Rare Disease Topic Submissions

Received by PCORI as of June 18, 2014

Advisory Panel on Rare Disease Summer 2014 Webinar – August 13

Is there any research out there about the effects of hyperthyroidism or hypothyroidism on ARVD (arrhythmogenic right ventricular dysplasia) progression? I have had a thyroidectomy for hyperthyroisim-and I have ARVD. I would like to see more research geared to those people with "other" disorders (hyperthyroidism/hypothyroidism) in combination with ARVD to see if there is any correlation to the thyroid levels, and oral thyroid meds taken and the way the disease may or may not progress. Do some of the antiarrhythmic drugs that patients may have to take affect thyroid levels? How does hyper- or hypothyroidism affect the structure of the ARVD heart? Knowing how other disease process that may affect vessels or structure of the heart could also progress ARVD would be beneficial in how ARVD is treated- and then patients could be educated about the need to monitor thyroid levels. Also IF some of the antiarrhythmic drug do affect the thyroid- this would be good info for the "young" ARVD patients.	Cardiovascular and Peripheral Vascular Disease	Arrhythmogenic Right Ventricular Dysplasia	Prioritized by the AP on APDTO on 1/13-14/2014 - Low priority
How to screen for abdominal aortic aneurysms (AAAs)? Currently AAAs are found incidentally during imaging for other medical problems. Very few people get a screening aortic ultrasound examination. Advances at the genetic level have identifed AAAs have a genetic pathophysiology. There is a need for blood tests that would then lead high risk individuals to ultrasound examinations for confirmation. AAA is a silent killer with approximately 15,000 deaths per year in the United States from this disease. The disease is "curable" with new minimally invasive aortic stent graft procedures when identified. The key is to identify the disease before it ruptures. Ruptured AAAs have an overall mortality rate of about 85%.	Cardiovascular and Peripheral Vascular Disease	Abdominal Aortic Aneurysms	
Basically anything to do with ARVD (arrhythmogenic right ventricular dysplasia) prevention so that children of patients can possibly avoid this largely-genetic disease; new meds to treat the condition; finding a cure; how much exercise is too much? I have ARVD and, more importantly, I would like to prevent my children from getting ARVD. I have friends who have lost loved ones to this condition. It's also frustrating at the apparent paucity of research into this disease, especially considering many of us were avid athletes doing what we thought was healthy for us. It's frustrating to compare the research attention we get when compared to other afflictions which are largely to individual choice/lifestyle.	Cardiovascular and Peripheral Vascular Disease	Arrhythmogenic Right Ventricular Dysplasia	Prioritized by the AP on APDTO on 1/13-14/2014 - Low priority
Can someone with a heart transplant due to ARVD (arrhythmogenic right ventricular dysplasia) still get the disease? Is the genetic disorder in the heart itself or in the blood? For people deciding if they want a heart transplant or not.	Cardiovascular and Peripheral Vascular Disease	Arrhythmogenic Right Ventricular Dysplasia	Prioritized by the AP on APDTO on 1/13-14/2014 - Low priority
One of the first things people suspected of having ARVD/C (arrhythmogenic right ventricular dysplasia/cardiomyopathy) are told to do is "stop exercising". But a life with no exercise at all is itself not healthy. We want to remain active, we want to maintain a level of fitness, but there is no information on what we really can and cannot do. There are people's opinions and ideas, but nothing founded in research. I want to know what effective and safe forms and parameters of exercise ARVD/C patients can engage in to maintain their quality of life. I have been diagnosed with ARVD/C, and on the advice of Johns Hopkins doctors have stopped exercising. And I have gained weight, have less energy, and feel my quality of life is lower. I want to know what I can do to safely remain fit and active with this disease.	Cardiovascular and Peripheral Vascular Disease	Arrhythmogenic Right Ventricular Dysplasia/Cardiomyopathy	Prioritized by the AP on APDTO on 1/13-14/2014 - Low priority
Because this particular form of pemphigoid is so rare, are there treatments that could apply to OCP (ocular cicatricial pemphigoid) patients as well as the broader patient base? Could studies on other imunosuppressant medications be applied to OCP? I have had this condition for almost 4 years and often feel completely isolated. Just knowing that someone is working to include us in studies, even though we are so few that an individual study is probably not practical, if we could be included in a broad approach, that would be helpful.	Eyes, Ears, Nose, and Throat Disorders	Ocular Cicatricial Pemphigoid (OCP)	
