CELEBRATING TWENTY YEARS
HONORING FOUNDER JANET SEGALL

HAPPY 20TH ANNIVERSARY TO THE IPPF!
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ONCE I FOUND THE IPPF I KNEW I WOULD BE OKAY
more on p. 8

21ST CENTURY CURES & ADVOCATING FOR CHANGE
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TELL US YOUR STORY!
The IPPF Awareness Campaign is looking for patient testimonials on your ORAL diagnostic experience.
For more information, email Kate at awareness@pemphigus.org or call 855-473-6744 x125.
Stories should be 300-800 words and emailed as a MS Word attachment or in the body.

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CHICAGO WAS A BLAST! I want to thank everyone who made it a success! The support from our sponsors, conference committee, staff and volunteers was tremendous! If you missed it, information is available on our website, www.pemphigus.org, or you can email Noelle Madsen at noelle@pemphigus.org and she will help you find what you’re looking for.

This issue is a good one (and a few pages thicker than usual). IPPF Founder Janet Segall was in Chicago for the Patient Conference and provides us with a summary of the weekend’s events (p. 4). This event was extra special because we celebrated the IPPF’s 20th Anniversary (p. 5). Congressman Danny Davis (Illinois 7th District) stopped by and said some wonderful things about the Foundation. Dr. Grant Anhalt presented Janet with a commemorative 20th Anniversary plate. And then Janet presented Rebecca Oling with the Founder’s Award. Rebecca has been amazing over the years and 2013 was no exception! Congratulations Rebecca!

William Zrnchik, MBA, MNM
IPPF Chief Executive Officer
will@pemphigus.org

person at a time, or auditoriums filled with medical or dental students. Find out how you can make a difference on pages 6 and 17. Dr. Terry Wolinsky McDonald tells us more about chronic illnesses (p. 7), while Dr. Mirella Bucci looks at the latest in P/P research (p. 8).

William Zrnchik, MBA, MNM
IPPF Chief Executive Officer
will@pemphigus.org

Voting is B.E.T.T.E.R. and our actions are even more important. We cannot afford to be complacent.

The IPPF has improved the lives of thousands of people since 1994. Programs like our Awareness Campaign and our Patient Education Series help medical professionals, patients, and caregivers. Thanks to donors, family members, friends, and of course, our sponsors and partners, the IPPF will be here for at least 20 more!

I'm glad that you're a part of this effort. We're in this together!

Celebrating Twenty Years.
1994-2014

People donate to the IPPF for a variety of reasons. On page 9, IPPF Treasurer Bill Gerstner shares his reasons for donating time and money to our cause.

Do you remember the scared feeling of your diagnosis? How about the relief after talking to Janet, Marc, Noelle, or any of our Peer Health Coaches? Diana’s story is all too familiar, but a call to the office and attending the Patient Conference in Chicago have her singing a new tune (p. 9).

Advocacy, Awareness, and Education are cornerstones of the IPPF. We talk with members and staff in the House and Senate advocating for favorable rare disease legislation. We increase awareness one person at a time, or auditoriums filled with medical or dental students. Find out how you can make a difference on pages 6 and 17. Dr. Terry Wolinsky McDonald tells us more about chronic illnesses (p. 7), while Dr. Mirella Bucci looks at the latest in P/P research (p. 8).

Questions?

Comments?

If you have a question for the IPPF, want to comment on a previous article, or recognize someone in our community, contact us and we’ll get you an answer or response. . . and maybe use it in a future issue of the Quarterly!

Quarterly related: editor@pemphigus.org
Foundation related: info@pemphigus.org
Letters to the Editor: editor@pemphigus.org
Disease, Treatment, Lifestyle: phc@pemphigus.org

www.pemphigus.org
Will Zrnchik, IPPF CEO, along with former board member Dr. Sahana Vyas, kicked off the weekend late Friday afternoon. Will provided an overview of the weekend to come and emphasized the importance of community participation in the IPPF. Volunteerism, fundraising, and participation in programs are all ways everyone can show support.

