



Common Data Elements

VERSION 211120

1. Informed consent:
 - a. Patient/parent(s)/legal representative(s) has/have given informed consent for the data being included in the ERN eUROGEN registry [Radio button, one selection]
 yes no
(If no, answer question 1b and continue with 1e. If yes, answer question 1d and further)
 - b. Why is there no informed consent signed? [Radio button, one selection]
 opt-out procedure other
(If other, answer question 1c)
 - c. Specification other reason [String]
 - d. Who signed the informed consent form? [Tick-box, multiple selection]
 Patient that reached the age of legal majority Date [Date, dd/mm/yy format; at/after DoB]
 Parent(s) / legal representative(s) Date [Date, dd/mm/yy format; at/after DoB]
 - e. Consent to the reuse of pseudonymized data to support commercial projects aimed to improve healthcare [Radio button, one selection]
 yes no unknown
 - f. Consent to the transfer of pseudonymized data to non-EU countries, in compliance with GDPR, to support projects aimed to improve healthcare [Radio button, one selection]
 yes no unknown
 - g. Consent to the linking of pseudonymized data to existing databases/registries to improve healthcare [Radio button, one selection]
 yes no unknown
 - h. Agree to be contacted by medical doctor about research projects and/or clinical studies related to condition [Radio button, one selection]
 yes no unknown
 - i. E-mail address filled out for future surveys [String]
2. Personal information:
 - a. SPIDER Pseudonym: [String]
 - b. Date of birth: [Date, dd/mm/yy format]
 - c. Birth sex: [Radio button, one selection]
 Female Male
 Undetermined Foetus (unknown)
3. Patient Status:
 - a. Patient's status: [Radio button, one selection]
 Alive Dead
 Lost in follow-up Opted-out
(If dead, answer question 2b)
 - b. Date of death: [Date, dd/mm/yy format; at/after DoB]
4. Care pathway:
 - a. First contact with specialised centre: [Date, dd/mm/yy format; at/after DoB]

5. Disease history:
- a. Moment of onset (at which symptoms/signs first appeared): [Radio button, one selection]
 Antenatal At birth
 After birth Undetermined
(If after birth, answer question 5b)
- b. Date of onset: [Date, dd/mm/yy format; at/after DoB]
- c. Is there a diagnosis? [Radio button, one selection]
 yes no
(If yes, answer question 4d, f, g and h. If no, answer question 5a)
- d. Moment of diagnosis: [Radio button, one selection]
 Antenatal At birth
 After birth Undetermined
(If after birth, answer question 4e)
- e. Date of diagnosis: [Date, dd/mm/yy format; at/after DoB]
- Rare disease's diagnosis: [Drop-down menus with one selection each]
- f. Workstream
g. Sub-thematic area of expertise
h. Rare or complex disease
6. Other information about diagnosis:
- a. Phenotype of undiagnosed case: [String]
phenotype (HPO)
- b. Is there genetic information about this patient? [Radio button, one selection]
 yes no
(If yes, answer question 5c)
- c. Genetic diagnosis retained by the specialized centre: [String]
*international classification of mutations (HGVS, strongly recommended - see link**) /HGNC/OMIM code*
7. Biological sample available: [Radio button, one selection]
 yes no unknown

*: <http://www.orphadata.org/cgi-bin/inc/product1.inc.php>

** : <http://www.hgvs.org>

***: <http://www.who.int/classifications/icf/whodasii/en/>



https://ec.europa.eu/health/ern_en



<http://eurogen-ern.eu/>

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