<table>
<thead>
<tr>
<th>Study Events</th>
<th></th>
</tr>
</thead>
<tbody>
<tr>
<td>Consent Information</td>
<td>3</td>
</tr>
<tr>
<td>Family History</td>
<td>5</td>
</tr>
<tr>
<td>Registry Visit</td>
<td>8</td>
</tr>
<tr>
<td>Visit Information</td>
<td>11</td>
</tr>
<tr>
<td>Vitals / Measures</td>
<td>13</td>
</tr>
<tr>
<td>Clinical History</td>
<td>15</td>
</tr>
<tr>
<td>Immunizations, Infusions, Transfusions</td>
<td>31</td>
</tr>
<tr>
<td>Medications &amp; Supplements</td>
<td>36</td>
</tr>
<tr>
<td>Surgeries &amp; Procedures</td>
<td>45</td>
</tr>
<tr>
<td>Transplantation &amp; Gene Therapy</td>
<td>50</td>
</tr>
<tr>
<td>Survival &amp; QoL</td>
<td>55</td>
</tr>
<tr>
<td>Core Lab Test Results</td>
<td>56</td>
</tr>
<tr>
<td>Molecular Information</td>
<td>61</td>
</tr>
</tbody>
</table>
Study Events

- There are 3 events in which you will enter data
  - **Consent**: required before entering clinical data
  - **Family History**: captures history of disease (PI or otherwise)
  - **Registry Visit**: captures all clinical data for your subject

- **Registry Visit** is the only event which collects longitudinal data.
  - Family History & Consent may be updated as necessary, but **do not** need to be updated with every registry visit.
• **Consent type:** indicate the IRB used at your enrolling site.
• Please specify reasoning for any IRB waivers.
Future Contact & Data Sharing

1. Per consent, patient allows USIDNET to contact him/her.
2. Per local protocol, patient allows USIDNET to contact his/her physician for additional studies.
3. Consent allows sharing of data with CIBMTR, PIDTC and related research data platforms (provide ID if available)

💡 If your consent form does not include these questions, answer NO.
Family History of PI

- Please complete this section, even if the answer is Unknown.
- You will also be asked to enter the family member and their PI diagnosis, if applicable.

- Are there other patients with a primary immunodeficiency in the family?
  - Yes
  - No
  - Unknown

- How many?

- Did a positive family history prompt testing of this patient?
  - Yes
  - No
  - Unknown

- Are the parents of the patient related to each other?
  - Yes
  - No
  - Unknown
Family History of other Diseases

• We would also like to know GENERALIZED family history of the following:
  – Autoimmune Diseases
  – Inflammatory Diseases
  – Malignancies
Family History of Genetic Testing

For family members *without primary immunodeficiency*, USIDNET collects the following:

- Relation to Patient
- Gene Tested
- Test Results
What is a **Registry Visit**?

- **Registry Visit**: time period in which a patient was seen by a provider for their immunodeficiency
  - **INITIAL VISIT**: time period up to (and including) official PI diagnosis
    - **HISTORY**: symptoms which prompted an immune evaluation
    - **LABS**: laboratory tests completed to confirm diagnosis
    - **TREATMENT**: How were symptoms cared for? What therapy was administered?
  - **FOLLOW UP VISIT**: encounters with info that is either new or has changed
    - **HISTORY**: new symptoms or infections that have developed
    - **LABS**: recent lab reports
    - **TREATMENT**: any changes in the way the PIDD is treated
What if data near diagnosis is unavailable?

If you do not have data near the time of diagnosis, follow these guidelines.

- **INITIAL VISIT:** all clinical information pertinent to the patients PIDD history
  - **HISTORY:** symptoms linked to this patient’s immunodeficiency
  - **LABS:** recent laboratory test results
  - **TREATMENT:** How did your institution decide to treat this patient?

- **FOLLOW UP VISIT:** Any information that is either new or has changed
  - **HISTORY:** have new infections developed? Have symptoms changed?
  - **LABS:** the most updated lab reports available.
  - **TREATMENT:** any changes in PI treatment?
Registry Visit

- Registry Visit contains 10 Core CRFs
  - Capture uniform data fields for all participants in the registry.
- A Disease-Specific CRF may also be visible.
  - Automatically detected and assigned based upon the patient diagnosis

- Visit Information
- Vitals Measures
- Clinical History
- Immunizations Infusions Transfusions
- Medications and Supplements
- Surgeries and Procedures
- Transplantation and Gene Therapy
- Survival and QoL
- Core Laboratory Test Results
- Molecular Information
Visit Information

- This tab documents the following:
  - Time period referenced in this visit
  - Subject’s PI diagnosis.

