kosek

Nationale Koordination Seltene Krankheiten
Coordination nationale des maladies rares
Coordinazione nazionale malattie rare

RECOGNITION PROCESS CENTRE FOR RARE DISEASES

Questionnaire

Application for recognition as a kosek Centre for Rare Diseases

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Inhaltsverzeichnis

1. Centres for Rare Diseases: general instructions for the questionnaire

The application documents must be filled in and submitted via this electronic template. In addition to the electronic documents, an application letter with original signature must be sent by mail to the following address:

Kosek
Coordination Rare Diseases Switzerland
c/o Unimedsuisse
Haus der Akademien Laupenstrasse 7
Postfach
3001 Bern

- → The requested enclosures must be submitted in electronic format.
- → Applicants can complete the questionnaire in German, French or English, as they wish. Once chosen, please maintain your language choice within the procedure as much as possible.
- → The specifications for the maximum number of characters are to be understood as "characters with spaces".
- → The required support documents are summarised in a table. Please enter the titles of your support documents and number your attachments according to this table.

Note: all questions must be answered (exception: applicants which do not have a fully implemented Centre for Rare Diseases do not have to answer questions 22, 23 and 25). Any incomplete or missing documents will be returned and the application will need to be resubmitted.

For simplification purposes, the Centre for Rare Diseases will be abbreviated as CRD.

*= core criterion

2. Centres for Rare Diseases: Questionnaire

2.1. Questionnaire: general information

1. Name of the institution(s) that coordinate(s) the Centre for Rare Diseases (CRD) *

(Maximum characters: 250)
Explanatory note: Centres for Rare Diseases can be coordinated by different institutions (multi-site, e.g. adult hospital and paediatric hospital) or by one institution (single-site). In any case, the CRD must coordinate paediatric as well as adult health care.

2. Medical Director(s) or Directorate(s) of your institution(s)*

(Maximum characters: 250)

3. Name of the Centre for Rare Diseases (CRD) *

(Maximum characters: 250)

4. Address(es) of the CRD (address(es), email(s), telephone number(s)) *

(Maximum characters: 250)

5. Name and contact details (email address(es) and telephone(s)) of the contact person(s) of the CRD*

(Maximum characters: 250)

6. Is your centre part of the CRD coordination group? * *(please tick the appropriate answer)*

☐ Yes ☐ No

Explanatory note: The coordination group for Centres for Rare Diseases is a Swiss national group constituted by the coordinator of each CRD in Switzerland.

7. Do you allow kosek to transfer your application to Orphanet, once your centre is recognized, in order to keep the Orphanet database updated? *(please tick the appropriate answer)*

☐ Yes ☐ No

Explanatory note: kosek works in collaboration with Orphanet Switzerland in order to simplify and to document the recognition processes.

2.2. Management and coordination of the centre

8. Does your institution have a coordinator for the Centre for Rare Diseases? *(please tick the appropriate answer)*

☐ Yes ☐ No

Explanatory note: the coordinator/contact person of the CRD is a member of the CRD staff. His or her role is to represent the CRD on a national and international level and maintain the contact with kosek.

a. If yes, please give his/her name and contact details (email address, telephone number and address)

(Maximum characters: 250)

b. If no, is your institution considering employing a coordinator? *(please tick the appropriate answer)*

☐ Yes ☐ No

c. In what time-frame does your institution plan to do it?

(Maximum characters: 250)

9. Tick the main activities of the CRD.

Explanatory note: The compulsory activities are listed first and marked with an asterisk. All activities can be distributed within the staff of the CRD (exception: the coordinator of the CRD remains the contact point for kosek).

- ☐ Coordination of diagnostic processes. *
- ☐ Coordination of clinical follow-up*
- ☐ Contact person for kosek*
- ☐ Drafting and distribution of the annual report (with statistics) to kosek and Orphanet *
- ☐ Coordination of an exchange between rare disease professionals (including all disciplines and reference centres / networks) within hospitals (rare disease boards). *
- ☐ Setting up and monitoring of CRD statistics *
- ☐ Coordination of patient registration with Orpha codes, once implemented*
- ☐ Coordination of the co-operation with the Swiss Rare Disease Registry*
- ☐ Coordination of the recording and updating of activities (clinical, scientific, etc.) in Orphanet for the entire hospital (with collaborating hospitals each hospital has its own contact person). If necessary, contact Orphanet Switzerland
- ☐ Coordination with the helpline(s) and existing information portals for rare diseases
- ☐ Information and promotion of intra-hospital co-operation concerning rare diseases (communications, newsletters, conferences, etc.)
- ☐ Management of the CRD team
- ☐ Editorial responsibility for the CRD website
- ☐ Coordination of the CRD's further training programmes
- ☐ Provision of useful resources/documents for the paediatric-adult transition for rare diseases in general
- ☐ Other *(please specify)*:

