



Note: << text >> gives information on technical details and conditions for automatic checks. It will not be visible for users

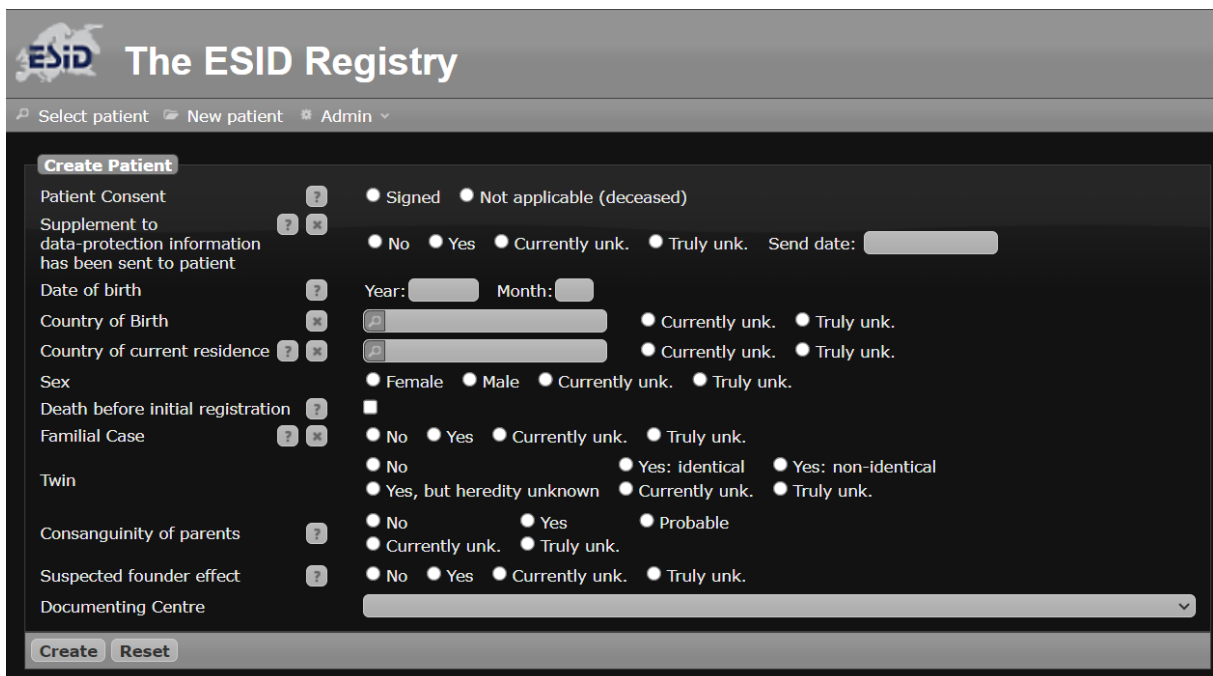
General notice: A value **must** be entered/selected for every field. If the information is not known (or currently not reachable), select one of the “**unknown**” options. Otherwise, the form cannot be stored.

Patient

<i>Field</i>	<i>Manual</i>
Date of registration: [Date] → Initial Documentation (Doc.) date	<<This date is automatically stored by the system (based on the server clock)>>
Patient consent: <input type="checkbox"/> signed <input type="checkbox"/> not applicable (deceased)	Patients have to give their written consent before you enter data. For minors, parents or the legal guardian have to give their written consent. Indicate what kind of consent the patient has given. “Not applicable” can only be selected for patients who have died before initial registration. Please check with your local data protection laws whether you can report deceased patients without consent.
Supplement to data-protection information has been sent to the patient: <input type="checkbox"/> yes/no/unknown plus date (depricated)	If for some reason a previous consent version has been signed, it has to be amended by a supplement, reflecting the GDPR. For newly registered patients a current GDPR-compliant consent version must be used.
Date of birth [Year] [Month]	If patient < 12 years old: Enter year AND month If patient >= 12 years old: Enter ONLY the year This restriction is necessary because of data protection regulations. << The month will remain stored when the child turns 12 years old >>
Country of birth: [List of countries, unknown]	
Country of current residence: [List of countries, unknown]	Select the country of current residence for this patient. This should be the country where the patient has his permanent residence, i.e. where he lives for the majority of the year. If the patient stays in the current country for a longer period, but only temporarily (e.g. for specialized medical treatment or seasonal work , select his country of origin.
Sex: <input type="checkbox"/> female <input type="checkbox"/> male <input type="checkbox"/> unknown	Select the genetic sex at birth
Death before initial registration: <input type="checkbox"/> (check box)	In general, only living patients should be (newly) reported. However, you can report deceased patients if a) your centre or national registry has a specific policy or runs a specific study that makes this necessary b) if the patient died shortly before registration. <<If selected, the “Death report form” will open. Centres will not be asked to update deceased patients afterwards >>
Familial case: <input type="checkbox"/> yes <input type="checkbox"/> no <input type="checkbox"/> unknown	Indicate if there is another patient with a diagnosed primary immunodeficiency in the genetic family (e.g. parents, siblings, grandparents). If there was no clear diagnosis of a PID, select “unknown”.

If yes, index patient ESiD ID: _____ <input type="checkbox"/> This is the index patient <input type="checkbox"/> more than one index patient	<<only visible if “familial case” is “yes”>> If available, enter the ESiD patient ID of the first patient diagnosed with this immunodeficiency in the respective
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	family (index case) If the current patient is this index case, select the checkbox "This is the index patient". Select "more than one index patient" e.g. if the patient forms the "missing link" between two index patients.
Index patient is the patient's: <<Drop down: grandson/daughter, son/daughter, niece/nephew, brother/sister, half-brother/sister, cousin, wife/ husband, father/mother, uncle/aunt, grandfather/-mother, granduncle/-aunt, great-grandfather/-mother, other, unknown >>	<<only visible if index patient ESiD ID is entered>> Indicate the relationship of the current patient to the index patient. <<Family branch only appears for options from "uncle/aunt" to "other">> Indicate which side of the family the index patient belongs to (paternal = father's side, maternal = mother's side)
Additional index patient ESiD ID:	<<only visible if "more than one index patient" has been selected>>
Twin: <input type="checkbox"/> no <input type="checkbox"/> yes: identical <input type="checkbox"/> yes: non-identical <input type="checkbox"/> yes, but heredity unknown <input type="checkbox"/> unknown	
Consanguinity of parents: <input type="checkbox"/> yes <input type="checkbox"/> no <input type="checkbox"/> unknown <input type="checkbox"/> probable	Indicate whether the parents or other ancestors (e.g. grandparents) of the patient are genetically related.
Suspected founder effect: <input type="checkbox"/> yes <input type="checkbox"/> no <input type="checkbox"/> unknown	<<Founder effect: If a population arises only from a small set of individuals, their genetic variability is limited and might inherit more commonly genetic defects if they were present in the 'founding' individuals. When ancestors of a patient come from the same small genetically isolated region a founder effect can be suspected.>>



