As of Sep 2018, 275 patients or caregivers registered and 196 contributed data to the registry; 74% of participants.

**2. THE OMS REGISTRY**

Patients may not be diagnosed or accurate diagnosis may not occur in a timely manner. Development of OMS-related challenges/symptoms for a given patient is often unclear. Best practices for diagnosed patients are present but not universally practiced. Patients may not be diagnosed or accurate diagnosis may not occur in a timely manner. Development of OMS-related challenges/symptoms for a given patient is often unclear. Best practices for diagnosed patients are present but not universally practiced.

**3. PATIENT DEMOGRAPHICS**

As of Sep 2018, 196 of 275 registered participants contributed data to the OMS Registry. However, the OMSLife Foundation is working with roughly 1,000 patients worldwide. As of Sep 2018, 196 of 275 registered participants contributed data to the OMS Registry. However, the OMSLife Foundation is working with roughly 1,000 patients worldwide.

**4. SUMMARY**

To better understand the natural history and experience of OMS patients, we solicited data from the National Organization for Rare Disorders (NORD) and the US Food and Drug Administration (FDA). This registry is sponsored by the National Organization for Rare Disorders (NORD) and the US Food and Drug Administration (FDA). This registry is sponsored by the National Organization for Rare Disorders (NORD) and the US Food and Drug Administration (FDA).
1. BACKGROUND AND OBJECTIVES

OMS is a rare disease affecting an estimated 1 in 100,000 people, with no known cure. Symptoms can manifest at any age, often soon after birth, and are characterized by ataxia, fever, tremors, opsoclonus, and myoclonus. The OMS Life Foundation and NORD are collaborating on the OMS Registry to better understand the natural history and experience of OMS patients.

2. METHODS

The OMS registry was launched in February 2017 and has enrolled 275 patients, of whom 137 have provided clinical and demographic information. Registry data are collected through an automated web-based tool, and patients can self-enter data at any time. The registry collects data on symptom onset, diagnosis, treatment, and quality of life.

3. SYMPTOMS AT ONSET

The aggregate score is based on 6 individual symptom scores. Arrows indicate significant (p<0.05) associations (Bonferroni correction). The most common symptoms at onset are ataxia (87%), opsoclonus (82%), and myoclonus (73%). Symptoms with significant associations include sleep disturbances (28%) and temper tantrums (16%), fever and vomiting (13%), and myoclonus and opsoclonus (28%).

4. SYMPTOMS AT ONSET

Of 115 respondents, 96% received IVIG (IVIG), 96% received ACTH/steroids (ACTH/Steroids), and 49% received chemo/immunotherapies (Chemo/Immunotherapy). Of 113 respondents, 3% had an unknown type of therapy (C/I, ST), 3% had a combination of therapies, and 14% received no therapy.

5. TREATMENT UTILITY

Patients received different Medication Types, with 41% receiving 3 or more different types of medications (C/I, IVIG, ST; C/I, ST; ST; 3%; C/I; 1%; C/I, ST; C/I; ST; 1%; C/I; ST; 3%; C/I; ST; ST; 3%).

6. TREATMENT UTILITY

The majority of respondents received 1 type of therapy (83%), followed by 2 types (10%), 3 types (6%), and 4 or more types (1%). Respondent perception of therapy effectiveness was generally positive, with 49% indicating that their therapy was very effective or moderately effective.

7. SUMMARY

The OMS registry is the first national patient registry for a rare disease affecting 1 in 100,000 people, with no known cure. The registry collects data on symptom onset, diagnosis, treatment, and quality of life. The registry is open to all OMS patients and is being used to generate evidence to guide future research and treatment options.

