New Stripes for Zebras

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Improving Health
Powering Research

A Patient-Powered Research Network
Mission Statement

Founded in 1980, the Immune Deficiency Foundation is the national patient organization in the United States dedicated to improving the diagnosis, treatment and quality of life of persons with primary immunodeficiency diseases through advocacy, education and research.
IDF wants you to

THINK ZEBRA!

“When you hear hoof beats, think zebras, not horses.”

In medical school, many doctors learn the saying, “when you hear hoof beats, think horses, not zebras” and are taught to focus on the likeliest possibilities when making a diagnosis, not the unusual ones. However, sometimes physicians need to look for a zebra. Primary immunodeficiency patients are the zebras of the medical world.
About Primary Immunodeficiency Diseases (PI)

- There are approximately 250,000 people who are diagnosed with PI in the U.S., and thousands more go undetected.
- Primary immunodeficiency diseases are a group of more than 240 rare, chronic disorders in which part of the body’s immune system is missing or functions improperly.
- While not contagious, these diseases are caused by hereditary or genetic defects, and although some disorders present at birth or in early childhood, the disorders can affect anyone, regardless of age or gender.
Survey Research

More than 35 surveys of patients and medical professionals since 1995

Highlights include:

  - 2009 IDF Patient Survey (Web-based)
- Prevalence Survey (2005)
- Physician Surveys
  - Pediatrician Survey (2007)
  - Family Practitioner Survey (2009)
  - Pulmonologist Survey (2011)
- Early vs. Delayed Diagnosis of SCID (2010)
- Primary Immunodeficiency & Women’s Reproductive Health Survey (2012)
Average Time to Diagnosis from Symptom Onset: All Diagnoses

- **2002**: Median = 9.2 months (N=1,397)
- **2007**: Median = 6.0 months (N=1,137)
- **2012**: Median = 7.5 months (N=1,413)

Q21. What kind of healthcare professional does the patient see most often for his/her healthcare? N=1427

- Allergy/Immunology: 38%
- General/Family Practice: 26%
- Hematology: 6%
- Immunology: 18%
- Infectious Disease: 5%
- Internal Medicine: 15%
- Nurse Practitioner: 2%
- Pediatrics: 13%
- Physician Assistant: 2%
- Other: 14%

Source: IDF 2012 National Patient Survey
Q19b. Please tell us about any diagnosed conditions the patient CURRENTLY has. N=1427

Sources: 2012 IDF National Patient Survey
Q26. Has the patient ever received any of the following treatments for PIDD?

N=1427

Source: IDF 2012 National Patient Survey
IDF Treatment Survey - SF-12 QoL Component & General Health Score Comparisons

Physical Component Summary (PCS)  
- All Respondents: 40.94  
- SCIG: 41.09  
- IVIG: 40.85

Mental Component Summary (MCS)  
- All Respondents: 46.26  
- SCIG: 46.66  
- IVIG: 45.96

General Health (GH)  
- All Respondents: 42.8  
- SCIG: 43.48  
- IVIG: 42.29

Source: IDF 2013 National Treatment Survey

* Significant difference between population with PI and the U.S. general population
eHealth Evolution for PI

- 2011- IDF launched an online electronic personal health record (ePHR)
  - Developed for individuals and families living with primary immunodeficiency diseases
  - Patients track their symptoms, record medications and log health information
  - No cost for patients and family members
- Fall 2014 - IDF transformed ePHR with new features, including cutting edge tracking and reporting tools:
  - Sync with Microsoft HealthVault
  - Set health and fitness goals and track progress
  - Quality of Life survey instruments
  - Connection to PI CONNECT

www.idfePHR.org
Blended Data: Inflammation in XLA

- Two sources: USIDNET and Patient Reported IDF Survey
  - Asked IDF to perform a survey of patients with XLA
  - USIDNET data on XLA patients
  - Use of both a patient survey and USIDNET Registry data was important in gaining a better understanding of the association of inflammatory disease in XLA.