Dr. Animesh Sinha (University of Buffalo) began with his session on pemphigus. He discussed the clinical features of the disease, and how the specific antibodies that attack the glue in your skin cells are formed, as well as what it looks like when the cells come unglued under a microscope. Dr. Sinha talked about the genetic markers for pemphigus and how the incidences are seen more often in certain groups of people than others. He gave newly diagnosed patients a really good picture of what it looks like to live with the disease. In closing, Dr. Sinha encouraged pa-

(1-3) Tim and Becky Strong with Rebecca and Scott Oling. Becky and Rebecca (a.k.a. B2) are active members of the IPPF community.
Happy 20th!

Will Zrnchik, CEO

At the IPPF’s Patient Conference in Chicago, the IPPF celebrated a major milestone: 20 years helping P/P patients around the world. Congressman Danny Davis (district) talked about how important foundations like the IPPF are in advocating for patients, and how proud he was to speak at our event.

Dr. Grant Anhalt was one of the original supporters of the Foundation. Early on, he helped make industry connections, meet physicians and researchers, and provided expert advice that she shared with anyone who would listen.

Dr. Anhalt and IPPF CEO Will Zrnchik surprised IPPF Founder Janet Segall with a commemorative plate signifying the IPPF’s 20 successful years. The importance of having the IPPF there for newly diagnosed patients cannot be underestimated, and with the momentum in place, the next 20 years will be even more successful.

Founding the IPPF was no easy task and required extra effort, steadfast dedication, and undeniable passion. There are few people who have embodied the IPPF like Janet has. When a person exhibits similar traits, they are recognized with the IPPF Founder’s Award. This year, Janet and Will presented Rebecca Oling, IPPF Board of Directors, with the award. In 2013, to add to Rebecca’s hours of in-person and online peer support, membership on the Awareness Campaign steering committee, she appeared on the morning show Good Morning New Haven and starved in an 11-video compilation about the IPPF on the Giving Library (www.givinglibrary.com).

It took a moment for it to sink in as Rebecca was at a loss of words. With applause filling the room, Rebecca accepted the award and said how honored she is to be considered as dedicated as Janet was and is to our cause.

Afterwards, everyone enjoyed the Casino Night fundraiser and live DJ. At the end of the night, after iPads, autoographed books, and assorted prizes were claimed, $18,404 was raised for patient support!

Thank you everyone! Here’s to the next 20 years!
How to Build Awareness

Kate E. Frantz, MPH
IPPF Awareness Program Manager
awareness@pemphigus.org

The average P/P patient sees five doctors over 10 months to obtain a diagnosis. The IPPF believes five doctors is too many, and 10 months is too long. If you are a patient, you probably have a similar story to tell.

Think back to when you were seeking a diagnosis. How could this experience have been easier on you and your family? Your story can help other patients avoid these delays to get a timely diagnosis. Here are some ways you can help spread awareness.

Awareness Ambassador

Awareness Ambassadors are IPPF community members who spread awareness locally. Activities may include sending articles to local newspapers, talking with your dentist, or organizing local outreach activities. The IPPF wants to use your passions and strengths to spread awareness. Stay tuned for future Awareness Ambassador application and training announcements. If you are interested in getting involved, or just want more information, email me at awareness@pemphigus.org.

Tell someone

Maybe you are not ready to become an Awareness Ambassador, but you still want to help spread awareness. There are other ways to do this. Start by telling someone you know about P/P. Perhaps your friends or family members know you have P/P, but don’t know much about it. Educate them. Start with what feels comfortable, but don't be afraid to go outside your comfort zone.

Send us your story

The IPPF needs patient testimonials for our website. These stories will bring awareness to patients’ experiences on getting diagnosed. Stories should address the following questions:

- What is your diagnosis?
- What type of doctor diagnosed you?
- How many doctors did you see?
- How many months did it take?
- What may have made the diagnosis process easier?
- What feelings or emotions surrounded your diagnosis process?

Stories should be 300-800 words. Anonymity will be respected, if requested. Please email your story (in a Word document if possible) to awareness@pemphigus.org.

Social Media

Social media is an incredible way to build awareness. For those of you using it, keep posting, liking, sharing and commenting. For those of you new to social media, jump on in!