The screenshot shows the 'Create Patient' form in the ESiD Registry. The form is organized into a grid with labels on the left and input fields on the right. The labels include 'Patient Consent', 'Supplement to data-protection information has been sent to patient', 'Date of birth', 'Country of Birth', 'Country of current residence', 'Sex', 'Death before initial registration', 'Familial Case', 'Twin', 'Consanguinity of parents', 'Suspected founder effect', and 'Documenting Centre'. The input fields consist of radio buttons for many options (e.g., 'Signed', 'Not applicable (deceased)', 'No', 'Yes', 'Currently unk.', 'Truly unk.', 'Female', 'Male', 'Yes: identical', 'Yes: non-identical'), dropdown menus for dates and countries, and a 'Documenting Centre' dropdown menu. At the bottom of the form, there are 'Create' and 'Reset' buttons.

Fig 1. Initial registration of a patient, User interface, centre is automatically selected/set for standard user

Consent

Consent version: : [List of consent versions applicable for the given centre]	Available versions for a given centre are selected in the backend via the admins, if consent is renewed a list will build up with all previous consents.
Research option: <input type="checkbox"/> yes <input type="checkbox"/> no	As selected in the patient consent form
Pharma option: <input type="checkbox"/> yes <input type="checkbox"/> no	As selected in the patient consent form
Non-EU option: <input type="checkbox"/> yes <input type="checkbox"/> no	As selected in the patient consent form
Date of signature: [Date]	Enter the date of the patient's signature. {calendar function implemented}