8. CONCLUSIONS

Multiple therapeutic approaches are employed in the care of OMS patients, with no known cure. Further research is needed to identify disease-modifying therapies. The OMS registry is an important step towards understanding the natural history and experience of OMS patients and improving outcomes for this rare disease.
THE USE OF PATIENT REPORTED REGISTRY DATA TO IMPROVE HEALTH OUTCOMES FOR PATIENTS WITH OPSOCLONUS MYOCLONUS SYNDROME (OMS)  

Michaelis M1, Khoury-Dennis C1, Boulanger V2, Seebald A2, Rossov S2, Milligan K3, Pacholski L1  

1. BACKGROUND AND AIM  
Opsoclonus Myoclonus Syndrome (OMS) is a rare, orphan disease primarily impacting children between the ages of 1-5 years. Lack of disease awareness by healthcare stakeholders results in diagnosis and treatment deficiencies. In 2017, The OMS Foundation and the National Organization for Rare Disorders (NORD) launched a patient reported registry to collect patient and caregiver-provided information to help with global OMS awareness.

2. METHODS  
As of September 2018, 175 patients or caregivers have registered and 1347 surveys have been completed in the OMS registry. Surveys are designed to include patient demographics, diagnosis, symptoms, disease severity, and treatments. For this study, we analyzed data collected at the time of the registry’s initial launch in March 2018 for 272 participants who completed a longitudinal survey.

3. PARTICIPANT RACE AND SEX  
Participants in the registry are mostly White/Caucasian (87%), Black or African American (5%), Hispanic or Latino (4%), and Asian (3%). Females (56%) are over-represented compared to males (44%).

4. COUNTRY OF RESIDENCE  
Participants in the registry are located in 53 countries, with the majority of participants in the US (98%, 1347/1347). Five countries comprise over 60% of the patient population: the United States (65%), Canada (13%), France (4%), Ireland (4%), and Japan (3%).

5. DIAGNOSIS  
73% of participants were diagnosed under the age of 18. Nearly 50% of patients were diagnosed less than 4 weeks after onset of symptoms and 57% within 1 year.

6. TREATMENT  
The recommended initial medication, IVIG + two other medications, was utilized by 48% patients in 2017-2018. Patients not using IVIG decreased to only 2%.

7. QUALITY OF LIFE – DURING THE FIRST 6 MONTHS  
The percentage of participants who are somewhat, good, very good, and excellent in terms of their overall health is 35%, 31%, 26%, and 9%, respectively. The percentage of participants who are somewhat, good, very good, and excellent in terms of their general health is 33%, 31%, 21%, and 15%, respectively.

8. CONCLUSIONS  
The OMS Registry provides insight into the natural history of OMS, the challenges in diagnosis and disease awareness, the patient experience, and real-world efficacy of both pharmaceutical and non-pharmaceutical therapies. Continuous collection of registry data will broaden disease awareness, inform treatments, and facilitate the development of further research into OMS.

The PKU Patient Registry:  
a Step for Better Disease Awareness and Improved Patient Outcomes

Elena Buldyreva, Chiao-Han Jeng, Odile Boudacq, Cameron T. Jones, Eberhard Buchler, C. C. Paige McCombs, Mark O. C. Christie, and Jeanne B. Quint

The PKU Patient Registry was established by the Phenylketonuria Genetic Health Alliance (NPKUA) and the Phenylketonuria Hart Foundation (PHF) in 2011. The PKU Patient Registry is a multi-institutional PKU-specific medical data registry designed to provide a unique, longitudinal data source for PKU patients to use to support research, enhance diagnosis and treatment, and provide education for clinicians and patients.

Launched in January 2017, the PKU Patient Registry is a secure web-based survey tool facilitating unparalleled patient reported outcomes, lab results, and clinical symptoms from patients with PKU or their caregivers. The registry contains data for 525 unique respondents of 862 patients who enrolled as of September 2018.

• The PKU Patient Registry represents a unique longitudinal data source that may help better understand the natural history of PKU.
• Participation in the registry may lead to better patient and caregiver awareness of the disease and its impact.
• The registry offers a unique environment where patients and families of PKU can learn about the disease and discuss it with other patients and family members.

• The registry can help the medical community better understand the natural history of PKU to define eligibility criteria for clinical trials.

• The registry can help identify areas for further research and the development of additional clinical trials.

• The registry can help identify potential biomarkers for PKU.

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• The registry can help identify potential biomarkers for PKU.

