

Procedures: Orphanet Advisory Board on Genetics

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I. Introduction

1. Purpose/objectives

This procedure describes the membership of the Orphanet Advisory Board on Genetics, its mandate and its general rules of procedure

2. Disclaimer

- This procedure is part of the RD-ACTION joint action 677024 which has received funding from the European Union's Health Programme (2014-2020).
- The content of this procedure represents the views of the author only and is his/her sole responsibility; it can not 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.

3. Range of application

This procedure is intended for the Orphanet Advisory Board of Genetics members and for the Orphanet audience.

4. References

- [Orphanet SOPs](#)
- **Orphanet Advisory Board on Genetics member list**

5. Definitions

Accreditation: Formal recognition of technical competence to perform specific types of testing by complying with specific management and technical requirements

External quality assessment (EQA): A system of objectively assessing the laboratory performance by an external agency (known as EQA providers) through the comparison of the lab performance against an agreed independent standard.

Orphanet Advisory Board on Genetics (GAB): is in charge of advising Orphanet on topics related to the database of genes and the database of medical laboratories and genetic tests, it reports to the Orphanet Management Board (MB) and issues comments and recommendations which will enable the MB to define changes to be introduced to the project. This Board was established in the context of the joint action RD-ACTION 2015-2018.

Orphanet Management Board (MB): This Board is composed by Orphanet Country Coordinators and it is chaired by the scientific coordinator of the project at the Inserm.

Orphanet Coordinating Team: Orphanet is a consortium of 40 countries coordinated by the Inserm-based team in France

Quality management data: Orphanet registers two types of data to assess the quality management of laboratories: Accreditation and external quality assessment.

Technical procedure of the test: Test methodology defined with a three level representation: specialty, objective and technique. For some specialties, objective and technique are not further developed (e.g. parasitology). For more information on the technical procedure representation and the terms used, please refer to the annex of this document or to [the Glossary and representation of terms related to diagnostic tests](#)

6. Filing and updates

This procedure is updated at least every year by the Orphanet International Coordinator and as often as required. The most up-to-date version is available on the Orphanet website: www.orpha.net.

II. Membership

Members of the Orphanet Advisory Board on Genetics are either geneticists members of the MB joining the advisory board on a volunteer basis or geneticists not belonging to the MB. These external members are invited under proposal of the Orphanet Management Board. In that case, external candidates are nominated if the majority of the Orphanet Management Board agrees.

Membership lasts for one year. Members can be reconducted if they postulate again every year, provided they have shown to be active during their membership.

III. Mandate

Board Members advise Orphanet on topics related to the database of genes and the database of medical laboratories and genetic tests.

Subjects concerning the database of genes can include the methodology to include a gene in the database, the way they are displayed in the website, the eventual communication products around genes database. Members can be solicited to help in the validation of particularly difficult cases in order to decide whether to include or not a particular gene related to a given disease.

Subjects concerning the database of medical laboratories and genetic tests can include: the dataset; the technical procedures of the tests; data on quality management; the way the tests and laboratories are displayed and the search functionalities in the website; the eventual communication products around the genetic tests database; and database quality control methods and means.

IV. General Rules of Procedure

1. Members of the Orphanet Advisory Board on Genetics will be contacted by mail by the Orphanet Coordinating team whenever the team needs advice in one of the subjects previously mentioned.
2. Questions and advices provided by the Orphanet Advisory Board on Genetics will be then discussed at the following MB meeting in order to make decisions on the eventual evolutions of the database or the website if relevant.
3. Members can also bring new subjects to be discussed either at the advisory board and/or at the MB level.

V. Additional information

Compensation:

The members are not compensated for the time they dedicate to the project.

Sessions:

Members meet at least twice a year by conference calls, or more frequently if needed.

Members:

http://www.orpha.net/orphacom/cahiers/docs/GB/Orphanet_GAB_members_2016.pdf

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

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http://www.orpha.net/orphacom/special/eproc_GAB_V01.pdf