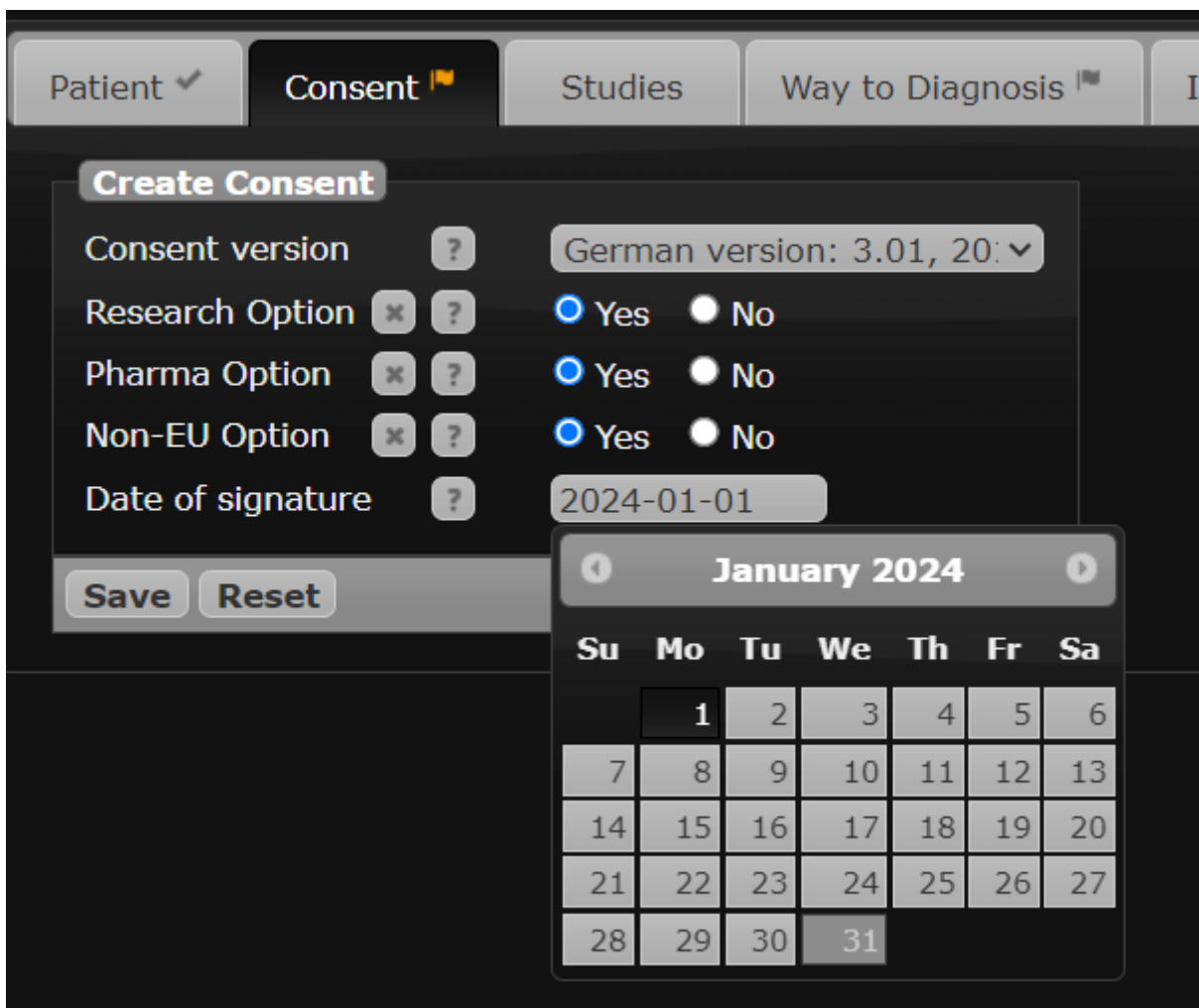


Fig 2-1: Documentation of (new) consents



Fig 2-2: Representation of (list) of consents (history of consents) and options

Studies

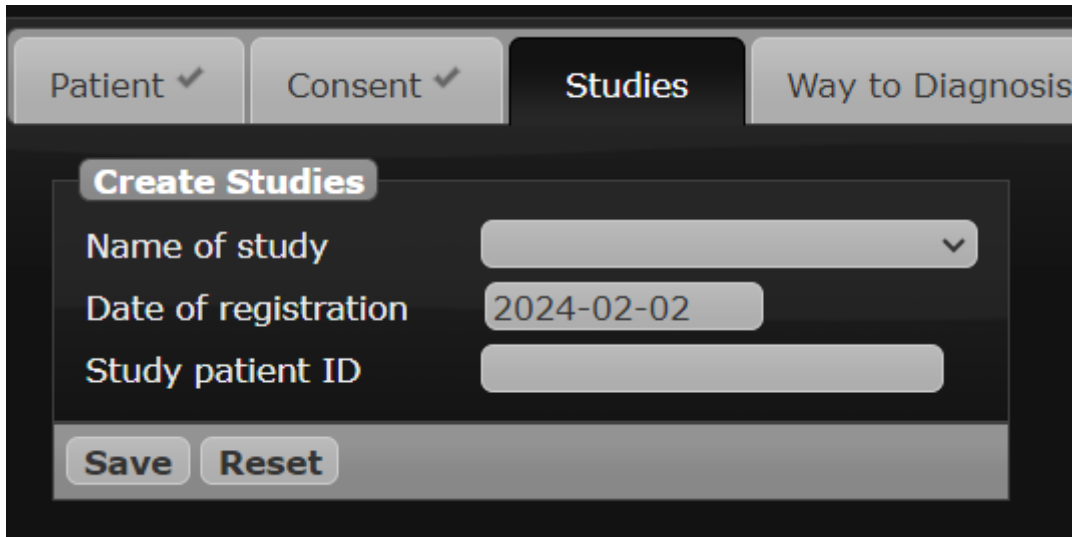


Fig 3: Enter the study details here. The study ID is generated by the system, or provided by a registration office (depends on study)

Way to diagnosis

<p>Date of first clinical diagnosis of IEI [Year] [Month] [Day] <input type="checkbox"/> unknown <input type="checkbox"/> Only genetically diagnosed</p>	<p><<Cannot be previous to date of birth>> Enter the date when this patient was first diagnosed with a primary immunodeficiency based on clinical features and laboratory values. If month and/or day are unknown, leave them open. If the date is completely unknown, select “date unknown”. If the patient has been given a genetic diagnosis before developing any clinical symptoms, select “only genetically diagnosed”.</p>
<p>First IEI-related symptom(s): <<one or several of>> <input type="checkbox"/> Infection <input type="checkbox"/> Immune dysregulation <input type="checkbox"/> Malignancy <input type="checkbox"/> Syndromal manifestations <input type="checkbox"/> other (if selected) Description: _____ <<or>> <input type="checkbox"/> First symptoms unknown <<or>> <input type="checkbox"/> no IEI-related symptoms at all → If “no IEI related symptoms” is selected the field “Diagnosis by lab abnormalities only” will appear, and “Date/Age of onset of symptoms” will disappear</p>	<p>Indicate the first clinical symptoms suggestive of a IEI in this patient (which is not necessarily the symptom leading to diagnosis). Select one or several. If the type of symptoms is unknown, select 'First symptoms unknown'. If there were no IEI-related symptoms at all, select 'no IEI-related symptoms'. Enter the information based on the physician's judgement (as noted in the patient chart) and not based on the patient's opinion. Definitions: Immune dysregulation: lymphoproliferation (splenomegaly, hepatomegaly, lymphadenopathy), granuloma, autoimmunity (e.g. cytopenia, thyroid disease, joint disease, hepatitis, vitiligo, alopecia, diabetes), inflammatory bowel disease, celiac disease, vasculitis, eczema, autoinflammatory disease Syndromal: 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</p>



<p>Date/Age of onset of symptoms [Year] [Month] or approximate age in years: [list with <1 , 1-5 , 6-10 , 11-15 , 16-20 , 21-25 , 26-30 etc.(in steps of 5), ..., 51-100] or <input type="checkbox"/> unknown</p>	<p><<Will not be visible if “no symptoms” has been selected. Cannot be later than genetic and clinical diagnosis. Exception: If later than one of these, the form can only be stored if “yes” is selected in the next field “absence of symptoms” >> Enter the year and month when the first symptoms suggestive of a IEI (see above) appeared in this patient. If the month is unknown, leave it open. If you only know the date approximately, select an entry from the list. Enter the date based on the physician's judgement (as noted in the patient chart) and not based on the patient's opinion. If only the frequency of infections raises the suspicion of a primary immunodeficiency, select the date when the suspicion came up, analogous to what is outlined in the chapter above related to first symptom. E.g. if only the third otitis media within a year raised the suspicion, please document the date of that third otitis. <<only one of [date, list, unknown] can be selected >></p>
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<p>Diagnosis by lab abnormalities only? <input type="checkbox"/> yes <input type="checkbox"/> no <input type="checkbox"/> unknown</p>	<p>Examples for this are: (1) The patient's blood was investigated due to non-PID related symptoms (2) The patient's blood was screened due to familial cases of PID <<Will only be visible if “no symptoms” has been selected OR if date of clinical diagnosis is prior to date of onset of symptoms. Symptoms and date of onset can also be entered in the latter case.>></p>
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<p>If yes, “Type of lab abnormalities” will appear: <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"/> Newborn screening <input type="checkbox"/> Hypogammaglobulinaemia <input type="checkbox"/> Hypergammaglobulinaemia (other than elevated IgE) <input type="checkbox"/> elevated complement <input type="checkbox"/> complement deficiency <input type="checkbox"/> Other, (if selected) Description:_____</p>	<p><<Will only be visible if the previous question has been marked “yes”>> Select one or several. If there was another kind of lab abnormality leading to the diagnosis of PID not given here, select “other” and description. Definition for hypogammaglobulinemia: values for IgA, G or M below age-related normal range.</p>
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The screenshot shows the 'Way to Diagnosis' form with the following fields and options:

- Date of first clinical diagnosis of IEI:** Year, Month, Day fields and radio buttons for 'Currently unk.', 'Truly unk.', and 'Only genetically diagnosed'.
- First IEI-related symptom(s):** A list of checkboxes including 'Infection', 'Immune dysregulation', 'Malignancy', 'Syndromal manifestations', 'Other', 'First symptoms currently unk.', and 'First symptoms truly unk.'. A 'Description:' field is also present.
- No IEI-related symptoms at all:** A checkbox.
- Date of onset of symptoms:** Year and Month fields.
- Approximate age of onset:** A dropdown menu with options: 'First year of life', '1 to 5 years', '6 to 10 years', '11 to 15 years', '16 to 20 years', '21 to 25 years', '26 to 30 years', '31 to 35 years', '36 to 40 years', '41 to 45 years', '46 to 50 years', and '51 to 100 years'.
- Age of onset unknown:** A checkbox.
- Buttons:** 'Save' and 'Reset'.

