

QUESTIONNAIRE

FOR THE DESIGNATION OF

ASSOCIATED CENTRES FOR RARE METABOLIC DISEASES

Author: Extended core group for the pilot project on rare metabolic diseases

Date: 02.03.2021

This document was elaborated with the collaboration of kosek in the context of its pilot project on rare metabolic diseases. This document will help the SGIEM answer kosek's questionnaire II, concerning the network, of the recognition process.



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A. Definition and core criteria of an Associated Centre (AC):

- I. Clinical health care:
Associated Centres can treat metabolic patients by themselves and follow them without much input from the Reference Centre. Associated Centres have a specialty consultation for inborn errors of metabolism (IEM) run by at least one professional that should have received training in the care of IEM patients, ideally under supervision of a fully trained paediatric or adult IEM specialist, has dedicated worktime for it and attends regularly metabolic meetings.

Associated Centres offer emergency on-call service and emergency treatment, as well as consultations of specialized dieticians. Associated Centres also have to ensure transition from paediatrics to adult medicine.
- II. Continued training:
Associated Centres should provide some general training in IEM for the hospital staff, but do NOT have to train specialists in IEM.
- III. Research activities:
Associated Centres actively participate in research projects of Reference Centre (potentially all sites).
- IV. Volume of activity:
No minimum activity level is required to define an Associated Centre. The volume of activity gives an overview about where patients are treated, in order to have a full picture of the care provision landscape in Switzerland. It is needed for documentation purposes.

B. Instructions for completing the questionnaire

All questions must be answered, either in English, French or German. Incomplete questionnaires will be returned.

Your answers should be as precise as possible.

Please use continuous text without line breaks or bullet points, this helps with an efficient evaluation of the forms.

Please submit the questionnaire with the requested annexes, inclusive an application letter from your institution signed by the medical director or directorate (*see template attached*) via e-mail (electronic format) to the following address:

Network.iem@kosekschweiz.ch

Please submit the complete application with all related documents by **22nd March 2021**.

Thank you.

For any question, you may contact us via e-mail (see e-mail address above).



C. General information about the candidate structure

1. Name of the institution(s) responsible for the candidate Associated Centre

2. Name(s) of the medical director(s) or directorate(s) of your institution(s)

3. Name of the candidate Associated Centre

4. What clinics/units constitute your Associated Centre? Please list them

5. Address of the Associated Centre (including general email and telephone number)

6. Name and contact details of the coordinator/head of the Associated Centre (email address and telephone)

Explanatory note: the coordinator is responsible for the good functioning of the Associated Centre and coordinates the clinical health care of patients.



7. For which diseases / (main) groups of diseases is your structure a candidate for designation by SGIEM? Please list them.

Explanatory note: SGIEM works with the classification of rare diseases as used in the European Reference Networks (ERNs) and provided by Orphanet. The diseases are listed in the Excel sheet sent as a separate document (see Excel sheet ERN disease group). Please note that the document has different tabs.

Whenever possible, please list main groups of diseases. Please avoid listing diseases singularly (column G and onwards in the attached Excel sheet). If you need help with this specific question, please contact network.iem@kosekschweiz.ch before answering.

ADD A DOCUMENT: Please list the diseases your structure is covering in a separate document (**Annex I.1**).

8. To which Reference Centre Site is your structure associated?



D. Clinical health care within your health care structure

9. Please describe how your candidate Associated Centre ensures multidisciplinary clinical health care for its patients with regard to

a. Diagnostics and therapy

b. Follow-up of the patient



- 10.** Please describe the non-medical services or infrastructures for patients in your Associated Centre (i.e. care management, dietary consultation, psychosocial services, etc.) that are specific to the disease group your centre is candidate for.

- 11.** How is the collaboration with the Reference Centre (RC) organised? Describe your collaborations with the different RC Sites? How is the supervision organised? Please describe it.



12. What forms of cooperation does your structure have at a regional / local level? Please describe the stakeholders and details of the cooperation.

Explanatory note: the goal of this question is to know whether and how the Associated Centre works with local family doctors, local specialists, local hospitals, etc.

13. Does your candidate Associated Centre have a rare disease board that is specifically for the disease group you are providing care for?

Explanatory note: the RD board does not have to be based in your institution. It can be organised within the national network / at a national or international level.