• The registry can help identify areas for further research and the development of additional clinical trials.

• The registry can help identify potential biomarkers for PKU.
The PKU Patient Registry: Evaluating Treatment Paradigms of Patients with PKU

Eileen Blakely1, Christine Brown1, Olaf Bodamer2, Harvey Levy2, Hilary Feldman, Rhonda Connelly1, Kathryn Moseley1, Vanessa Boulanger3, Allison Seebald3, Suzanne Rossov3, Scott Milligan4, Janna Radtchenko4

The majority of the PKU patients follow PKU diet and generally feel well; although keeping food diary is

METHODS

Patient diagnosed through newborn screening, n (%)

Yes
45%
Sometimes
30%
No
25%

Some patients reported that they are only able to follow the PKU diet during part of the year; 10% reported that they follow a different diet approach altogether. The research team would like to thank patients and families for data contribution to the registry.

Further data collection is needed to better understand maternal PKU.

To date the registry has enrolled 862 patients and collected information from 525 unique

© 2019 Trio Health Advisory Group, Inc.; NORD - National Organization for Rare Disorders, Inc.
Pemphigus and Pemphigoid

The Pemphigus-Pemphigoid Registry: Real-World Diagnostic and Treatment Patterns

**BACKGROUND AND OBJECTIVE**
- The Pemphigus-Pemphigoid Registry is a secure web-based survey launched in January 2017, providing insight into patient demographics, family and medical history, disease characteristics, treatment patterns, and quality of life.

**DESIGN**
- The registry includes surveys completed by PKU patients or caregivers, examining data collected as of March 2019 for 563 participants who completed surveys at various intervals. Data source documentation, including clinic notes and lab results, is used to understand patient demographics, treatment patterns, outcomes, and disease burden.

**PATIENT CHARACTERISTICS N=138**

<table>
<thead>
<tr>
<th>Region</th>
<th>n (%)*</th>
</tr>
</thead>
<tbody>
<tr>
<td>North America</td>
<td>116 (85)</td>
</tr>
<tr>
<td>Asia</td>
<td>4 (3)</td>
</tr>
<tr>
<td>Middle East (Israel, Qatar)</td>
<td>2 (1)</td>
</tr>
<tr>
<td>Other</td>
<td>12 (9)</td>
</tr>
</tbody>
</table>

**AGE AT DIAGNOSIS**
- 55% (203/368) of participants were diagnosed within a week of birth.

**AGE OF DEPRESSION OR ANXIETY ONSET**
- The median age at which participants began experiencing depression was 17 years (mean of 19.5), and the median age at which participants began experiencing anxiety was 13 years (mean of 15.6).

**TREATMENT PATTERNS**
- Forty-three percent of the patients take one medication, 13% take two medications, 44% take 3 or more medications for their condition.

**NOTE:** The research team would like to thank patients and families who contributed their information to the registry. Although the majority of the patients are interested in research participation, very few of them currently participate in clinical trials.

**REFERENCES:**
- 1. International Pemphigus & Pemphigoid Foundation (IPPF), 2. National Organization for Rare Disorders (NORD), 3. Trio Health Analytics

**CONTRIBUTORS:**
- Few patients self-report symptoms of anxiety, depression, or both.
- The number of participants who said they were very or extremely worried about how PKU might affect their future health for those with both anxiety and depression was significantly different compared to those with neither (significance level of 0.05, z=2.352, p=0.019).

**FIGURE 2. PATIENT DISTRIBUTION BY REGION**

**FIGURE 3. PATIENT DISTRIBUTION BY RACE**

**FIGURE 4. PATIENT DISTRIBUTION BY AGE AT DIAGNOSIS**

**FIGURE 5. PATIENT DISTRIBUTION BY TREATMENT PATTERNS**

**SUMMARY:**
- The Pemphigus-Pemphigoid Registry offers a unique opportunity to understand patient experiences with their disease, treatment, and quality of life.

**ACKNOWLEDGEMENTS:**
- The research team would like to thank patients and families who contributed their information to the registry.

