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Procedural document:
Data collection and registration of expert centres in Orphanet

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I. Introduction

1.1 Purpose/objectives

Orphanet offers, amongst a range of expert resources on rare diseases, a directory of expert centres and networks of expert centres dedicated to the medical management and/or genetic counselling for one particular rare disease or a group of rare diseases.

This directory is intended to improve the referral of patients towards expert centres and ease the process of second opinions between professionals.

This document aims to explain the set of criteria used to select, register and update the expert centres and the network of expert centres.

1.2 Disclaimer

- This procedural document is part of the OrphaNetWork Direct Grant (831390) which has received funding from the European Union's Health Programme (2014-2020).
- The content of this procedural document represents the views of the author only and is his/her sole responsibility; it cannot be considered to reflect the views of the European Commission and/or the Consumers, Health, Agriculture and Food Executive Agency or any other body of the European Union. The European Commission and the Agency do not accept any responsibility for use that may be made of the information it contains.

1.3 Range of application

The present procedure applies to all the expert centres and the networks of expert centres registered in Orphanet. The registration and update of the expert centres is performed by the Orphanet national teams (ONT) having signed a Network Agreement and a Data Transfer Agreement (DTA) with the Orphanet Coordinating Team (OCT).

1.4 References

- Orphanet Standard Operating Procedures
- EUCERD recommendations on quality criteria for centres of expertise for rare diseases in member states.
- Core competences for genetic counsellors
- Professional and educational standards for genetic counsellors
- Code of professional practice for genetic counsellors

1.5 Definitions

Data transfer agreement (DTA): Contract between the providing and recipient institutions that governs the legal obligations and restrictions, as well as compliance with applicable laws and regulations, related to the transfer of such data between the parties.

European Reference Networks (ERNs): Virtual networks involving healthcare providers across Europe, aiming to facilitate discussion on complex or rare diseases and conditions that require highly specialised treatment, and concentrated knowledge and resources.

Genetic counselling expert centre: Centre delivering genetic advice either for all rare genetic diseases or for a rare genetic disease/group of diseases.

Information scientist (IS): Member of an Orphanet national team, responsible for the collection, assessment and registration of data.

Medical management expert centre: Specialised centre for a rare disease (or a group of rare diseases) organised for the medical management of patients or recognised as an expert centre for consultation. It delivers a service of indisputably higher quality for rare diseases than a standard hospital service in the relevant specialty.

National Advisory Board: Orphanet national teams can decide to set up a National Advisory Board, its members being nominated by the appropriate legitimate institutions (learned societies, national authorities, etc.), which are defined at country level. National Advisory Board members contribute with their expertise to Orphanet at country level and validate any database content concerning resources listed for the country in question as well as Oprhanet Rare Disease Nomenclature production in national language if relevant.

Network of expert centres: Clinical centres with expert knowledge in the same rare disease or group of rare diseases and officially established as a network.

Orphadata: Website providing the scientific community with comprehensive, high-quality and freely-accessible datasets related to RD and orphan drugs, in a reusable format.

Orphanet coordinating team (OCT): It is the French US14 Inserm based team who coordinates the Orphanet Network, produces the English Orphanet Nomenclature and its scientific annotations and is also responsible for coordination of the production of the scientific content and of all Network activities including translation.

Orphanet national teams (ONT): Teams located in each participating country of the Orphanet network, and endorsed by national authorities. An ONT is composed, at least, of a country coordinator who is responsible for the national Orphanet activities including translation. It can also include one or several information scientists, translation staff and a project manager.

Orphanet national validator: Professional designated to assess at the national level the appropriateness to register a specific expert resource in the Orphanet database. It can be the national coordinator, a member of the national Advisory Board or of the Health Authorities.

Orphanet national websites: Each Orphanet national team maintains a national entry point to Orphanet, providing information on the team and on the latest news and updates concerning national activities, in the national language of the country concerned.

Orphanet Management Board (MB): Decision-making body of the Orphanet Network. It is composed of the National Coordinators (one per member of the Orphanet Network). The MB is in charge of identifying funding opportunities, of guiding the project to provide an optimum service for the end-users, and of considering the inclusion of new teams as well as ensuring the continuity of Orphanet. It is chaired by the project coordinator at the Inserm. The Board is coordinated by Inserm.