10. Is the directorate of your institution willing to ensure financial continuity for your centre? *
(please tick the appropriate answer)

☐ Yes ☐ No

a. If no, what strategy does your institution pursue to ensure sustainability of the CRD? *

(Maximum characters: 1000)

11. Is a specific budget for your centre allocated? **(please tick the appropriate answer)*

☐ Yes ☐ No

a. If yes, what is included in this budget (in terms of content and duration)?

(Maximum characters: 1000)

12. If your centre is already operational and funded, please fill in the template on your workforce (with training/qualification and working time percentage) *

ADD A DOCUMENT
Please provide an organisational chart and fill in the template with the current workforce of your Centre for Rare Diseases (see *3. Checklist of the documents to enclose with the application*).

a. Comments on the situation (contracts - fixed-terms vs permanent, changes, positions, etc.)

Explanatory note: the answer for this field is not mandatory

(Maximum characters: 1000)

13. What links and co-operation does your centre have with other Centres for Rare Diseases at national level?

(Maximum characters: 1000)
Explanatory note: Please describe only activities and collaborative projects concerning care (diagnostic processes and general patient pathways), further training, research, information and administration your centre pursues in collaboration with other national Centres for Rare Diseases. Please do not include disease specific collaborations.

14. What links and cooperation does your centre have with other Centres for Rare Diseases at European or other international level?

(Maximum characters: 1000)
Explanatory note: Please describe only activities and collaborative projects concerning care (diagnostic processes and general patient pathways), further training, research, information and administration your centre pursues in collaboration with other Centres for Rare Diseases in Europe and the world. Please do not include disease specific collaborations.

15. Which patient organisation(s) does your centre engage with and what kind of collaboration do you pursue? *

(Maximum characters: 1000)
Explanatory note: please name mainly associations on rare diseases on the general level (umbrella organisations like ProRaris, Maraval, Associazione Malattie Genettiche Rare Svizzera Italiana, Unirares). Patients organisations which are oriented on specific rare diseases do not have to be listed exhaustively, as the collaboration with these lies within the responsibility of the clinics and reference centres).

2.3. Clinical health care

16. Does your centre have one or several entry points that accommodate and coordinate the clinical health care for patients without a diagnosis? * *(please tick the appropriate answer)*

☐ Yes ☐ No

a. Please give the location, address, email and phone number of the entry point(s).

(Maximum characters: 250)

b. If no, in what time-frame will it be set up? *

(Maximum characters: 250)

17. Has your centre implemented a structure or structures providing a multidisciplinary approach? * *(please tick the appropriate answer)*

☐ Yes ☐ No

a. If yes, please describe your structure(s)

(Maximum characters: 2000).
Explanatory note: structures like rare disease boards, genetic consultations, round tables, etc., ensuring that the patient has access to the most competent professional(s).

ADD A DOCUMENT
Please add the composition of your rare disease structure(s) (with permanent members), agenda and, if existing, a template of documentation of their decisions (see *3. Checklist of the documents to enclose with the application*).

18. Does your CRD have a structured pathway for patients without a diagnosis? *
(*please tick the appropriate answer.)*

☐ Yes ☐ No

a. If yes, please describe the pathway to diagnosis and its follow-up. *

(Maximum characters: 1000)

b. If no, in what time-frame will it be set up? *

(Maximum characters: 250)

ADD A DOCUMENT
Please provide a flowchart of the consultation (see *3. Checklist of the documents to enclose with the application*).

19. Does your institution have any specific technical platform for diagnosis and/or for medical management?

☐ Yes ☐ No

a. If yes, please tick the technical platform(s) you have in place in your institution *(several answers possible)*

- ☐ Diagnostic
- ☐ Laboratories
- ☐ Pathological laboratories
- ☐ Specialised radiological investigations
- ☐ Access to a genetic laboratory
- ☐ Trained geneticist in your organisation
- ☐ Other (*please specify*):

20. Does your centre use Orphacodes? **(please tick the appropriate answer)*

☐ Yes ☐ No

a. If no, is your centre considering implementing them? **(please tick the appropriate answer)*

☐ Yes ☐ No

b. If yes, in what time-frame? *

(Maximum characters: 250)

21. Does your centre collect data on patients without a diagnosis or with a suspicion of a rare disease? *(please tick the appropriate answer)*

☐ Yes ☐ No

a. What coding system(s) for patients without a diagnosis or with a suspicion of a rare disease does your centre use? *(please tick the appropriate answer(s) – several answers possible)*

☐ Human Phenotype Ontology

☐ Orphacodes

☐ Other *(please specify)*:

b. If you do not use a coding system for patients without a diagnosis, is your centre considering implementing one? *(please tick the appropriate answer)*

☐ Yes ☐ No

c. If yes, which one and in what time-frame?

