kosek

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

DEFINITION AND CONCEPT OF A CENTRE FOR RARE DISEASES

A process for the recognition of health care structures by kosek

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1. Background

As the national coordinating body for rare diseases, kosek is responsible for recognising institutions according to certain criteria. The aim is to improve the health care provided to patients.

This document defines the concept and process in order to be recognised as a Centre for Rare Diseases by kosek. The definition of a Centre for Rare Diseases is drawn from the detailed concept design (upcoming document).

The key terms are explained below and apply to the recognition procedures.

The structures reflect two different levels:

1. If the diagnosis of a rare disease is known, patients will be cared for at a disease-specific level, i.e. in reference centres and care networks.
2. If there is no diagnosis, or if the patient is an isolated case (meaning the patient cannot be assigned to a specific disease or disease group, and therefore cannot be cared for at any disease-specific level), or the patient does not know which institution he or she should turn to, then clarifications are made on a cross-disease level: Centres for Rare Diseases constitute this level.

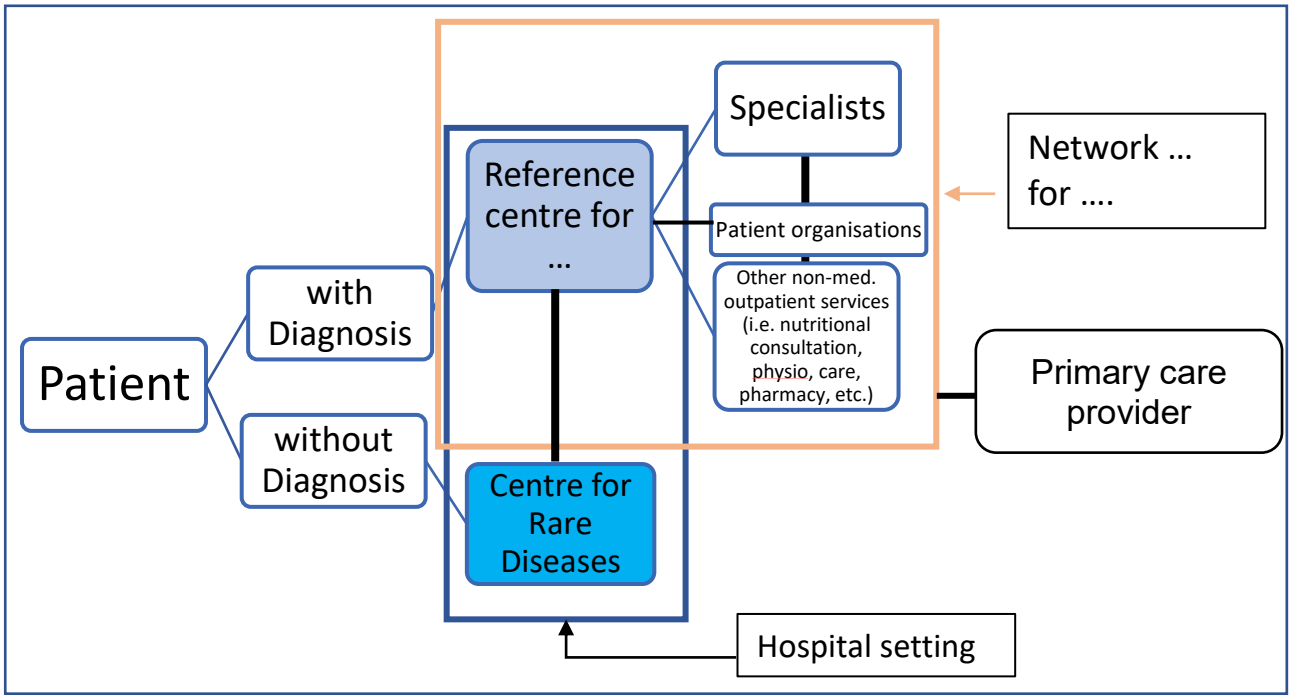

Figure 1: Overview of the health care landscape

In the case of patients without diagnosis, organisational structures known as **Centres for Rare Diseases** shall provide care, as shown in the diagram above:

This health care concept provides patients with alternative structures, according to the status of their diagnosis. The diagram also illustrates the network which is built around reference centres. Importance is laid on the coordination of health care between the various participating members of the network, which remains open to integrate expertise from outside the reference centres.