How can we address the cascade of disparities facing people with intellectual and developmental disabilities such as autism, Down's syndrome, and fragile X?	Functional Limitations and Disabilities	Fragile X	

My wife (recently deceased), and I, belong to an HMO. It is the only thing offered by the employer, so there really wasn't a choice about other health insurance. When my wife was diagnosed with a rare form of cancer (a sarcoma), we wanted to go to a specialist in her particular disease for a second opinion and consultation. Of course, the HMO required we go to one of their doctors for a second opinion, which we did. This seems somewhat unethical (although legal) because the doctor we went to shared office space with the oncologist we were seeing at the time. How truly independent can a patient expect that second doctor will be, since they are both employed by the same organization? For diseases in which the HMO doctors lack sufficient treatment experience in rare cancers or disease, why can't HMO's at least provide a doctor that isn't influenced by the doctrine of the HMO so a patient can truly get an unbiased opinion, without having to pay an exhorbitant sum for the service? This is important because we had to fight three rounds of appeals to get authorization to go see a specialist. This was particularly stressful on myself but on my wife as well, whose sole purpose (in my opinion) was to beat this dreaded disease. As I thought about this, and the fact that the appeals process is extremely intimidating, I became aware that patients/caregivers who have less resources to fight these kinds of battles are at a great disadvatage and must simply accept what the HMO mandates. While I understand there are agencies that can help patients deal with appeals, many patients have no idea where to turn for help in these circumstances.	Health Care Delivery System	Sarcoma	
The goals: Access to a timely, quality diagnosis that minimizes time for patients bouncing around between specialists (translational gap between PCP and specialty care). Prevention inherently suggests potential to prevent disease; this is only applicable to a subset of rare disease causing etiologies. A big challenge to answering this question is that rare disease CER is not everywhere well defined (CER relies on comparison of known options, many patients do not have known or validated options and are still working in the dark to manage their – oftentimes – chronic condition).	Health Care Delivery System	Rare Disease (General)	
Is it safe enough for me to start to transition my 18-year old with sickle cell anemia from a pediatric hematologist that is passionate about the disease to an adult system where care is fragmented and adult patients are still stigmatized for requiring narcotics to manage debilitating pain? The conversation needs to take place between all parties involved. Parents work too hard to keep their sick children alive throughout the early years of battlin with this disease, only to have to see their had work go down the drain when their children transition to an adult provider that may not have had the opportunity to treat this disease very often. Some doctors are reluctant to admit their lack of expertise or are unwilling to consult or take advice from their patients, who know their bodies best. They are often unwilling to get a consult from the patients prior hematology team when asked to do so.	Health Care Delivery System	Sickle Cell Anemia	
How can we get more funding for gene therapy or zinc finger nuclease research? This funding will help so many disorders, such as CGD (chronic granulomatous disease), muscular dystrophy, etc. One of my dear sons died of complication from CGD at the age of 32. A bone marrow transplant is too dangerous for adult CGD patients. If there was a cure for CGD through gene therapy or zinc finger nuclease, he might be alive today. I have another son with CGD who is now age 31. Most CGD patients do not live past 30 years old. Most likely he will die also without a cure. I head the CGD Association www.cgdassociation,.org and I'm aware of three CGD patient have died this past fall that were in their 20's. CGD is a rare disorder 1/200,000 We need research money now to cure this terrible disease which will also help many other diseases. This is government money well spent.	Immune System, Connective Tissue, and Joint Disorders	Chronic Granulomatous Disease	
