Activities and needs of EU registers for RD

About this survey

Dear Colleague,

The European Commission has identified rare diseases registers as one of the priorities of the EU policy framework for rare diseases in Europe. According to the Commission Communication COM (2008) 679/2 on Rare diseases, registers and databases are key instruments to increase knowledge on rare diseases and develop clinical research.
Also, the recently launched initiative International Rare Disease Research Consortium (IRDiRC) has identified registers, databases and biobanks as one of the priority means to foster transatlantic cooperation among EU, USA, Canada, China, Japan and other countries.

In 2010 the European Commission funded the EPIRARE Project (www.epirare.eu), in order to build consensus and synergies to address regulatory, ethical and technical issues associated with the registration of rare diseases patients and to elaborate possible policy scenarios.

Through this survey the EPIRARE project intends to explore the functioning, resources, problems, needs and expectations of existing registers on the European territory.
The final aim is to develop tools and services in support of existing registers and to favour the creation of new ones where needed.
The survey is directed to both active and expired RD registers.

Your participation to the survey is of paramount importance as we believe that through joint efforts great advances can be contributed to the rare disease field.
Your participation to this survey will ensure your prompt access to the preliminary results and you will be further consulted on the preparation of support services and tools for RD Registers.

Any specific information on your register or database will be kept confidential.

Thank you in advance for your cooperation!

The EPIRARE coordinating team

1. You will need about 25 minutes to fill up the questionnaire. However you can save a partially filled questionnaire and complete it at another time. Do you wish to procede?

- ○ Yes
- ○ No, I really have no time to reply
- ○ No, I don't believe questionnaires are useful
- ○ No, I am not informed enough to reply
- ○ No, I am not authorized to reply

Activities and needs of EU registers for RD

2 New adress

2. Do you know a person who can reply to the questionnaire? IF YES, please write his/her name, surname and email adress here:

Activities and needs of EU registers for RD

General characteristics of the Register

*3. Contact details

Name and Surname:	
Name of the Register:	
Address:	
City/Town:	
State/Province:	
Country:	
Email Address:	
Phone Number:	

4. Year of starting activity (first case collected)

- ○ Before 1960
- ○ 1960-9
- ○ 1970-9
- ○ 1980-9
- ○ 1990-9
- ○ 2000-11

*5. Is your register currently active (collecting data)?

- ○ yes
- ○ no

Activities and needs of EU registers for RD

General Characteristics (2)

6. How many active cases/patients are included in your register? (prevalence)

- 10 to 50
- 51 to 200
- 201-500
- 501-1000
- 1001-2000
- 2001-5000
- 5001-10000
- >10000

7. How many new cases have been entered during the last year? (incidence)

- None
- 5 to 10
- 11 to 50
- 51-100
- 101-200
- >200

8. How many rare diseases are included?

- Just one
- A group of related RDs
- Several RDs (or groups of RDs) not related among them
- All rare diseases

Other (specify):

Activities and needs of EU registers for RD

Aims and scope of the register

9. What are the aims of the register? (select all that apply)

- [ ] Natural history of the disease
- [ ] Epidemiological research
- [ ] Clinical research (patient recruitment for clinical trials)
- [ ] Disease surveillance
- [ ] Treatment evaluation (efficacy)
- [ ] Treatment monitoring (safety)
- [ ] Mutation database
- [ ] Genotype-phenotype correlation
- [ ] Social planning
- [ ] Healthcare Services planning

Other (specify):

10. Are your data used for pharmacovigilance?

- ○ Yes, they are currently used for pharmacovigilance
- ○ They could easily be used for pharmacovigilance in their current form
- ○ They could be used for pharmacovigilance if integrated with new information
- ○ No, the register has very different aims form pharmacovigilance

Other (specify)

11. What is the geographical coverage of the register?

- ○ International
- ○ European
- ○ National
- ○ Regional
- ○ Local (i.e.: Hospital, Primary health care centre, …)

Other (specify)

Activities and needs of EU registers for RD

12. What is the target population of the register?

- ○ Population-based
- ○ Hospital-based
- ○ Case series
- ○ Family case series
- ○ Cohort of cases (all cases are linked by some variable different from the diagnosis)

Other (specify)

