

EVALUATION OF POPULATION NEWBORN SCREENING PRACTICES FOR RARE DISORDERS IN MEMBER STATES OF THE EUROPEAN UNION

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Background

As a result of a tender launched by the European Commission, an activity has started with the aims of identifying and evaluating all aspects deemed relevant to the implementation of a public health action in newborn screening (NBS), taking into consideration the views of professionals, patients and health authorities. Newborn screening practices and policies will be mapped in the whole European Union and analysed on the basis of current expert methodologies and stakeholders' views. Within the perspective that NBS is implemented as a public health initiative, a range of aspects will be considered, which ensure the feasibility and sustainability of the screening program and its efficacy in improving population health, as well as patient care and quality of life. Moreover, challenges and opportunities resulting from NBS implementation will be identified and solutions proposed, accompanied with the information necessary to let national authorities to make their own free but informed choices. Finally, the feasibility of supporting actions at the Community level will be explored in order to identify the strategies which the European Commission can adopt to promote the establishment and improvement of NBS programmes in the EU.

Outcomes

The expected deliverables are:

- **Report on the practices of NBS for rare disorders.**
 - This report will describe the main features of the existing NBS practices, which are of relevance to inform the development of national policies on NBS in the EU
- **Expert opinion, including a decision-making matrix, on the development of European policies in the field of newborn screening for rare diseases.**
 - This document is intended to provide a reference set of criteria, encompassing both general issues and specific health system conditions, which the national health authorities in EU may use to make their decisions on the development of national NBS programmes.
- **A European Union Network of Experts on Newborn Screening (EUNENBS).**
 - This network is made of experts designated by the national health authorities of each of the 27 EU Member States, and can be extended with a directory including other experts from the academic community, who may prove useful for ad hoc consultation, e.g. on specific diseases, tests and issues of NBS. [Information on how to register in this directory is provided below.](#)
 - This network will be presented to the EU Committee of Experts on Rare Diseases and may provide advice to them for the best development of the NBS activities. A core group supervises the preparation of the tender deliverables.
- **A European Experts Consensus Workshop on Newborn Screening.**
 - This Workshop will finally approve the expert opinion and the decision-making matrix.

Progress status

- A first meeting has been convened with a number of experts active in European scientific and professional associations, which are operationally involved in NBS, and representatives of the key European alliances of rare disease patients.
- A questionnaire is being developed in collaboration with the participants to the first meeting to launch in June a survey on current practices.
- Invitations to Member States are being prepared for the set up of the health authority component of EUNENBS.
- The criteria for the selection of experts to be included in the EUNENBS directory are being discussed. **Interested experts, who comply with the selection criteria, are kindly invited to apply for the registration in the EUNENBS Directory** following the instructions provided in the application form. Selection criteria and application form will be made available as soon as possible in the web page: <http://www.iss.it/cnמר/prog/cont.php?id=1582&lang=1&tipo=64>

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