What is mucous membrane pemphigoid/cicatricial pemphigoid/bulbous pemphigoid? How is it identified, how long does that take, how does it affect the body, and what are the treatments and how long should each be tried and what should be expected from each treatment (side affects, benefits, how long before moving to next treatment? What happens when it is in remission? It took 4 biopsies and a lot of money and discomfort and stress (luckily my dermatologist thought he recognized what it probably was...my dentist recognized it but couldn't diagnose me, and that was frustrating!). Then treatment started and I never knew what to expect from each drug or how long to stay on it and the side effects were worse than disease! And then the doctors don't really know what it is and some don't want to know. I was never even sure of the proper name of the disease MMP, CP, or BP until my contact at IPPF who has the disease explained it to me. The IPPF website has some informative information but I wanted a more indepth study of the treatments and results to be expected on each.	Immune System, Connective Tissue, and Joint Disorders	Mucous membrane pemphigoid/cicatricial pemphigoid/bulbous pemphigoid	

EDS (Ehlers Danlos syndrome) affects many people, but it is under-diagnosed and there is no specific treatment or cure. EDS has to do with "faulty collagen". Major symptoms are hypermobility, and issues with every organ, system in the body b/c collagen is everywhere. Basic layman description: the collagen is too stretchy. Collagen is everywhere, in every organ, system,etc." This means that a person w/ EDS also may suffer with POTS- postural orthostatic tachycardia syndrome, gastroparesis, chiari, spinal cord issues, bone issues including degenerative disc disease , cervical instability problems with the cardiovascular system. EDS can kill. It is very painful and debilitating. My daughter, son and I have this, the accompanying conditions,and she has Fabry disease:not producing enough of the enzyme alpha-galactosidase which allows for a build-up of GL3, a lipid. It also is very painful. Who can I contact to get research done on EDS etc to try to find effective treatments and/or cures? My family is all affected by EDS. It is terribly upsetting to have my child crying in pain and telling me it hurts and that I cannot fix it. She has been describing/crying with pain since she could talk and express this. We have tried all the natural approaches, have spent years in PT, etc. She is now taking prescription pain meds to try to knock it back enough to function. She misses a lot of school due to the pain and fatigue. We are continually going to Drs to try to find answers, and to deal with the pain. She has had several surgeries to correct torn cartilage in her hips. We have been told she needs brain surgery. this surgery is very risky and not very good outcomes. I am also a volunteer representative with CEDSA- Center for Ehlers Danlos Syndrome Alliance. We work with many EDS patients try to navigate the school system with their kids and medical issues. We also try to advocate, and support the EDS community with medical and emotional issues. IT is devastating to many people.	Immune System, Connective Tissue, and Joint Disorders	Ehlers Danlos syndrome	
How to prevent brain damage and neurodevelopmental disabilities due to excessive jaundice/hyperbilirubinemia in newborn infants? (Including severe damage – kernicterus – and subtle damage causing learning disabilities, dyslexia, ADHD, and autism.)	Liver and Biliary Tract Disorders	Excessive jaundice/hyperbilirubinemia, kernicterus	
I am caregiver for my 60-yr old sister diagnosed wtih Lennox-Gastaut seizure disoder. Because of multiple foci in the brain from which seizures precipitate, current meds are not effective for total seizure control. Currently, she has the best seizure control since the age of 11. But it has been a "hit and miss" project over the years trying different meds and wondering if we were missing just the right combination of meds that would be effective for her. Onfi, the latest additon to her regime (Lamictal, Keppra and Banzel) has resulted in the most notable seizure reduction. She now averages a 3-10 partial seizures weekly and 1-2 cluster seizures monthly. It would be helpful to be able to access information from other patients with the same diagnosis regarding meds, diet, exercise, prevention techniques that have been effective as well as "triggers" for seizure activity. Although each patient is different, I suspect there are some similarities within similar diagnoses. Thank you. It's important b/c the information is not currently available and choosing the best med is largely trial and error process, even for neurologists.	Neurologic Disorders	Lennox-Gastaut Seizure Disoder	