Activities and needs of EU registers for RD

Collected data

13. Does a case definition exist for the RDs of your interest?

- ○ Yes
- ○ No

14. Are standardised inclusion/exclusion criteria defined for the RD cases?

- ○ Yes
- ○ No

15. What kind of disease coding systems are in use? (select all that apply)

- ☐ ORPHAcodes
- ☐ MIM codes
- ☐ ICDO (for oncology)
- ☐ ICD9 codes
- ☐ ICD10 codes
- ☐ SNOMED codes
- ☐ MESH (Pubmed) codes
- ☐ Your own code system
- ☐ No coding system is used, just the disease name

Other (specify):

Activities and needs of EU registers for RD

16. What kind of data are collected? (select all that apply)

- [ ] Anagraphical data
- [ ] Diagnosis
- [ ] Anthropometric information
- [ ] Socio-demographic information
- [ ] Genetic data
- [ ] Clinical data
- [ ] Medications, devices and health services
- [ ] Patient-reported outcomes (e.g. quality of life data, Health status, etc)
- [ ] Family history
- [ ] Birth and reproductive history
- [ ] Clinical research participation and bio-specimen donation
- [ ] Patient's preferences for communication

Other (specify):

17. Is the date of the patient death collected?

- ○ Yes, directly
- ○ Yes, by linking with other data source
- ○ No

18. In case of termination of the register, data will be:

- ○ Destroyed
- ○ Archived for an undetermined amount of time
- ○ Archived for a determined amount of time
- ○ Made available to other registries or to the research community
- ○ There is no policy for termination

Other (specify):

Activities and needs of EU registers for RD

Register's sources

19. Which are your data providers? (select all that apply)

- [ ] Clinical units
- [ ] Clinical Genetic units
- [ ] Laboratories/central services (biochemistry, pathological services, genetic, Rx, etc)
- [ ] Hospital databases (Discharge registers)
- [ ] Patients and families
- [ ] Patients' groups (associations/federations)
- [ ] Disability registers
- [ ] Mortality registers
- [ ] Birth registers
- [ ] Centres of expertise
- [ ] Other registers

Other (specify):

20. How are data entered in the register? (select all that apply)

- [ ] Information is entered on-line by data providers
- [ ] Information is entered on-line by patients (patient reported outcomes)
- [ ] Information is sent electronically by data providers and entered by the register's staff
- [ ] Information is sent on paper by data providers and entered by the register's staff
- [ ] Information is downloaded from other primary databases

Other (specify):

21. Are data periodically updated?

- ○ Yes
- ○ No

Activities and needs of EU registers for RD

Quality of data

22. What methods do you use to avoid data entry mistakes? (select all that apply)

- [ ] Information is typed twice (manual control)
- [ ] An internal program automatically checks the type of response (automatic control)
- [ ] Information is entered electronically but there is no control for mistakes
- [ ] There is no specific method to avoid data entry mistakes

Other (specify):

23. Has the register a set of quality indicators?

- ○ Yes
- ○ No

24. if YES, could you briefly list the indicators used, please?

25. Has the register been checked for reliability (when a data provider always interprets the same question in the same way)?

- ○ Yes
- ○ No

26. Has the register been checked for agreement (when different data providers interpret the same question in the same way)?

- ○ Yes
- ○ No

27. Is internal validity checked in the register (information corresponds to the explored reality)?

- ○ Yes
- ○ No

28. Are quality tests/surveys periodically performed?

- ○ Yes
- ○ No

29. Do you have methods to avoid duplication of the registered cases?

- ○ Yes
- ○ No

Activities and needs of EU registers for RD

30. Are instructions for use of the register available?

- ○ Yes, publicly available
- ○ Yes, available for partners
- ○ Not available but under construction
- ○ No, they are not available

31. Do you train or provide a training kit for new users? (select all that apply)

- ☐ There is a structured training course
- ☐ Supervised training is made
- ☐ A training kit is provided
- ☐ A training kit is under construction
- ☐ There is no structured training for new users

Other (specify)

Activities and needs of EU registers for RD

Ethical and Legal issues

32. Your register has been established:

- By law
- To comply with regulatory requirements
- As part of a research project
- Following autonomous initiatives (clinicians' initiatives, patient driven registries, etc…)

Other (specify):

33. What kind of data do you collect?

- Irreversibly anonymised
- Reversibly anonymised
- Identifiable

34. Has the register protocol been approved by an Ethics Committee?

- Yes, when it was set up
- Yes, for every new research
- Not yet
- Not required

*35. Do you ask for patient's written and informed consent to include his/her identifiable data in the register?