Since both reference centres and multi-disciplinary Centres for Rare Diseases need to provide a range of services that hospitals offer conjunctually (i.e. common to all cases) (e.g. complaint management, quality management, support for family members), it makes sense to integrate them into the hospital setting.

The aim of the health care organised on this basis is to ensure that patients are assigned as quickly as possible to the right specialists, who possess the appropriate disease-specific knowledge.

1.1. Guiding principles behind the recognition process

The project of the recognition of healthcare structures is based on a number of parameters which are context-sensitive:

1. Switzerland does not have a legal framework that applies specifically to rare diseases, and which enables specific planning, or the designation of service providers and related contracts. The general legal provisions for health care therefore apply – whether at national, cantonal or intercantonal levels. The consequence of this is that recognition of structures does not constitute binding official service contracts which oblige suitable service providers to supply these services, while excluding others. To the contrary, recognition entails more a consensual agreement between service providers in the field of rare diseases, to commit themselves, in cooperation with their network partners, to ensuring good health care. The creation of networks and structures cannot therefore be a legal requirement, and similarly, recognition decisions cannot be subject to legal challenge.

   The linking of service providers into networks and the recognition of structures (reference centres and Centres for Rare Diseases) are based on the degree to which those involved are willing to coordinate care throughout Switzerland. Service providers can and should therefore jointly decide how to implement the networks and care structures. This corresponds to a "bottom-up" approach, which gives participants the necessary flexibility, self-organisation and co-responsibility in the structure-building process, so that any differences in the current organisation of health care and existing structures can be taken into account.

2. This project follows a participatory approach. The service providers involved jointly coordinate the care of patients with a rare disease and formulate this in a mutually satisfactory agreement (a so-called "gentlemen's agreement").
   Participation includes patients, whose role goes beyond that of passive subjects, to that of central to the field of rare diseases. This participation is enacted through patients being represented, mainly through patient umbrella organisations, throughout the process.

3. Recognition of the structures thus created is not certification or accreditation (which would require an official accreditation body).

2. Centres for Rare Diseases: Definition

Centres for Rare Diseases are cross-disease care structures, or units which fulfil the function of interdisciplinary contact points for clarification and coordination of treatments, as well as performing information tasks and coordinating specialists (SAMS, 2016).

Centres for rare diseases are thus multi-disease centres which provide patients with a diagnosis as quickly as possible, and then refer them to the appropriate disease-specific network or reference centre. In this function, Centres for Rare Diseases play an important central hub role in the health care system. The centres should provide patients with information regarding referrals for their case, and serve as cross-disease centres, which bring together specialists from different fields in the one institution.

Centres for Rare Diseases need the right kind of expertise, an organisational environment, and a minimal infrastructure. See the description of tasks under Chapter 2.1.

The Centres for Rare Diseases function as interdisciplinary and cross-disease contact points – primarily for patients with no diagnosis. Centres for Rare Diseases should not be seen as separate organisational units of a hospital, with their own beds, but rather as a structure through which the expertise of a hospital in different fields can flow. Centres for rare diseases do however need a small

core team that acts as a contact point, organises interdisciplinary work, is responsible for the continuity of services, and, last but not least, coordinates with hospital internal and external players.

2.1. Responsibilities of the Centres for Rare Diseases

The Centres for Rare Diseases must fulfill five core tasks (see Figure 2).