Start by creating a Facebook, Twitter, or Instagram account. Then connect with the IPPF. You can do this by visiting Facebook.com/HealOurSkin, Twitter.com/HealOurSkin, or on Instagram, search for @HealOurSkin. This is where you will find up-to-date information on the Awareness Campaign and the IPPF. We encourage you to use hashtags. A hashtag is a way to categorize a post by assigning a special topic to it, making it easily found in searches. Examples of hashtags to use are: #pemphigus, #pemphigoid, #awareness, #IPPFawareness, and #IPPF.

Using social media might help lead someone new to the IPPF website.

Confused? No worries, the IPPF is here to help. Email our social media guru, Noelle Madsen, at Noelle@pemphigus.org. She will help you to start using social media and have you posting, tweeting and sharing in no time.

The IPPF encourages our Community to get involved with the Awareness Campaign. If you are interested in learning more about the campaign or getting involved, please contact Kate Frantz at awareness@pemphigus.org.
Chronic illnesses are generally misunderstood. How is that for an understatement? What is the most misunderstood part? How about the word “chronic”? This fact cannot be overstated. For those of you with chronic illnesses (not just those with pemphigus or pemphigoid), how many times have you wanted to tell people to look up the word “chronic”? Believe me, I have plenty of empathy and sympathy for those of us with these disorders; no one wants or asks for one (or more) of these. There is, however, a lot to be said about feeling misunderstood. How much can friends and loved ones deal with hearing about all the things wrong with you? News flash: There is a limit.

Before diagnosis, most people are more likely to talk about how they are feeling than to write about it. They will often tell those closest to them how they are feeling and describe their basic profile of symptoms. While it may be almost all that the ill person can think and talk about, others—no matter how caring and loving—can only hear so much before shutting down their ability to hear the actual meaning of the words. To the listener, those important feelings and difficulties, pain and frustration will likely become “white noise” after a while.

We all know how difficult it can be to get properly diagnosed, although the IPPF continues to make great progress in reeducating professionals and reducing the time to diagnosis. However, a lot of patients get misdiagnoses, biopsies, and other tests that are misinterpreted or performed incorrectly. Eventually a proper diagnosis is made, and while many treatments can be difficult or even intolerable, being diagnosed represents a new chapter.

However healthy you identified yourself before the diagnosis, you will no doubt change your outlook, unless you are still in the denial stage and having difficulty accepting the change in your health. This is Your “new normal” and has a lot of variability. Add to this the often roller coaster effect of constant medication on both mood and...
Molecular Snapshot of Pemphigus

How identifying genetic markers could shine new light on PV

Mirella Bucci, PhD
Secretary, IPPF Board of Directors

Pemphigus vulgaris (PV) is a disease in which the immune system has gone awry. In the normal case, specialized proteins called antibodies allow the immune system to defend itself against foreign organisms like viruses and bacteria and also altered cells within us (such as cancer cells). The root cause of PV is the generation of antibodies that target “self” proteins that are normally ignored by the immune system.

These problematic antibodies, called autoantibodies, are found in the blood but can target the skin where they recognize specific proteins (desmogleins) located at the junctions between cells. This recognition ultimately causes, by mechanisms not fully understood, weakening of cell-cell junctions (acantholysis) and associated blistering that is the hallmark of PV.

While there is strong support that there are specific genes that predispose individuals to PV, most of these have not been identified. It is very likely, as with many human diseases, that alterations in multiple genes are required to develop PV. As well, there are very likely environmental factors, such as medications, viral and bacterial pathogens, environmental chemicals and toxins, and physical and/or emotional stress, that are contributing factors to disease development. Ultimately, there appears to be a complex interplay of genetic and environmental factors that leads to the misdirection of the immune system and a resultant attack on one’s own skin cells.

In an attempt to dissect the unique molecular environments that are formed within PV patients when all of these variables are added and to better understand the molecular basis of disease in the context of PV, the research group led by Dr. Animesh Sinha characterized the collection of RNAs found in the blood of patients with PV and compared those to healthy patients. The Sinha lab recently published their work in a paper in the journal *Genes and Immunity* (doi:10.1038/gene.2013.44).

Having a great interest in both the genetic and molecular bases of PV, the Sinha group focused their current work on the genes that are misregulated in PV. By comparing the collections of genes that are “turned on” or “turned off” during PV with those of healthy individuals, they could define molecular signatures that provide information about the molecular processes and biological pathways that are perturbed in PV.

