ESID Registry Level 1 Dataset (work in progress)
Baseline form

Note: << text >> gives information on technical details and conditions for automatic checks. It will not be visible for users
This form shall be filled at the initial documentation of a patient (**registration in the system).**
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.

Baseline form / Patient

Field	*Manual*
Date of registration: [Date] ➔ **Documentation Date**	<<This date is automatically displayed and stored by the system (based on the server clock)>>
Patient consent: ☐ signed ☐ 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: ☐ **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 only 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: ☐ **female** ☐ **male** ☐ **unknown**	
Death before initial registration: ☐ **(check box)**	In general, only living patients should be 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: ☐ yes ☐ no ☐ 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: _____ ☐ This is the index patient ☐ 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

	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: ☐ no ☐ yes: identical ☐ yes: non-identical ☐ yes, but heredity unknown ☐ unknown	
Consanguinity of parents: ☐ **yes** ☐ **no** ☐ **unknown** ☐ **probable**	Indicate whether the parents or other ancestors (e.g. grandparents) of the patient are genetically related.
Suspected founder effect: ☐ **yes** ☐ **no** ☐ **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.>>

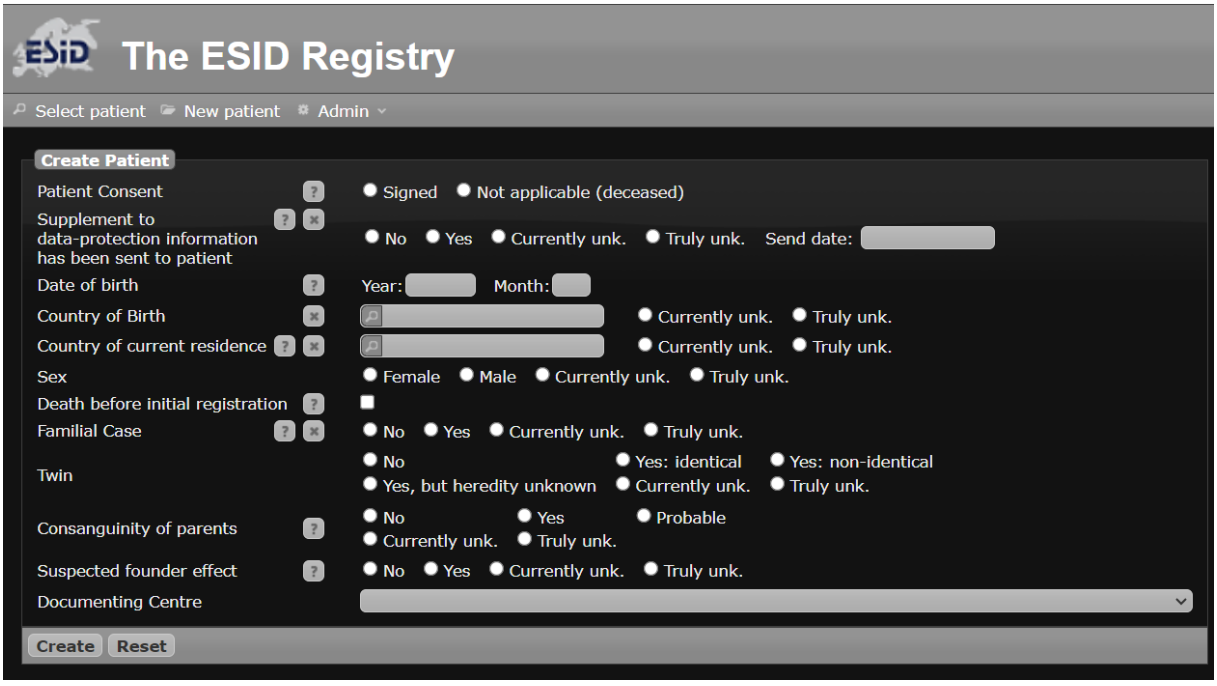

Fig 1. Initial registration of a patient, User interface, centre is automatically selected/set for standard user

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: ☐ **yes** ☐ **no**	As selected in the patient consent form
Pharma option: ☐ **yes** ☐ **no**	As selected in the patient consent form
Non-EU option: ☐ **yes** ☐ **no**	As selected in the patient consent form
Date of signature: [Date]	Enter the date of the patient's signature. {calendar function implemented}