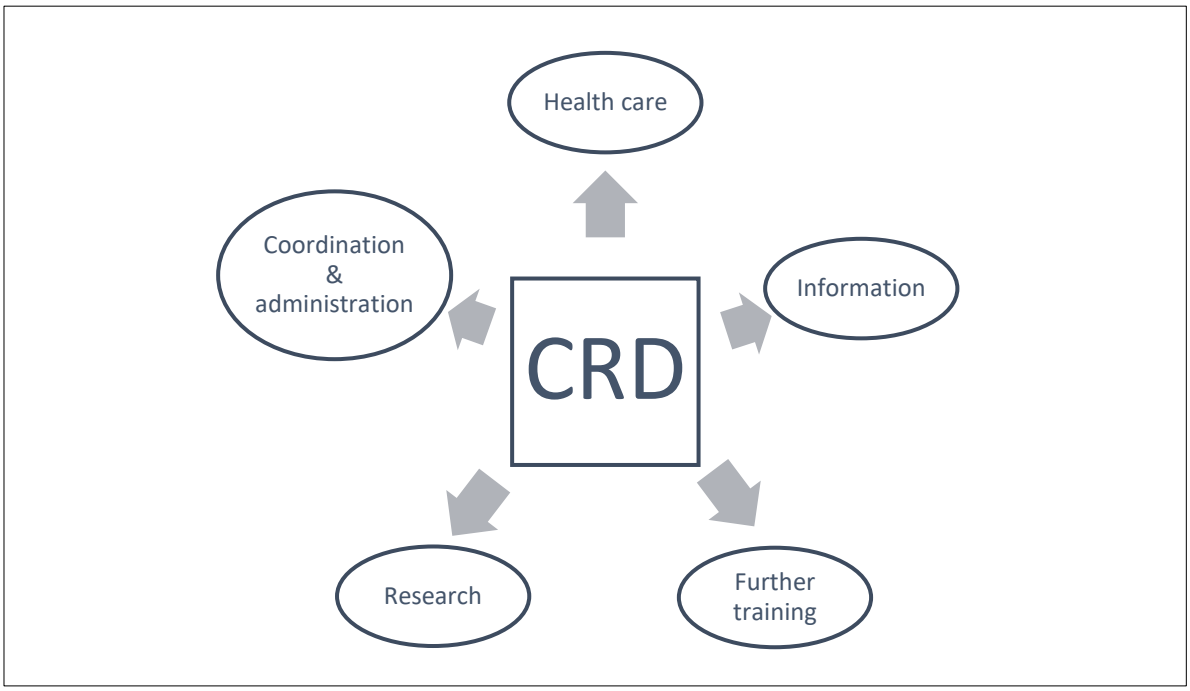

Fig. 2: Core tasks of the Centres for Rare Diseases (CRD)

a) Coordination and administration

" Coordination and administration " is a core function which brings together the activities of the Centre for Rare Diseases. It ensures the continuity and sustainability of the structure and profiles the role of the Centre for Rare Diseases within the health care system. In order to ensure a high level of quality of care, coordination is needed within the hospital, between the Centres for Rare Diseases and reference centres, and between individual experts and related services.

At the same time, a nation-wide level of cooperation between the Centres for Rare Diseases, as cross-disease contact points, is also foreseen, facilitated through a contact person from each Centre for Rare Diseases. Whether there is a need within Switzerland for a "leading house", to coordinate the cooperation centrally, is still an open question, and yet to be agreed between the Centres for Rare Diseases. The roles can be jointly defined and allocated at national level, so that a broad sharing of tasks is possible. Co-ordination topics include, but are not limited to:

- Implementation of target group-specific information packages (e.g. helplines, homepages, patient brochures)
- Education, training and further training on rare diseases
- Research cooperation
- Allocation of common tasks
- Defining meaningful patient paths.

Administration and coordination also covers the involvement of major patient groups or their umbrella organisations. A structured contact with patient organisations for rare diseases is therefore also a

criterion for the recognition of Centres for Rare Diseases. This can be done, for example, by an advisory committee with patient representatives participating.

The introduction of Orphacode as a common coding system for the documentation of rare disease cases should be followed personally by the directors of the Centres for Rare Diseases. The corresponding projects require close cooperation with those responsible for the clinic information system, IT services and medical coding. The Centre for Rare Diseases must be involved in this work so that the documentation facilitates cooperation within the hospital. At the same time, the Centre for Rare Diseases ensures the clinics and reference centres are also able to participate in the development of specific documentation. For co-ordination purposes, the various Centres for Rare Diseases should ideally use Orphacode coding tools that can be inter-connected.