Unlike the search for genetic mutations that cause disease, which requires determining the chemical sequence of the DNA of patients and
Why I Donate

William Gerstner
Treasurer, IPPF Board of Directors

I am frequently asked the question of why I donate to the IPPF, especially because I don’t have one of the P/P diseases.

Originally the answer was simple: a good friend of mine did so I supported the organization for her. As I got more and more involved the answer changed.

I started to see all that the IPPF can do for everyone affected by this disease. It became clear to me that in donating to this organization my dollars could do more here than almost anywhere else. There is so much to do to help those in need, so it is nice to know that my donations make a difference.

And everyone’s donations can make a difference, no matter how small or large. I hope that everyone affected can make a meaningful contribution. It’s not just about donating money. Time and dedication are also things I proudly donate, and that time can be more valuable than cash.

Bill Gerstner is the Treasurer for the IPPF Board of Directors and is a financial professional living in Chappaqua, New York.

Once I found the IPPF, I Knew I Would be Okay

Diana
Newly Diagnosed in Florida


Like most people, I had never heard the name. Looking it up online a few hours later brought tears and the shakes as pictures and definitions scrolled across my screen.

In my web travels that day, I also found the IPPF website. I hopped from link to link reading information that I assumed was trustworthy simply because it seemed like some of the people on the site had PV. But if I’m being honest? That first day was all about processing. It was enough to know that a foundation existed and that they had a website for future reference. And...well, there seemed to be a conference in Chicago. So I left the website and didn’t go back, preferring to take things day by day, make a plan with my doctors, and stay relentlessly positive. Thankfully, the PV started to stabilize.

But the idea of the conference lingered. I debated with myself. Did I want to go to and meet a bunch of other sick people? Would what I learned bring me down? Or

CONTINUED ON PAGE 13...
patients and their relatives to donate blood to further his research on the causes of pemphigus and to create better treatments.

Dr. Amit Shah (University of Buffalo) presented on the IPPF Registry and what the data tells us. Pemphigus and pemphigoid are rare diseases so having a registry helps promote better understanding of the diseases around the world. A primary goal of the study is to investigate different characteristics of patients enrolled. The registry shows gender prevalence, average age, and racial/genetic breakdowns are. The registry data tells us more women diagnosed, and the average age of onset is 40-60 years. The data indicates women have mucosal activity more than men, while men are more prone to skin lesions. These findings will help researchers and physicians expand their knowledge of the disease.

Dr. Razzaque Ahmed (Boston Blistering Disease Clinic) rounded out the evening with an overview of pemphigoid. He explained how pemphigoid was different from pemphigus by location and look of the blisters. He said mucous membrane pemphigoid (MMP) and cicatricial pemphigoid (CP) typically affect middle-aged (and older) individuals. He explained the differences between bullous pemphigoid (BP) & MMP stating with ocular MMP the trachea can also be affected. Dr. Ahmed emphasized that early diagnosis and treatment is essential, especially with MMP (individuals can lose their sight or breathing capacity due to scarring).

Sahana and Will opened Saturday’s session with a warm welcome and were followed by IPPF Board President, Dr. Badri Rengarajan. Badri began with the importance of the IPPF to people with P/P - newly diagnosed, in a flare, in remission, and everywhere in between. He told the audience the Foundation makes all its resources available to patients, caregivers, family members, and medical professionals free of charge. Knowing this, it is equally important for the Foundation going forward to continue to help others for years to come. Badri mentioned four ways the Foundation helps patients: improving quality of life;
reducing diagnostic time; understanding and coping with flares; and supporting new diagnostic methods and research. The asked the audience to reach out to the Foundation when they need help and to support the Foundation to increase our services.

**Dr. Sergei Grando** (University of California - Irvine) discussed prednisone (what corticosteroids are commonly known as) and how steroids work. He mentioned side effects of steroids and affects on patients. He suggested the treatment process should be a team effort. Dr. Grando also spoke on adjuvant drugs (to reduce steroid doses) and the use of IVIg and an immunosuppressive to reduce disease activity.