Fig 4-1: Way to diagnosis with symptoms

The screenshot shows the 'Way to Diagnosis' form with the following fields and options:

- Date of first clinical diagnosis of IEI:** Year, Month, Day fields and radio buttons for 'Currently unk.', 'Truly unk.', and 'Only genetically diagnosed'.
- No IEI-related symptoms at all:** A checked checkbox.
- Diagnosis by lab abnormalities only:** Radio buttons for 'Yes', 'No', 'Currently unk.', and 'Truly unk.'.
- Type of lab abnormalities:** A list of checkboxes including 'Lymphopenia', 'Neutropenia', 'Thrombocytopenia', 'Anaemia', 'Monocytopenia', 'Elevated IgE', 'Newborn screening', 'Hypogammaglobulinemia', 'Hypergammaglobulinemia (other than elevated IgE)', 'elevated complement', 'complement deficiency', and 'Other lab abnormalities'. A 'Description:' field is also present.
- Buttons:** 'Save' and 'Reset'.

Fig 4-2: Way to diagnosis with lab abnormalities only

IEI Diagnosis

<p>Current IEI diagnosis</p>	<p><<Users can select a IEI diagnosis by entering a search string either for the IEI diagnosis or the available genes. The entry is stored permanently and can only be replaced by a new entry at a later documentation timepoint>></p> <p>Select the most recent IEI diagnosis for this patient (corresponding to the most recent visit date or date of last news). If you cannot find the appropriate disease, or if a disease is missing, please send an email to registry@esid.org</p>
<p>Affected gene: [List of genes] <<or>></p> <p><input type="checkbox"/> Genetically tested, but no mutation found</p> <p><input type="checkbox"/> Not genetically tested</p> <p><input type="checkbox"/> Results pending</p> <p><input type="checkbox"/> History of genetic tests unknown</p>	<p><<Relevant genes appear as a list according to the selected diagnosis; in addition, “no mutation found” and “not genetically tested” (where appropriate). Note that there is no “unknown” option! >></p> <p>Select the gene in which disease-causing mutation(s) have been found in this patient. If you have sequenced one or more of the known genes but have found no mutation, select “no mutation found”. If no molecular analysis has been performed at all, select “not genetically tested”.</p> <p>If a gene is missing, send an email to registry@esid.org</p>
<p>Additional genes: _____</p>	<p>If more than one IEI-causing gene mutation has been found in this patient, or if other gene mutations have been found, you can enter these here.</p>
<p>Date of genetic diagnosis: [Year] [Month] [Day] <<or>></p> <p><input type="checkbox"/> unknown</p>	<p><<Will only be visible if an affected gene has been selected. Cannot be previous to date of birth. Exception: If previous to date of birth less than 9 months, form can only be stored if “Prenatal diagnosis” in the next field is selected. >></p> <p>If applicable, enter the date when the genetic diagnosis was confirmed (date of molecular analysis). If month and/or day are unknown, leave them open. If the date is completely unknown, select “Date unknown”</p>
<p>Lab that performed the genetic analysis: <<Drop down list>></p>	<p><<Only visible if a gene or “no mutation found” has been selected >></p> <p>If the lab that performed the analysis is not in the list, please send an email to esid-registry@uniklinik-freiburg.de</p>
<p>Sequencing method:</p> <p><input type="checkbox"/> Gene sequencing</p> <p><input type="checkbox"/> Whole exome/genome sequencing</p> <p><input type="checkbox"/> Non-genetic definitive test</p> <p><input type="checkbox"/> Unknown</p>	<p><<Will only be visible if an affected gene has been selected>> Select the sequencing method applied. If the molecular analysis was performed as a candidate gene testing (using the “traditional” method of Sanger sequencing), select “Gene sequencing”. If the whole genome or exome was sequenced, select that option. If a non-genetic test like 22q11 FISH for DiGeorge syndrome was used, select ‘Non-genetic definitive test’.</p>
<p>Reason for genetic analysis:</p> <p><input type="checkbox"/> Analysis following clinical diagnosis</p> <p><input type="checkbox"/> Family screening</p> <p><input type="checkbox"/> Prenatal diagnosis</p> <p><input type="checkbox"/> Diagnosis by neonatal screening</p> <p><input type="checkbox"/> unknown</p>	<p><<Will only be visible if an affected gene has been selected. Only one can be selected.>></p> <p>Select the reason for the molecular analysis in this patient.</p>

<p>Optional (depending on selected diagnosis) Do you want to add L2 documentation? <input type="checkbox"/> yes <input type="checkbox"/> no <input type="checkbox"/> Ask again later</p>	<p><<Only visible if a respective diagnosis has been selected. On “Yes” additional Level 2 unPAD forms will be presented. On “No”, you won’t be asked again.>> If you need to change the decision, please contact: esid-registry@uniklinik-freiburg.de</p>
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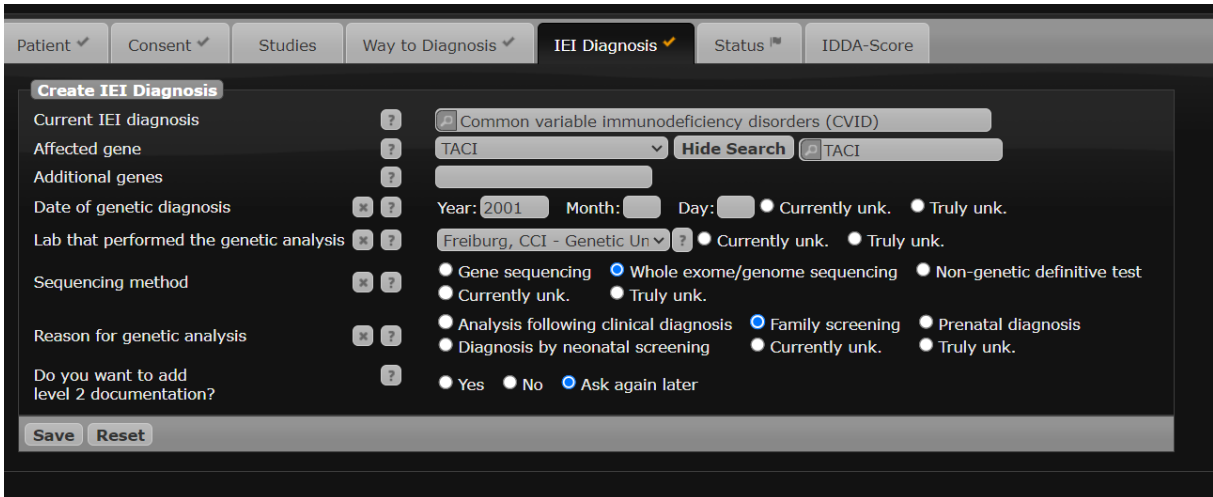


Fig 5; IEI diagnosis

Status:

This data shall be entered for the initial documentation date and updated on every subsequent documentation date (slightly different set of items for baseline/follow-up). *Depending on the selected options, additional TABs may appear (please see below).*