The area of responsibility also includes interdisciplinary tasks, such as quality assurance and improvement, complaint management, maintaining expertise, and internal and external information. As in the case of direct health care services, these can be organised in conjunction with other services covering more than one illness. As each hospital is organised differently, no uniform organisational forms are required. This pragmatic approach makes it possible to manage the operation of a Centre for Rare Diseases efficiently in terms of expenditure.

b) Health care

Clinical services include initial and follow-up care for patients suspected of having a rare disease or without a diagnosis. The Centres for Rare Diseases organise the expertise between different disciplines. In order to ensure a correct diagnosis is arrived at, interdisciplinary boards are convened and/or consultations undertaken, and where necessary second opinions obtained. Central to the phase up to diagnosis is the function of care management, which involves coordinating appointments and tests, so that those affected can keep their appointments as concentrated as possible, and the results of the findings are promptly and completely available to the responsible panel of experts.

Since rare diseases are not usually emergencies, but chronic diseases, there is no need for special emergency services or 24h/day coverage. The Centres for Rare Diseases therefore operate on a day-to-day basis, and any emergency or after-hours requirement can be covered by the hospital emergency ward.

Patients are usually referred to disease-specific centres of expertise after diagnosis, but Centres for Rare Diseases can coordinate initial treatment after diagnosis so that the patient is not exposed to any unnecessary gaps in treatment. The Centre for Rare Diseases also ensures that patients have access to concrete support options: e.g. disease-specific support, psycho-social services (psychological and social counselling, etc.), and overnight accommodation for parents of hospitalised children, or other accompanying persons. These services are organised by the hospital where the Centre for Rare Diseases is located.

Not all of the services provided by Centres for Rare Diseases apply exclusively to people with rare diseases, so the Centres for Rare Diseases may also coordinate and share resources with similarly located services. What is important is that the staff is aware of and trained in the needs of patients with rare diseases, including such factors as coordinating appointments, managing cases, and social counselling.

Another important aspect is the cooperation between paediatrics and adult medicine. This is ensured by the Centre for Rare Diseases itself, since each Centre for Rare Diseases has within it specialists from both paediatrics and adult medicine.

The requirements for Centres for Rare Diseases are described in the 2016 SAMS report. However, the professionals involved do not have to form their own organisational unit, and do not have to be exclusively responsible for rare diseases. What is important is an appropriate staffing level with clear responsibilities:

- A first contact person and coordinator who provides information and puts the patient in contact with the appropriate specialist (triage function, coordination of appointments/diagnostic tests)
- Medical paediatrics specialist
- Medical specialist for adult medicine
- A specialist in nursing or hospital social work
- A contact person for the kosek projects and coordinating committees shall be designated from among the professionals named above
- Contact and connection to a genetics laboratory which can carry out the necessary genetic analyses to clarify the diagnosis.

c) Information

The following are part of the information task:

- Information concerning own services (structures and expertise in the clinics of the institution)
- Co-ordination of the records and their updating with Orphanet
- Cooperation with or operation of a Helpline

Rare diseases also have a specific information task. Centres for rare diseases should inform patients about the services offered by their hospital and act as a hub. As they work at a cross-disease level, they must also be able to provide general information on rare diseases to interested parties, patients, relatives and professionals. They must make patients aware of the services they are entitled to (medical care and psychological support, social counselling, access to patient organisations, etc.). This requires a broad knowledge of the existing services and landscape in the field of rare diseases.

Helplines for rare diseases are also part of the information resources.

