

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 birth: _____ 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

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:

- | | | |
|---|---|---|
| <input type="checkbox"/> Lymphopenia | <input type="checkbox"/> Neutropenia | <input type="checkbox"/> Thrombocytopenia |
| <input type="checkbox"/> Anaemia | <input type="checkbox"/> Monocytopenia | <input type="checkbox"/> Elevated IgE |
| <input type="checkbox"/> Hypogammaglobulinaemia | <input type="checkbox"/> 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:

- | | | |
|--|---|--|
| <input type="checkbox"/> Anaphylaxis | <input type="checkbox"/> Aseptic meningitis | <input type="checkbox"/> Fever |
| <input type="checkbox"/> Headache | <input type="checkbox"/> Local side effects (rash, swelling...) | |
| <input type="checkbox"/> Renal failure | <input type="checkbox"/> Venous thrombosis | <input type="checkbox"/> Arterial thrombosis |
| <input type="checkbox"/> 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!