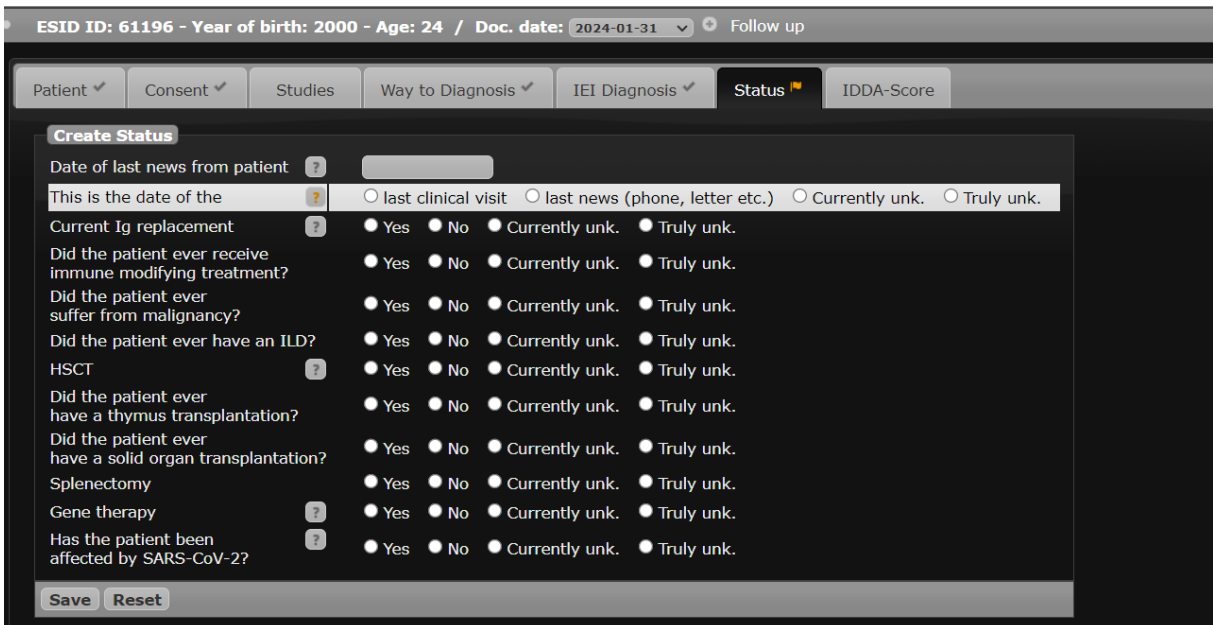
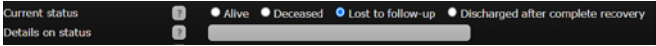


Fig 6-1: Status overview at **initial** documentation (first Doc. date), for more details on the items please refer to the next section on Follow-up.

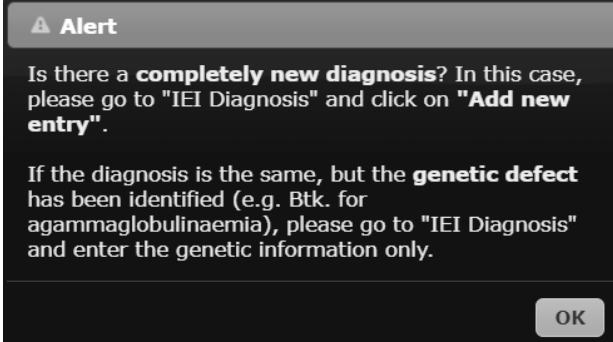
Follow-up


<p>Date of last news from patient: [Date] <<or>> <input type="checkbox"/> No news from patient since last documentation</p>	<p><< Can only store a date that comes after the most recent documentation date. >> Enter the date when you last received news on the patient. If you have no news at all from the patient, please select 'No news from patient since last documentation'.</p>
<p>This is the date of the: <input type="checkbox"/> last clinical visit <input type="checkbox"/> last news (phone, letter etc.) <input type="checkbox"/> unknown</p>	<p>Indicate whether this is the date of the last patient visit to the physician or hospital, or whether information was received e.g. by telephone from the patient, the patient's family or a physician.</p>

All following questions refer to the **timepoint of the patient's last visit or date of last news**.

<p>Current status: <input type="checkbox"/> Alive <input type="checkbox"/> Deceased <input type="checkbox"/> Lost to follow-up <input type="checkbox"/> Discharged after complete recovery</p>	<p>Indicate whether the patient is still alive based on the latest information you have. If you have no information on the current status of the patient because you have lost contact to him/her and do not expect to hear from him/her again, select "lost to follow-up". If the patient is not followed anymore because the immunodeficiency has resolved, select 'discharged after complete recovery'. << If "deceased" is selected, the "Death report form" (TAB) will open. Please refer to Annex A. Centres will not be asked to update patients with status "deceased", "lost to follow-up" and "Discharged" afterwards. For patients with status 'alive' the system will create an e-mail reminder for F/U documentation one year after the last Doc. date >></p>
<p>Details on status: _____ [free text]</p>	<p><<optional. Appears if "Lost to follow-up" is selected>> If the patient is lost to follow-up, you may enter details here (e.g. name of physician the patient was referred to)</p> 

Changes to diagnosis

<p>Changes to diagnosis: <input type="checkbox"/> No change << default value >> <<or>> <input type="checkbox"/> PID diagnosis has changed <<or>> <input type="checkbox"/> No PID after all</p>	<p>If the IEI diagnosis has changed or if you have identified the affected gene, select "PID diagnosis has changed" and fill in the respective fields in TAB "IEI diagnosis:"</p>  <p>If the diagnosis of an IEI has been incorrect and there is no IEI in this patient at all, please select "No PID after all".</p>
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<p>New IEI diagnosis << new entry >></p>	<p><<if “PID diagnosis has changed” has been selected. If the affected gene has been identified, this is stored with the existing entry. Otherwise, the system creates a completely new PID Diagnosis entry (thereby creating a “diagnostic history”)>></p> <p>Select the most recent PID diagnosis for this patient (corresponding to the most recent visit date or date of last news). If you cannot find the appropriate disease, or if a disease is missing, please send an email to registry@esid.org</p> 
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ESID ID: 61196 - Diagnosis: CVID - Year of birth: 2000 - Age: 24 / Doc. date: 2024-02-02

Patient ✓ Consent ✓ Studies Way to Diagnosis ✓ IEI Diagnosis ✓ Status ■ IDDA-Score

Create Status

Date of last news from patient ? No news from patient since last documentation

This is the date of the ? last clinical visit last news (phone, letter etc.) Currently unk. Truly unk.