As an overarching unit of a hospital, Centres for Rare Diseases should coordinate and enter hospital records on Orphanet, and keep them updated on an annual basis. Orphanet therefore becomes a comprehensive instrument that fulfils its purposes as fully as possible and offers easy access to information.

d) Further training

Interdisciplinary education and training in rare diseases is one of the core activities of the Centres for Rare Diseases. It ensures that the Centres for Rare Diseases maintain a sufficient level in expertise, and that this expertise is passed on, and is sustainable in the long term. Thus, the Centres for Rare Diseases should carry out interdisciplinary training in the field of rare diseases, if possible, in co-ordination between the different Centres for Rare Diseases. The further training also makes it possible to network the specialists in this field, as it facilitates the exchange of specialist know-how and disseminates new research findings.

e) Research

Centres for Rare Diseases participate, to the extent of their possibilities, in clinical research, health care research and basic research, with the aim of improving diagnosis. This may include, for example, participation in national and international studies, as well as publication in scientific journals and the organisation of national and international conferences.
Research at the Centres for Rare Diseases is cross-disease oriented. Disease-specific research is undertaken at the corresponding reference centre / network

3. Centres for Rare Disease: Phases of the recognition process

There are a number of project phases in the recognition process. These are described in more detail below.

Project phase	Description	Est. time period
PRELIMINARY PHASE		**5-7 Months**
1 – Initiation of the cooperation between CRDs	Interested institutions are invited to a kickoff launch, i.e. to form a working group to coordinate the work between the Centres for Rare Diseases (self-organisation). Support by kosek	1 Month
2 – Formulation of services on offer, and coordination between the CRDs concerning common tasks	Self-organisation by the working group. Exchange of organisational models and allocation of roles between the interested institutions. Support by kosek Important: The members of the working group must inform their institutions/their governance and obtain the relevant permits (the process must be supported by the institution's governing authorities)	4-6 Months
APPLICATION PHASE		
3 – Application phase for recognition as a Centre for Rare Diseases	Individual Centres for Rare Diseases submit their application (questionnaires with operational requirements).	Deadline: Submission of the application: 30th September each year (An extension period of one month may be considered on written application to kosek, including reasons.)
4 - Evaluation of the application	kosek evaluates the applications. If clarification is needed, a dialogue is sought. Rejections are possible, leaving options for finding solutions. **Conclusion:** the Centre for Rare Diseases is assessed and deemed recognised / not recognised, or recognised subject to conditions.	Duration: 3-6 Months
COMMUNICATION PHASE		
5 – Documentation and publication of recognition	- Documentation of expert reports in Orphanet, - Transparent documentation procedures and decisions via kosek website, - Communication concerning recognition to the FOPH, - Publication and formulation of recognition decisions by FOPH and	Max. 2 months following the published decision.

	cantons (e.g. by means of GDK recommendations).	
6 – Reports	Annually, agreed with Orphanet	Annually, certainly by May the following year
7 – Audits/Re-evaluation	Re-evaluation of the institution by kosek	Every 4 years, as of the positive recognition decision

Fig. 3: Project phases for the recognition of Centres for Rare Diseases

Phase 1: Initiation of cooperation between Centres for Rare Diseases

This phase begins with a 'Kick-Off' Event, attended by representatives of the interested institutions. This phase is the official launch of a nationwide and multi-institutional cooperation between all Centres for Rare Diseases.

Centres for Rare Diseases can take the opportunity at this event to consider where they stand, and what they want to achieve together. kosek planned an initial exchange of experience and a description of the tasks that the Centres for Rare Diseases (collectively) need to accomplish.

Conclusion: Creation of Working Group Centres for Rare Diseases.

Phase 2: Design of service range and co-ordination between the CRDs concerning common tasks

The initiation phase is followed by the design phase. On the one hand, this is undertaken by the providers themselves, who formulate their respective centre service range within the framework conditions prevailing. The self-organisation of the Centres for Rare Diseases is to be discussed in the coordination group. This comprises:

- a discussion of roles and allocation of responsibilities
- definition and organisation of Helpline services

Role of kosek: procedural support, support for self-organisation.

