PEMPHIGUS and PEMPHIGOID
What is PEMPHIGUS AND PEMPHIGOID?

Overview

Pemphigus and pemphigoid are rare autoimmune blistering diseases of the skin and/or mucous membranes. There is currently no cure for either, only remission. Pemphigus is used specifically to describe blistering disorders caused by autoantibodies that recognize components of the epidermis (for instance cellular desmoglein 1 and desmoglein 3). This in turn leads to disruption of the intercellular junctions and loss of integrity (leading to bullae formation).

Pemphigoid is a group of subepidermal, blistering autoimmune diseases that primarily affect the skin, especially in the lower abdomen, groin, and flexor surfaces of the extremities. Here, autoantibodies (anti-BPA-2 and anti-BPA-1) are directed against the basal layer of the epidermis and mucosa.

A person’s immune system makes antibodies to attack viruses and harmful bacteria. In the context of pemphigus and pemphigoid, however, the immune system is over-active and antibodies instead attack healthy cells in the skin or mucous membranes. As a result,

- Skin cells separate from each other
- Fluid collects between skin layers
- Blisters form and may cover a large area of skin

Men and women are equally affected and both conditions are known to affect people across racial and cultural lines. There are, however, certain groups of people (Ashkenazi Jews, people of Mediterranean, North Indian, and Persian decent) who have a higher incidence of the diseases. Pemphigus and pemphigoid are not genetically transmitted (hereditary), though there may be a genetic predisposition to develop the diseases. It is not currently possible to predict, however, who may get these diseases. The conditions themselves are rarely fatal, and most deaths occur from infections of compromised tissues. If left untreated, these diseases may be fatal.

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Lisa

I was a full-time professional photographer and marketing consultant who realized one day that it took almost 3 days to recover from a 10-hour wedding or event every week. I didn’t have 3 days to recover or plug through the week and Sunday I would start to feel like a normal person again. It would be months before PV would actually show itself in the form of red patches covering one unable to think, work, or pay my bills. It took 6 months and 7 physicians to finally get a proven diagnosis. It also took choosing between paying for health insurance or car insurance and eventually between paying my rent and having a car. That’s what happened to my life.

Lisa

Meet Pemphigus Warrior Lisa

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Pemphigoid vs Pemphigus

Pemphigoid affects a lower layer of the skin, between the epidermis and the dermis, creating tense blisters that do not break easily, while pemphigus affects the upper layer within the epidermis and causes lesions and blisters that are easily ruptured. Pemphigus does not cause permanent scarring unless there is an infection associated with the sore.

Pemphigus Types

PEMPHIGUS VULGARIS (PV)

PV is the most common of these conditions. Blisters are soft and fragile and may form at the mouth first, then spread to the skin and even the genitals. Blisters are frequently painful but not itchy, and those in the mouth make chewing and swallowing difficult. PV does not cause permanent scarring unless there is an infection associated with the sore.

PEMPHIGUS FOLIACEUS (PF)

PF is less severe than PV. Blisters may form on the scalp and face first, then spread to the chest and back. They do not occur in the mouth and are not usually painful, as the blisters are superficial and form crusts.

PEMPHIGUS VEGETANS

With this form of the disease, thicker sores mainly form in the groin and under the arms.

IGA PEMPHIGUS

This strain of the disease is caused by IgA antibodies binding to epidermal cell proteins. When it first presents it may resemble PF or may appear as small pustules.

PARANEOPLASTIC PEMPHIGUS (PNP)

This type is associated with certain forms of cancer. Blisters form inside the mouth and may affect the lungs, leading to a fatal outcome. Sores of the mouth, lips, and esophagus are almost always present, and skin lesions of different types occur. In some cases, the diagnosis of the disease will prompt physicians to search for a hidden tumor. If the tumor is benign, the PNP will improve if the tumor is surgically removed.