(Maximum characters: 1000)

22. What is the total number of patients seen for diagnostic clarification and second opinion in the centre in the past 12 months?

Explanatory note: This number does not include all patients seen in the hospital but only those that went through the CRD. For institutions, that haven't implemented their centre(s) for rare diseases yet, please go to question 24.

(Maximum characters: 1000)

23. Please fill in the table in the appendix I (p. 21) in order to provide an overview of the origins of the patients in your Centre for Rare Diseases

Explanatory note: Institutions that haven't implemented their centre(s) yet, do not have to fill out the table in the appendix I, p. 21.

24. For institutions, that haven't implemented their Centre(s) for Rare Diseases yet, please describe how you will collect the number of these patients in your centre in the future. *

(Maximum characters: 1000)

25. What is the total number of diagnoses you could make during the past 12 months in your Centre for Rare Diseases?

(Maximum characters: 250)
Explanatory note: For institutions that haven't implemented their centre(s) yet, please skip this question (including a. b, and c.)

a. What is the total number of **rare disease** diagnoses you made during the past 12 months in your Centre for Rare Diseases?

(Maximum characters: 250)

b. What is the total number of **other diagnoses** (other than rare diseases) you made during the past 12 months in your Centre for Rare Diseases?

(Maximum characters: 250)

c. What is the total number of patients without a diagnosis during the past 12 months in your Centre for Rare Diseases?

(Maximum characters: 250)

26. Does your CRD organise psychological and/or social support?
(please tick the appropriate answer)

☐ Yes ☐ No

a. If yes, please describe how the CRD implements psychosocial support

(Maximum characters: 1000)

b. If no, do you consider implementing it? *(please tick the appropriate answer)*

☐ Yes ☐ No

c. If yes, in what form and time-frame do you consider implementing it?

(Maximum characters: 250)

27. Does/do your institution(s) have a transcultural translation service? *(please tick the appropriate answer)*

☐ Yes ☐ No

28. Does/do your institution(s) have an internal quality control system (at hospital level)?
(please tick the appropriate answer)

☐ Yes ☐ No

a. Please name the head of the unit

(Maximum characters: 250)

b. Please tick the systems used and the external bodies that certify the quality of your institution(s) (i.e. syst. ANQ, syst. ISO, …) *(several answers possible)*

Explanatory note: the systems and bodies measure the general quality assurance and apply on a general hospital level

- ☐ ANQ (Swiss National Association for Quality Development in Hospitals and Clinics)
- ☐ IQM (Initiative Quality Medicine)
- ☐ ISO (International Organization for Standardization)
- ☐ SanaCERT (Swiss Foundation for the Certification of Quality Assurance in Health Care)
- ☐ JCI (Joint Commission International)
- ☐ EFQM (European Foundation for Quality Management)
- ☐ SAS Accredited lab (Swiss Accreditation Service)
- ☐ FOPH lab (Federal Office for Public Health)
- ☐ EQA (External Quality Assessment)
- ☐ Other *(please specify)*:

29. Does your centre measure any quality indicators specifically for your rare disease activities within your CRD? *(please tick the appropriate answer)*

☐ Yes ☐ No

a. If yes, please describe them

(Maximum characters: 1000)

30. Does your centre participate in the Swiss Rare Disease Registry (once it is installed)? * *(please tick the appropriate answer)*

☐ Yes ☐ No

a. If no, in what time-frame are you considering participating?

(Maximum characters: 250)

31. Does your centre have a general transition concept? *(please tick the appropriate answer)*
Explanatory note: the term "transition" means here the shift from paediatric to adult medicine.

☐ Yes ☐ No

a. If yes, please describe your concept, the tools and your use of them.

(Maximum characters: 1000)

b. If your centre does not have a transition concept (yet), in what form and time-frame are you considering implementing one?