- **MUST BE COMPLETED FIRST:** no other tabs are accessible until this section is completed
Visit Information

Please answer the following question for each visit:

Visit covers up to the following date

This date is the most recent encounter used to abstract information for this entry.
Vitals / Measures

• This tab collects the vitals recorded during a clinical encounter.

• When entering data, please keep the following in mind:
  • **METRIC OR STANDARD:** when entering height and weight, you will first choose a set of units.
  • **BMI:** USIDNET automatically calculates BMI based upon height and weight. You cannot manually enter a BMI.
Vitals / Measures

- Date (or age)
  - Initial Visit: closest to diagnosis
  - Follow-Up: most recent visit

- Height and Weight
  - Feet and lbs are integer values.

- BMI: automatically calculated

- Head Circumference: if available
Clinical History

• This tab collects the following pieces of information:
  – Newborn Screening
  – History of Disease
  – Infections
  – Conditions
  – Allergies
  – Malignancies
Clinical History

- **Newborn Screening:** complete if known
- **Onset of Symptoms:** when did patient start having symptoms of PIDD?
- **Diagnosis Established:** when was the PIDD diagnosis clinically determined?
Clinical History: Infections

- Has patient experienced infections?
- Infection Name(s)
- Infectious Organism(s)
- Infection Details
  - Present BEFORE OR AT PI diagnosis?
  - New instance or a continuing issue?
**Clinical History: Infections**

<table>
<thead>
<tr>
<th>Infection Name</th>
<th>Presented before or at PI diagnosis</th>
<th>This infection is a...</th>
<th>Organism</th>
<th>Organism</th>
<th>Organism</th>
</tr>
</thead>
</table>
Example: Patient had pneumonia infections which prompted the PI evaluation. Even with IVIG, pneumonia is still a frequent problem.
**Clinical History: Infections**

<table>
<thead>
<tr>
<th>Infection Name</th>
<th>Presented before or at PI diagnosis</th>
<th>This infection is a...</th>
<th>Organism</th>
<th>Organism</th>
<th>Organism</th>
</tr>
</thead>
<tbody>
<tr>
<td>Chicken Pox</td>
<td>[ ] Yes</td>
<td>[ ] New instance</td>
<td>Varicella-zoster</td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td>[ ] No</td>
<td>[ ] Continuing Issue</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td>[ ] Unknown</td>
<td>[ ] Unknown</td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

**Example:** Patient contracted *chicken pox* from brother. Documented in a follow-up encounter.
Clinical History: Conditions

All **non-malignant** systemic & organ-specific conditions are collected in this section.

- Has patient experienced conditions?
- **Condition Name(s)**
- **Condition Details**
  - Present BEFORE OR AT PI diagnosis?
  - Persists at this visit?
Clinical History: Conditions

Example: Patient has moderate asthma. First asthma attack was in 2005, and he was diagnosed with a PI in 2007.
Clinical History: Conditions

Example: a patient presents for annual follow up and reports newly-diagnosed steatosis.
## Clinical History: Conditions

<table>
<thead>
<tr>
<th>Condition</th>
<th>Present before or at PI diagnosis</th>
<th>Condition persists at visit</th>
</tr>
</thead>
<tbody>
<tr>
<td>Acne</td>
<td>Yes</td>
<td>Yes</td>
</tr>
</tbody>
</table>

**Example:** a newly-diagnosed adult patient mentions a history of teenage acne that has since resolved.
Clinical History: Allergic Reactions

Please answer the following

- Has patient experienced allergic reactions?
- Reaction Name(s)
- Agent Name(s)
Clinical History: Allergic Reactions

<table>
<thead>
<tr>
<th>Agent</th>
<th>Specify</th>
<th>Reaction</th>
<th>Reaction Unknown</th>
</tr>
</thead>
<tbody>
<tr>
<td>Food</td>
<td>Pineapple</td>
<td>Anaphylaxis</td>
<td></td>
</tr>
</tbody>
</table>

**Example**: patient exhibits anaphylaxis if exposed to pineapple.
Clinical History: Allergic Reactions

<table>
<thead>
<tr>
<th>Agent</th>
<th>Specify</th>
<th>Reaction</th>
</tr>
</thead>
<tbody>
<tr>
<td>Environmental</td>
<td></td>
<td>Allergic Rhinitis</td>
</tr>
</tbody>
</table>

Example: patient has allergic rhinitis in Spring and Autumn
Clinical History: **Allergic Reactions**