**Dr. Razzaque Ahmed** returned to the stage for a talk on treatment side effects. He commented on how extreme cases of P/P can end up in burn units – not an appropriate treatment. Dr. Ahmed suggested a patient’s treating physicians should be told what drugs are being taken so treatment for additional problems is carefully coordinated. He talked about prednisone side effects and the importance of keeping track of them to share with your physician. He discussed side effects of immunosuppressives (such as Imuran®, CellCept®, and Cytoxan® and their link to cancer), IVIG, Rituxan®, and other treatments. At the end, Dr. Ahmed emphasized open communication with all a patient’s physicians to ensure the best possible care.

Did you know 13 million liters of plasma are collected each year, and the antibodies extracted from this plasma is what makes IVIG? **Dr. Michael Rigas** (Kabafusion) explained this, and more in his talk. He told the audience how the drug is made, where it comes from, and why it costs what it does. Dr. Rigas then explained how it is administered to a patient, and what patients should expect after the infusion. He said IVIG as a P/P treatment is not approved by the United States FDA. He closed by saying there are many fac-
energy. Without some measure of resilience, a good support system, and doctors who are able to both treat you and communicate with you, this upheaval in your life can be a recipe for disaster.

A supportive network of peers, such as health coaches, social media, local support groups, and patient forums, all can make the journey less difficult. Still, chronic illness is now part of your life, most likely forever. In contrast to other equally debilitating conditions, you will not undergo surgery and then recover and be able to put it behind you. To make matters more grim, having one chronic illness does not play any part in your susceptibility to another chronic disease. You may in fact be more susceptible to additional autoimmune or other diseases. The message here is to not allow yourself or your doctors to lose perspective or to have tunnel vision.

“Although the world is full of suffering, it is also full of the overcoming of it.”

Helen Keller

Once you know that you have a chronic illness, invisible though it may be to others, you will now be living with whatever the illness and treatment is doing to your body and psyche. You will eventually come to accept certain limitations that are inevitable in a body at war with itself. You will likely feel that your own body has betrayed you. You will be angry and may feel punished, scared, sad, anxious, or all of the above. If unable to get all these natural feelings under control, you may fall into a downward spiral. Be aware.

It is also important for those with chronic illnesses to become educated patients rather than “good” patients. I am not in any way advocating noncompliance, but rather urging you to be active and proactive in your treatment. It is your body, and once diagnosed with chronic illness you are the expert on
...continued from ONCE I FOUND THE IPPF, page 9

was knowledge and togetherness power? I started re-
searching the conference agenda and realized that not
going wasn’t an option.

The session content included information that I desper-
ately needed to learn, from PV 101 to treatments to nutri-
tion. Better still, the sessions were presented by research-
ers and specialists from the nation’s top medical centers
and universities as well as by patients and IPPF staffers
who have personal insight into these diseases. I learned so
much in one weekend!

I can now say that the Chicago conference was the
turning point for my taking control of this disease pro-
cess rather than just passively listening to my doctors and
following directions. I’m full of questions. Now, because I
have a fuller understanding, I can be a partner in my own
care.

the Chicago conference was
the turning point for my
taking control of this disease
process

Most importantly, I learned that I’m not alone. The ex-
perience of meeting other people with PV has been pro-
found. Everyone asked questions of each other and shared
their stories. The conference was a little under a month
ago, and I find myself wistful now for Chicago and wishing
the dates for New York were already set. But meanwhile,
the connections remain.

I’ve talked to wonderful people who, like me, are pa-
ients. Two people from my state shared doctor’s refer-
ces with me. Another attendee has checked in with me
twice and we’re well on the way to becoming friends.

I’ve had several contacts from IPPF staff in response
to questions I’ve had. The IPPF’s work continues, because
life and this illness continue. It’s comforting to know that
there is a central hub for those of us with PV and related
diseases where we are understood and cared for as part
of a larger community. Thank you to everyone involved
in making this organization so vibrant and responsive.

Tips for Better
Office Visits

Write your questions down. This will help you organize your thoughts and reduce
the chances of forgetting to ask an important question. Ask the most important ones first.
You should bring an extra copy in case you can’t get them all answered during your visit.

