

Proposal of Common Data Elements for the European Platform of rare disease ...

Dear Colleague,

this questionnaire focuses on a limited number of registry data elements, which are relevant either to link information from different rare disease patient registries and to perform a number of predefined analyses of interest to public health institutions, patients and their association, pharmaceutical industry and any other interested party which could financially support registries and the associated research activities.

This questionnaire proposes definitions* and, as relevant, formats and permissible values of this select of data elements and asks about their feasibility in your registry.

Answers are given mostly by ticking boxes in a same set of standard answers, so that your time dedicated to reply to the questionnaire will be kept to the minimum.

Please complete your answer by 22 March 2013.

We thank you for your kind collaboration.

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*: for the sake of wider connectivity, proposed features take into consideration the data elements identified by the US Office for Rare Disease Research, initiatives undertaken by some EU Countries and other specialized research platforms.

Identification of the Registry

* 1. Please, identify yourself

Name of your Registry

Respondent's name

Respondent's e-mail address

2. Which rare disease (or related group of rare disease) is in the scope of your Registry?

One rare disease, please specify the disease's name:

Related group of rare diseases, please specify the group's name:

Other, please specify:

3. Please, indicate the Registry aims: (select all that apply)

	Epidemiology and Public Health	Clinical research	Treatment
-	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
-	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
-	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
-	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
-	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
-	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>

If you have any comments:

4. Is your Registry part of a Network?

- Yes
- No

5. If YES, please indicate the Network:

On patient's personal data

6. Is your Registry able to collect the patient's National Social Security Code?

- Yes, the Registry is
- No, the Registry is not
- The Registry could be adapted to collect this information

7. Could your Registry collect the "First name of patient" defined as following:

DEFINITION: "First name of patient as recorded in birth certificate, passport or identity card"

FORMAT: full name, not initials

- This information is not collected
- This information is already collected as proposed
- This information could be adapted as proposed AND data already collected CAN be made compatible
- This information could be adapted as proposed, BUT data already collected CANNOT be made compatible
- This information cannot be adapted as proposed

8. Could your Registry collect the "Last name of patient" defined as following:

DEFINITION: "Last name of patient as recorded in birth certificate, passport or identity card"

FORMAT: full name, not initials

- This information is not collected
- This information is already collected as proposed
- This information could be adapted as proposed AND data already collected CAN be made compatible
- This information could be adapted as proposed, BUT data already collected CANNOT be made compatible
- This information cannot be adapted as proposed

9. Could your Registry collect the "Gender of patient" defined as following:

DEFINITION: "Patient's physical sex at birth"

PERMISSIBLE VALUES: male, female, other (in any format)

- This information is not collected
- This information is already collected as proposed
- This information could be adapted as proposed AND data already collected CAN be made compatible
- This information could be adapted as proposed, BUT data already collected CANNOT be made compatible
- This information cannot be adapted as proposed

10. Could your Registry collect the "Patient's city of birth" defined as following:

DEFINITION: "Name of city/town/village where the patient was born as it appears on the birth certificate, passport or identity card"

FORMAT: full name of city

- This information is not collected
- This information is already collected as proposed
- This information could be adapted as proposed AND data already collected CAN be made compatible
- This information could be adapted as proposed, BUT data already collected CANNOT be made compatible
- This information cannot be adapted as proposed

On patient's personal data

**11. Could your Registry collect the "Patient's country of birth" defined as following:
DEFINITION: "Name of country where the patient was born as it appears on the birth certificate, passport or identity card"**

FORMAT: full name of country

- This information is not collected
- This information is already collected as proposed
- This information could be adapted as proposed AND data already collected CAN be made compatible
- This information could be adapted as proposed, BUT data already collected CANNOT be made compatible
- This information cannot be adapted as proposed

**12. Could your Registry collect the "Patient's city of residence" defined as following:
DEFINITION: "Name of city/town where the patient usually lives"**

FORMAT: full name of city

- This information is not collected
- This information is already collected as proposed
- This information could be adapted as proposed AND data already collected CAN be made compatible
- This information could be adapted as proposed, BUT data already collected CANNOT be made compatible
- This information cannot be adapted as proposed

13. Could your Registry collect the "Patient's country of residence" defined as following:

DEFINITION: "Name of country where the patient usually lives"

FORMAT: full name of country

- This information is not collected
- This information is already collected as proposed
- This information could be adapted as proposed AND data already collected CAN be made compatible
- This information could be adapted as proposed, BUT data already collected CANNOT be made compatible
- This information cannot be adapted as proposed

14. Could your Registry collect the "Date of patient's birth" defined as following:

DEFINITION: "Date of patient's birth recorded in birth certificate, passport or identity card"

FORMAT: complete date (year, month, day) in any format

- This information is not collected
- This information is already collected as proposed
- This information could be adapted as proposed AND data already collected CAN be made compatible
- This information could be adapted as proposed, BUT data already collected CANNOT be made compatible
- This information cannot be adapted as proposed