Orphanet Quality Criteria Questionnaire (OQCQ): Questionnaire aimed at evaluating the appropriateness of registering in Orphanet a medical management expert centre that is not officially-designated. It is is based on the EUCERD recommendations on quality criteria for expert centres for rare diseases.

Orphanet quality questionnaire for Genetic Counselling Units / Clinics: Questionnaire aimed at evaluating the appropriateness of registering in Orphanet a genetic counselling expert centre that is not officially-designated. It is based on peer reviewed literature about genetic counselling practices in Europe and the European Board of Medical Genetics documentation for European unified competency standards for ensuring patient safety.

Rare disorder (RD): A rare disorder is defined according to the European legislation defining a prevalence threshold of not more than 5 affected persons per 10'000 (Regulation (EC) N°141/2000 of the European Parliament and of the council of 16 December 1999 on orphan medicinal products).

Service contract: Contract, established by Inserm-Transfert, allowing for-profit companies/organisations to access Orphanet data via a for-fee Orphadata account. The contract stipulates the conditions of use of the data.

1.6 Filing and updates

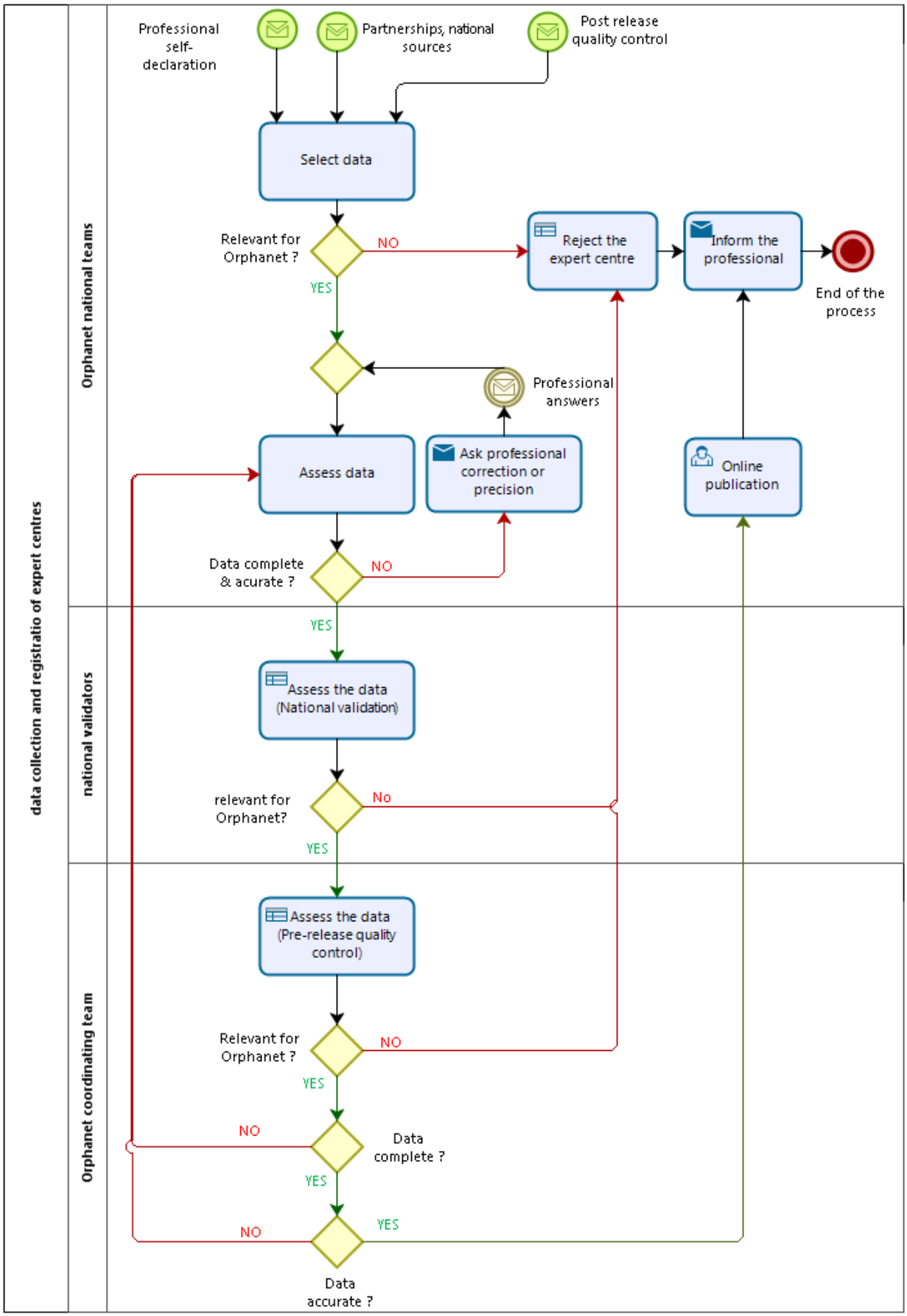
This document is updated by the coordinating team as often as necessary and at least once a year. The most up-to-date version is available on the Orphanet website:

https://www.orpha.net/orphacom/cahiers/docs/GB/eproc_expert_centres_R2_PatCar_Cns_EP_02.pdf

II. Methodology

2.1 Flowchart

The general process for expert centres data collection, registration, validation and its quality control is presented below:



[1] Powered by Bizagi

2.2 Description

The process of registration/update of expert centres and networks of expert centres starts with:

- Professionals declaring their activity through the Orphanet online registration service or in any form (e-mail, phone calls, etc.).
- An exchange of data through a partnership with a source of data (i.e. national Authorities, Expert networks)
- A post-release quality control task focused on expert centres or network of expert centres

An annual update is organised and launched by the OCT. All the professionals responsible of expert centres / networks of expert centres registered in the database are invited to review and update their activities through the Orphanet online registration service. National teams are responsible for the follow-up of their feedback.

2.3 Sources of information

National teams are in charge of identifying the sources of information for expert centres and networks of expert centres in their countries, and are advised to establish partnerships with them to be as **exhaustive** as possible.

In case of establishing a partnership, national teams must inform the coordinating team, as some types of partnerships require the signature of a data transfer agreement (DTA).

The mains sources of information are:

a) For officially designated expert centres

- National list of centres of expertise, or reference centres, recognised by the Health authorities, if applicable.
- European Reference Networks (ERN).

b) For non-officially designated expert centres

- Learned societies, foundations and other networks
- Expert centres involved in clinical trials
- Scientific publications
- Patient organisations
- Websites of Human Genetics societies (for genetic counselling centres)
- Pharmaceutical and biotechnology companies involved in orphan drugs
- Professionals declaring an expert centre through the Orphanet online registration service.

2.4 Data Selection

National teams are involved in collecting and registering information on expert centres and networks of expert centres located in their own country. National teams and the coordinating team are involved in collecting information on international networks, but only the coordinating team can register them.

National teams start the data selection by verifying that the expert centre meets the inclusion criteria for Orphanet. An expert centre can offer medical management and/or genetic counselling services. Both options have specific selection criteria.

a) Inclusion and exclusion criteria for the medical management centres

i. Inclusion criteria

In order to be registered in Orphanet, a medical management expert centre should be a specialised centre for a rare disease (or a group of rare diseases) either officially designated or complying with the Orphanet Quality Criteria Questionnaire (OQCQ, see annex 1). It should have an adapted technical platform, a multidisciplinary team (when necessary) and clinical research expertise.

The OQCQ is based on the EUCERD recommendations on quality criteria for expert centres for rare diseases, which are:

1. Capacity to produce and adhere to good practice guidelines for diagnosis and care.
2. Quality management in place to assure quality of care, including National and European legal provisions, and participation in internal and external quality schemes when applicable.
3. Capacity to propose quality of care indicators in their area and implement outcome measures including patient satisfaction.
4. High level of expertise and experience documented, for instance, by the annual volume of referrals and second opinions, and through peer-reviewed publications, grants, positions, teaching and training activities.
5. Appropriate capacity to manage RD patients and provide expert advice.
6. Contribution to state-of-the-art research.
7. Capacity to participate in data collection for clinical research and public health purposes.
8. Capacity to participate in clinical trials, if applicable.
9. Demonstration of a multi-disciplinary approach, when appropriate, integrating medical, paramedical, psychological and social needs (e.g. RD board).
10. Organisation of collaborations to assure the continuity of care between childhood, adolescence and adulthood, if relevant.
11. Organisation of collaborations to assure the continuity of care between all stages of the disease.
12. Links and collaboration with other CE at national, European and international level.
13. Links and collaboration with patient organisations where they exist.
14. Appropriate arrangements for referrals within individual Member States and from/to other EU countries if applicable.
15. Appropriate arrangements to improve the delivery of care and especially to shorten the time taken to reach a diagnosis.
16. Consideration of eHealth solutions (e.g. shared case management systems, expert systems for tele-expertise and shared repository of cases).