Conclusion: Roles are defined and allocated, including executive positions as necessary. The preparation for the application phase is completed

Phase 3: Application for recognition as a Centre for Rare Diseases

Once the roles have been defined and allocated among the Centres for Rare Diseases, these can apply for recognition by kosek. For this purpose, kosek provides a recognition questionnaire that contains all operational requirements for the Centres for Rare Diseases. It is the responsibility of the hospitals wishing to apply to fill in the questionnaire as precisely and independently as possible, and to submit it to kosek within the specified timeframe.

At the initial application stage, Centres for Rare Diseases may not yet be able to satisfy all the requirements, or may only do so for some or all of them at a later date. These centres may still submit an application, but must explain how they intend to meet the criteria and in what timeframe. Recognition will then be granted on condition that the relevant requirements are met at the appropriate time.

Role of kosek: transparent communication of the framework conditions for recognition (procedures, organisation, timeframe and criteria), answering questions.

It is important that kosek only answers (concrete) questions on individual points of the application when the hospitals apply. The kosek office must not anticipate the evaluation.

Conclusion: the Centres for Rare Diseases have completed the recognition questionnaire and submitted their application for recognition to kosek within the specified time frame.

Phase 4: Evaluation of the Application

In this phase, all application dossiers are carefully analysed and assessed in detail so that recognition decisions can be made by kosek.

Where there are points that are problematic or not or only partially fulfilled, an interview will be called with the applicant structure. Applications for recognition of structures may be turned down, with justification. In such cases, the structures involved may seek other solutions, for which kosek may be involved.

The rejection of an application by kosek has no financial or legal consequences for the institution.

Role of kosek: Implementation of the recognition procedure. kosek alone is responsible for this procedure.

Conclusion: The Centres for Rare Diseases are assessed and recognised, or recognised under conditions, or not recognised. The health care providers have allocated the roles among themselves and coordinate their work. In the case of non-recognition, the decision and reasons are communicated clearly to the structure in question.

Phase 5: Documentation and publication of recognitions

Following recognition by kosek, the decision will be documented and communicated:

- On the kosek website: transparent communication procedures and decisions
- Inclusion in Orphanet database
- The centres themselves can communicate the recognition decision through their own communication channels.

The recognition will be acknowledged by the federal government, the GDK/CDS (Cantonal Health Directors Conference), and the cantons:

- Publication on the website of the Federal Office of Public Health (FOPH)
- The form of communication by the GDK/CDS and the cantons is yet to be decided. One option is for the GDK/CDS to draw up recommendations for the cantons on this subject.

Role of kosek: kosek is also responsible for this phase, and must therefore inform all partners involved in the dissemination or officialization of the recognition promptly of the decision.

Conclusion: the recognition decision has been officially communicated and is visible on the various communication platforms. The recognition is acknowledged by FOPH and GDK/Cantons, and will be communicated and promoted in an appropriate way.

Phase 6: Reporting

The recognised Centres for Rare Diseases report annually. This report is sent to both kosek and Orphanet Switzerland (no double reporting) and includes:

- A presentation of the activities performed in the 5 core tasks (health care - incl. number of patients, further training, information, research, coordination and administration)
- The status of development projects at the Centre for Rare Diseases
- Objectives for the following year

- Contact details of the coordinator of the Centre for Rare Diseases

Conclusion: annual reports and up-to-date data for Orphanet.

Phase 7: Audit/ Re-evaluation

The recognition is valid for 4 years as of the positive recognition decision. At the expiry of this period, the institution is required to undergo a re-evaluation, by completing or updating the recognition questionnaire, and submitting it to kosek within the deadline (30 September). kosek will review the Centre for Rare Diseases and decide whether the institution will remain recognised for another four years, and whether this recognition is subject to any conditions.

The terms of the audit phase will be communicated to the institutions in due time.

4. Bibliography

Schweizerische Akademie für Medizinische Wissenschaften (2016). *Nationales Konzept Seltene Krankheiten.* ***Vorschlag zur Umsetzung von Massnahme 1. «Ein Prozess zur Errichtung von Referenzzentren wird definiert».*** Bericht der SAMW zuhanden des Bundesamtes für Gesundheit