## USIDNET vs. XLA Survey
### Reported Inflammation & Autoimmunity

<table>
<thead>
<tr>
<th>Condition</th>
<th>USIDNET Registry (N=149)</th>
<th>XLA Survey (N=128)</th>
<th>p value</th>
</tr>
</thead>
<tbody>
<tr>
<td>Aches, malaise or fatigue</td>
<td>22 (14.8)</td>
<td>44 (34.4)</td>
<td><strong>P &lt; .001</strong></td>
</tr>
<tr>
<td>Pain, swelling, arthralgia</td>
<td>18 (12.1)</td>
<td>28 (25.0)</td>
<td><strong>P = .035</strong></td>
</tr>
<tr>
<td>Abdominal pain</td>
<td>10 (6.7)</td>
<td>22 (17.2)</td>
<td><strong>P = .008</strong></td>
</tr>
<tr>
<td>Diarrhea</td>
<td>13 (8.7)</td>
<td>27 (21.1)</td>
<td><strong>P = .005</strong></td>
</tr>
<tr>
<td>Chills</td>
<td>1 (.7)</td>
<td>17 (13.3)</td>
<td><strong>P &lt; .001</strong></td>
</tr>
<tr>
<td>Shortness of breath</td>
<td>1 (.7)</td>
<td>21 (16.4)</td>
<td><strong>P &lt; .001</strong></td>
</tr>
<tr>
<td>Constipation</td>
<td>3 (2.0)</td>
<td>14 (10.9)</td>
<td><strong>P = .002</strong></td>
</tr>
</tbody>
</table>
A patient-researcher collaboration
For primary immune deficiencies

Sponsored by PCORI
So much data!

- Advent of Electronic Health Record (EHR)
  - Systematic collection of comprehensive individual data
- Digital registries and databases
  - Accumulate knowledge on disease
- eHealth era
  - 16,000-plus healthcare apps in the Apple store.

This should be transformative!
Imagine!

- Harnessing large data sets to learn what treatments work
- Engaging patients and researchers collaboratively
- Cost effective research on outcomes
Unique challenges of rare diseases

- Sample size
- Limited funding
- Data is skewed to single center experiences
Hurdles and challenges of research registries

- Expensive infrastructure (funding)
- Time commitment for data entry
- Regulatory burden
- Lack of cross registry compatibility
- Finding the right questions
The ideal

- Patient input is critical
  - Prioritize research questions
- Broad representation is essential
- Unbiased data collection
- Varied modes of health care delivery
  - Private practice
  - Subspecialist
  - Academic centers
PI CONNECT

- Collect data on patients with primary immune deficiencies
  - Diverse clinical problems
  - >250 diseases
- Facilitate dialog between patients and researchers
- Support research on primary immune deficiency
- Help patients live BETTER
2011- IDF launched an online electronic personal health record (ePHR)
  - No cost for patients and family members
  - >1000 patients

September 2014 – IDF new ePHR
  - Sync with Microsoft HealthVault
  - Set health and fitness goals and track progress
  - Quality of Life survey instruments
    - PROMIS 29

www.idfePHR.org
Outreach to Patients and Families

- IDF social media
  - Facebook
  - IDF Friends – IDF’s custom-built social network
- e-newsletters, newsletters and blogs
- Patient Education Meetings
- New patient packets
- Directed email
- IDF website
- Videos
IDF ePHR and PI CONNECT Info Card

Improving Health  Powering Research

ePHR
Immune Deficiency Foundation

Take charge of your health with the IDF ePHR, the electronic personal health record designed for the primary immunodeficiency (PI) community.
- Easy-to-use tools to help you improve your health and organize your info.
- Track your health and fitness goals, set reminders and notifications.
- Access IDF ePHR from anywhere—your computer, tablet or phone.
- Keep your medical info safe, secure and private at no cost to you.

An added benefit of IDF ePHR is the opportunity to be a part of research by joining PI CONNECT, the IDF Patient-Powered Research Network. See reverse side for more information about PI CONNECT.

Create an account! www.IDFePHR.org
For more information, e-mail info@idife.org or call 800-236-4483.

PI CONNECT
IDF Patient-Powered Research

PI CONNECT
Finally, there is a tool for you to make your voice heard in primary immunodeficiency (PI) research—PI CONNECT, the IDF Patient-Powered Research Network. PI CONNECT empowers you to help others living with PI by simply logging your experiences and joining the conversation.