<table>
<thead>
<tr>
<th>Agent</th>
<th>Specify</th>
<th>Reaction</th>
</tr>
</thead>
<tbody>
<tr>
<td>Drug</td>
<td>Bactrim</td>
<td>Reaction Unknown</td>
</tr>
</tbody>
</table>

**Example:** a Bactrim allergy is listed in the patient record.
Clinical History: Malignancies

Please answer the following:

• Does patient have a history of malignancy?
• Name of Cancer(s)
• Cancer Details
  • Presented before or at PI diagnosis?
  • Persists at this visit?
  • Treatments
### Clinical History: Malignancies

<table>
<thead>
<tr>
<th>Name of Cancer</th>
<th>Presented before or at PI diagnosis</th>
<th>Condition persists at visit</th>
<th>How treated?</th>
</tr>
</thead>
<tbody>
<tr>
<td>Melanoma</td>
<td>Yes/No/Unknown</td>
<td>Yes/No/Unknown</td>
<td>Surgery, Radiation</td>
</tr>
</tbody>
</table>

**Example:** patient is undergoing post-operative radiation therapy for melanoma. Patient did not have melanoma when diagnosed with PI.
The registry uses **conditional logic** to only display vaccine names if the user selects “Yes”.
Similarly, the **Vaccine Reactions Table** will only display names of vaccines that were selected.
Immunizations, Infusions, Transfusions

- Immune Globulin Replacement Therapy
  - All questions in this section utilize conditional logic to display relevant questions.
  - On the next slide, please reference the flowchart to see which fields are applicable to your patient.
Immunizations, Infusions, Transfusions

- Has patient EVER received Ig Therapy?
  - Yes
    - Is patient currently on Ig Therapy?
      - Yes
        - Start Date (or age)
        - Route
        - Dose
        - Frequency
        - Reactions to Treatment
      - No
        - End Date (or age)
        - Reactions to Treatment
  - No
    - Reactions to Treatment
If your patient received a **blood transfusion**, please mark the appropriate checkboxes.
Patient medications are divided into 3 sections

- Anti-Infective Medication
- Immunomodulators
- Other Medications and Supplements
Medication: Anti-Infectives

In this section, you will answer the following:

- Has patient used anti-infectives in the past year?
- How many courses of antibiotics were used for active infection in the past year?

If patient has used anti-infectives, you will be asked to enter:

- Prophylactic Anti-Infectives
- Non-Prophylactic Anti-Infectives
**Medication: Prophylactic Anti-Infective**

<table>
<thead>
<tr>
<th>Drug Name</th>
<th>Course</th>
<th>Adverse Reaction?</th>
<th>Reaction</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>Continuous</td>
<td>Yes</td>
<td>Unknown</td>
</tr>
<tr>
<td></td>
<td>Intermittent</td>
<td>No</td>
<td></td>
</tr>
<tr>
<td></td>
<td>Rotating</td>
<td>Unknown</td>
<td></td>
</tr>
</tbody>
</table>

- Drug Name is linked to [RXNORM](http://rxnorm.nlm.nih.gov), a medical dictionary.
- RXNORM accepts **brand and generic** drug names.
- You **do not** need to provide **dose**, but you can if you wish.
**Medication: Other Anti-Infective**

<table>
<thead>
<tr>
<th>Drug Name</th>
<th>Indication</th>
<th>Specify</th>
<th>Outcome</th>
<th>Adverse Reaction?</th>
<th>Reaction</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>☐ Acute Infection</td>
<td>☐</td>
<td>☐Resolved</td>
<td>☐Yes</td>
<td>☐</td>
</tr>
<tr>
<td></td>
<td>☐ Chronic Infection</td>
<td>☐</td>
<td>☐Unresolved</td>
<td>☐No</td>
<td>☐</td>
</tr>
<tr>
<td></td>
<td>☐ Unknown</td>
<td>☐</td>
<td>☐Unknown</td>
<td>☐Unknown</td>
<td>☐</td>
</tr>
</tbody>
</table>

- Please make sure that you are entering anti-infectives in the appropriate section (prophylactic vs. non)
- Drug Name is also linked to **RXNORM**, a medical dictionary.
Medication: Immunomodulators

In this section, you will answer the following

- Has patient received immunomodulator medication within the past year?