Current status ? Alive Deceased Lost to follow-up Discharged after complete recovery

Changes to diagnosis ? No Change PID diagnosis has changed No PID after all

Current Ig replacement ? Yes No Currently unk. Truly unk.

Did the patient ever receive immune modifying treatment? Yes No Currently unk. Truly unk.

Did the patient ever suffer from malignancy? Yes No Currently unk. Truly unk.

Did the patient ever have an ILD? Yes No Currently unk. Truly unk.

HSCT ? Yes No Currently unk. Truly unk.

Did the patient ever have a thymus transplantation? Yes No Currently unk. Truly unk.

Did the patient ever have a solid organ transplantation? Yes No Currently unk. Truly unk.

Splenectomy Yes No Currently unk. Truly unk.

Gene therapy ? Yes No Currently unk. Truly unk.

Has the patient been affected by SARS-CoV-2? ? Yes No Currently unk. Truly unk.

Fig 6-2: Status at **follow-up** documentation (please note additional items: “Current status” and “Changes to diagnosis”, prefilled with default values where possible). If there are no further changes to previously documented items, the follow-up is completed with updating this.

Immunoglobulin (Ig) replacement, *additional TAB*

<p>Does the patient currently receive Ig-replacement? <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> unknown</p>	<p>Current Ig replacement ? <input checked="" type="radio"/> Yes <input type="radio"/> No <input type="radio"/> Currently unk. <input type="radio"/> Truly unk.</p>
<p>Date of first Ig replacement</p>	<p><< Cannot be previous to the date of birth>></p>

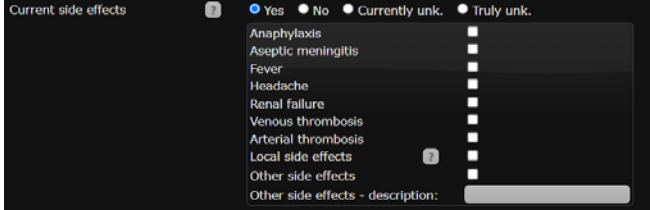
<p>[Year] [Month] [Day] <<or>> <input type="checkbox"/> Date unknown</p>	<p>Give the date when Ig replacement was first applied in this patient. If month and/or day are unknown, leave them open. If the date is completely unknown, select "Date unknown".</p>
<p>Current brand name [List of brand names] <<or>> <input type="checkbox"/> unknown</p>	<p>Which brand of immunoglobulins does the patient currently use?</p>
<p>Current route of administration <input type="checkbox"/> Subcutaneous <input type="checkbox"/> Intravenous <input type="checkbox"/> Intramuscular <input type="checkbox"/> Push</p>	<p><<Route is automatically selected by the system if a respective brand name is selected>></p>
<p>Current place of administration <input type="checkbox"/> home <input type="checkbox"/> hospital <input type="checkbox"/> hospital: inpatient <input type="checkbox"/> hospital: outpatient <input type="checkbox"/> both (home&hospital) <input type="checkbox"/> Office outpatient <input type="checkbox"/> unknown</p>	<p>Select the place of administration of Ig replacement. If the infusion is given in a hospital, indicate whether this is done during an inpatient or outpatient stay. If this is not known, choose the option "hospital".</p>
<p>Patient's current weight [integer] kg <input type="checkbox"/> unknown</p>	<p><< If brand name, weight, and absolute dose are entered, the system will calculate and store the mg/kg dose. If the mg/kg is entered manually in addition and differs from the calculated value, the system asks the user which value should be used.>></p>
<p>Current dose [integer] mg/kg body weight or [decimal] <input type="checkbox"/> g <input type="checkbox"/> ml <input type="checkbox"/> dose unknown</p> <p>Interval for this dose Every [integer] <input type="checkbox"/> week(s) <input type="checkbox"/> day(s) <<or>> [integer] times per <input type="checkbox"/> week <input type="checkbox"/> month <input type="checkbox"/> year <input type="checkbox"/> interval unknown</p>	<p>Indicate the current dose and frequency of Ig replacement in this patient. Enter the relative dose (per kg body weight) or the absolute dose, or both, if available.</p> <p>Enter the actual interval, e.g. if the patient applies 10 ml of SCIg every second day, enter "10 ml every 2 days", and NOT "35 ml every week".</p> <p>In the case of alternating doses (e.g. "10 ml one week and 20 ml the next week"), calculate the mean value (e.g. "15 ml every week").</p>
<p>Current side effects <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> unknown</p>	<p>Indicate whether the administration of the current Ig replacement has ever caused or is still causing side effects.</p>
<p>If yes, type of side effects <<one or several possible>> <input type="checkbox"/> Anaphylaxis <input type="checkbox"/> Aseptic meningitis <input type="checkbox"/> Fever <input type="checkbox"/> Headache <input type="checkbox"/> Renal failure <input type="checkbox"/> Venous thrombosis <input type="checkbox"/> Arterial thrombosis <input type="checkbox"/> Local side effects <input type="checkbox"/> Other, specify: _____</p>	<p><<Only visible if "side effects" has been answered with "yes">></p> 

Fig 6-3: Optional Ig-Replacement TAB

Immune Modifying Treatment, additional fields inline

Fig 6-4: Options for immune modifying treatment can be selected by clicking on the respective item in 'available options' and will then be moved to the list of selected options