Be a courteous patient. Delays hap-
pen, visits run long, and emergencies arise. Bring a book or magazine with you. If you have
a tablet or smart-phone, bring that. The wait
may be longer than you anticipated, but at
least you’ll get the most out of it.

Personal Physician Network. We see
many physicians. As the patient, you should
share your treatment information with each of
them. If there is a conflict, say the need to
come off of your P/P medication because you
need something else for a different condi-
tion, encourage them to talk to each other and
come to an agreement that benefits you.

Do you have your own tips?
If you do, send them to Noelle Madsen at
noelle@pemphigus.org.

SMOOTHIE RECIPE
Vicky Starr vstarr@medprorx.com

INGREDIENTS
• Vanilla Whey Protein powder
• Truvia®/Stevia®
• Jarred Peaches in white grape juice (in a glass jar at
Trader Joe’s® or Whole Foods®)
• 1 large Strawberry
• Coconut Milk
• Ice
• Blender

HOW TO PREPARE
1. Fill blender with Ice ¾ full (less if you desire)
2. 2 Large Scoops of vanilla whey protein
3. 6 halves of peaches and ¼ cup of the juice
4. 1 large washed Strawberry
5. 3 packets of Truvia®/Stevia®
6. Pour the coconut milk over the ingredients
   until it covers ½ the ice.
7. Blend until smooth and ENJOY!!

Diana is a newly diagnosed patient living in Florida.
tors to be considered before a patient gets IVIG and to talk with your doctor if you have questions.

Dr. Grant Anhalt (Johns Hopkins University) presented on the physiology of PV. He explained how and why the cells detach from one another. He said many currently-prescribed anti-inflammatory drugs do nothing to inhibit antibody production. He provided a recap of Imuran®, CellCept®, IVIG, and rituximab and how they work on P/P. He has found rituximab has been very successful in the treatment of PV without the side effects commonly found in cancer drugs. Dr. Anhalt described how rituximab destroys maturing B-cells for 6-9 months and how results of several studies showed the success of rituximab in early stages of PV.

Victoria Carlan (IPPF Board member and founder of the Canadian Pemphigus & Pemphigoid Foundation) talked about personal support networks. She opened with her personal PV journey explaining the importance of her support network, and how she used it to live successfully with P/P. This enabled her to find answers and find encouragement. She explained how support networks can build-up physical, mental and emotional strengths.

IPPF Awareness Program Manager

Kate Frantz talked about the IPPF’s Awareness Campaign. Building awareness in the medical community is important to reducing diagnostic time for patients. She said we can all help with awareness in our own ways. One way is becoming an Awareness Ambassador in your community. Awareness Ambassadors will go into their community to spread P/P awareness. Others can write to newspapers, speak at professional gatherings, and engage others in your community. She stressed the importance of spreading awareness through social media to help create a “brand” others can relate to the IPPF and P/P.

One of the IPPF Awareness Campaign’s Patient Educators, Rebecca Strong, discussed additional ways to spread awareness. People can write to their federal, state, and local representatives encouraging them to be involved with improving your health and supporting legislation that benefits all of us. Be your own advocate and ask those you know who might be able to help advocate for you. There really truth to the Power of One.

Dr. Firdaus Dhabhar (Stanford University) presented on stress & autoimmunity. Dr. Dhabhar discussed the biological responses that happen with stress are not always negative, but can be positive. Short-term, acute stresses (such as surgery, vaccinations, etc.) can enhance positive immune response. However, chronic, long-
long-term stresses have negative effects on the body. With long-term stresses, the goal is to minimize their effects with better sleep, nutrition, exercise, calming activities, or whatever works for you.

On Sunday, the patients took center stage for a patient panel discussion. Our panelists included IPPF Senior Peer Health Coach Marc Yale (MMP/OCP), Becky Strong (PV), Peer Health Coach Mei Ling Moore (PV), IPPF Board member Rebecca Oling (PV), and Janet Segall (PV). Questions covered personal best practices, dealing with side effects, and product recommendations.

This successful segment was followed up with a 90-minute teleconference in May 2014 where over 80 people registered with 40 people on the call at any given time.

Following the patient panel, the IPPF hosted several workshops. These smaller, focused sessions were on topics such as different stress reduction methods, diet and nutrition, oral care, ocular concerns, IVIG, and reimbursement issues. There was also a successful focus group centered on the Awareness Campaign.