MUCOUS MEMBRANE PEMPHIGOID (MMP)

This form of the disease affects the eyes, mouth, and throat. A clinical form—ocular cicatricial pemphigoid (OCP)—may result in blindness if it involves the eyes. If it involves the deeper parts of the throat, it may cause respiratory compromise.

BULLOUS PEMPHIGOID (BP)

This strain is limited to the skin. Blisters present predominantly on the abdomen, groin, back, arms, and legs, and they may itch and be painful.

Meet Pemphigus Warrior MARC

Marc is a 69-year-old American Ashkenazi Jew (97% DNA) whose battle with pemphigus began in 2011.

A Chicago businessman, I had to close my company of more than 40 years in 2010 due to the US economic collapse. In anticipation of that, I moved to Florida, and have acquired licenses as a general residential contractor, home inspector, and mold assessor/inspector.

In July 2011, my chest began itching and growing blisters. As the blisters multiplied and scabbed on my back, face, hands, and chest, I made multiple visits to a dermatologist, but her cosmetic background failed to find a diagnosis until I quickly grew worse. At the point where I was sleeping on a black plastic bag so I wouldn’t stick to the sheets, in September of that year, when my daughter, a physician assistant, saw me, she immediately called an immunologist at the University of South Florida (USF). When the physician saw me, he immediately admitted me to Tampa General Hospital for 100+ mg of prednisone, and to clean up my skin, I gradually cleared up over 8 months to recover. Despite the treatment, PF and PV came back in November of 2012. The following January I received IVIg treatment, followed by Rituxin® in February. My pain began in April, and three more IVIs in June and July. My fight against pemphigus required dermatologists at USF to see my story complete with photos, to elucidate how dermatologists respond in the news. I moved to LA in 2013 while taking treatment for PV and the IVIg in the US. In December 2017, although I did not have PV blisters in my mouth, a throat problem occurred. What was initially diagnosed as esophageal thrush, turned out to be PV in my throat. Coping with the constant pain, managing the care of open wounds, and sharing the story, over the long call, was a gigantic challenge. For me, the solution has been correct medical treatment, a strong personal faith, and great support from people close to me. For me, 2019 began with USF advances, to hopefully put all varieties of pemphigus into remission once again.

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**GESTATIONAL PEMPHIGOID (GP)**
Typically presenting in the second trimester of pregnancy, GP is a blistering rash starting around the navel and spreading to the entire body.

**EPIDERMOLYSIS BULLOSA ACQUISITA**
The skin is prone to blistering on the skin without involvement of mucosal surfaces. The blisters are usually smaller than those seen in pemphigoid.

**Diagnosis**
Pemphigus and pemphigoid are diagnosed through special testing and clinical presentation. Types of testing include:

- Lesion Biopsy—a sample of the blistered skin is removed and examined under a microscope. Additionally, the layer of skin in which cell-to-cell separation occurs may be determined.
- Direct Immunofluorescence—the skin sample is treated to detect desmoglein autoantibodies in the skin. The presence of these antibodies indicates pemphigus.
- Indirect Immunofluorescence or Antibody Titer Test—this measures desmoglein autoantibodies in the blood serum. It may be used to obtain a more complete understanding of the course of the disease.

**Standard Therapies**
The treatment of pemphigus and pemphigoid is directed toward suppressing the skin and mucosal lesions of the diseases and preventing complications potentially associated with treatment. Although there is no cure yet for pemphigus, the disorder may usually be controlled. Most patients will eventually enter a complete remission in which they are off all therapy and there is no evidence of the disease. Generally, the less widespread the pemphigus is, the easier it is to control. The development, severity, and progression of the diseases are not uniform, and the response to therapies may vary among individuals. Consequently, physicians will take several different factors into account when planning an individual's treatment, which will be tailored to the patient’s specific needs and situation.

Treatment is usually separated into 3 phases: control, consolidation, and maintenance. In the control phase, high-intensity therapy is used to bring the disorder under control by initiating clearance of current lesions, reducing and/or suppressing new lesion formation, and improving other symptoms such as itch relief. In the consolidation phase, a consistent dose of medication is used until a significant portion of lesions have healed. In the maintenance phase, the dose of medication is gradually reduced until a minimal level is achieved that successfully prevents the development of new lesions.