If yes, you will be asked to enter:

- Non-Transplant Immunomodulators
- For-Transplant Immunomodulators
**Medication: Immunomodulators**

Immunomodulator Medications include (but are not limited to):

- Anti-Inflammatory Drugs
- Biologic Therapy
- Cytokines
- Immune Suppressives
**Medication: Non-Transplant Immunomodulators**

<table>
<thead>
<tr>
<th>Drug Name</th>
<th>Indication</th>
<th>Improvement</th>
<th>Adverse Reaction</th>
<th>Reaction</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>Prophylaxis</td>
<td>Yes</td>
<td>Yes</td>
<td></td>
</tr>
<tr>
<td></td>
<td>Treat Disease</td>
<td>No</td>
<td>No</td>
<td></td>
</tr>
<tr>
<td></td>
<td>Unknown</td>
<td>Unknown</td>
<td>Unknown</td>
<td></td>
</tr>
</tbody>
</table>

- Drug Name is linked to RXNORM, a medical dictionary.
- RXNORM accepts **brand and generic** drug names.
- You **do not** need to provide **dose**, but you can if you wish.
Medication: For-Transplant Immunomodulators

- Please make sure that you are entering immunomodulators in the appropriate place (transplant vs. non)
- Drug Name is linked to RXNORM, a medical dictionary.
**Medication & Supplements: Other**

<table>
<thead>
<tr>
<th>Name</th>
<th>Adverse Reaction?</th>
<th>Reaction</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>☐ Yes</td>
<td>☐ No</td>
</tr>
</tbody>
</table>

- If participant regularly takes **ANY ADDITIONAL** medications or supplements to manage their health, please include here.
- Drug Name is also linked to **RXNORM**, a medical dictionary.
Surgeries & Procedures

- This CRF collects data on
  - Surgeries
  - Non-Surgical Support
  - Other Treatments
In the **Surgeries** section, please provide:

- Name of Surgery
- Date of Surgery

Surgeries are linked to **Medical Coding**
The *Procedures* section is divided into 3 categories:

<table>
<thead>
<tr>
<th>Nutritional Support</th>
<th>• Parenteral Nutrition</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>• Enteral Nutrition</td>
</tr>
<tr>
<td></td>
<td>• Others</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Respiratory Support</th>
<th>• ECMO</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>• Intubation</td>
</tr>
<tr>
<td></td>
<td>• CPAP</td>
</tr>
<tr>
<td></td>
<td>• BiPAP</td>
</tr>
<tr>
<td></td>
<td>• Oxygen Supplementation</td>
</tr>
<tr>
<td></td>
<td>• Others</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Therapy &amp; Educational Support</th>
<th>• Individualized Educational Plan (IEP)</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>• Mental Health Services</td>
</tr>
<tr>
<td></td>
<td>• Occupational / Physical / Speech Therapy</td>
</tr>
<tr>
<td></td>
<td>• Others</td>
</tr>
</tbody>
</table>
## Surgeries & Procedures

For each category, we would like to know:

- Has patient received this support?
- Type of Support
- Date Began
- Still Needed?
## Procedures: Other Treatments

<table>
<thead>
<tr>
<th>Does this patient regularly use any of the following to manage their primary immunodeficiency?</th>
</tr>
</thead>
<tbody>
<tr>
<td>(Select all that apply)</td>
</tr>
</tbody>
</table>

- Chiropractor
- Acupuncture
- Herbal Remedies
- Homeopathic Practitioners
- Other (specify)

If patient uses any of the treatments above to manage their PI, please complete this question.
Transplantation & Gene Therapy

This section collects data on the following treatments:

- Hematopoietic Stem Cell Transplant
- Solid Organ Transplant
- Gene Replacement Therapy

Information entered in this section will automatically transfer over to every new Registry Visit.
Transplantation: Stem Cell

USIDNET collects the following data points:

- Date (or age)
- Reason
- Donor
- Graft
- Purification of Stem Cells
- Conditioning
- Engraftment
- Chimerism
- Acute GVHD
- Chronic GVHD
Transplantation: **Solid Organ**

USIDNET collects the following data points:

- Date (or age)
- Reason
- Donor
- Organ
- Immunosuppression
- Outcome
Transplantation Considered, not Performed

If patient did not have a transplant:
- Transplant considered & not performed?
- Why not?

<table>
<thead>
<tr>
<th>Solid Organ</th>
</tr>
</thead>
<tbody>
<tr>
<td>- Lack of donor</td>
</tr>
<tr>
<td>- Age or condition of the patient</td>
</tr>
<tr>
<td>- Unfavorable probability of success</td>
</tr>
<tr>
<td>- Religious objections</td>
</tr>
<tr>
<td>- Insurance denial</td>
</tr>
<tr>
<td>- Hyper-immune status</td>
</tr>
<tr>
<td>- Other</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Stem Cell Transplant</th>
</tr>
</thead>
<tbody>
<tr>
<td>- Lack of donor</td>
</tr>
<tr>
<td>- Age or condition of the patient</td>
</tr>
<tr>
<td>- Unfavorable probability of success</td>
</tr>
<tr>
<td>- Religious objections</td>
</tr>
<tr>
<td>- Insurance denial</td>
</tr>
<tr>
<td>- Other</td>
</tr>
</tbody>
</table>
USIDNET collects the following data points