15. Could your Registry collect the "Patient's date of death"?

FORMAT: complete date (year, month, day) in any format

- This information is not collected
- This information is already collected
- This information could be adapted AND data already collected CAN be made compatible
- This information could be adapted, BUT data already collected CANNOT be made compatible
- This information cannot be adapted

16. Could your Registry collect "Patient's contact" defined as following:

DEFINITION: "Any patient contact information, collected by the registry at the central or peripheral level, that allow to contact the patient"

- This information is not collected
- This information is already collected as proposed
- This information could be adapted as proposed AND data already collected CAN be made compatible
- This information could be adapted as proposed, BUT data already collected CANNOT be made compatible
- This information cannot be adapted as proposed

On disease data

17. Please, indicate if your Registry collects data on:

- Patients with confirmed diagnosis only
- Both patients with confirmed and with suspected diagnosis

18. Is your Registry able to collect data on "Genetic features of the rare disease"?

- Yes, the Registry is
- No, the Registry is not
- The Registry could be adapted to collect data on "Genetic features"

19. If Yes, please indicate what genetic data you collect:(select all that apply)

- Gene (HGNC Gene Symbol)
- Chromosome number
- Chromosomal reference sequence accession and version number
- RefSeqGene accession and version number
- Locus Reference Genomic (LRG)
- Variant description in HGVS format
- Variant description in other format
- Other

Other, please specify

20. Could your Registry collect "Genetic features" on the patient's family members?

- This information is not collected
- This information is already collected
- This information could be adapted AND data already collected CAN be made compatible
- This information could be adapted, BUT data already collected CANNOT be made compatible
- This information cannot be adapted

21. In case of disease subject to neonatal screening, could your Registry collect data on the "screening results" as following:

DEFINITION: Results of laboratory screening test

PERMISSIBLE VALUES: positive, negative, not tested, not known (in any format)

- This information is not collected
- This information is already collected as proposed
- This information could be adapted as proposed AND data already collected CAN be made compatible
- This information could be adapted as proposed, BUT data already collected CANNOT be made compatible
- This information cannot be adapted as proposed

22. Could your Registry collect the "Clinical data of disease" defined as following:

DEFINITION: "Any symptoms, clinical signs or surgery due to the patient's rare disease"

- This information is not collected
- This information is already collected as proposed
- This information could be adapted as proposed AND data already collected CAN be made compatible
- This information could be adapted as proposed, BUT data already collected CANNOT be made compatible
- This information cannot be adapted as proposed

On disease data

23. Could your Registry collect the "Date of diagnosis" defined as following:

DEFINITION: "Date when the rare disease diagnosis was confirmed"

FORMAT: complete date (year, month, day) in any format

- This information is not collected
- This information is already collected as proposed
- This information could be adapted as proposed AND data already collected CAN be made compatible
- This information could be adapted as proposed, BUT data already collected CANNOT be made compatible
- This information cannot be adapted as proposed

24. Could your Registry collect the "Date of disease onset" defined as following:

DEFINITION: "Date when patient first began experiencing symptoms or signs of the rare disease"

FORMAT: complete date (year, month, day) in any format

- This information is not collected
- This information is already collected as proposed
- This information could be adapted as proposed AND data already collected CAN be made compatible
- This information could be adapted as proposed, BUT data already collected CANNOT be made compatible
- This information cannot be adapted as proposed

25. Could your Registry collect the "Date of first report to the Health Service" defined as following:

DEFINITION: "Date when patient first reported symptoms or signs of the rare disease to physician"

FORMAT: complete date (year, month, day) in any format

- This information is not collected
- This information is already collected as proposed
- This information could be adapted as proposed AND data already collected CAN be made compatible
- This information could be adapted as proposed, BUT data already collected CANNOT be made compatible
- This information cannot be adapted as proposed

26. Could your Registry collect the "Co-morbidity" defined as following:

DEFINITION: "Other diseases observed in the patient"

FORMAT: any format and coding system

- This information is not collected
- This information is already collected as proposed
- This information could be adapted as proposed AND data already collected CAN be made compatible
- This information could be adapted as proposed, BUT data already collected CANNOT be made compatible
- This information cannot be adapted as proposed

27. Could your Registry collect the "Hospitalization" defined as following:

DEFINITION: "Cumulative number of patient's admissions to the hospital due to the rare disease"

FORMAT: number

- This information is not collected
- This information is already collected as proposed
- This information could be adapted as proposed AND data already collected CAN be made compatible
- This information could be adapted as proposed, BUT data already collected CANNOT be made compatible
- This information cannot be adapted as proposed

28. Could your Registry collect data on "Assistive devices" defined as following:

DEFINITION: "Type of assistive devices used by patient"

FORMAT: type of device

- This information is not collected
- This information is already collected as proposed
- This information could be adapted as proposed AND data already collected CAN be made compatible
- This information could be adapted as proposed, BUT data already collected CANNOT be made compatible
- This information cannot be adapted as proposed

On data of disease

29. Is your Registry able to collect data on "Patient's disability" caused by rare disease?

- Yes, the Registry is
- No, the Registry is not
- The Registry could be adapted to collect data on "Patient's disability"