Malignancy, additional fields inline



Fig 6-5: List of Malignancies

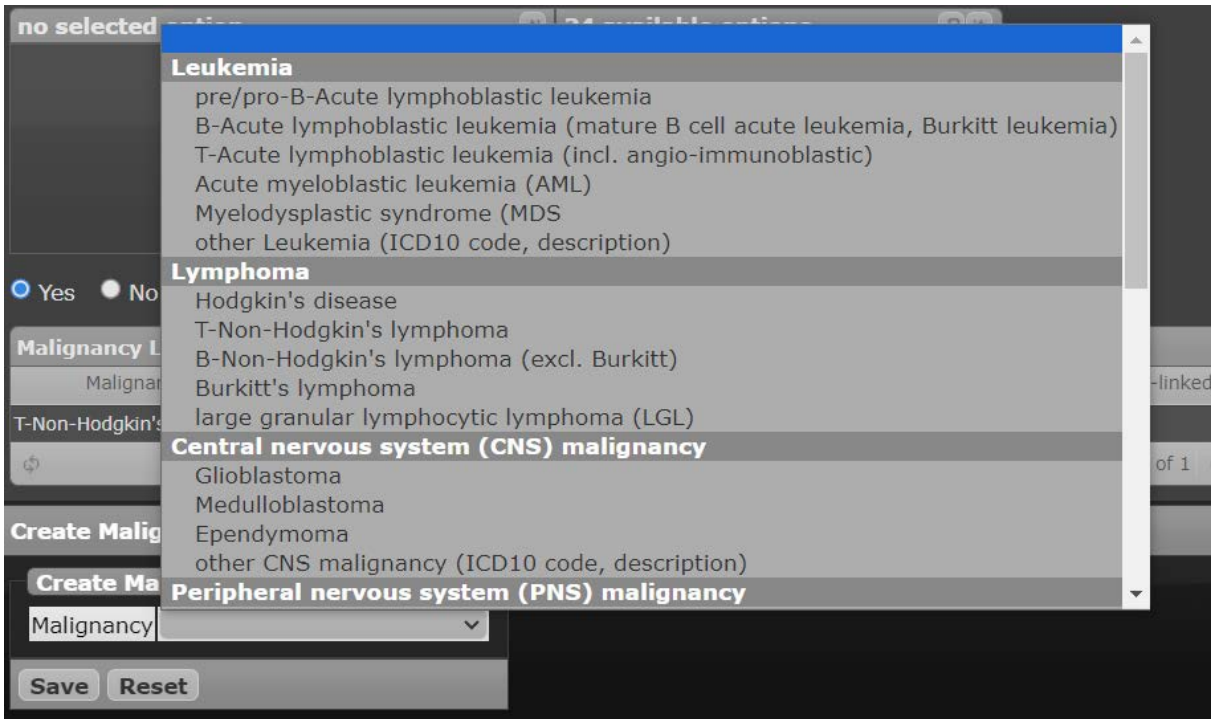


Fig 6-6: Dialogue to add more malignancies from a hierarchically ordered list or by providing ICD10 code and -text

ILD, additional TAB, not shown deprecated/under revision

HSCT, additional fields inline for SCETIDE ID and EMBT ID, Additional Tab for more detailed information on HSCT

<p>Haematopoietic stem cell transplantation (HSCT): <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Unknown</p>	<p>Indicate whether haematopoietic stem cell transplantation (HSCT) has ever been performed in this patient.</p> <p>HSCT <input type="radio"/> Yes <input checked="" type="radio"/> No <input type="radio"/> Currently unk. <input type="radio"/> Truly unk.</p>
<p>If yes, SCETIDE ID: EBMT ID:</p>	<p>If the patient has been reported to SCETIDE, provide the ID number. SCETIDE is the registry for Stem Cell Transplants for primary Immune Deficiencies in Europe, in collaboration with ESID. Contact info.scetide@nck.aphp.fr for more information. If the patient has been reported to the EBMT Registry, provide the ID number.</p>
<p>If yes, enter for each transplantation:</p>	<p><<several entries possible>></p>
<p>Date of transplantation [Year] [Month] [Day] <input type="checkbox"/> Date unknown</p>	<p><<Cannot be previous to the date of birth. >></p>

<p>Type of donor</p> <input type="checkbox"/> MSD (Matched sibling donor) <input type="checkbox"/> MUD (Matched unrelated donor) <input type="checkbox"/> MMUD (Mismatched unrelated donor) <input type="checkbox"/> Haplo-identical (parent) donor <input type="checkbox"/> Autologous <input type="checkbox"/> Other related donor <input type="checkbox"/> Unknown	<p>Indicate the type of stem cell donor.</p>
<p>Source of CD34 stem cells <<one or several of>></p> <input type="checkbox"/> bone marrow <input type="checkbox"/> peripheral blood <input type="checkbox"/> cord blood <input type="checkbox"/> fetal liver <input type="checkbox"/> unknown	<p>Indicate the source of the stem cells used in this HSCT.</p>



Fig 6-7: List of haematopoietic stem cell transplantations (HSCT) and dialogue window for entering new/additional HSCT procedures

Thymus transplant, additional fields inline

Did the patient ever have a thymus transplantation? Yes No Currently unk. Truly unk.

Date of thymus transplantation Year: Month: Day: Currently unk. Truly unk.

Type cryopreserved fresh Currently unk. Truly unk.

Fig 6-8: Fields for thymus transplantation

Solid organ transplantation, additional fields inline

Did the patient ever have a solid organ transplantation? Yes No Currently unk. Truly unk.

Date of first solid organ transplantation Year: Month: Day: Currently unk. Truly unk.

Organ(s) of first SOT

Fig 6-9: Fields for Solid organ transplantation, List of organs freetext

Splenectomy, additional field inline

Splenectomy Yes No Currently unk. Truly unk.

Date of Splenectomy Year: Month: Day: Currently unk. Truly unk.

Fig 6-10: Fields for Splenectomy

Gene therapy, additional TAB

<p>Gene therapy</p> <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Unknown	<p>Indicate whether gene therapy has ever been performed in this patient.</p> <p>Gene therapy <input type="text"/> <input checked="" type="radio"/> Yes <input type="radio"/> No <input type="radio"/> Currently unk. <input type="radio"/> Truly unk.</p>
<p>If yes, enter for each gene therapy:</p>	<p><<several entries possible>></p>
<p>Date of gene therapy</p>	<p><<Cannot be previous to the date of birth.>></p>

[Year] [Month] [Day] <input type="checkbox"/> Date unknown	Enter the date when the gene therapy was initiated.
---	---



Fig 6-11: List of gene therapies and dialogue window for entering new/additional procedures

IDDA-Score: TAB is always visible, documentation optional
 IDDA (immune deficiency and dysregulation activity) score, Version 2.1 as of July 2020