In addition to the (myxopapillary ependymoma) tumor, patient has kyphoscoliosis. Should deformity correction be undertaken, or are there conditions under which deformity correction is NOT adviseable with residual tumor present? Currently facing this decision.	Neurologic Disorders	Myxopapillary Ependymoma and Kyphoscoliosis	
Why is physical therapy not used more often, in conjunction with botox injections, with people with dystonia? Why have there been no clinical trials to attest to its effectiveness? Another question: Why is so little known about the number of people affected by dystonia? If there was more information on how many people affected, maybe more funding for research on cause/treatments could be generated. On the various dystonia advocacy websites you will see various numbers proposed but there really has been no systemic study in this regard. For many people with dystonia, treatment options can be frustratingly limited. Medications can have side effects that overwhelm the benefits. Botulinum injections typically help but may only address part of the problem. Deep brain stimulation (DBS) is a serious, expensive and still experimental procedure, typically reserved for certain forms of dystonia. Genetic research holds some promise but is a long time down the road and may not be relevant to all forms of dystonia. Fortunately, there has been progress in one treatment approach- that approach is physical therapy. Especially when combined with botulinum injections and provided by a qualified therapist, the "new" physical therapy offers great potential for dystonia patients. I could say a lot more in this regard but space is limited. Would love to discuss further.	Neurologic Disorders	Dystonia	

How can we improve prompt identification and treatment of PANDAS/PANS to limit adverse effects on patients and the costs associated with treating these effects? PANDAS (Pediatric Autoimmune Neuropsychiatric Disorder Associated with Strep) is a subset of PANS (Pediatric Acute Onset Neuropsychiatric Syndrome). For a description, please see: http://intramural.nimh.nih.gov/pdn/web.htm. Children stricken with this illness show a sudden onset of OCD, tics, and/or significant behavior changes following a strep, mycoplasma pneumonia or similar infection. If untreated, children with this disorder may struggle in school, suffer socially, and, if older, may resort to drugs or end up in the juvenile justice system. Undiagnosed and untreated (or undertreated) PANDAS/PANS children often spend years in the mental health system, with repeated stays on psych wards and frequent ER visits. I have two children who suffered from having this disorder undiagnosed and untreated. My son was first affected at age 6 and again at ages 7 and 9, after which he became chronic until age 13. In each case his antecedent strep was symptomless and identified only because his sister was symptomatic and he was tested. His treatment, however, was the standard 10 day antibiotic course, insufficient to address his severe symptoms. He lost his childhood to this disorder. His sister did not suffer from this until adolescence, when it took the form of severe agoraphobia and anxiety, and went unidentified for a number of years during which she went to many mental health professionals who could only deem her complicated. Pediatricians and mental health professionals should consider PANDAS/PANS when dealing with a child or adolescent experiencing sudden onset of OCD, tics, or severe behavioral changes and have them tested promptly for strep even in the absence of the usual symptoms.	Neurologic Disorders	PANDAS/PANS	
What interventions for aphasia are most effective? What is the optimal dose of aphasia therapy for maximizing patient outcomes? What neural and cognitive factors most accurately predict outcomes?	Neurologic Disorders	Aphasia	
Do more comprehensive/longer aphasia evaluations result in more positive treatment outcomes (e.g., faster and greater amount of cognitive-linguistic progress)? Health care facilities and insurance companies keep pressuring clinicians to complete their evaluation of aphasic patients (as well as patients with other communication disorders) in shorter and shorter sessions. There is no evidence base for this practice. An incomplete or cursory evaluation may result in an inappropriate or inefficient treatment plan.	Neurologic Disorders	Aphasia	