30. If your Registry is already collecting data on "Patient's disability", please indicate what assessment questionnaire is used:

31. Is your Registry able to collect data on "Patient's quality of life" ?

- Yes, the Registry is
- No, the Registry is not
- The Registry could be adapted to collect data on "Patient's quality of life"

32. If your Registry is already collecting data on "Patient's quality of life", please indicate what assessment questionnaire is used:

33. Is your Registry able to collect the following data on "Centre which made diagnosis"?

	Yes	No	The Registry could be adapted to collect this data
Full name of the Diagnosing Centre	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
City/Town where the Diagnosing Centre is	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Contact data of the the Diagnosing Centre	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>

34. Is your Registry able to collect the following data on "Treating Centre"?

	Yes	No	The Registry could be adapted to collect this data
Full name of the Treating Centre	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
City/Town where the Treating Centre is	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Contact data of the Treating Centre	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>

On treatments

35. Could your Registry collect data on “Current orphan drug treatment” defined as following:

DEFINITION: "A list of all current orphan drugs that a patient is currently taking"

FORMAT: name of all active ingredient

- This information is not collected
- This information is already collected as proposed
- This information could be adapted as proposed AND data already collected CAN be made compatible
- This information could be adapted as proposed, BUT data already collected CANNOT be made compatible
- This information cannot be adapted as proposed

36. Could your Registry collect data on “Side effects of orphan drug treatments” defined as following:

DEFINITION: "A list of confirmed or suspected side effects observed during the current orphan drug treatments"

- This information is not collected
- This information is already collected as proposed
- This information could be adapted as proposed AND data already collected CAN be made compatible
- This information could be adapted as proposed, BUT data already collected CANNOT be made compatible
- This information cannot be adapted as proposed

37. Could your Registry collect data on “Current drug treatment” defined as following:

DEFINITION: "A list of all current drugs (different from orphan drugs) that a patient is currently taking"

FORMAT: name of active ingredient

- This information is not collected
- This information is already collected as proposed
- This information could be adapted as proposed AND data already collected CAN be made compatible
- This information could be adapted as proposed, BUT data already collected CANNOT be made compatible
- This information cannot be adapted as proposed

38. On which other types of treatment (different from medications) does your Registry collect data? (select all that apply)

- This information is not collected
- Cognitive Rehabilitation
- Physical Rehabilitation
- Surgery
- Other

Other, please specify

On other data of the Registry

39. Is your Registry able to collect repeated observations to support longitudinal studies?

- Yes, The Registry is
- No, the Registry is not
- The Registry could be adapted to update its data

40. If YES, please indicate what data are updated:

	Yes	No
"Patient's city of residence"	<input type="radio"/>	<input type="radio"/>
"Patient's country of residence"	<input type="radio"/>	<input type="radio"/>
"Clinical data of disease"	<input type="radio"/>	<input type="radio"/>
"Co-morbidity"	<input type="radio"/>	<input type="radio"/>
"Hospitalization"	<input type="radio"/>	<input type="radio"/>
"Assistive devices"	<input type="radio"/>	<input type="radio"/>
"Patient's disability"	<input type="radio"/>	<input type="radio"/>
"Patient's quality of life"	<input type="radio"/>	<input type="radio"/>
"Treating Centre"	<input type="radio"/>	<input type="radio"/>
"Current drug treatment"	<input type="radio"/>	<input type="radio"/>
"Current orphan drug treatment"	<input type="radio"/>	<input type="radio"/>
"Side effects of treatments"	<input type="radio"/>	<input type="radio"/>

41. Is your Registry able to collect the following data on "patient's recruitment" and biospecimen donation?

	Yes	No	The Registry could be adapted to collect this data
Patient's willingness to be contacted to participate in a future trial	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Patient's willingness to be contacted about donating biological samples for research	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>

42. If patient has already donated biological samples, could your Registry collect data on "Donation of biological samples"?

- This information is not collected
- This information is already collected
- This information could be adapted AND data already collected CAN be made compatible
- This information could be adapted, BUT data already collected CANNOT be made compatible
- This information cannot be adapted

43. If your Registry collects data on "Donation of biological samples", please indicate which data are collected:

	Yes	No
Type of biological sample (i.e. tissue or body fluid or other biospecimen)	<input type="radio"/>	<input type="radio"/>
Name of the Biobank where the biological sample is stored up	<input type="radio"/>	<input type="radio"/>
Contact details of Biobank where the biological sample is stored up, for communication purposes	<input type="radio"/>	<input type="radio"/>

Other, please specify

44. Is your Registry able to collect data on "Transplantations"?

- Yes, The Registry is
- No, The Registry is not
- The Registry could be adapted to collect data on "Transplantations"

45. If YES, please indicate which data the Registry collects:

	Yes	No
Transplant material (i.e. organ or cells or other material)	<input type="radio"/>	<input type="radio"/>
Date of transplantation	<input type="radio"/>	<input type="radio"/>