**WEBLINKS:**
Socio-demographics

• Management of care
• Treatment and disease progression

Pemphigus and pemphigoid (P/P) are rare, autoimmune blistering diseases that affect a very small percentage of the population, thus real-world data is needed to better understand diagnosis and patient demographics, treatment patterns, outcomes, and disease burden. The registry retrieves patient data for the following purposes:

- To characterize and describe the pemphigus and pemphigoid population as a whole
- To assist the pemphigus and pemphigoid community with the development of recommendations for treatment and disease progression
- Towards better representing female patient population (Figure 2), possibly due to better openness among female patients

As of September 2018, 259 patients enrolled and 138 provided their demographic information and medical history to the registry. Median age at patient diagnosis in the registry is 51.0 years (mean 49.1, SD 14.3). The overall incidence and prevalence of pemphigus varies depending upon the specific population studied. Pemphigus is estimated to affect anywhere from 0.7 -5 people per 1,000,000 per year in the general population. The incidence and prevalence of pemphigoid is 10 times lower than pemphigus. Approximately 40% of patients with pemphigus develop pemphigoid at some point in their disease course and the opposite occurs in 20%. The lower incidence and prevalence of pemphigoid may be due to patients with pemphigus being more likely to die before developing pemphigoid or because pemphigoid is a rare entity. Here, autoantibodies (anti-Dsg and anti-Dplg) are directed against desmoglein-1 and desmoglein-3 proteins. The pemphigus and pemphigoid antibodies can be measured using an indirect immunofluorescence test (IIF) on tissue explants. The antibodies can also be measured in plasma or tissue homogenates using a solid-phase enzyme-linked immunosorbent assay test (ELISA).

The Pemphigus-Pemphigoid Registry:
First Report of Patient Quality of Life Measures

Marc L. Yale 1, Vanessa Boulanger 2, Allison Seebald 2, Suzanne Rossov2, Scott Milligan 3, Janna Radtchenko 3

FIGURE 1. PATIENT QUALITY OF LIFE WITH PATIENT ENJOYMENT OF LIFE

FIGURE 6. PATIENT MENTAL HEALTH

FIGURE 7. TROUBLE DOING ALL USUAL WORK INCLUDING WORK AT HOME

CONCLUSIONS

- Pemphigus and pemphigoid are rare, autoimmune blistering diseases. The registry was created to address the knowledge gaps and to help in understanding the disease burden of P/P.
- The registry is supported by the patient-driven nonprofit organization NORD (National Organization for Rare Disorders).
- The Pemphigus-Pemphigoid Registry is the only worldwide registry that focuses on P/P.

REFERENCES


2. Inadequate sleep quality and fatigue are the most frequently reported symptoms negatively affecting patients lives.

3. Nearly 70% report poor or fair quality of sleep and the same proportion reports that they have to always or usually limit fun activities that they do with others.

4. As a result, 19% of the patients report fair or poor satisfaction with their social activities and relationships. One-fourth of patients report poor or fair mental health.

5. The overall incidence and prevalence of pemphigus varies depending upon the specific population studied. Pemphigus is estimated to affect anywhere from 0.7 -5 people per 1,000,000 per year in the general population.
**Pemphigus and Pemphigoid**

**UNDERSTANDING REAL-WORLD TREATMENT PATTERNS AND QUALITY OF LIFE IN PATIENTS WITH PEMPHIGUS AND PEMPHIGOID USING A PATIENT REGISTRY DATABASE**

**1. BACKGROUND AND AIM**

Pemphigus and pemphigoid (PP) are rare, autoimmune blistering diseases; real-world data is needed to better understand diagnosis, treatment patterns, and quality of life in these difficult to diagnose patients. The PP Registry was launched in March 2016 to fill this urgent need and bring awareness to healthcare providers. Here, we describe characteristics of registry patients, their diagnosis and treatment, and their quality of life.

**2. METHODS**

As of September 2018, the registry has enrolled 359 patients. Patient demographics, diagnosis type, treatment patterns, and quality of life (QoL) measurements including physical, mental, and social health, and efficacy participation were analyzed for patients who provided diagnostic and clinical information. For this study, we examined data collected as of March 2018 for 167 participants who completed 764 surveys.