My daughter, an OT, works with an 8 year old girl who has a condition doctors at NIH say is related to CMT (Charcot-Marie-Tooth). In addition this child has severe swelling which inhibits movement and comfort. Is any effort being directed to conditions such as this one? The folks at NIH really did not come up with a diagnosis. Some kind of infantile CMT was the closest description. Thank you. The whole family is suffering and have pretty well exhausted every avenue medical knowledge available to them. They need some encouragement that someone, somewhere is working on information that would help this child.	Neurologic Disorders	Charcot-Marie-Tooth	
Can PCORI fund a study on ketogenic diet therapy for infants who are diagnosed with the serious and debiliating diagnosis of a seizure disorder called infantile spasms? I am a clinical dietitian who specializes in ketogenic diet therapy. I'm especially impressed by the excellent results that this temporary diet has on infants with the seizure disorder of infantile spasms (IS). The alternative treatment is a steriod called ACTH which costs over $250,000 per infant. Several studies have published very postitive outcomes including a recent study; 65% of infants with IS became seizure-free on ketogenic diet formula and the cost savings are immense.	Neurologic Disorders	Infantile Spasms	
If I need a clinical trial will my doctor be willing to give me the name of other doctors who will be doing trials? Who follows through from start to finish when going into a trial? It seems that is the patient's responsibility while in a confused state of mind. From fellow patients that go back and forth between origianal treating drs and trials. Confusion as to who is in charge, who patients are comfortable with, and some feelings of being left out of the loop. Mesothelioma is one cancer that has to addressed by clinical trials. There is only palliative treatment at this time.	Oncology and Hematology	Mesothelioma	

I am President of the Huisman Sickle Cell Foundation of Augusta, Inc. My question relates to sickle cell patients. Question: If sickle cell patients are serviced through a local support group vs. a clinic, what is the knowledge basic and compliance with medical protocol? My hypothesis: Support groups are more supportive of their patients vs. a clinic associated with a hospital. Thus, they may understand their medical life long regime and may be able to navigate their health better with fewer hospital admissions. I believe sickle cell patients as well as any other chronic disease patient will comply greater with knowledge and support.	Oncology and Hematology	Sickle Cell Anemia	
Barriers to utilization of hydroxyureanational utilization of hydroxyurea, a medicine that helps patients with sickle cell anemia is very low and there is an urgent needs to improve outcomes in this population. Understanding the barriers to underutilization of this medication can help us in enhancing HR-QOL of this population.	Oncology and Hematology	Sickle Cell Anemia	
Why is there no drug or chemotherapy that will put PTCL-NOS (peripheral T-cell lymphoma not otherwise specified) into remission? Because I have been fighting PTCL-NOS for a year.	Oncology and Hematology	Peripheral T-cell Lymphoma (not otherwise specified)	
With regard to cutaneous T-cell lymphoma (mycosis fungoides): What treatments are generally most successful for patients with stage 1a disease--light therapy, topical skin medications, or systemic drugs? I am at this stage and it is unclear which treatments are optimal. It seems to depend on your doctor's past experiences.	Oncology and Hematology	Mycosis Fungoides	
How can we allow the public access to medial research journal articles without charging them generally $30 or more per article? It is quite difficult to afford this when you're already caring for someone with a complex medical condition, but one for which understanding the medical research can be quite helpful in their care. My son has the rare disease Hunter syndrome or mucopolysaccharidosis II and there is little research going on and few patients. Patients often fend for themselves or band together in Facebook groups to try to answer questions and better care for their children since we often have more expertise in our child's condition than doctors for whom this may be the only case they've seen in their career. Medical journal articles could allow us to be more of the experts our children need, especially since their doctors do not have time to read all of the articles about all of the rare conditions faced by only maybe one patient in their care.	Other	Hunter Syndrome	