- Yes
- No, but the patient has the possibility to opt out of the register
- No, informed consent is not collected/required
- Not applicable as data are collected anonymously

Other (specify):

Activities and needs of EU registers for RD

Informed Consent

36. What information is provided on the patient information sheet? (select all that apply)

- [ ] Register's general scope
- [ ] Register's specific research objectives
- [ ] That the register is part of a network
- [ ] Type of coding and access to the register
- [ ] Type of users accessing the data
- [ ] Level of data protection from intrusions (encryption systems)
- [ ] Right to withdraw
- [ ] Information on data property rights
- [ ] Information on entitlement to intellectual property
- [ ] Possibility of the register's termination
- [ ] Possibility to be contacted for participating in clinical trials
- [ ] Name and contact of the data processor

Other (specify):

37. The patient information sheet has been revised:(select all that apply)

- [ ] By a local ethics committee
- [ ] By a patients association
- [ ] Revision not required

Other (specify):

38. Is the patient's information sheet publicly available and easily accessible? (e.g. on the register website)

- ○ Yes
- ○ No

Activities and needs of EU registers for RD

39. If a participant decides to withdraw from the register, he/she may: (select all that apply):

- [ ] Have data anonymised for future research (irreversible destruction of participant's identity)
- [ ] Withdraw the authorisation to future uses of the data (except already aggregated or published data)
- [ ] Have data destroyed
- [ ] Withdraw the authorisation to be re-contacted
- [ ] Withdrawal is not possible as this is a mandatory public health register
- [ ] Information not available

Activities and needs of EU registers for RD

Governance

40. Has the register a main governing board?

- ○ Yes
- ○ No

41. IF YES, How is the governing board composed? (select all that apply)

- ☐ Internal members
- ☐ Patient representatives
- ☐ External experts

Other (specify):

42. IF YES, What are the main functions of the governing board? (select all that apply)

- ☐ Financial and administrative issues
- ☐ Ethical and legal issues
- ☐ Database content, research objectives, epidemiology, biostatistics, etc.
- ☐ Communication with the funding source, health care providers, patients, etc.
- ☐ Data access and use by internal and external researchers
- ☐ Coordination of all parties involved in the register

Other (specify)

43. Has the register other governing bodies?

- ○ Yes
- ○ No

Activities and needs of EU registers for RD

44. IF YES, how are they composed, and what are their functions?

	Internal members	Patient representatives	External experts
Financial and administrative issues	☐	☐	☐
Ethical and legal issues	☐	☐	☐
Database content, research objectives, etc.	☐	☐	☐
Communication with the funding source, health care providers, patients, etc.	☐	☐	☐
Data access and use	☐	☐	☐
Coordination of all parties involved in the register	☐	☐	☐
Other (specify)	☐	☐	☐

Specify here:

Activities and needs of EU registers for RD

Communication

45. Who is informed of the register activities? (select all that apply)

- [ ] Data Providers
- [ ] Local Ethics Committee
- [ ] Hospitals/Centres of Expertise
- [ ] University
- [ ] Personal Data Protection Authority (or similar body)
- [ ] Funding organisations, public or private
- [ ] Public health policy makers
- [ ] Patients' Associations
- [ ] Social Health Insurances ("payers")
- [ ] None

Other (specify):

46. How do you communicate your activities? (select all that apply)

- [ ] Direct contact
- [ ] Website
- [ ] Newsletter
- [ ] Newspaper
- [ ] Scientific journals
- [ ] Communication to scientific meetings
- [ ] Institutional bulletins
- [ ] None

Other (specify):

47. Has the register been acknoledged in any peer reviewed scientific publication?

- ○ No
- ○ Yes, 1-5 times
- ○ Yes, 6-10 times
- ○ Yes, 11-15 times
- ○ Yes, >15 times

Activities and needs of EU registers for RD

Access to data and security

48. Is your register collaborating and sharing data with (select all that apply)

- [ ] Other registers
- [ ] Biobanks
- [ ] Centres of expertise
- [ ] None of them

Other (specify):

*49. Do you make your data available outside the register?