Once the workshops concluded, attendees gathered back in the main room for a Q&A with some of the weekend’s speakers. Questions were asked, debated, and answered by experts from different specialties.

Will and Badri reminded everyone we all can be involved in making sure newly diagnosed patients get the help they need by participating in IPPF programs and donating to our cause. And during his closing remarks, Will announced the 2015 Patient Conference will be in New York, and information will flow as it is available.
One thing that we all can agree upon in this day and age, with modern technology rapidly moving forward, is if we want to find a cure or even what triggers pemphigus and pemphigoid we need to accelerate the pace of research. The 21st Century Cures Initiative does just that.

Introduced to Congress in May 2014 by Representative Fred Upton, a U.S. congressman representing Michigan’s 6th District, and Representative Diana DeGette, Chief Deputy Whip, the initiative is a bipartisan effort that creates an opportunity to bring researchers, caregivers, and patients together to keep discovery and medicine a top priority for our country.

The collective action of the initiative will drive basic research, streamline drug development, and introduce new technology in digital medicine and social media at the delivery phase which could save pain and suffering for patients in the pemphigus and pemphigoid community. The twenty-first century has seen many scientific and technological breakthroughs, such as the mapping of the human genome and personalized medicine, and now it is time to focus this same intensity on rare diseases.

If you ask the patients in our disease community if there are gaps in diagnosis and treatment, you will hear a resounding, “Yes!” Unfortunately, there are also gaps in the regulatory processes and numerous opportunities to improve these processes, which will be all be explored through this initiative.

Faster access to treatment, improving the clinical trials system, development of new innovative treatments, and collaboration on new technological resources are just a few of the highlights. By bringing together all of the parties invested in these rare diseases, we are sure to improve the lives of many patients living with them.

No legislation has been proposed yet, as the 21st Century Cures Initiative is just in its early stages. One way for you to get involved now is to write your congressional representatives and ask them to support the initiative.

You can find your congressional representatives’ contact information at www.govtrack.us/congress/members.

Marc Yale is a pemphigoid patient living in Ventura, California. He has been a Certified Peer Health Coach with the IPPF since 2008. Marc advocates for our patient community both on the State and Federal levels. Marc has contributed regularly the Quarterly newsletter in his column “Coaches Corner”. Marc can be reached at marc@pemphigus.org.
how you feel, even if you have not attended medical school.

This is where “journaling” comes in handy. You will find it helpful to track your symptoms, medications, and positive and negative changes. Some people journal daily. This is ideal, but extremely difficult for most people.

Just do the best you can, and know ahead of time that there will be great days and not such great days. Also, keep a written (or digital) record of your own medical history—past, present, and ongoing. This can help both you and your doctors evaluate your progress and your reactions to treatment, and determine the next steps.

When journaling, I also recommend keeping track of your mood. Events, certain medications, and even the season of year (and number of hours of sunlight) may trigger mood changes. If you keep track of your moods, you will be more likely to find possible patterns and learn what to do to manage symptoms and flares. This applies to both your physical health and emotional health.

I highly recommend giving journaling a try for at least six months. This does not necessarily mean daily entries, but you should consider it a commitment. Sometimes just writing that you have nothing to write or don’t feel like writing is important—for your own records.

By recognizing and recording your feelings, you will be able to identify, through your journal, the often intense feelings associated with chronic illness. Doing so will give you more control in your new normal.

In these “invisible” chronic illnesses there is no one perfect way to proceed. The care of your expert physicians will be critical. However, your instincts are equally important. If you have given a doctor the benefit of the doubt, and it just does not feel right, speak up for yourself. This is someone you will be working with for a long time. If you have given the partnership your best shot and are not satisfied, remember you have a choice. You can always seek a second opinion if you do not want to change doctors.

As for your emotional well-being, everyone does not need psychological intervention, but it does not hurt to talk to a professional if you feel overwhelmed. Sometimes validation of overwhelming feelings is useful, and given the circumstances, that may be all that you need. Just be careful about who you see. I have written articles on chronic illness clients for clinical psychologists: Those articles are usually Chronic Illness 101!