What plans does PCORI have to fund research questions for rare diseases, specifically rare chromosome disorders that do not have a syndrome name and rarely, if ever, get funding, but have a multitude of body system impacts? Many individuals are affected by rare chromosome disorders, but because they don't have a name, they aren't funded for research, but have many of the same symptoms as other named diseases.	Other	Rare Chromosome Disorders	
Rare diseases – don’t always have #’s still PRI.	Other	Rare Disease (General)	
For rare diseases, how do we inlcude the "Zebra" in differential diagnosis? How to communicate rare instance to patients- how to identify symptoms to look for?	Other	Rare Disease (General)	
How to fund research into rare diseases?	Other	Rare Disease (General)	
Why is it that it takes so long to diagnose a rare disease? A recent report (Shire 2013) reported that in the US it took on an average 8 physicians over 7 years to get a proper diagnosis.	Other	Rare Disease (General)	
My daughter has co-existing PTSD with medical procedure phobia and several significant health conditions, i.e. cystic fibrosis and common variable immune deficiency. We have found it impossible to find her care anywhere in the United States where they don't make both of these problems worse by holding and restraining her down for her medical care instead of providing sufficient destraction or a mild sedative. I feel like I have to choose between her mental and physical health care needs. We need a physician who will take into account her mental health problems as well as her physical ones.	Psychiatric Disorders	Cystic Fibrosis	

How can we best demonstrate to our care teams the struggles of living with a chronic illness (such as CF) so they fully understand what it is they are asking their patients to do when they flippantly suggest adding a new treatment/medication option that will extend an already lengthy 2 hour per day treatment regime to 2.5 hours a day and the impact that has on the patient and the family? The practicalities of living with a chronic illness and treating a chronic illness are two very different things. Doctors who really are trying to help their patients do not seem to see the impact it has on a patient or family when they suggest a new treatment that now requires more time and more effort from the patient and family. I am not suggesting they do not add treatments but they need to be more aware of what it is they are asking and show the patients they know that what they are asking is a lot but that the benefits far out-weigh the extra time and all that entails.	Respiratory Disease	Cystic Fibrosis	
Does treating bronchopulmonary dysplasia (BPD) in preterm neonates with diuretics improve their long term outcomes including mortality, length of ventilation, length of oxygen dependence, and long-term neurodevelopmental outcomes as compared to no treatment? If so, which specific diuretics improve outcomes in patients with BPD the most, with the least serious side-effects (osteopenia, electrolyte depletion, nephrocalcinosis)? BPD is commonly treated with long-term diuretic therapy. A soon to be published manuscript (in revision- Pediatrics) will show enormous variation between institutions in the use of diuretic for BPD patients even after controlling for risk factors/confounders. There is no evidence to support the use of long-term diuretics for BPD and these drugs have the potential for harmful side-effects (osteopenia, electrolyte depletion, nephrocalcinosis). A comparative effectiveness investigation is needed to determine if this common practice (diuretic administration) is helpful or harmful for infants with BPD.	Respiratory Disease	Bronchopulmonary Dysplasia	
Can Zyflamend be taken safely by women with LAM (lymphangioleiomyomatosis) disease? Can Zyflamend be effective in slowing the progression of LAM Disease? Reading about this herbal compound from Memorial Sloan Kettering website, I wonder if it's possible this herbal compound made up of primarily turmeric and curcumin would be beneficial to slowing the progression of lymphangioleiomyomatosis. My daughter was diagnosed two years ago. Would there be any reason not to try this? (http://www.mskcc.org/cancer-care/herb/zyflamend) There is no cure for this deadly disease that strikes down young women in the absolute prime of their lives. This is a non invasive food/herb-based supplement that has a natural anti-inflammatory effect and may possibly interrupt the disease process on a cellular level. It would be worthy of investigation.	Respiratory Disease	Lymphangioleiomyomatosis Disease	