- ○ Yes
- ○ Not yet
- ○ No

Activities and needs of EU registers for RD

Access to data and security (2)

50. Data are made available to:

	Anonymous	De-identified	Identifiable
Public Authorities	☐	☐	☐
Public Institutions	☐	☐	☐
Private institutions/citizens	☐	☐	☐
Companies/Industries	☐	☐	☐
Patients Associations	☐	☐	☐

Other (specify)

51. Are data transferrable:

	Anonymous	De-identified	Identifiable
Inside your Country	☐	☐	☐
Inside EU	☐	☐	☐
Worldwide	☐	☐	☐

52. Does your register apply a specific procedure for access to data? (select all that apply)

- ☐ Registration
- ☐ Approval by an internal committee
- ☐ Approval by an external committee
- ☐ Contract agreement for data analysis/transfer
- ☐ (Pre-) Pay access or transfer fees (for industry)
- ☐ (Pre-) Pay access or transfer fees (for all users)

Other (specify):

53. How do you ensure security of your register? (select all that apply)

- ☐ Passwords are renewed periodically and systematically
- ☐ The register data are hosted in a dedicated server
- ☐ The register have a reliable back up strategy
- ☐ The register has an intrusion detection system
- ☐ A commercial anti-virus is used

Other (specify):

Activities and needs of EU registers for RD

Register's sustainability

54. The register was set up with funding coming from: (select all that apply)

- [ ] No specific fund
- [ ] Regional Authority
- [ ] National Authority
- [ ] University/Research Institute
- [ ] Hospital
- [ ] Patients Association
- [ ] Foundation
- [ ] Industry/Industrial Association
- [ ] EU Commission/EU Agency
- [ ] Information not available

Other (specify)

55. Who is funding the register today? (select all that apply)

- [ ] No specific funds
- [ ] Regional Authority
- [ ] National Authority
- [ ] University/Research Institute
- [ ] Hospital
- [ ] Patients Association
- [ ] Foundation
- [ ] Industry/Industrial Association
- [ ] EU Commission/ EU Agency
- [ ] Information not available

Other (specify):

Activities and needs of EU registers for RD

56. Please indicate your average yearly budget over the past 3 years:

- ○ No funds
- ○ < 50K€
- ○ 51 - 100 K€
- ○ 101 - 200 K€
- ○ 201 - 500 K€
- ○ 501 K€ - 1M€
- ○ > 1M€
- ○ Information not available
- ○ I do not wish to answer this question

57. Are there policies/decisions in place to ensure the long-term sustainability of the register?

- ○ Yes
- ○ No

58. How many persons are payed by the register?

	persons (n)
Medical staff	
Technical staff	
Administrative staff	
Volunteers	
Other	

Other (specify)

Activities and needs of EU registers for RD

Needs and Expectations

59. What are the main needs of your register? (select max 5 answers)

- [ ] Gather financial support
- [ ] Update your data collection form
- [ ] Widen the geographical coverage of the register
- [ ] Motivate data providers
- [ ] Recruit new data providers
- [ ] Assess the quality of your data
- [ ] Improve training of the register's staff
- [ ] Revise the informed consent form
- [ ] Gather expert legal advice
- [ ] Establish a more robust system of governance
- [ ] Strengthen the relation with patients associations
- [ ] Hire new employees
- [ ] Gather technical help to refurbish your IT system
- [ ] Improve the data protection/security system
- [ ] Communicate and publicise your results
- [ ] Link your register with other registers
- [ ] Link your register with biobanks and bioinformatics

Other (specify):

60. Do you find desirable that the European Commission (EC) draws new legislations to uniformly regulate registries across the EU?

- ○ Yes
- ○ No
- ○ I have no opinion on the subject

Other (please specify)

Activities and needs of EU registers for RD

61. Do you expect your country to provide public funding for a centralised national register on RD?

- ○ Yes
- ○ No
- ○ It already exist in our Country

Other (please specify)

62. Do you find desirable that the EC and Member States build a single portal to access all existing RD registries in the EU?