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Meet Pemphigus Warrior

FRED

Fred

When I first showed symptoms of PV, I was beginning to wrap up a 37-year career as a writer and manager in a variety of public and private sector organizations, and looking forward to a work-at-my-own-pace retirement. I knew something was going wrong with my mouth and my skin, but it took 7 months and several doctors to get on the road to remission. By the time my drug regimen started, I had lost 30 pounds, my scalp was covered with crusting wounds, and I had 23 areas of my body showing active lesions. Most of my battle with PV took place before the advent of rituximab as a front-runner in treatment options, so I spent 9 years on various dosages of prednisone and a few different steroid-sparing drugs. I'd always been an active, high-energy individual, so the crushing fatigue from prednisone was especially difficult for me. In the months leading up to my retirement, it was disheartening to finish a day's work and have nothing left in the tank for outside activities. My wife and friends were a great source of hope during the darker times.

I'm happy to report that I have been in remission—no meds and no new lesions—for a little more than 3 years now. I am back to an active schedule; some think I'm trying to make up for lost time. Any fatigue I feel nowadays is more likely the result of being in my seventies. I have resumed my writing, and published several articles and short stories in a number of magazines. I play bass guitar and sing in a classic rock band and have become a winning poker player. I work with central New Jersey's homeless and immigrant populations, have helped edit 4 novels, serve on the advisory board for the Rutgers University Writers' Conference, and provided human resource consulting services for small businesses. My wife and I enjoy traveling; we have visited 17 countries and journeyed extensively in the United States. We also have a 2-year-old grandson to spoil. Despite the curves thrown at me by this disease, I've got to say life has been good to me. There are many others whose pemphigus or pemphigoid is more severe than mine. I'd like to do what I can to let them know that things can, and will, get better and that they should never feel they are alone on their path toward remission.

The mainstay of treatment is the use of corticosteroids such as prednisone, anti-inflammatory medications that also suppress the normal function of the immune system. Steroids may be applied directly to the affected areas (topically) or may be taken by mouth or given by injection (systemic steroids). Topical therapy is generally given to reduce pain and prevent or treat infection. Most individuals will receive systemic steroids to bring about control of pemphigus. The dose of steroids used can be tapered once control of the disorder is achieved.

Rituximab is now considered a first-line therapy for pemphigus; it was approved by the FDA in June 2018 for this indication. Rituximab, which can prevent new autoantibodies from forming, takes 3 to 4 months for the existing autoantibody levels to fall, during which time some dose of steroids may be required. For patients with severe disease, IVlg may be added to help clear existing autoantibodies. Other medications that may be used in combination with corticosteroids to treat individuals with pemphigus include immunosuppressive drugs such as mycophenolate mofetil, azathioprine, methotrexate, or cyclophosphamide; immunomodulating drugs like dapsone; or antibiotics such as doxycycline. Use of these medications may allow physicians to lower the overall dose of steroids. Some individuals respond quickly to therapy; others respond more slowly or do not respond at all. In severe cases or in cases where individuals fail to respond to other therapies, pulse steroids, plasma exchange, and/or IVlg may be used. Research has shown that IVlg therapy may markedly decrease levels of the abnormal antibodies associated with pemphigus without decreasing levels of normal healthy antibodies. IVlg is normally given with other therapy, such as steroids and immunosuppressive drugs, to prevent rebound of disease as the therapy is tapered.

Pulse-steroid therapy refers to the administration of extremely high levels of steroids given for a short period of time. Plasmapheresis is a method for removing unwanted substances (eg, autoantibodies) from the blood, and is not used as much now because of increased risk of infections. Blood is removed from the patient and blood cells are separated from plasma. The patient’s plasma is then replaced with other human plasma and the blood is transfused into the patient. These approaches are most frequently used now only if rituximab is not tolerated or is ineffective.