- Gene Therapy (Y/N)
- Date (or age)
- Gene
- Vector Type
- Cells Transduced
- Conditioning
- Results
- Complications
Survival and QoL

In this section you will enter the following

• **Is patient alive?**
  – If patient is deceased, please enter date and cause if available

• **Patient Disabilities**
  – Please describe conditions and severity of disability

• **Karnofsky & Lansky Indexes**
  – Classifies patients by functional impairment.
  – **Karnofsky** used for adults, **Lansky** used for pediatrics
Core Lab Test Results

To enter lab results, please follow these steps:

1. Check the **Data Available** boxes for all tests that have been performed

2. **Initial Visit**: Enter diagnostic labs if available (i.e. before treatment initiated)
   - If diagnostic labs are not available, enter any available data.

3. **Follow-up Visit**: Enter any available updated labs
<table>
<thead>
<tr>
<th>Complete Blood Count</th>
<th>Lymphocyte Phenotype</th>
<th>Lymphocyte Phenotype</th>
<th>Immunoglobulins</th>
</tr>
</thead>
<tbody>
<tr>
<td>WBC</td>
<td>Abs Lymph Count</td>
<td>Total Memory B</td>
<td>IgG</td>
</tr>
<tr>
<td>Platelets</td>
<td>CD3 T Cells</td>
<td>IgM Memory B</td>
<td>IgA</td>
</tr>
<tr>
<td>RBC</td>
<td>CD4 Helper T</td>
<td>Switched Memory B</td>
<td>IgM</td>
</tr>
<tr>
<td>Hgb</td>
<td>CD8 Cytotoxic T</td>
<td></td>
<td>IgM</td>
</tr>
<tr>
<td>Lymphocytes</td>
<td>CD19 B Cells</td>
<td></td>
<td>IgE</td>
</tr>
<tr>
<td>PMN</td>
<td>CD20 B Cells</td>
<td></td>
<td>IgD</td>
</tr>
<tr>
<td>Eosinophils</td>
<td>CD56/CD16</td>
<td></td>
<td>IgG1</td>
</tr>
<tr>
<td>Basophils</td>
<td>CD4 Subsets</td>
<td></td>
<td>IgG2</td>
</tr>
<tr>
<td>Monocytes</td>
<td>CD8 Subsets</td>
<td></td>
<td>IgG3</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td>IgG4</td>
</tr>
</tbody>
</table>
Core Lab: Vaccine Responses

• Was Testing Done?
• Date of Test
• Result

<table>
<thead>
<tr>
<th>Vaccines</th>
<th>Results</th>
</tr>
</thead>
<tbody>
<tr>
<td>Diphtheria</td>
<td>Chicken Pox</td>
</tr>
<tr>
<td>Tetanus</td>
<td>Measles</td>
</tr>
<tr>
<td>HIB Polysaccharide</td>
<td>PHI X 174</td>
</tr>
<tr>
<td>Isohemagglutinin - Anti A</td>
<td>Protein Conjugated HIB</td>
</tr>
<tr>
<td>Isohemagglutinin - Anti B</td>
<td>Mumps</td>
</tr>
<tr>
<td>Diphtheria</td>
<td>Rubella</td>
</tr>
</tbody>
</table>
Core Lab: Pneumococcal Vaccine

For both Conjugated & Unconjugated Vaccines

- Testing Done?
- Date of Test
- # of protective serotypes
- Total # of serotypes tested
- Vaccine Name
- # of times administered
Core Lab: Additional Responses

- Autoantibodies
- Complement Function
- Delayed Hypersensitivity Skin Testing
- Lymphocyte Function
- TRECsg
- TRC V-Beta Repertoire
Molecular Information

This section collects all genetic information

- Pattern of Inheritance
- Gene Mutations
- Enzyme Testing
Molecular Info: Gene Mutation

Is this patient’s PI linked to a Gene Mutation?

- Gene
- Type of Mutation
- DNA Change
- Protein Change
- Zygosity

You may enter up to 3 different genes
## Molecular Info: Enzyme Analysis

- **Protein Name**
- **Protein Expressed?**
- **Test Method** *(Western, FACs, Other)*
Don’t Forget!

• When finished entering data in a CRF, **mark the CRF as complete**.

• The registry will make sure that **required fields** are filled & data passes **validation checks**.

• Only click the checkbox after **all sections** in a form have been completed, as it applies to the entire CRF.