ERN-RND Registry: Data-Set Manual 2025

This manual gives you a short overview which data is needed for the ERN-RND registry and explains how to fill in the different columns in the registry template.

Please submit all patients of the relevant disease groups seen in your centre the previous year. Also those without a genetically determined diagnosis!

You can upload your document here:
https://uktcloud.medizin.uni-tuebingen.de/index.php/s/WDjitbZFhPcvpdB
Password: ERN-RND2024
The UKT cloud is in German, please find a translation at the end of this document.

If there are any questions please contact:
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1. Pseudonym: For data protection reasons the information has to be pseudonymised, coded with numbers, letters or a combination of both. You can use the SPIDER tool for that (https://eu-rd-platform.jrc.ec.europa.eu/spider/). **It is required that the patient has the same pseudonym the following years. Please use the same pseudonym like the years before if he/she has already been submitted once.**

2. Year of Birth: For confidentiality reasons the registry will restrict the information only to Year of birth. It is important not to detail the Date of birth. **Please only insert a four-digit number.**

3. Gender: This item is essential for the assessment of sex specific aspects of the diseases.
Choose one of the following codes in exactly the format printed in bold:
(i) female: **f**
(ii) male: **m**
(iii) unspecified: **u**
(iv) unknown: **unk**

4. Patient Status: To clarify the accessibility to patients, information is needed regarding the patients status.
Choose one of the following codes in exactly the format printed in bold:
(i) alive: **a**
(ii) dead: **d**
(iii) unknown: **unk**

5. Year of death: Fill in only if applicable.

6. Year of first contact with specialized centre: This data may be of interest to inform on availability of longitudinal data in retrospective. **Insert here the year as a four-digit number.**

7. Age of onset: This data is essential information in the course of the disease. **Insert here a one- or two-digit number.**

If not exactly known please set
(i) for infancy: 1
(ii) for childhood: an estimated value
(iii) for asymptomatic: asymp
(iv) for presymptomatic: p
(v) unknown: unk

8. Age at diagnosis: This data is of interest to assess delay in diagnosis.
Insert here a one- or two-digit number. If unknown: unk

9. Disease Group: There are six main disease groups in the ERN-RND.
Please choose one group from the dropdown list:

(i) Ataxia/HSP
(ii) Chorea/Huntington
(iii) Dystonia/NBIA/Paroxysmal movement disorders
(iv) FTD
(v) Leukodystrophy
(vi) Parkinsonism

10. Orphacode 1: Orphacode is an internationally accepted diagnostic standard for the specification of rare diseases. Here we collect the Orphacode of the disease groups.
Please use one of the following codes from the dropdown list:

(i) Ataxia: **102002**
(ii) HSP: **685**
(iii) Chorea: **306715**
(iv) Huntington: **399**
(v) Dystonia: **68363**
(vi) NBIAs: **385**
(vii) Paroxysmal movement disorders: **306768**
(viii) FTD: **282**
(ix) Leukodystrophy: **68356**
(x) MSA: **102**
(xi) Other PSP subtypes: **683**
(xii) CBD: **454887**
(xiii) Genetic Parkinson syndrome: **307052**
(xiv) without determined diagnosis after full investigation: **616874**

11. Orphacode 2: Here we collect the Orphacode on the level of groups of disorders. **This field is optional. You can use one of the following codes from the dropdown list or another one.**

(i) FTD; Behavioral variant of frontotemporal dementia: **275864**
(ii) FTD; Primary progressive aphasia: **95432**
(iii) PPA; Agramatic var. of primary progressive aphasia: **100070**
(iv) PPA; Semantic primary progressive aphasia: **100069**
(v) PPA; Primary progressive apraxia of speech: **314566**
(vi) MSA-C: **227510**
(vii) MSA-P: **98933**
(viii) PSP-CBS: **240103**
(ix): PSP-P: **240085**
(x) Other PSP subtypes: **683**
(xi) Richardson syndrome: **240071**

12. OMIM Code: Online Mendelian Inheritance in Man (OMIM) is an internationally accepted coding system for genetic diseases. **Here OMIM-code for the gene is required** (not for the disease!).
All codes can be found at the following address: https://www.omim.org
Insert the six-digit number for all patients with a genetically determined diagnosis (only with pathogenic or likely pathogenic variants, not with variants of unkown significance (VUS)).

13. HPO Terms: Human Phenotype Ontology (HPO) provides lists of internationally accepted key features for the standardized description of phenotypes. **Insert here only the belonging code for the main symptom, if the diagnosis is not determined (unsolved cases):**

(i) Ataxia: **HP_0001251**
(ii) HSP: **HP_0001258**
(iii) Chorea: **HP_0002072**
(iv) Dystonia: **HP_0001332**
(v) NBIA: **HP_00012675**
(vi) Paroxysmal dyskinesia: **HP_0007166**
(vii) FTD: **HP_0002145**
(viii) Leukodystrophy: **HP_0002415**
(ix) Parkinsonism: **HP_0001300**

14. Agreement: This item provides information whether the patient agrees to be contacted for research purposes. **`Yes´ is default, please check**.