**3. PARTICIPANT RACE AND SEX**

Participants in the registry are mostly white (80%, 132/165), non-Latino (90%, 149/165), and female (70%, 116/165).

**4. COUNTRY OF RESIDENCE**

Participants in the registry are located in 56 countries, with the majority of participants in the US (80%, 132/167).

**5. AGE AT DIAGNOSIS**

53% (74/127) were diagnosed with Pemphigus Vulgaris.

**6. TREATMENT**

61% (67/109) of patients used oral corticosteroids and 53% (67/127) topical steroids.

**7. QUALITY OF LIFE**

The average age of diagnosis was 48 years, and most participants (94%, 167/175) were diagnosed within 12 m of symptoms onset.

**8. CONCLUSIONS**

PP creates significant burden and negative impact on physical and mental health. Future studies and continued collection of this registry data will continue to help raise awareness amongst medical, research, and patient communities.

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**SYNGAP1-Related NSID**

**UNDERSTANDING DEMOGRAPHICS AND DIAGNOSIS IN SYNGAP1-RELATED NON-SYNDROMIC INTELLECTUAL DISABILITY (NSID) PATIENTS USING A PATIENT REGISTRY DATABASE**

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1Bridge the Gap – SYNGAP Education and Research Foundation, Cypress, TX, USA; 2National Organization for Rare Disorders (NORD), Danbury, CT, USA; 3TRIO Health Analytics, La Jolla, CA, USA

**1. BACKGROUND AND AIM**

SYNGAP1 mutations cause a variety of developmental and neuropsychiatric symptoms, including intellectual disability, seizures, autism, and movement delay. SYNGAP1 mutations are found in approximately 1 in 100,000 people and result in SYNGAP1-NSID, a non-syndromic intellectual disability (NSID) in people with no other known causes. As such, confirmation of SYNGAP-related NSID is through genetic testing. SYNGAP1-NSID is thought to result from limited functional levels of SynGAP, a protein critical in proper brain development and function. Predominantly affecting children, SYNGAP1 mutations lead to developmental delay, intellectual disability, and additional symptoms that are common with other causes. As such, SYNGAP1-NSID is a heterogeneous condition with a broad range of phenotypes.

**2. METHODS**

In December 2018, SYNGAP1-NSID was designated as an Orphan Disease by the US National Institutes of Health. SYNGAP1-NSID is an Orphan Disease of National Significance, which allows for the SYNGAP1-NSID Registry to leverage funds from the US Food and Drug Administration. Patients referred to the SYNGAP1-NSID Registry have received support from the SYNGAP Research Foundation, a 501(c)(3) nonprofit organization. The SYNGAP1-NSID Registry was created to provide a comprehensive database of data to help facilitate future research.

**3. PATIENTS**

The registry contains 13 surveys covering diagnostics, disease, treatment, care, management, and quality of life. As of December 2018, 105 patients have provided data for 717 survey submissions.

**4. RACE AND GENDER**

73% (78/105) of patients were White, 13% (14/105) were Latino, and 3% (3/105) were American Indian. 40% (42/105) of patients were male, 35% (37/105) were female, and 25% (26/105) were Assigned Male at Birth (AMAB). 54% (58/105) of patients had documented genetics.

**5. AGE AT DIAGNOSIS**

46% (49/106) of patients had delayed or impaired motor milestones, and 54% (57/106) had delayed or impaired cognitive milestones. 46% (49/106) were diagnosed with congenital heart defects. 58% (62/107) had feeding problems, and 42% (45/107) had prematurity. Nearly all participants (97%, 57/59) were diagnosed with delayed or impaired speech. 67% (72/108) had behavioral problems. 61% (67/109) were diagnosed with seizures, and 76% (83/109) had self-injurious behavior.

**6. TREATMENTS**

42% (43/102) were on immunosuppressants (Cyclophosphamide, Azathioprine, Methotrexate, Mycophenolate Mofetil, Prednisone), and 39% (40/103) were on plasma exchange.

