Questionnaire to be filled in by patient and physician

Dear patient, dear parent,
Please fill in this questionnaire as far as possible. Please mark the fields which you cannot answer yourself and ask your physician to complete them during your visit today.

Name: _____ **Visit date**:_____

Date of brith: _____ Country of birth: _____

Gender: ❑ female ❑ male Weight: _____

Which primary immunodeficiency (PID) have you been diagnosed with?

_____

Does the diagnosis fulfil the new ESID Registry diagnostic criteria*? ❑ yes ❑ no ❑ not in the list ❑ unknown
(*See PDF-Datei Registry diagnostic criteria auf http://esid.org/Working-Parties/Registry/Diagnosis-criteria.)

Do you have **relatives** affected by a PID? ❑ no ❑ yes: _____ ❑ unknown

Do you have a **twin sibling**? ❑ no ❑ yes ❑ identical twin ❑ non-identical twin

Are your **parents** related by blood? (e.g. cousins of first or second degree) ❑ no ❑ yes ❑ possible ❑ unknown

1. Date of first **clinical** diagnosis of **a primary immunodeficiency**: _____
❑ Date unknown ❑ Only genetically diagnosed

Genetics: ❑ No genetic analysis performed
Affected gene: _____ ❑ No mutation found

Date of genetic diagnosis: _____ ❑ Date unknown
Lab that performed the genetic analysis (name, town):_____

Reason for genetic analysis:
❑ Analysis following clinical diagnosis ❑ Family screening ❑ Prenatal diagnosis
❑ Diagnosis by neonatal screening ❑ unknown

Sequencing method
❑ Gene sequencing ❑ Whole exome/genome sequencing ❑ Non-genetic definitive test ❑ unknown

2. **What were your first symptoms?**
Please tick the first symptoms **suggestive of a PID**.

❑ **Infection**

❑ **Immune dysregulation** such as:
lymphoproliferation (splenomegaly, hepatomegaly, lymphadenopathy), granuloma, autoimmunity (e.g. cytopenia, thyroid, joint disease, hepatitis, vitiligo, alopecia, diabetes), inflammatory bowel disease, celiac disease, vasculitis, eczema, autoinflammatory disease

❑ **Malignancy**

❑ **Syndromal manifestations** such as:
Dysmorphic features such as short stature, facial abnormalities, microcephaly, skeletal abnormalities, other organ manifestations such as albinism, hair or tooth abnormalities, heart or kidney defects, hearing abnormalities, primary neurodevelopmental delay, seizures

❑ **Others**: please specify: _____
❑ **unknown**
❑ **no symptoms**

Version : Sep 8, 2014

3. When did the PID present for the first time (onset of symptoms)?
Date or age _____ _____
Or approximate age: ❑ 0, ❑ 1-5 years ❑ 6-10 y. ❑ 11-15 y. ❑ 16-20 y. or older: _____

❑ Date completely unknown ❑ No symptoms at all

4. Was the patient diagnosed in the absence of symptoms on the basis of lab abnormalities? (i.e. you had no symptoms related to a PID)
❑ yes ❑ no ❑ unknown

If yes, which kind of lab abnormalities:
❑ Lymphopenia ❑ Neutropenia ❑ Thrombocytopenia
❑ Anaemia ❑ Monocytopenia ❑ Elevated IgE
❑ Hypogammaglobulinaemia ❑ Other, please specify:
_____

5. Are you currently being treated with immunoglobulin replacement? ❑ yes ❑ no
If yes,
Current brand name: _____
Route of application: ❑ subcutaneous ❑ intravenous ❑ intramuscular
Place of application?: ❑ home therapy ❑ in-patient clinic ❑ out-patient clinic
Dose: _____ (_____mg/kg) How often? _____

Side effects: ❑ yes ❑ no ❑ unknown

If yes, type of side effects:
❑ Anaphylaxis ❑ Aseptic meningitis ❑ Fever
❑ Headache ❑ Local side effects (rash, swelling…)
❑ Renal failure ❑ Venous thrombosis ❑ Arterial thrombosis
❑ Other, specify: _____

When did you receive immunoglobulin replacement **for the first time**? _____

6. Have you ever received a stem cell transplant (HSCT)? ❑ yes ❑ no ❑ unknown

If yes, date of transplantation _____

Donor: ❑ MSD (Matched sibling donor) ❑ MUD (Matched unrelated donor)
❑ MMUD (Mismatched unrelated donor) ❑ Haplo-identical (parent) donor
❑ other related donor ❑ Autologous ❑ unknown

Source of stem cells: ❑ bone marrow ❑ cord blood ❑ peripheral blood ❑ unknown

7. Have you ever been treated with gene therapy? ❑ yes ❑ no ❑ unknown

If yes, date of therapy: _____

If more than one HSCT or gene therapy has been performed, please write the details on a separate sheet.

_____ _____
Date, patient's or parents' signature Date, physician's signature

Thank you for taking the time for the ESID Registry! ☺