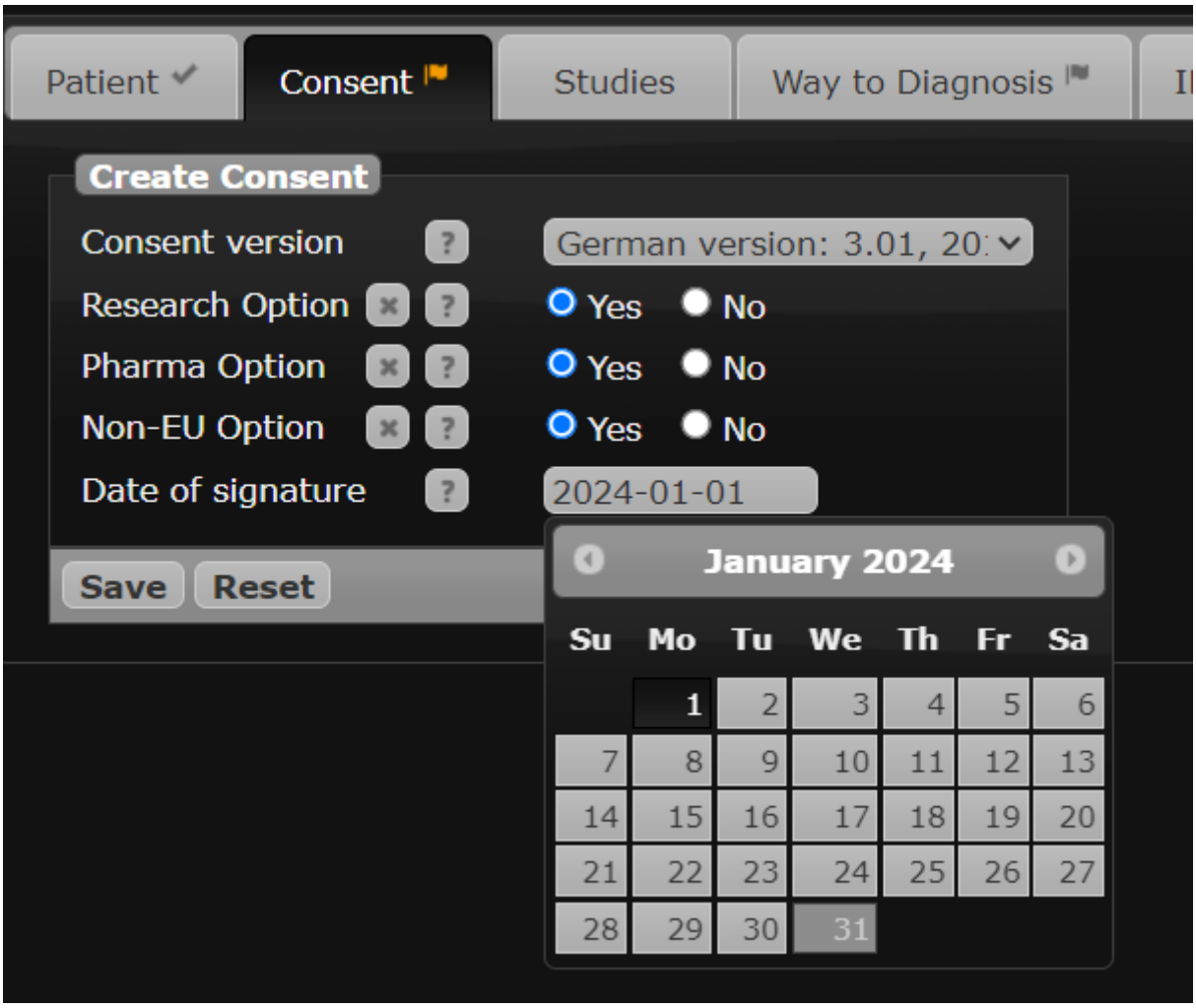

Fig 2: Documentation of (new) consents

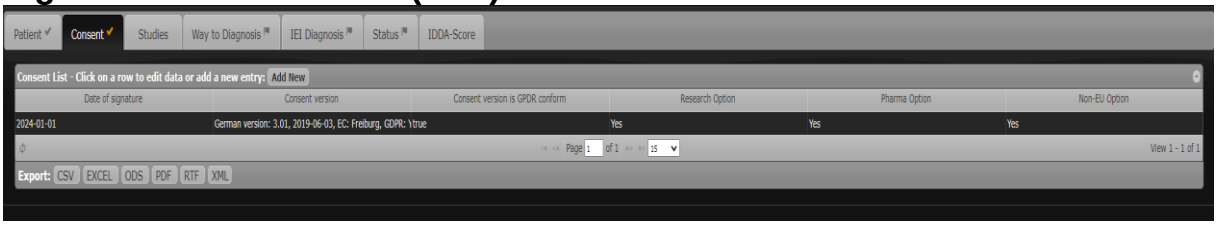

Fig 3: Representation of (list) of consents

Way to diagnosis

Date of first clinical diagnosis of PID [Year] [Month] [Day] ☐ date unknown ☐ only genetically diagnosed	<<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”.
First PID-related symptom(s): <<one or several of>> ☐ Infection ☐ Immune dysregulation ☐ Malignancy ☐ Syndromal manifestations ☐ other (indicate): _____ <<or>> ☐ First symptoms unknown <<or>> ☐ no PID-related symptoms	Indicate the first clinical symptoms suggestive of a PID 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 PID-related symptoms at all, select “no PID-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

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)] ☐ unknown	<<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 PID (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 both – date or list – can be selected >>
Was the patient diagnosed in the absence of PID-related symptoms on the basis of lab abnormalities? ☐ yes ☐ no ☐ unknown	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.>>
If yes, which of the following? ☐ Lymphopenia ☐ Neutropenia ☐ Thrombocytopenia ☐ Anaemia ☐ Monocytopenia ☐ Elevated IgE ☐ Hypogammaglobulinaemia ☐ Other, specify: _____	<<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". Definition for hypogammaglobulinemia: values for IgA, G or M below age-related normal range.