What are the specific drug classes or individual prescription drugs that induce or trigger pemphigus in humans? There have been a few in vitro studies in addition to some anecdotal data, but expanding this field of study might be of value in preventive regimens or therapies, in addition to assisting pharmaceutical companies and the FDA in developing and approving drugs that do not carry these risks. While pemphigus in all its forms remains a rare disease, there seems to be an increase in the number of reported cases in developed countries. Some of this may be due to better diagnosis, but the role of prescribing practices and the process of approving drugs for human use should be explored. Identifying specific triggers will allow future researchers and advocates to focus their efforts on education and prevention.	Skin Disorders	Pemphigus	
Taking into account the physical and emotional impact, what are the needs of newly diagnosed pemphigus vulgaris patients? What strategies are available and what new support mechanisms could be introduced? The disease is rare and information available can be distressing. Health professionals sometimes have little knowledge of the disease. The patient may appear healthy, while struggling both physically and mentally. Patients, careers and health professionals could have a clearer picture of coping strategies and sources of support. Sometimes patients may need more than a course of treatment to cope with the realization of what this disease entails. It may help if the primary health professional is aware of this.	Skin Disorders	Pemphigus Vulgaris	

Lichen sclerosus is a little understood condition that has significant morbidity, especially for women. While super potent steroids appear to be a good first line treatment, their longterm safety is unknown. In addition, there is no information regarding the use of CAM treatments either in place of or adjuvant with topical steroids or the less widely used immune system modulators. Women are told that this is a chronic condition with no known cure while at the same time, they are told that science cannot yet tell us a specific cause. Prospective cohort studies need to be conducted with rigor to ascertain cause, prevention and treatment. The available evidence is of poor quality on anything but super potent topical steroid which helps many, but certainly more should be done to prevent the mobidity casued by this horrifying condition. Morbidity and impact on quality of life.	Skin Disorders	Lichen Sclerosis	
What research is/has been done to determine if amalgam poisoning correlates with PV (pemphigus vulgaris)? Because I was diagnosed with PV, but symptoms disappeared with removal of amalgams.	Skin Disorders	Pemphigus Vulgaris	
I have been diagnosed with pemphigus vulagaris since 2004. Since 2006 I have been taking rituximab infusions regularly. At the moment on average 2-3 times a year along with mycophenalate moefetil and prednisolone. My question is what is the long term effect of retuximab? I am 40 years now. Any suggestions on this topic would be really helpful. My question is helpful to patients like me who wants the disease under control but worried about future. What sort of adverse effect we need to face because we are in that 40 years age group. If rituximab is used for cancer paitients their cycle finished with maximum of 4-5 infusions. But patients like me we are taking year after year because it is the only one helps us to control the disease. Some suggestion on this topic would help.	Skin Disorders	Pemphigus Vulgaris	
What is the best option for remission: methotrexate or Rituximab? I was diagnosed with PV (pemphigus vulgaris) in 2009 after I have been prescribed methotrexate since the beginning of this year. I have a very plain soft diet, leading towards vegetarian. The Pemphigus almost goes into remission but it doesn't take much to damage my mouth and throat. I am wondering what would be the best treatment or other options. Or do i have to be more patient? I have heard of the Rituximab injection having good results. I am concerned that the longer I am on these harsh drugs methotrexate and prednisone the more damage it is doing to me.	Skin Disorders	Pemphigus Vulgaris	
I have lichen sclerosis, so does my 26 year old daughter. I wish there was more research on this disease. I went 18 years being misdiagnosed, then found out what I had by searching the internet. This disease is not curable, and hinders so many women, it is painful, and prevents many from having sexual relations. Why is this disease so misunderstood and proper treatment protocols are so difficult? My 26 year old daughter was diagnosed with lichen sclerosis in August of 2012. She has 2 daughters, my granddaughters, I fear that someday they will develop lichen sclerosis. This disease is very unpleasant and very painful. How would you like to take a shower and towel dry your vulva and have our skin rip open. That happened to me for years. My vulva skin would rip open if I sat down too hard in a chair, my perineum tore every time I had sex with my husband. In my support groups there are dozens of new members each week that have been recently diagnosed.	Skin Disorders	Lichen Sclerosis	