How Does PI CONNECT Work?
- Create an IDF ePHR account and join PI CONNECT. See reverse side for more information about IDF ePHR.
- Your account information becomes a part of the USDNET® patient-consented registry, which contains clinical data on several thousand patients with PI. Your information is secure, aggregated and de-identified.
- PI CONNECT** is your opportunity to consent into the registry with just a few simple clicks!

Create an IDF ePHR account and join PI CONNECT!
www.IDFePHR.org

How Can I Power Research?
- Through PI CONNECT you can help power research by providing real-time information and giving researchers a first-hand account of the true patient experience.
- You also have access to an exclusive research forum where you will be able to discuss and offer your opinions about PI research.
- All of this can lead to improved diagnosis and treatment of PI. Your participation can really make a difference!

* USDNET, the United States Immunodeficiency Network, is a program of IDF and funded in part by the National Institute of Allergy and Infectious Diseases (NIAID) and the National Institutes of Health (NIH) through the Department of Health & Human Services.
** PI CONNECT is funded by the Patient Centered Outcomes Research Institute (PCORI) and is a part of the National Patient-Centered National Clinical Research Network (PCORnet).
IDF ePHR Users

- Original IDF eHealthRecord supported almost 1,000 users with 48 different PI types.

- Demographics
  - 22% 17 years or younger
  - 35% 18-44 years
  - 32% 45-64 years
  - 10% 65 years or older

- Gender
  - 63% Female
  - 37% Male
PI CONNECT

IDF ePHR
Track Your Health

USIDNET
Advance Scientific Research

PI CONNECT
The United States Immunodeficiency Network (USIDNET) oversees a Registry of Patients with PI.

- Funded by NIAID
- Administered as a program of IDF

Physician-validated clinical data on over 3,800 patients with PI

- Laboratory results
- Genetic and molecular information
- Diagnostic criteria

Longitudinal Data

- Incumbent upon clinicians updating patient records over time
USIDNET

- **Strengths**
  - High quality data
  - Depth to laboratory data

- **Weaknesses**
  - Labor intensive
  - Academic center skew
  - Limited longitudinal data
  - High regulatory burden

ePHR

- **Strengths**
  - Patient reported outcomes
  - Longitudinal data
  - Good kinetic capture

- **Weaknesses**
  - Diagnoses not validated
  - Limited laboratory data
PI CONNECT

- Anticipate 1250 patients will be involved with PI CONNECT
- Both USIDNET registry and IDF ePHR use the same servers and platforms, making transfer of data possible and secure
PI CONNECT

HealthVault Partners

HealthVault

IDF Reporting requirements
IDF Research

EMR

Computer/Tablet

Mobile App

ePHR

ETL

ePHR Data Warehouse/cube

PI Connect Consented

USIDNET Data Warehouse/cube

PopMedNet

USIDNET, a program of IDF

Immune Deficiency Foundation
Regulatory aspects

- **USIDNET Patient Consent Process**
  - IRB-approved
  - Traditional consent language on 11 page paper form
  - Time burden on patient and clinician

- **PI CONNECT Electronic Consent Process**
  - IRB-approved with waiver of documentation
  - Simplified and plain-language e-consent online
    - Patients can read and truly understand what they’re agreeing to do
    - One click
    - No staff time required
PI CONNECT is a tool designed to directly connect patients to research being conducted in the field of primary immunodeficiency (PI) diseases.

By joining PI CONNECT, you agree that the information you enter in your IDF ePHR, electronic personal health record, will be shared with the United States Immunodeficiency Network (USIDNET). USIDNET is an established registry containing medical data on thousands of patients with PI.

PI CONNECT allows you to:

- Share your experiences to help advance research of PI.
- Track and share your health information with researchers to broaden the understanding of PI.

What Do We Promise?

We promise to safeguard your privacy.

Your data is de-identified which keeps your identity confidential.

What Are the Benefits?

Contribute to research to ensure better care in the future.

Receive exclusive access to the PI CONNECT Research Forum.

What Do You Do?

Once you have created your ePHR and consented to PI CONNECT, simply use IDF ePHR to manage your health info.

We’ll take it from there!

What Are Your Rights?

It’s completely voluntary! You may choose to stop participating at any time.

Know that you can choose not to participate now or even in the future.

PI CONNECT REGISTRATION

PI CONNECT Consent

What is PI CONNECT?

