

QUESTIONNAIRE

FOR THE DESIGNATION OF

CARE FACILITIES FOR RARE METABOLIC DISEASES

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

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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 a Care Facility (CF):

- I. Clinical health care:
Patient care should be done in collaboration with or under supervision of a Reference Centre Site as agreed with the latter. Regular patient consultations have to be done, but not mandatorily in a specialty consultation, but rather as a local partner of the lead institution (Reference Centre Site). Description of the integration into the local and regional network is important.
- II. Continued training:
Regular participation in national continuing training offered by the SGIEM and/or other opportunities (congresses, meetings etc).
- III. Research activities:
Participation to research activity is optional but not mandatory for inborn errors of metabolism (IEM).
- IV. Volume of activity:
No minimum activity level is required to define a Care Facility. The volume of activity gives an overview about where patients are treated. 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 Care Facility

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

3. Name of the Care Facility

4. What clinics/units constitute your Care Facility? Please list them

5. Address of the Care Facility (including general email and telephone number)

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

***Explanatory note:** the coordinator is responsible for the good functioning of the Care Facility 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 Care Facility 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 Care Facility (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 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 Care Facility works with local family doctors, local specialists, local hospitals, etc.

13. 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):*



14. 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**).

15. **Optional question:** During consultations with patients, do professionals in your Care Facility structure share information about patient organisations with patients? (i.e. brochures, address of patient organisations/groups, etc.)? *(please tick the appropriate answer)*
- Yes
- No

Explanatory note: This question is optional.

E. Continuing training of the candidate structure

16. Does the staff in your structure receive regular continuing training? *(please tick the appropriate answer)*
- 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.3**).



F. Research activities

17. Does your candidate structure actively take part in the Swiss Rare Disease Registry by regularly entering data? *(please tick the appropriate answer)*
- Yes
 - No

G. Documentation in Orphanet

18. Are your structure's activities registered in the Orphanet database? *(please tick the appropriate answer)*
- 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
19. Do you allow SGIEM to transfer your application to Orphanet in order to keep the Orphanet database updated? *(please tick the appropriate answer)*
- Yes
 - No
20. If your structure fulfils the criteria for being a Care Facility 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? *(please tick the appropriate answer)*
- Yes
 - No



H. Volume of activity

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

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

I. General remarks

23. 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 your staff receives , including level of training (local/regional, national or international level). |
| Annex I.4 | 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> |