In my opinion, health psychologists are often the best choice, but not the only choice, and many of them are more oriented towards research than clinical care. There are always choices. Again, trust your gut, and ask people you trust and respect.

In a future article, I will review some current symptoms and diagnoses as per American Psychiatric Association criteria. If there are specific symptoms anyone would like me to review, please contact me terry@pemphigus.org with your questions.

“Whatever course you decide upon, there is always someone to tell you that you are wrong. To map out a course of action and follow it to an end requires courage.”

Ralph Waldo Emerson

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comparing them to healthy “control” patients, searching for molecular signatures of disease, as was done in the Sinha lab, involves monitoring gene activity (also called gene expression).

Gene activity describes the process from which the genes that are found on chromosomes (made from the DNA blueprint) are made into RNA: their destiny is essentially “turned on.” These RNAs are ultimately translated into the proteins that represent the code of the unique segment of DNA from which they came. The collection of expressed genes (RNAs) that are present at any given time is called the transcriptome. It is the transcriptomes that Sinha’s group captured in four different groups of people, two with PV and two without.

The authors collected RNA samples from 31 people: 13 had active PV, eight were in remission from PV symptoms, and 10 were healthy people that have no PV diagnosis. A subset (four) of this control group were “HLA-matched” to PV patients. These people were chosen because of an important finding that Dr. Sinha had made about PV previously.

From studies of the genetic basis of PV, it is known that 95% of PV patients have a specific type of one specific group of genes called HLA. There are many HLA types within the population, and these are inherited and encode for important parts of the immune system.

So, it is not surprising that a subset of these types is connected with autoimmune disorders and other diseases: in PV, those 95% of patients have the DRB1*0402 or DQB1*0503 subtypes of HLA. This is striking, indeed, but very few people who have these HLA subtypes actually have PV. Part of the mystery that needs to be unraveled in understanding the genetic and molecular bases of PV is determining how these HLA variants are involved. Once they collected their data from the four patient groups using cutting-edge micro-array technology that allows for the simultaneous screening of >50,000 RNAs, the analysis began (see figure below).

Comparing the PV patient group (active or remittent) with the HLA-matched healthy group, the authors found a disease signature of 1,203 genes that were mostly more active (as opposed to less active) in PV patients than in the healthy group.

Several of the specific genes they found (like ADAM9 and ATP2C1) had already been linked to PV previously, which instills confidence that the approach was finding genes that are relevant to PV. The authors also defined an activity signature of 252 genes when they compared active to remittent PV patients. This is the signature that gives some insight into what differentiates someone who has active blistering and someone in remission, so it may hold some clues to the molecular environment required for a flare.

On closer inspection of the genes that are in the activity signature, the authors noted that the genes are involved in several known molecular pathways including inflammation and apoptosis as well as other immune-related pathways. Apoptosis, a mechanism whereby unnecessary cells can commit suicide, has recently seen a lot of attention in PV research as it plays an important role in acantholysis [link to Spring Quarterly Research Highlight].

The authors defined a third signature, a control signature, of
534 genes that differ between the HLA-matched people and healthy people who do not have the PV-linked HLA. This signature was particularly useful when compared to the disease signature, since the authors reasoned that genes that overlapped between these two signatures constitute a protection signature—155 genes that might help to understand why HLA-matched people with either the DRB1∗0402 or DQB1∗0503 HLA subtype never develop PV.

Three of these genes (IL13RA1, OGFRL1 and CNPY3) were particularly interesting to the authors and deserve further study to determine their role in disease protection and prevention of lesions.

Finally, the Sinha group compiled the data from all of the signatures and found that among the thousands of genes they defined as being differentially turned on or off in the three experimental signatures, a high proportion of them are found in the same chromosomal regions, called “hot spots.”

Such hot spots, where genes that are highly dysregulated in disease are concentrated, may point to chromosomal regions where PV researchers can narrow their search for the genetic causes of PV in future work. Overall, the recent work by the Sinha group reveals new insights regarding the molecular mechanisms that underlie the development of PV.

A more detailed understanding of the genetic and immunological factors that predispose and protect individuals to autoimmune disease advance us towards the development of more specific and effective therapies. 

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