PI CONNECT is a tool designed to directly connect patients to research being conducted in the field of primary immunodeficiency (PI) diseases.

By joining PI CONNECT, you agree that the information you enter in your IDF ePHR, electronic personal health record, will be shared with the United States Immunodeficiency Network (USIDNET). USIDNET is an established registry containing medical data on over 3500 patients with PI. Combining your information with USIDNET will help provide the most complete picture of your health. Physicians and researchers use what's known as "de-identified" data, which means it cannot be traced back to you, from the registry to gain a better understanding of PI diseases, their outcomes, and treatments.

By joining PI CONNECT, you allow us to:

- Store the data from your ePHR in the USIDNET Registry.
- Combine the information you enter with medical information provided by your physician.

**Participant First Name:** Bin

**Participant Last Name:** RecordTest

**Participant consent (for users 18 years of age and older) or Parent/Guardian Consent (for users under 18 years of age):**

- [ ] I consent to participate or to allow my child/ward to participate in PI CONNECT
- [ ] I do not consent to participate or allow my child/ward to participate in PI CONNECT

**Participant assent (for users 13 through 17 years of age):**

- [x] This research study has been explained to me and I agree to be in this study
- [x] This research study has been explained to me and I do not agree to be in this study
- [ ] The participant is under 13 years of age or older than 17 years of age

**SAVE**
Research forum

- Research blog
  - Researchers and patients blog about topics

- Research bulletin board
  - Patients post comments

- Research updates
  - Videos posted as a resource

- Research round table
  - IDF meetings foster collaboration
Welcome!

The PI CONNECT Research Forum serves as the portal for consented PI CONNECT users to engage with researchers and others to exchange and discuss research ideas. Please remember that the information on this site should not be used in place of a visit, consultation, or advice of your healthcare provider.

Should you have any healthcare-related questions, you should call or see your clinician.

Share ideas, comments, and observations in the following forums:

General Discussion
Disease Specific Research
Treatments

Topics Being Discussed
- advance
- alternative
- autoimmune
- common
- common variable immune deficiency
- conditions
- CVID
- CVID Aspergillosis
- Pseudomonas
- gene therapy
- human genome
- Immunoglobulin
- inflammation
- medicine
- nasal rinse
- non-traditional progress
- prophylactic antibiotics
- research
- secondary complication
- symptoms
- treatment
- x-linked agammaglobulinemia
- XLA
Imagine! New Stripes!

- Large validated data sets
- Patients and researchers collaborating
- Cost effective
Hurdles and challenges

- **Expensive infrastructure**
  - USIDNET (first registry) $160,000
  - ePHR (second registry) $27,000

- **Time commitment for data entry**
  - Pay, cajole, reward
  - Blue button technology

- **Regulatory burden**
  - Paper consents
  - Electronic consent
  - Waiver of documentation

- **Cross platform compatibility**
  - Individual mapping
  - PCORI mandates certain fields be shared
  - EPIC compatibility

- **Finding the right questions**
  - Researchers ask “their questions”
  - Collaborate
The “right” question

- Meeting held at NIH in 2013
- Multiple questions generated by patients and researchers
- Patients focused on optimal treatments
- What dose of immunoglobulin is right for me?
IVIG dosing: 10X range

- No clear guidance on the most commonly used treatment for immune deficiencies
PI CONNECT

- Ask whether patients feel differently on different doses
  - PROMIS 29
    - NIH-validated QOL instrument
- Collect antibiotic use information
- Personalized medicine

This dose is best for THIS patient!
Patient involvement is essential

- Provide data
- Focus research questions
- Engage researchers

How to keep them engaged?
- Rewards for continued data input
- Patients compare their QOL to others
- Prompts to enter data
- Video resources
- Invitations to round tables and webinars
- Focused staff member
Physician engagement is essential

- Newsletters
- Surveys
- Data reports
- Authorship on papers
- Payments
- Recognition of participating physicians
- Webinars
Summary

- What is working?
  - Technology infrastructure
  - Data (amount and standards)
  - Online consent
  - Patient engagement (so far!)

- What is evolving?
  - Blue button technology
  - Physician engagement
  - Best formats for the collaborative dialog
  - Engagement across registries
  - Long term patient engagement

- Disappointments?
  - None! It’s been a great experience!
Thank you!