook page. Online resources such as the MSA coalition. Without them, we would be lost and alone.

- How do you learn about new treatments and cures? How do you communicate...? Again, the facebook group and online resources. Twitter. Clinicaltrials.gov.

- What can we learn from your experiences with clinical trials...? You can learn that the process is undervalued, underfunded and takes too long. It takes an average 10 years for a new therapy to get to the patients who need it. Institute a Federal "Right to Try" law, so that patients may attempt treatments that are showing promise. Right now, my mother could be having stem cell therapy that could save her life. I can't tell you the helplessness that we feel that we have nothing to offer her. Nothing.

- The role of government and the regulators questions I'll leave to the researchers, who are the experts. I hope some respond to this questionnaire.

- What is the role of public and private funding in the research? It is the only way treatments or cures will be found. Right now, the MSA stem cell trial is being funded solely by a private donation.

- Are there success stories the committee can highlight and best practices? This, I don't know. There haven't been any in MSA that I'm aware of. Have you read Amy Dockser Marcus' fantastic piece of journalism in the WSJ called "Trials"? It highlights researcher/patient collaboration. I think this is essential.

- How have you worked with other patients to support one another? Through social media, we've developed "pen pals" for emotional support. We alert others to any clinical trials or information that we hear. We offer condolences and prayers when a member of the MSA community passes.

- What is the financial burden of your disease? We're just beginning to find out. I fear that the cost of assistive devices and long term care will deplete my parents' savings. My mother is 60 years old, so she does not qualify for Medicare. Also, she was a stay-at-home mom, so she does not qualify for Social Security benefits (not enough work credits), even though my father has worked for more than 40 years to support our family. My father is 62. If he retires now to care for my mother, his retirement benefits will be less than if he waits until full retirement age, putting their financial stability in jeopardy. Also, if he retires, my mother loses her health insurance. Obviously, this is not possible. There are several things Congress can do right now to help people with a terminal rare disease: 1) make Medicare available to everyone under 60 with a rare disease even if they are not able to be approved for SS benefits because of a lack of work credits. 2) eliminate the two-year waiting period for Medicare benefits for those with compassionate allowance diseases.

- How can Congress help? Funding, funding, funding. Give everyone with rare diseases under 65 Medicare (mothers should not be penalized because they stayed home to raise their children and didn't work outside the home) without a waiting period. This is already done for people with ALS. And finally, don't make funding and prioritizing rare disease research a partisan issue or a bargaining chip that ends up getting lost in politicians' bid to keep their jobs and keep their parties in power. Make this about what's right: helping the 30 million Americans and their families who have rare diseases and who are, in nutshell, currently without hope.

Realistically, I know that none of this will be done in time to help my mother. I can't convey to you how much it hurts for me to type those words. But I am writing to you because I know that you and I can help someone else, someone in the future. I do not want you or anyone else ever to have to go through what we're going through right now.

Thank you for your time.

My very best,  
Vicki [REDACTED]

[REDACTED]







To Whom This May Concern:

"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." - *Jackie D. Hancock, Jr., CEO of the PKD Foundation*

I implore you to please consider the lack of treatments for PKD and help find a cure under the 21st Century Cures program.

Thank you.

**All the Best,**  
Yossi [REDACTED]

To Whom It may concern:

I am the mother of four children. My youngest, an eight-year-old -child was born with PKU. My husband and I both work, our combined incomes do not allow us to receive any state or federal help. While we have been able to provide my daughter with the best foods we can afford, it has been very difficult and costly to do so these last few years when we are also paying college bills for our older boys. Our insurance coverage for her required medical food is only limited to \$2500 per year! This amount usually runs out in 5 months! All other expensive medical food we provide for our daughter is out of pocket. Don't get me wrong, I will continue to provide the best possible for my daughter. I'd much rather go without fancy designer shoes, so she can have her expensive low-protein ice cream. However, it is imperative that government officials take into consideration higher-earning-working-class citizens, and vote to pass the H.R. 3665 Medical Foods Act. Thank You.

Yvonne [REDACTED]

[REDACTED]

[REDACTED]

Hello,

My name is Zoe [REDACTED] and I'm the mother of 2 beautiful children who are living with a rare disease called Friedreich's Ataxia. I just wanted to first say thank you for taking the time to read our story. And for giving me the opportunity to share with you some of the challenges we face.

When our daughter [REDACTED] was born in 2000, she was perfect. She reached all of her developmental milestones with ease, and was such a happy and sweet baby. In 2005 our son [REDACTED] was born, he was perfect and our family was complete. Life was very very good. In 2006 when [REDACTED] was in Kindergarten she started to develop some puzzling symptoms. Her balance and coordination began to decline, it took us several years and many many tests and doctors appointments to finally receive the proper diagnosis. When she was 9 we were told that she had Friedreich's Ataxia, a rare life-shortening neurodegenerative disorder with no current treatment options. We were told that there was nothing we could do for her, and that she would slowly lose the ability to walk, talk, see, and that she would not live into her 30's. We were also told that FA is a hereditary disease and that our son [REDACTED] could also be affected. We had him tested 2 years later and were devastated to learn that he also has FA.

My husband and I had never heard of FA, we knew no one that had even had it. We had no idea that each one of us was a carrier of this horrible disease.

FA is rare and affects approx. 16,000 people worldwide. The gene for FA was only discovered in 1998 but since then an organization called FARA (The Friedreich's Ataxia Research Alliance) has been working tirelessly to fund research and collaboration efforts between scientists. Just in the past few years, we have seen a big jump in interest in the scientific community because they know what causes the disease, they truly believe this is a disease that can be treated and possibly cured. What they need is funding and for the FDA to fast track drug candidates. I once read an article that said the average time period for a new drug from idea to availability to patients was something like 18 years. We do not have 18 years. My daughter [REDACTED] has already lost the ability to walk, her speech is slurring more and more each day, her heart is affected. My son [REDACTED] is still very "normal" but we see that his energy level is dropping, he's tripping over his feet and his spine is starting to curve due to the scoliosis that accompanies FA. We need a treatment now. There are several drugs in trials right now that may be a treatment, but it could take years before they are available. And once they are available we have no guarantee that they will be affordable. I'm terrified that a drug will become available and that the cost will be out of our grasp financially. We struggle now to stay on top of the doctor bills. We have private insurance through my husbands company. We have a \$6,000 deductible, which we have met every year for the past 5 years. We've also had to make changes to our home to make it accessible for [REDACTED] and her wheelchair. We are also going to need to purchase a van with a ramp. My husband makes just enough money that we do not qualify for any federal or county assistance. I am not able to work because [REDACTED]'s disease makes it so that she misses a lot of school. The emotional and financial burden on our family is a heavy one. But we are hopeful for the future. We know that a cure will come, we just need it NOW.

Zoe [REDACTED]