- ○ Yes
- ○ No
- ○ I have no opinion on the subject

Other (please specify)

63. Would you find it feasible and useful to have a EU platform providing services for RD registries?

- ○ Yes
- ○ No
- ○ I have no opinion on the subject

Other (please specify)

64. Please indicate which of the services below should be offered by a EU platform for registries (select only 3):

- ☐ IT tools (e.g. Database software and secure data exchange),
- ☐ Legal advice
- ☐ Model documents (e.g. Informed consent form)
- ☐ Expert technical advice
- ☐ Quality control systems, quality experts advice, etc.
- ☐ Tools for networking among partners and among registries
- ☐ Facilitated access to useful data sources

Other (specify):

Activities and needs of EU registers for RD

Expired registers

65. If no, why?

- [ ] Lack of funding
- [ ] Study concluded
- [ ] Loss of commitment from data providers

other (specify)

66. When the register was set up, it was funded by: (select all that apply)

- [ ] No specific fund
- [ ] Regional Authority
- [ ] National Authority
- [ ] University/Research Institute
- [ ] Hospital
- [ ] Patients Association
- [ ] Foundation
- [ ] Industry/Industrial Association
- [ ] EU Commission/EU Agency

Other (specify)

67. Year of ending activity (last case collected)

- ○ Before 1960
- ○ 1960-9
- ○ 1970-9
- ○ 1980-9
- ○ 1990-9
- ○ 2000-2011

Activities and needs of EU registers for RD

68. How many active cases/patients were included in your register? (prevalence)

- 10 to 50
- 51 to 200
- 201-500
- 501-1000
- 1001-2000
- 2001-5000
- 5001-10000
- >10000

69. How many rare diseases were included?

- Just one (specify)
- A group of related RDs(specify)
- Several RDs (or groups of RDs) not related among them
- All rare diseases

Specify here:

Activities and needs of EU registers for RD

Aims and scope of the register

70. What were the aims of the register? (select all that apply)

- [ ] Natural history of the disease
- [ ] Epidemiological research
- [ ] Clinical research (patient recruitment for clinical trials)
- [ ] Disease surveillance
- [ ] Treatment evaluation (efficacy)
- [ ] Treatment monitoring (safety)
- [ ] Mutation database
- [ ] Genotype-phenotype correlation
- [ ] Social planning
- [ ] Healthcare Services planning

Other (please specify)

71. What was the geographical coverage of the register?

- ○ International
- ○ European
- ○ National
- ○ Regional
- ○ Local (i.e.: Hospital, Primary health care centre, …)

Other (specify)

72. What was the target population of the register?

- ○ Population-based
- ○ Hospital-based
- ○ Case series
- ○ Family case series
- ○ Cohort of cases (all cases are linked by some variable different from the diagnosis)

Other (specify)

73. Did a case definition exist for the RDs of your interest?

- ○ Yes
- ○ No

Activities and needs of EU registers for RD

74. Were standardised inclusion/exclusion criteria defined for the RD cases?

- ○ Yes
- ○ No

75. What kind of disease coding systems were in use? (select all that apply)

- ☐ ORPHAcodes
- ☐ MIM codes
- ☐ ICDO (for oncology)
- ☐ ICD9 codes
- ☐ ICD10 codes
- ☐ SNOMED codes
- ☐ MESH (Pubmed) codes
- ☐ Your own code system
- ☐ No coding system is used, just the disease name

Other (specify):

76. What kind of patient's data were collected? (select all that apply)

- ☐ Anagraphical data
- ☐ Diagnosis
- ☐ Anthropometric information
- ☐ Socio-demographic information
- ☐ Genetic data
- ☐ Clinical data
- ☐ Medications, devices and health services
- ☐ Patient-reported outcomes (e.g. quality of life data, Health status, etc)
- ☐ Family history
- ☐ Birth and reproductive history
- ☐ Clinical research participation and bio-specimen donation
- ☐ Patient's preferences for communication

Other (specify):

77. Was the date of the patient death collected?

- ○ Yes, directly
- ○ Yes, by linking with other data source
- ○ No

Activities and needs of EU registers for RD

Quality of data

78. What methods did you use to avoid data entry mistakes? (select all that apply)

- [ ] The information was typed twice (manual control)
- [ ] The type of response was checked automatically by an internal program (automatic control)
- [ ] Information was entered electronically but without control for mistakes
- [ ] There was no specific method to avoid data entry mistakes
- [ ] Other, please specify

79. Did the register have a set of quality indicators?

- ( ) Yes
- ( ) No

80. if YES, could you briefly list the indicators used, please?