- Yes
- No

a. If yes, please describe how it functions, its regular members/specialties, and its decision-making procedure.



- 14.** Is/are there (a) specific platform(s) within your institution that your candidate structure uses to perform all necessary disease-related analyses for diagnosis and medical management?

Explanatory note: the word «platform» is to be understood as an infrastructure within your institution.

- Yes
- No

- a.** If yes, please tick the platform(s) that is/are in place in your institution (*several answers possible*):

- Biomedical/laboratories
- Genetic
- Pathological
- Other (*please specify*):

- b.** Briefly describe the specific platform(s) in regard to IEM care.

- 15.** What coding system does your structure use, to code the patients' diseases? (*please tick the appropriate answer; several answers possible*)

- Orphacode
- ICD-10
- Human Phenotype Ontology (HPO)
- Other (*please specify*):



16. Does your candidate structure follow existing disease-specific national or international guidelines and recommendations for the clinical health care of the patients?

- Yes
- No

a. If yes, please list them and specify if they are national or international.

17. Optional question: Is your structure regularly involved in activities in your area of IEM, such as the preparation of guidelines or recommendation for the clinical health care of the patients?

- Yes
- No

a. If yes, please list them and specify if they are national or international.



18. If the complexity of the disease makes it necessary, does your candidate Associated Centre have the required infrastructure (i.e. day hospital) and staff (i.e. a case manager) at your disposal?

- Yes
- No

a. If yes, please describe briefly the infrastructure and staff and how it is organised (including laboratories and genetics)

19. Does your candidate structure organise a 24/7 health care coverage for your patients with a rare disease?

- Yes
- No

a. If yes, please describe how it is organised (who is involved, procedure, etc.), including all available services that have to be in place in your centre (specialists, laboratories, products, non-medical staff, therapies, etc.)

Explanatory note: a 24/7 coverage can be organised within a network (or) at a national level. This question might be a core criterion for some diseases, but not for all, as not all diseases within a disease group require a 24/7 health care coverage.

20. How do you provide emergency care for your IEM patients? Describe whether your candidate institution has an emergency care unit and arrangements for patient transfers

ADD A DOCUMENT: please add a document, if you have a description of the emergency process or a tool available (**Annex I.2**).

21. **Optional question:** During consultations with patients, do professionals in your candidate structure share information about patient organisations with patients? (i.e. brochures, address of patient organisations/groups, etc.)?

- Yes
 No

E. Continuing training of the candidate structure

22. Does your centre provide general continuing training on inborn errors of metabolism?

- Yes
 No

ADD A DOCUMENT: Please list the **continuing training provided by your candidate centre** in a separate document. Please specify the target audience of the training (medical or non-medical staff) and at what level it took place (local/regional, national or international) (**Annex I.3**).



23. Does the staff in your structure receive regular continuing training?

- Yes
- No

ADD A DOCUMENT: Please list the **continuing training your staff receives** in a separate document. Please indicate whether the training was provided at a local/regional, a national (i.e. within an existing network) or an international level (**Annex I.4**).

F. Research activities

ADD A DOCUMENT: please list in one document the peer-reviewed publications of your centre for the past five years, the current research projects and clinical trials of your centre, the main competitive and / or non-competitive grants and awards/recognitions received in the five last years. (**Annex I.5**).

24. Has your candidate centre published on IEM in peer-reviewed journals in the last five years?

- Yes
- No

25. Does your candidate centre (currently) participate in research projects of a Reference Centre Site for the specific rare diseases / (main) groups the centre provides for?

- Yes
- No

26. Does your candidate centre (currently) take part in clinical trials?

- Yes
- No



27. Does your candidate centre actively take part in disease specific registers by entering data?

- Yes
- No

a. In which disease-related registers does your centre enter data actively? Please provide a list.