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Ellen

By 2006, I had taught English in China, worked for a global engineering company in San Francisco, and had just moved to Australia to start a new adventure. I prided myself on my adaptability and readiness to take on new challenges, but these attributes did not prepare me for what was to come. I was 45, and I looked and felt remarkably healthy when painful lesions started appearing on my gums and inside my mouth. The blisters were so painful that I had to avoid touching them, and fluids became my main source of nourishment. The treatments were not helping, and I didn’t want to treat my symptoms. I knew I had to find a new treatment, and I did. I was referred to a dermatologist in San Francisco who performed a skin biopsy and diagnosed me with pemphigus. I had never heard of this condition, but I knew it was serious. The pemphigus progressed quickly from my mouth to almost every part of my body. The first few years of treatment were very difficult and involved pill-popping and other medications. It took time to determine the most effective combination, along with the appropriate dosage, while minimizing adverse side effects. After a Port-a-Cath implant, I had close to 150 IVIg treatments and dozens of rituximab infusions. Oftentimes the side effects of the meds were worse than the blisters that covered my body. I learned that my treatment was an active process, and I worked with my physician and other specialists to find the right combination of medications and dosages. I adjusted my diet to avoid certain foods and altered my lifestyle to reduce stress. I learned to manage my disease and partner in determining the best course of action. Having a rare disease can be isolating at times, and when I tell people the name of my condition, I am usually met with confusion and concern. It is possible to live a normal, healthy, and active life with a chronic disease, but it requires patience, acceptance, and resilience.

Who we are: THE INTERNATIONAL PEMPHIGUS & PEMPHIGOID FOUNDATION (IPPF)

Vision
To find a cure for pemphigus and pemphigoid.

Mission
To improve the quality of life for all people affected by pemphigus and pemphigoid through early diagnosis and support.

IPPF Overview
The most important objectives of the IPPF are to provide patients and physicians worldwide with information about pemphigus and pemphigoid, and to provide patients and their caregivers much-needed comfort and support so they can continue to live active productive lives. To help fulfill those objectives, we:

- Offer a physician-referral service to help patients find the best medical care possible
- Provide a number of valuable and popular patient-support services
- Publish informational brochures, pamphlets, and a quarterly newsletter with news, useful information, medical updates, personal stories, and more
- Run an annual patient/physician meeting
- Collaborate with pharmaceutical companies on the leading edge of treating these diseases
- Provide up-to-date information about current clinical trials and research on the diseases in which patients may be able to participate
- Maintain relationships with congressional representatives and others who may be able to encourage or provide research funding

To ensure that we are able to provide the most current information about the diseases and treatments, we have developed and continue to maintain close relationships with physicians and leaders in the medical community, including the National Institutes of Arthritis, Musculoskeletal and Skin Diseases (NIAMS), part of the National Institutes of Health, and the American Academy of Dermatology (AAD). The IPPF is also an active member in a number of other organizations that help fulfill our role as patient advocates and enable us to have greater impact as we work together: NORD, the Coalition of Skin Diseases (CSD), the Derma Care Access Network (DCAN), and the International Alliance of Patient Organizations (IAPO).

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Diagnosis for patients in the registry was pemphigus vulgaris (52%).

Participant Gender (n=138): 75% of the patients in the registry are female.