Can a cause or cure for lichen sclerosis be found in my lifetime? Please research lichen sclerosis for a possible cause/cure. Currently the "Gold standard" for treatment is clobetasol and long term use can cause almost as many problems as the condition. There is also no known cause. Maybe if a cause were found a treatment could be devised. I remember when I was diagnosed my gyn termed it a nuisance condition, her remark is typical of doctors attitude when talking about this disease, it is much more than a nuisance. It IS life altering! I know little girls as young as 2 and men and women as old as 80 who are all desperately searching for better treatment or at long last a cure. You have the power to help.	Skin Disorders	Lichen Sclerosis	
What kind of research is being done to find the cause of or treatment/cure of lichen sclerosis? This question is very important to its patients because there are so many unanswered questions about this very scary disease. As of now, the only course of treatment is a topical ointment that has very serious side effects. LS sufferers would greatly appreciate it if you would consider funding for research about this life altering disease. Thank You!	Skin Disorders	Lichen Sclerosis	

Is neurotoxic spongy fibrous syndrome (NSFS) otherwise called Morgellons a disease or myth? Because alot of people are suffering from this disorder without due medical treatment or health care assistance/preventive measures as to control its infections. A research study would enforce scientific verification of its occurence, cause, infective patterns, cures and create healthy dialogue between the clinicians,researchers, patients and the public, etc.	Skin Disorders	Morgellons	
Why has more not been done for lichen sclerosis? This question is important to me and to many others because there is a huge lack of awareness and effective long term treatment methods for LS which is an extremely personal and chronic disease. Many woman go years without being diagnosed because doctors do not properly identify the issue and are left with painful and damaging results. For woman who are lucky enough to get a diagnosis, they are simply left with the direction to use a cortisone steroid, most likely for life. Though this is the standard method of treatment, it simply suppresses the symptoms and does not address the root cause of why the body is attacking itself. Patients need MORE!	Skin Disorders	Lichen Sclerosis	
Adrenoleukodystrophy (ALD) is an X-linked disorder with an incidence of 1 in 17,000 males. It is characterized by an increase in very long chain fatty acids (VLCFA) in plasma and brain. In approximately 40% of boys with ALD, an acute inflammatory process develops within the central nervous system (CNS). The median age of onset of this neuroinflammatory process is age 7. Untreated, it is rapidly progressive and lethal, generally within several years of onset. Early in the course of cerebral ALD (C-ALD), bone marrow transplantation is effective in achieving disease stabilization. While patients early in the course of their disease have good outcomes with transplantation, many boys are not diagnosed until they are more advanced, limiting the ability of transplantation to stabilize the disease process. The question therefore becomes "Can we tell parents making decisions for their affected boys who is likely to have a reasonable outcome, and who is not?" Imagine that you have a boy that you have always thought is absolutely healthy, but he starts doing worse in school, is complaining of vision issues or hearing issues, and at some point an MRI is done. The MRI scan suggests ALD, and the disease is confirmed with a blood test. Then the parents learn that not only does there son have an inherited disease affecting the brain, but that it is progressive and will be lethal within a few years. There is one potential intervention, which is bone marrow transplantation. However, in some situations transplant may stop the disease from getting worse, but in other situations transplant may lead to a boy that is devastated neurologically, and will require virtually total care, possibly for decades. It is extremely important to develop methods to determine who should be transplanted, and who should not. This is the goal of our proposed study.	Neurologic Disorders	Cerebral adrenoleukodystrophy (ALD)	Prioritized by the AP on APDTO on 4/19-20/2013 - Low priority