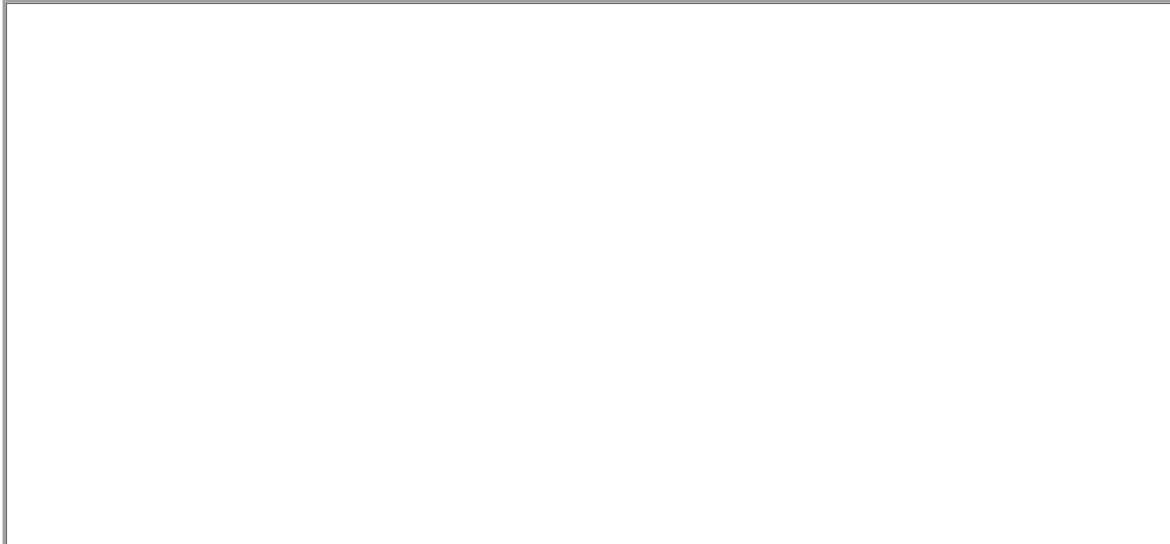
28. Does your candidate structure actively take part in the Swiss Rare Disease Registry by regularly entering data?

- Yes
- No

G. Coordination and administration

29. Please describe the structure of your candidate Associated Centre (organisation chart, names, functions and work percentage of the regular staff of the structure).

Explanatory note: You may add flowcharts, figures or tables if they help illustrate your answer.

A large, empty rectangular box with a thin black border, intended for the respondent to provide a detailed description of their candidate Associated Centre's structure, including an organisation chart, names, functions, and work percentages of regular staff.

ADD A DOCUMENT: If available, please add a description of the structure of your candidate centre (organisation chart, functions and staff of the centre etc.) (**Annex I.6**).

30. What is included in the budget of your structure? Does the budget of your structure cover specific IEM care (e.g. dedicated worktime for specialist)?

A large, empty rectangular box with a thin black border, intended for the respondent to describe the budget of their structure and whether it covers specific IEM care, such as dedicated worktime for a specialist.



H. Documentation in Orphanet

31. Are your structure's activities registered in the Orphanet database?

- Yes
- No

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

- Expert centres
- Research activities
- Clinical trials
- Diagnostic tests
- Registries
- Your centre's cooperation with patient organisations

32. Do you allow SGIEM to transfer your application to Orphanet in order to keep the Orphanet database updated?

- Yes
- No

33. If your structure fulfils the criteria for being an Associated Centre as defined above and documented in this questionnaire, the SGIEM will acknowledge this label and your structure will appear as such on the Orphanet (in the SGIEM network section) and on the SGIEM webpage. Do you agree?

- Yes
- No

I. Volume of activity

34. Total number of patients seen last year (absolute number)

35. Number of new cases last year (absolute number)



J. General remarks

36. 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 optional.

Checklist of the documents to enclose with the application

- | | |
|------------|---|
| Annex I.1 | List of the diseases the health care structure is covering <i>(see separate Excel sheet of the ERN disease group)</i> |
| Annex I.2 | Description of the emergency process or an emergency tool, if existing |
| Annex I.3 | List of the continuing training provided by your candidate structure , including target audience of the training and level (local/regional, national or international) |
| Annex I.4 | List of the continuing training your staff has received , including level of training (local/regional, national or international level). |
| Annex I.5. | List of the peer-reviewed publications of your structure for the past five years, the current research projects and clinical trials your structure is involved with, the main competitive and/or non-competitive grants and awards/ recognitions received in the five last years |
| Annex I.6. | Description of the structure of your candidate centre (organisation chart, functions and staff of the centre, etc), if available |
| Annex I.7. | Application letter from your institution signed by the medical director or directorate <i>(see templates in German and in French attached) Please add your letter in one of these languages or in English).</i> |