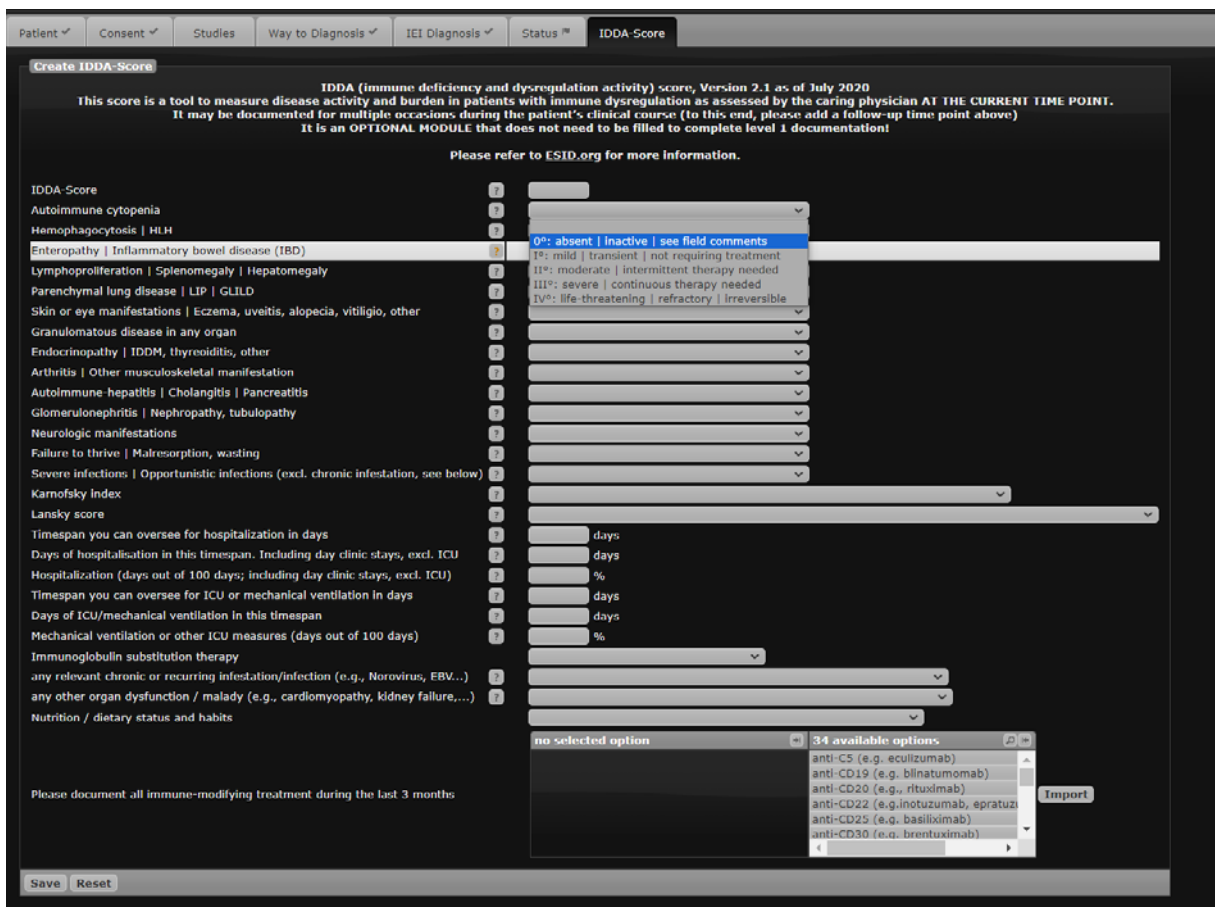


Fig 7: IDDA-Score, only for selected patients, IDDA-score may be optionally, additionally documented where suitable on documenting centres discretion.



ANNEX A Death report form:

Death report form, additional TAB

Current status ? Alive Deceased Lost to follow-up Discharged after complete recovery

<p>Date of death: [Year] [Month] [Day] <input type="checkbox"/> Date of death unknown</p>	<p><< The date of death must not be previous to the date of birth>> If day and/or month are unknown, leave these fields empty. If the date is completely unknown, select "Date of death unknown"</p>
<p>Main cause(s) leading to death: <input type="checkbox"/> Septic shock <input type="checkbox"/> Heart failure <input type="checkbox"/> Respiratory failure <input type="checkbox"/> Liver failure <input type="checkbox"/> Renal failure <input type="checkbox"/> Multiple organ failure <input type="checkbox"/> Haemorrhage <input type="checkbox"/> Thrombosis <input type="checkbox"/> Neurological complications <input type="checkbox"/> Surgical complications <input type="checkbox"/> Drug toxicity <input type="checkbox"/> Relapse of malignancy <input type="checkbox"/> Veno-occlusive disorder <input type="checkbox"/> Graft-versus-host Disease <input type="checkbox"/> Rejection/Poor graft function <input type="checkbox"/> Post-transplant lymphoproliferative disorder <input type="checkbox"/> Other - specify: _____ <input type="checkbox"/> Unknown</p>	<p>Indicate the main cause(s) of death (several can be selected).</p>
<p>Underlying morbid setting(s) associated with mortality: <input type="checkbox"/> Infection, specify: _____ <input type="checkbox"/> Malignancy, specify: _____ <input type="checkbox"/> Immune dysregulation, specify: _____ <input type="checkbox"/> Transplantation-related cause <input type="checkbox"/> Other - specify: _____ <input type="checkbox"/> Unknown</p>	<p>Indicate the general clinical context leading to occurrence of the death event (several can be selected). Definition of immune dysregulation: lymphoproliferation, (splenomegaly, hepatomegaly, lymphadenopathy), granuloma, autoimmunity (e.g. cytopenia, thyroid disease, joint disease, hepatitis, vitiligo, alopecia, diabetes), inflammatory bowel disease, celiac disease, vasculitis, eczema, autoinflammatory disease</p>
<p>ICD10 Categorisation <<several can be entered>> Code: Text:</p>	<p><<optional fields, entries are selected via searchable field from ICD10 dictionary>> Enter at least two characters in either the code or the text field. Matching entries are presented as a list that you can select from.</p>

Patient ✓ Consent ✓ Studies **Death info** Way to Diagnosis ✓ IEI Diagnosis ✓ Status ✓ Ig-Repl.

Create Death info

Date of death ?

Year Month Day

Currently unk. Truly unk.

Main cause(s) of death ?

<input type="checkbox"/> Septic shock	<input type="checkbox"/> Heart failure	<input type="checkbox"/> Respiratory failure	<input type="checkbox"/> Liver failure
<input type="checkbox"/> Renal failure	<input type="checkbox"/> Multiple organ failure	<input type="checkbox"/> Haemorrhage	<input type="checkbox"/> Thrombosis
<input type="checkbox"/> Neurological complications	<input type="checkbox"/> Surgical complications	<input type="checkbox"/> Drug toxicity	
<input type="checkbox"/> Relapse of malignancy	<input type="checkbox"/> Veno-occlusive disorder		
<input type="checkbox"/> Graft-versus-host Disease	<input type="checkbox"/> Rejection/Poor graft function	<input type="checkbox"/> Post-transplant lymphoproliferative disorder	

Other main factor Description:

Main factor currently unk.

Main factor truly unk.

Underlying morbid setting(s) associated with mortality ?

<input checked="" type="checkbox"/> Infection	Description:	<input type="text"/>
<input checked="" type="checkbox"/> Malignancy	Description:	<input type="text"/>
<input checked="" type="checkbox"/> ? Immune dysregulation	Description:	<input type="text"/>
<input checked="" type="checkbox"/> Transplantation-related cause		
<input checked="" type="checkbox"/> Other underlying cause:	Description:	<input type="text"/>
<input type="checkbox"/> Underlying cause currently unk.		
<input type="checkbox"/> Underlying cause truly unk.		

ICD-10 categorisation (optional) ?

Code ?

Text ?

Fig 8: Death report form