**7. QUALITY OF LIFE**

76% (85/111) of participants had used an assistive device to help them move around, communicate, or do things. 19% (19/102) of participants had used a wheelchair, and 17% (16/102) wore foot braces. 53% (58/109) of participants wore glasses.

**8. PHYSICAL AND MOVEMENT**

43% (43/98) of patients could only move their torso, 49% (49/99) only their arms and legs, and 38% (38/99) only their head. 53% (53/99) of patients had trouble feeding children, 43% (43/99) trouble feeding adults, 54% (54/99) could not walk, and 47% (47/99) could not run.

**9. SUMMARY**

Families of SYNGAP1-NSID patients have shared important information to help diagnose SYNGAP1-NSID, and the registry continues to expand to support research and help SYNGAP1-NSID patients and their families.
SYNGAP1-Related NSID

QUALITY OF LIFE IN PEDIATRIC PATIENTS WITH SYNGAP1-RELATED NON-SYNDROMIC INTELLECTUAL DISABILITY (NSID); DATA FROM THE SYNGAP1 (MRD5) PATIENT REGISTRY

Understanding Disease and Burden in SYNGAP1-Related Non-Syndromic Intellectual Disability (NSID) Patients Using a Patient Registry Database

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1Bridge the Gap Education and Research Foundation, Cypress, TX, USA; 2National Organization for Rare Disorders (NORD), Danbury, CT, USA, 3TRIO Health Analytics, La Jolla, CA, USA

1. BACKGROUND AND AIM

Predominantly affecting children, SYNGAP1 mutations lead to developmental delay, intellectual disability, and additional symptoms that are sometimes with other causes. As such, confirmation of SYNGAP-related NSID is through genetic testing. To improve awareness and understanding of SYNGAP-related NSID and better inform treatment development, the Bridge the Gap Education and Research Foundation, in partnership with the US Food and Drug Administration, launched the SYNGAP (MRD5) patient registry in 2017. Here, we describe patient demographics, diagnoses, and quality of life in registry patients.

2. METHODS

The registry contains 13 surveys covering demographics, disease, treatment, care management, and quality of life. As of March 2019, 152 patients have provided data for this survey submission.

3. PARTICIPANT RACE AND SEX

Participants in the registry are mostly white (80%, 98/111), non-Hispanic or Latino (76%, 85/111), and female (55%, 62/111).

4. COUNTRY OF RESIDENCE

Patients in the registry are located in 24 countries, with the majority of participants in the US (54%, 43/81).

5. DISEASE

All respondents indicated diagnosis before age 18, with 54% diagnosed before age 5. 43% of participants were diagnosed within one year of symptom onset.

6. TREATMENT

Most participants (84%) indicated that they took medications to treat side effects.

7. QUALITY OF LIFE - OVER THE PAST 4 WEEKS

Patients in the registry have severe disease burden and impaired quality of life. Data collection through the SYNGAP (MRD5) patient registry continues with the intent of raising awareness of the disease and enabling the development of treatments.

8. CONCLUSIONS

Patients in the registry have significant disease burden and impacted quality of life. Data collection through the SYNGAP (MRD5) patient registry continues with the intent of raising awareness of the disease and enabling the development of treatments.

ISPOR 2019 | May 18-22, 2019 | New Orleans, LA, USA
1. BACKGROUND AND AIMS
As of January 2019, 357 registry patients.

2. METHODS
The registry initiated February 2017 and contains 15 surveys covering diagnostics, care management, and quality of life. PARTICIPANTS

3. RACE (N=357)
- Male: 64%
- Female: 36%

4. SYMPTOMS LEADING PATIENT TO SEEK CARE
- Headaches: 5%
- Other: 22%

5. MISDIAGNOSIS BY INITIAL TUMOR LOCATION
- Abdominal wall: 61%
- Joint / extremity: 42%
- Head / neck: 9%

6. QUALITY OF LIFE (QOL) AND TUMOR LOCATION IN PATIENTS WITH DESMOID TUMORS;
DISEASE MANAGEMENT AND QUALITY OF LIFE

- Data regarding the diagnosis of their desmoid tumor was provided by 127 respondents: a comparison of symptoms between those who had tumors diagnosed in a head and neck location or in an extremity