The questionnaire must be filled-in by professionals when the expert centre is not officially designated, and it is assessed by national validators.

ii. Exclusion criteria

Orphanet does not register an expert centre if it is:

- A conventional specialised medical department without specific focus on rare diseases, even if it is a reputed one.
- A self-declared centre that does not fulfil the selection criteria.

b) Inclusion and exclusion criteria for genetic counselling centres

i. Inclusion criteria

In order to be registered in Orphanet, a genetic counselling centre should deliver genetic advice either for all genetic diseases or for a genetic disease/group of diseases, and be either officially

designated or comply with the Orphanet quality questionnaire for Genetic Counselling Units / Clinics (annex 2).

The Orphanet quality questionnaire for Genetic Counselling Units / Clinics (annex 2) is based on peer reviewed literature about genetic counselling practices in Europe and the European Board of Medical Genetics documentation for European unified competency standards for ensuring patient safety (2010-2013):

- Core competences for genetic counsellors
- Professional and educational standards for genetic counsellors
- Code of professional practice for genetic counsellors

The questionnaire must be filled-in by professionals when the genetic counselling centre is not officially designated, and it is assessed by national validators.

ii. Exclusion criteria

Orphanet does not register a genetic counselling expert centre if the center is:

- Not officially designated and the quality questionnaire for Genetic Counselling Units / Clinics is not validated.

c) Inclusion and exclusion criteria network of expert centre

i. Inclusion criteria for network of expert centre

- Clinical centres with expert knowledge in the same field and officially established as a network.
- The network should be focused on a rare disease or on a group of rare diseases.

ii. Exclusion criteria for network of expert centre

Orphanet does not register as network of expert centres:

- A self-declared network (same hospital or same disease without any funding).
- Clinical centres participating in a same clinical trial.
- Clinical centres with expert knowledge in the same field but without official recognition as a network.

2.5 Data assessment

If the centre complies with the Orphanet inclusion criteria for expert centres, the national IS analyse the information to check that the mandatory dataset (cf below) is provided and that it is coherent, and eventually introduce the necessary corrections before submitting to national and international pre-release quality control.

In case of inconsistency or missing information, the IS will contact the professional in order to clarify or obtain the information needed.

a) Orphanet dataset for expert centres

i. Mandatory dataset

- Name of the expert centre in local language and in English
- Name and address of the institution and of the department/service responsible of the expert centre

- Name and details (email address and phone number) of at least one expert centre coordinator[2])
- The disease(s) or group of diseases covered by the expert centre
- Indication whether the expert centre is officially designated by the national health authorities Or the Orphanet quality questionnaires (If the centre is not officially designated)
- Indication whether the expert centre is intended for children, adults or both.
- Indication whether the expert centre is a genetic counselling clinic, a medical management clinic or both.

ii. Optional dataset

- Website of the expert centre
- Team members (with a maximum of seven professionals)

b) Mandatory dataset for network of expert centres

- Name in local language and English
- Network website or description
- Type of network
- Geographical coverage
- Name and personal details of the coordinator of the network
- Disease(s) covered
- List of expert centres integrating the network

2.6 Pre-release national validation

A validation process at the national level is mandatory for expert centres:

- In the case of officially designated expert centres, the Orphanet national validator(s) confirm the fact that the centre has indeed received an official designation from the national health authorities.
- In the case of non-officially designated expert centres, the Orphanet national validator assesses if the centre meets the inclusion criteria, paying special attention to the Orphanet quality questionnaires. This a very important step given that the quality questionnaires should be appraised according to the national context.

In the case of the OQCQ, by decision of the **Orphanet Management Board**:

- The following questions are "major" criteria, although not mandatory:

Question 1: How many patients did you see with this disease or group of diseases last year?
Question 2: Do you provide expert advice/second opinion to other clinicians (mail, telephone)?
Question 7: Does the centre publish peer reviewed publications?