Participant Age at Diagnosis (n=136):
- 20-30 years: 30%
- 31-40 years: 25%
- 41-50 years: 20%
- 51-60 years: 15%
- 61+ years: 10%

Participant Diagnosis (n=118):
- Pemphigus Vulgaris: 52%
- Bullous Pemphigoid: 19%
- Mucous Membrane Pemphigoid: 5%
- Paraneoplastic: 3%
- IgA Pemphigus: 3%
- Other: 3%

Patient Distribution by Age at Diagnosis (n=136):
- 20-30 years: 24%
- 31-40 years: 28%
- 41-50 years: 30%
- 51-60 years: 10%
- 61+ years: 8%

Patient Distribution by Diagnostic Test Performed (n=160):
- Lesion Biopsy by Direct Immunofluorescence: 61%
- Indirect Immunofluorescence: 17%
- ELISA: 9%
- None: 3%

Patient Distribution by Insurance (n=125):
- Commercial: 55%
- Medicare: 22%
- Other Insurance: 17%
- Medicaid: 6%
- None: 2%

Patient Distribution by Medication Use (n=89):
- Oral Corticosteroids: 61%
- Topical Steroids: 55%
- Immunosuppressants: 43%
- Antibiotics: 12%
- Dapsone: 11%
- Colchicine: 9%
- Nicotinamide: 4%
- Rituximab: 3%
- IVIg: 2%
- Dapsone: 1%
- Others: 1%

Patient Distribution by Physical Health (n=75):
- Good: 75%
- Fair: 20%
- Poor: 40%

Patient Distribution by Mental Health (n=77):
- Good: 22%
- Fair: 23%
- Poor: 55%

Patient Distribution by Quality of Life (n=76):
- Good: 22%
- Fair: 24%
- Poor: 53%

Participant Distribution by Location:
- Live in the United States (80% [111/138]), while others live in the UK (7%, 9/138), Canada (2%, 3/138), India (1%, 2/138), Australia (1%, 2/138), and other countries around the world. Of the 138 patients, 55% (76/138) had commercial insurance.

Pemphigus and pemphigoid represent diseases with significant patient burden often requiring multiple frequently administered medications (pill burden of 2+ pills daily for 50% of the patients) and resulting in hospitalizations in a quarter of the managed population, despite continuous outpatient care. More than half the patients (52% [61/118]) were diagnosed with pemphigus vulgaris, 19% (22/118) with mucous membrane pemphigoid, 14% (16/118) with bullous pemphigoid, and the remaining 16% (19/118) with other P/P subtypes (the total of percentages is >100% due to rounding). The majority of patients (58%) were diagnosed by lesion biopsy by direct immunofluorescence, while 24% were diagnosed with indirect immunofluorescence.

4% of registry participants have had surgeries to treat their condition. Of patients reporting medication information, 61% (54/89) took oral corticosteroids, 55% (49/89) topical corticosteroids, 43% (38/89) immunosuppressants, and 25% (22/89) rituximab. Half of the patients receive their medication once daily, nearly 40% receive it twice a day, and the remaining patients take their medication more frequently. Half of the patients take their medication orally, a quarter use topicals, 19% intravenous (IV) drugs, 2% intramuscular (IM), 1% subcutaneous (subQ), and the remaining patients administer their medications via nasal, transdermal, or buccal administration routes. Although the majority of patients are interested in research participation, very few currently participate in clinical trials.

Quality of life was reported as fair or poor for 22% (17/76) of patients. Patients reported frequent fatigue (42%, 32/77) and depression (17%, 13/78). A quarter (25% [19/76]) of patients indicated pain greatly interfered with their enjoyment of life and 34% (24/71) reported fair or very poor quality of sleep. One-fifth of patients (20% [15/76]) rated their physical health as fair or poor, while 12% (9/77) of patients rated their mental health as fair or poor.

Clinical Study

Launched in March 2017, the IPPF registry is sponsored by NORD through a cooperative agreement with the FDA. The IPPF collects information on socio-demographics, medical and diagnostic history, treatment and disease progression, management of care, and quality of life. Patient respondents were predominately female (75%, 104/138) and white (77%, 101/138).

Median age at patient diagnosis in the registry is 51.0 years (mean 45.1, SD 9.4). Most of the patients in the registry currently live in the United States (80% [111/138]), while others live in the UK (7%, 9/138), Canada (2%, 3/138), India (1%, 2/138), Australia (1%, 2/138), and other countries around the world. Of the 138 patients, 55% (76/138) had commercial insurance.