(Maximum characters: 250)

2.4. Information and support

32. Does your centre have or participate in a single point of contact for patients and professionals that can provide information, support and orientation within a short time (e.g. helpline, e-mail, contact form, etc.) *(please tick the appropriate answer)*

☐ Yes ☐ No

33. Does your centre have or participate in a helpline? *(please tick the appropriate answer)*

☐ Yes ☐ No

a. If yes, please name the helpline and describe its organisation and functioning, including workforce, statistics, activity report, etc.

(Max. characters: 1000)

34. Is your helpline member of the European Network of Rare Diseases Helplines (Eurordis)? *(please tick the appropriate answer)*

☐ Yes ☐ No

35. Does your centre have a structured communication system on rare diseases within your hospital/ institution? *(please tick the appropriate answer)*

☐ Yes ☐ No

a. If yes, which one(s)? *(please tick the appropriate answer(s), several answers possible)*

☐ Intranet

☐ Newsletter

☐ Special events

☐ Other *(please specify)*:

b. If no, do you plan to establish a structured communication system on rare diseases? *(please tick the appropriate answer)*

☐ Yes ☐ No

c. If yes, please describe which one(s) and in what time-frame

(Maximum characters: 250)

36. Does your centre have other structured communication systems on rare diseases outside the hospital/institution (care partners or greater public)? *(please tick the appropriate answer)*

☐ Yes ☐ No

a. If yes, which one(s)? *(please tick the appropriate answer(s), several answers possible)*

☐ Internet/Webpage

☐ YouTube Channel

☐ Facebook /Twitter /Instagram specific page

☐ Newsletter

☐ Regular column in the mass media: which one(s)? *(please specify)*:

☐ Seminars or public presentations or conferences

☐ Special events (e.g. rare disease day)

☐ Other *(please specify)*:

37. Does your centre publish on rare diseases in journals (publications, articles, presentations, etc.)? *(please tick the appropriate answer and enclose the list of publications of the centre for the past 24 months)*

☐ Yes ☐ No

Explanatory note: the publications concern rare diseases in general, specifically diagnostic processes or patient pathways. They do not concern a specific rare disease or rare disease group.

ADD A DOCUMENT

Please provide a list of the publications and communications of your Centre for Rare Diseases as a separate document (see *3. Checklist of the documents to enclose with the application*)

Explanatory note: Please list only the publications of the Centre for rare diseases

2.5. Further and continuing training

38. Does your centre provide and/or coordinate further training on rare diseases in general? *(please tick the appropriate answer)*

☐ Yes ☐ No

a. If yes, please describe the further training of the past 12 months (including the number of participants, if the training was ISFM-approved or approved by professional society/societies)

(Maximum characters: 1000)

b. If your centre does not have further training yet, does your centre wish to implement it? *(please tick the appropriate answer)*

☐ Yes ☐ No

c. If yes, please describe your approach

(Maximum characters: 1000)

2.6. Research

39. Does your centre have or coordinate any research activity about rare diseases at the Centre for Rare Diseases? *(please tick the appropriate answer)*

☐ Yes ☐ No

a. If yes, please describe it

(Maximum characters: 1000)

2.7. Final comments

40. If you have a general comment or some elements to add to this questionnaire, you may do so in the comment section below

Explanatory note: This field is not mandatory and does not have to be answered.

(Max. characters: 1000)

3. Check-list of the documents to enclose with the application

Explanatory note: The documents, are all mandatory if not stated differently.

I. Organisational chart and list of the current workforce of the Centre for Rare Diseases (*See related question n°12, p. 7. See template in a separate document)*
II. Composition of your rare disease structure(s) (with permanent members), agenda, and if existing a template of documentation of their decisions (*See related question n°17, p. 10*)
III. Flowchart of the consultation procedures (*See related question n°18, p. 10-11*)
IV. List of publications and communications of the Centre for the past 24 months (if applicable – *see related question n°37, p. 18*) *Explanatory note: the publications and communications concern rare diseases in general, not a specific rare disease or rare disease group. Please list only the publications and communications of the Centre for Rare Diseases*
V. Commitment letter from your institution signed by the medical director or directorate *(see templates in German and in French in a separate document. Please send your letter in French OR German*)

Appendix I: Table of the number of patients seen in the CRD by origin in the last year

Patient's canton of residence	Number of patients (absolute number)
AG	
AI	
AR	
BE	
BL	
BS	
FR	
GE	
GL	
GR	
JU	
LU	
NE	
NW	
OW	
SG	
SH	
SO	
SZ	
TG	
TI	
UR	
VD	
VS	
ZG	
ZH	
Switzerland	
Foreign country/countries	