- The criteria should be appraised qualitatively by the national validator according to the disease(s) and the national context.
- Each country needs to indicate on their national website which other criteria are taking into account in order to be transparent on how the data selection is carried out.

2 Please note that Orphanet register a maximum of 2 expert centre coordinators

In the case of the Orphanet quality questionnaire for Genetic Counselling Units / Clinics, the criteria should be appraised qualitatively according to the disease(s) and the national context.

Each ONT has to establish and publish on their national website the list of the current national validators.

2.7 Pre-release quality control (PrRQC)

Once the candidate expert centre passed the national validation, the coordinating team performs a pre-release quality control to assess the relevance and correctness of data collected by the national teams.

This quality control is mainly focused on the disease(s) linked, the quality questionnaires for non-officially designated expert centres, and on the coherence of the whole dataset.

In case some information is missing or needs correction, the form is sent back to the national teams.

2.8 Data publication

Once all the quality control steps have been completed, the information on expert centres and networks of expert centres is accessible on the Orphanet website and can be retrieved from Orphadata after signature of a Data Transfer Agreement (DTA) or a service contract. Once published, the ONT are in charge of informing the professional(s) that the activity has been published.

2.9 Post-release validation

The post-release quality control for expert centres includes the quality control projects and the post-release national review.

Quality control projects: Projects organised by the coordinating team on a regular basis to check the completeness and consistency of the data.

Post release national review: At least once a year, the OCT makes a directory of all the expert centres registered for their country available to the national teams. National teams are invited to review their data, to send them to a national board of experts / national health authorities to ensure data are still up-to-date, accurate and comprehensive. The post-release validation process has to be defined by each team and communicated to the coordinating team. National teams can, at any time, perform an extraction from the database to perform this post release quality control.

III. Annexes

3.1 Annex 1 – Orphanet Quality Criteria Questionnaire (OQCQ)

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Questionnaire for medical management expert centres

(Fill in a separate form for each clinic)

For the Orphanet Coordinator only	*Return this form to:*
Checked on (date):	***Fax:***
Validation: ☐ Yes ☐ No	***E-mail:***

Form intended for the scientific committee to check whether the expert centre matches the quality criteria defined by the EU Committee of Experts on Rare Diseases (EUCERD) for a Centre of Expertise.

Name of expert centre: .....

Name of coordinator: .....

Diseases or (group of) managed in this expert centre:
.....
.....
.....

ACTIVITY OF THE EXPERT CENTRE FOR RARE DISEASES (RD)*

*A disease is considered to be rare when it affects less than 1 person in 2,000

Are you officially a centre of expertise?

☐ Yes => If yes, you do not need to answer the following questions

☐ No => If no, please answer the following questions

1. How many patients did you see with this disease or group of diseases last year?
Total number of patients seen last year: ……..
Number of new cases last year: ……..
Percentage of patients from other regions from the country: ……..
Percentage of patients from abroad: ……..

2. Do you provide expert advice/second opinion to other clinicians (mail, telephone)? ☐ Yes ☐ No
Number of expert opinion given last year: ……..

3. Is your centre multi-disciplinary, integrating medical, biological, paramedical, psychological and social needs (such as a rare disease bo: ☐ Yes ☐ No
Please explain your answer:
.....
.....
.....

4. **Does your centre organise collaborations to ensure the continuity of care between childhood, adolescence and adulthood, if this is releva** ☐ Yes ☐ No

5. **Does your centre have appropriate arrangements in place for referrals within your country and from/to other EU countries (if applica** ☐ Yes ☐ No

6. **Does your centre have quality management procedure(s) in place to ensure quality of care (including National and/or European legal provisions), and does it participate in internal and external quality schemes, if ap** ☐ Yes ☐ No

If yes, please describe:

.....
.....
.....

7. **Does the centre publish peer reviewed publications?**

Number of publications on the disease(s) over the past five years: ……..

Number of communications on the disease(s) over the past five years: ……..

8. **Have you obtained grants for studies on this disease or group of diseases?** ☐ Yes ☐ No

If yes, please describe:

.....
.....
.....

9. **Do you participate in systematic clinical data collection?** ☐ Yes ☐ No

If yes, please describe:

.....
.....
.....