7. MISDIAGNOSIS OF DESMOID TUMORS; INSIGHT FROM THE DESMOID TUMOR RESEARCH FOUNDATION (DTRF) NATURAL HISTORY STUDY

- Data collected through the DTRF patient registry provides a growing awareness of desmoid tumors

8. SYMPTOMS LEADING PATIENT TO SEEK CARE
- Headaches: 5%
- Other: 22%

9. SUMMARY
- Most participants (88%) were diagnosed within 5 years of onset of symptoms
- The mean age of diagnosis is 32.5 years
- Registry participants are mostly female (78%, 277/357)
- Patients with desmoid tumors have varied QOL and tumor locations. Data collection through the registry is ongoing and essential to raising awareness of this disease and improving care for these patients.

10. MISDIAGNOSIS BY INITIAL TUMOR LOCATION
- Misdiagnosis rates vary by initial tumor location and are highest for abdominal wall and joint / extremity. Assessment of approaches using clinical best practices focused on improving accuracy and effectiveness as more evidence accumulates.
Patients with the autoimmune disease ITP suffer from bleeding events as a result of low platelet counts. These events may manifest as bruises, petechiae, blood in the stool, or nosebleeds. In 2017, PDSA in collaboration with NORD launched the ITP Natural History Registry to understand patient characteristics, their disease, disease management, and quality of life. Here, we describe the demographics and quality of life for registrants to date.

### Quality of Life and Demographics of Patients with Immune Thrombocytopenia (ITP)

**Methodology**

The PDSA Registry contains 6 surveys covering patient demographics, medical and diagnostic information, treatment utilization, disease progression, and quality of life. Here, we describe the quality of life for registrants to date, as it differs significantly depending on whether the participant had had a splenectomy.

**Data Collection**

Data collection continues through the PDSA ITP Natural History Registry with the intent of raising disease awareness and understanding the impact of the disease.

**Characteristics of Patient Registrants**

- **Age at Diagnosis**: 32 years (median age)
- **Time from Onset of Symptoms to Diagnosis**: Median of 3 months
- **Race**: 62% White, 35% Black or African American, 1% Other or Unknown
- **Sex**: 53% Female, 47% Male
- **Splenectomy**: 17% of participants had a splenectomy
- **Medication Use**: Prednisone (42%), IVIG (23%), Steroids (12%)
- **Insurance Coverage**: 36% Commercial, 32% Medicaid
- **Severity of Disease**: 60% Excellent, 20% Very Much, 10% Some, 10% No

**Quality of Life**

- **Participant Anxiety in the Past 7 Days**: 53% reported feeling anxious
- **Participant Fatigue in the Past 7 Days**: 81% reported feeling fatigued

**Conclusion**

The number of participants who use TPO and did not report fatigue was not significantly different than the number of total participants who did not report fatigue (z=0.409, p=0.682, not significant at p < 0.05). Anxiety and fatigue are common concerns for patients with ITP, and understanding how these symptoms impact their quality of life is crucial for improving their care.
ACKNOWLEDGMENTS

The IPPF would like to acknowledge its Board of Directors and staff for all of their efforts in bringing the IPPF Natural History Study and this book to fruition. The foundation would especially like to thank all patients with pemphigus and pemphigoid who have the courage to share their stories, especially Marc, Hub, Lisa, Ellen, and Fred. Their advocacy and willingness to do whatever it takes to spread awareness about the burden of these diseases is an example of the hope that we can give others.

The DTRF would like to acknowledge its Board of Directors and staff for all of their efforts in bringing the DTRF Natural History Study and this book to fruition. The foundation especially would like to thank all patients with desmoid tumors who have the courage to share their stories, especially Rudy, Sandy, and Jack.

THE DESMOID TUMOR RESEARCH FOUNDATION

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THE DESMOID TUMOR RESEARCH FOUNDATION

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