15. Patient Consent: **`Yes´ is default, please check**.

16. Biological Sample: This data is principle information on availability of biomaterial of any type (DNA, blood, urine, CSF, etc.). **Choose one of the codes:**
(i) yes: **y** (ii) no: **n**

17. Link to Biobank: If there are biological samples available, here should be a hyperlink to the biobank where the samples are stored. **Insert here the hyperlink to the respective Biobank.**

18. Classification of disability: ERN-RND disease groups have agreed on disease group specific scores that are used in all patients. **Insert only the total score (number without acronym).** These are the scores:

(i) Ataxia: Scale for the Assessment and Rating of Ataxie (SARA)
(ii) HD and Chorea: Unified Huntington's Disease Rating Scale – Total Motor Score (UHDRS-TMS)
(iii) Dystonia: Modified Rankin Scale (mRS)
(iv) Frontotemporal dementia: CDR plus NACC FTLD global score
(v) HSP: Spastic Paraplegia Rating Scale (SPRS)
(vi) Leukodystrophy: Gross Motor Function Classification system (GMFC)
(vii) Atypical Parkinson: Hoehn & Yahr Scale

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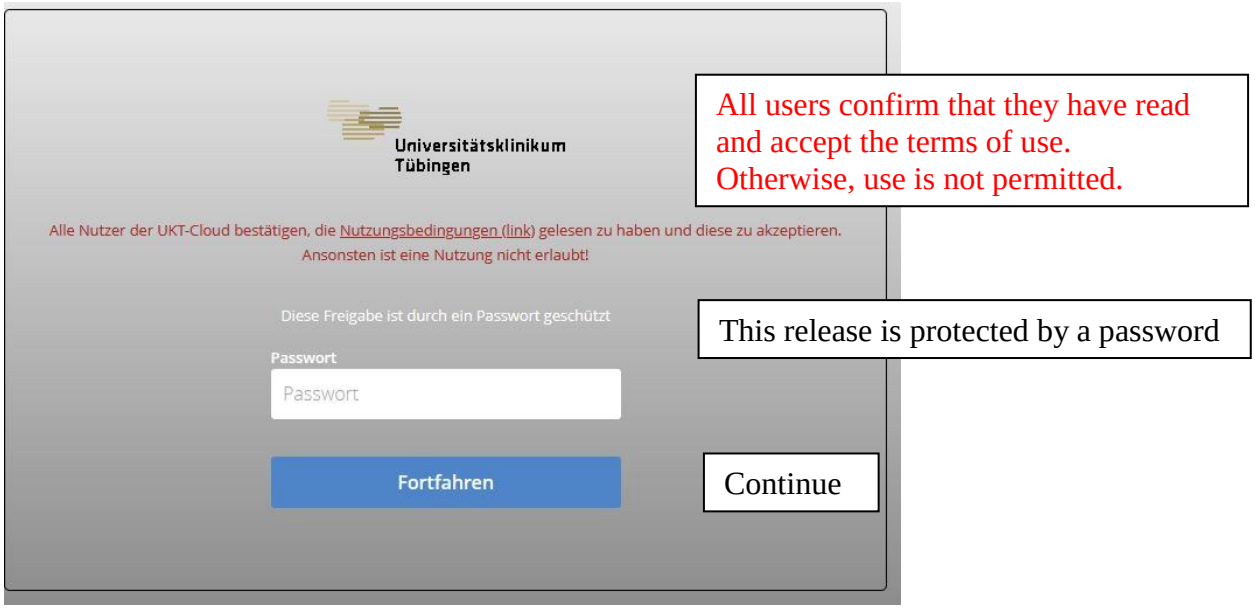

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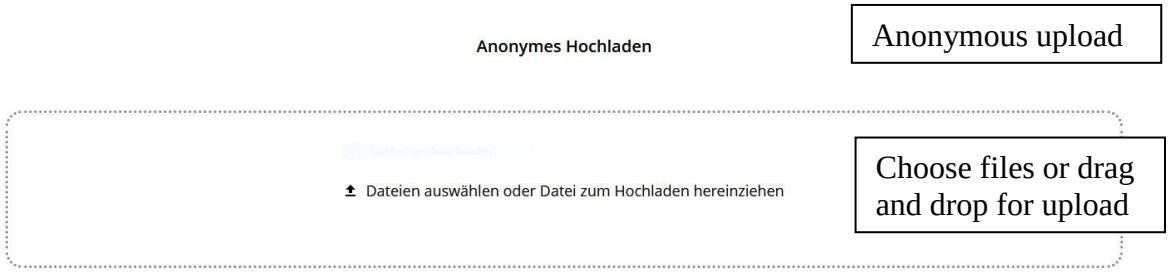

After uploading you see the file which is now in the cloud (Hochgeladene Dateien, Uploaded files). After that you can close the window. We will confirm the upload by email.