10. **Do you participate in clinical trials?** ☐ Yes ☐ No

If yes, please describe:

.....
.....
.....

11. **Do you teach and train for this disease or group of diseases?** ☐ Yes ☐ No

If yes, please describe:

.....
.....
.....

12. **Do you have links and collaborations with patient organisations?** ☐ Yes ☐ No

If yes, please describe:

.....
.....
.....

13. **Does the centre have links and collaborations with other centres at national, European or international** ☐ Yes ☐ No

If yes, please describe:

.....
.....
.....

3.2 Annex 2: Questionnaire for Genetic Counselling Units / Clinics

(Fill in a separate form for each unit/clinic)

For the Orphanet Coordinator only **Checked on (date):** **Validation:** ☐ Yes ☐ No	***Return this form to:*** ***Fax:*** ***E-mail***:

Form intended for the scientific committee to check whether the clinic/unit matches the quality criteria defined

ACTIVITY OF THE GENETIC COUNSELLING UNITS FOR RARE DISEASES (RD)*

*A disease is considered to be rare when it affects less than 1 person in 2,000

1. Patient population

How many patients with this disease or group of diseases did you provide genetic counselling, approximately per annum? (Your country's Orphanet Information Scientist can discuss disease groupings with you)

Total number of patients seen : ……..
Number of new cases: ……..
Time period: ……..

2. The team

a) The following professionals who provide genetic counselling includes (state number):

Clinical Geneticists ……..

Genetic counsellors with an MSC in genetic counselling ……..

Genetic nurses with an MSc in genetic nursing ……..

Genetic counsellors without an MSc ……..

Genetic nurses without an MSc ……..

How many of the above staff have current National or European registration:

Genetic counsellors: …….. Genetic Nurses ……..

Other health professionals (list please, with their genetic counseling qualifications)

……
……
……

b) Does your service include (i.e. in-house, positions funded by your service) :

Laboratory scientists, Psychologists, Social workers, Other medical specialists

Please describe

……
……

c) Does your service have designated pathways for collaboration with professionals external to your service?

Laboratory scientists, Psychologists, Social workers, Other medical specialists

Please describe……..
……

3. **<u>The scope of genetic counselling services provided</u>**

   a) **Do patients receive psychological support as part of the genetic counselling consultation? Yes …… No ……**
   From the genetic counselling professional during consultation Yes …… No ……
   From other counselling or psychologist team members as part of the service Yes …… No ……

   b) **Which of the following genetic counselling services does your team provide?**
   - ☐ Cascade genetic counselling and testing (where applicable) to at-risk family members
   - ☐ Pre-symptomatic genetic testing (where applicable) to at-risk individuals
   - ☐ A pathway for access to the appropriate clinical services (by medical specialty) for those who are shown to be carriers on pre-symptomatic testing
   - ☐ Access to prenatal diagnosis services (where applicable)
   - ☐ Patient education materials and links to patient support groups
   - ☐ Written information provided to patients after consultation
   - ☐ A pathway for access to further psychological support, when necessary

   c) **Do you provide expert advice or second opinions for genetic counselling issues to other clinicians by post or telephone?**
   Estimate number of expert opinions given last year: ……..

4. **<u>Quality management</u>**

   a) **Does your centre have quality management procedure(s) in place to ensure quality of care in genetic counselling (including National and/or European legal provisions),**
   If yes, please describe:
   ……

   b) **Does your centre participate in the CEQAS quality assurance scheme for genetic counselling?**
   Yes …… No ……

   c) D**oes your centre participate in other internal or external quality schemes for genetic counselling,?**
   Yes …… No ……_
   If yes, please describe: ……
   ……

   d) **Please describe how professional genetic counselling standards are maintained within the team (eg.: through formal case discussion, clinical and/or counselling supervision, continuing professional development participation, and other activities)**

……
……
……
……

5. **<u>Teaching and training</u>**

   a) **Does your centre offer an MSc Genetic Counselling training program? Yes …… No ……**

   b) **Does your centre offer clinical placements for trainee Genetic Counsellors / Genetic Nurses ?**
   Yes …… No ……

   c) **Does your centre teach other health care professionals about genetic counselling Yes …… No ……**

If yes, please describe:

……
……

For any questions or comments, please contact us: contact.orphanet@inserm.fr

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