PID Diagnosis

Current PID Diagnosis	<<Users can select a PID Diagnosis by entering a search string either for the PID Diagnosis or the available genes. The entry is stored permanently and can only be replaced by a new entry at a later documentation timepoint.>> 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, send an email to esid.registry@kenes.com.
Affected gene: [List of genes] <<or>> ☐ Genetically tested, but no mutation found ☐ Not genetically tested ☐ History of genetic tests unknown	**<<**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! >> 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”. If a gene is missing, send an email to esid.registry@kenes.com.
Additional genes: _____	If more than one PID-causing gene mutation has been found in this patient, or if other gene mutations have been found, you can enter these here.
Date of genetic diagnosis: [Year] [Month] [Day] ☐ Date unknown	<<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. >> 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”
Sequencing method: ☐ Gene sequencing ☐ Whole exome/genome sequencing ☐ Non-genetic definitive test ☐ Unknown	<<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'.
Lab that performed the genetic analysis: <<Drop down list>>	<<Only visible if a gene or “no mutation found” has been selected >> If the lab that performed the analysis is not in the list, send an email to esid.registry@kenes
Reason for genetic analysis: ☐ Analysis following clinical diagnosis ☐ Family screening ☐ Prenatal diagnosis ☐ Diagnosis by neonatal screening ☐ unknown	<<Will only be visible if an affected gene has been selected. Only one can be selected.>> Select the reason for the molecular analysis in this patient.

Stem cell / gene therapy

Has one of the following ever been performed in this patient?	
Haematopoietic stem cell transplantation (HSCT): ☐ Yes ☐ No ☐ Unknown	Indicate whether haematopoietic stem cell transplantation (HSCT) has ever been performed in this patient.
If yes, SCETIDE ID: **EBMT ID:**	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.
If yes, enter for each transplantation:	<<several entries possible>>
Date of transplantation [Year] [Month] [Day] ☐ Date unknown	<<Cannot be previous to the date of birth. >>
Type of donor ☐ MSD (Matched sibling donor) ☐ MUD (Matched unrelated donor) ☐ MMUD (Mismatched unrelated donor) ☐ Haplo-identical (parent) donor ☐ Autologous ☐ Other related donor ☐ Unknown	Indicate the type of stem cell donor.
Source of CD34 stem cells <<one or several of>> ☐ bone marrow ☐ peripheral blood ☐ cord blood ☐ unknown	Indicate the source of the stem cells used in this HSCT.
Gene therapy ☐ Yes ☐ No ☐ Unknown	Indicate whether gene therapy has ever been performed in this patient.
If yes, enter for each gene therapy:	<<several entries possible>>
Date of gene therapy [Year] [Month] [Day] ☐ Date unknown	<<Cannot be previous to the date of birth.>> Enter the date when the gene therapy was initiated.

Immunoglobulin (Ig) replacement

Does the patient currently receive Ig-replacement? ☐ Yes ☐ No ☐ unknown	
Date of first application [Year] [Month] [Day] ☐ Date unknown	<< Cannot be previous to the date of birth>> 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".
Current brand name [List of brand names] ☐ unknown	Which brand of immunoglobulins does the patient currently use?
Current route of administration ☐ intravenous ☐ subcutaneous ☐ intramuscular	<<Route is automatically selected by the system if a respective brand name is selected>>
Current place of administration ☐ home ☐ hospital ☐ hospital: inpatient ☐ hospital: outpatient ☐ both (home&hospital) ☐ unknown	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".
Current dose [integer] mg/kg body weight or [decimal] ☐ g ☐ ml ☐ dose unknown **Interval for this dose** Every [integer] ☐ week(s) ☐ day(s) <<or>> [integer] times per ☐ week ☐ month ☐ year ☐ interval unknown	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. 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". 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").
Patient's current weight [integer] kg ☐ unknown	<< 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.>>
Current side effects ☐ Yes ☐ No ☐ unknown	Indicate whether the administration of the current Ig replacement has ever caused or is still causing side effects.
If yes, type of side effects <<one or several possible>> ☐ anaphylaxis ☐ fever ☐ headache ☐ local side effects ☐ renal failure ☐ aseptic meningitis ☐ venous thrombosis ☐ arterial thrombosis ☐ Other, specify: _____	<<Only visible if "side effects" has been answered with "yes">>