Activities and needs of EU registers for RD

Ethical and legal issues

81. Your register was established:

- By law
- To comply with regulatory requirements
- As part of a research project
- Following autonomous initiatives (clinicians' initiatives, patient driven registries, etc…)

Other (specify):

82. Had the register protocol been approved by an Ethics Committee?

- Yes
- No
- Not required

Other (specify):

83. What kind of data did you collect?

- Irreversibly anonymised
- Reversibly anonymised
- Identifiable

84. Did you ask for patient's written and informed consent to include his/her identifiable data in the register?

- Yes
- No, but the patient could opt out of the register
- No, informed consent was not collected
- Not applicable as data were collected anonymously

Other (specify):

Activities and needs of EU registers for RD

Governance

85. Was there a governing board of the register? if yes, what were its functions?

- [ ] Financial and administrative issues
- [ ] Ethical and legal issues
- [ ] Database content, research objectives, epidemiology, biostatistics, etc.
- [ ] Communication with the funding source, health care providers, patients, etc.
- [ ] Data access and use by internal and external researchers
- [ ] Coordination of all parties involved in the registry
- [ ] There was no governing board

Other (specify)

86. Who was informed of the register activities, if any? (select all of those that apply)

- [ ] Data Providers
- [ ] Local Ethics Committee
- [ ] Hospitals/Centres of Expertise
- [ ] University
- [ ] Personal Data Protection Authority (or similar body)
- [ ] Funding organisations, public or private
- [ ] Public health policy makers
- [ ] Private Company
- [ ] Patients' Association
- [ ] Social Health Insurances ("payers")

Other (specify):

Activities and needs of EU registers for RD

Access to data and security

87. After termination of the register, data were:

- ○ Destroyed
- ○ Archived for an undetermined amount of time
- ○ Archived for a determined amount of time

Other (specify):

88. If your data were archived, would you be willing or in the position to make your data available for future studies?

- ○ Yes, data are already available
- ○ Yes, they could be made available in the future
- ○ No

89. Did your register apply a specific procedure for access to data? (select all of those that apply)

- ☐ Registration
- ☐ Approval by an internal committee
- ☐ Approval by an external committee
- ☐ Contract agreement for data analysis/transfer
- ☐ (Pre-) Pay access or transfer fees (for industry)
- ☐ (Pre-) Pay access or transfer fees (for all users)
- ☐ There was no specific procedure

Other (specify):

90. Was your register collaborating and sharing data with (select all those that apply)

- ☐ Other registries
- ☐ Biobanks
- ☐ Centres of expertise

Other (specify):

Activities and needs of EU registers for RD

91. How did you ensure security of your register? (select all that apply)

- [ ] Passwords were renewed periodically and systematically
- [ ] The register data were hosted in a dedicated server
- [ ] The register had a reliable back up strategy
- [ ] The register had an intrusion detection system
- [ ] A commercial anti-virus was used

Other (specify):

Activities and needs of EU registers for RD

Needs and Expectations

92. Do you find desirable that the European Commission (EC) draws new legislations to uniformly regulate registries across the EU?

- ○ Yes
- ○ No
- ○ I have no opinion on the subject

Other (please specify)

93. Do you expect your country to provide public funding for a centralised national register on RD?

- ○ Yes
- ○ No
- ○ It already exist in our Country

Other (please specify)

94. Do you find desirable that the EC and Member States build a single portal to access all existing RD registries in the EU?

- ○ Yes
- ○ No
- ○ I have no opinion on the subject

Other (please specify)

95. Would you find it feasible and useful to have a EU platform providing services for RD registries?

- ○ Yes
- ○ No
- ○ I have no opinion on the subject

Other (please specify)

Activities and needs of EU registers for RD

Final comments

***96. This is the end of the questionnaire.**

Choose YES if you consider the questionnaire finalised

Choose NO if you wish to revise the questionnaire later (replies will be saved and you will be redirected to the first page)

- Yes
- No

Comments

Activities and needs of EU registers for RD

End

Thank you for your participation!