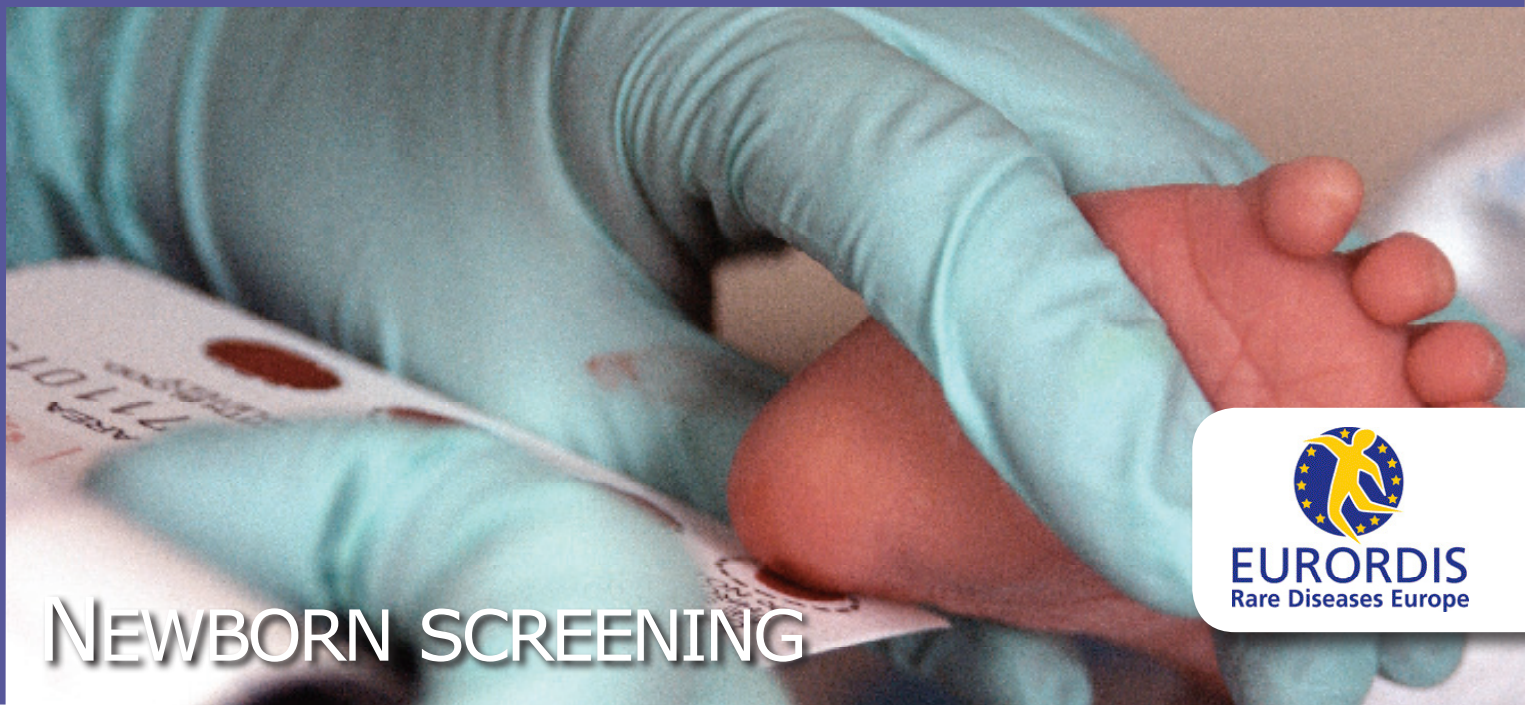

NEWBORN SCREENING

Newborn screening (NBS) is the process of systematically testing newborns (usually by taking a blood sample by heel prick) just after birth for certain treatable diseases. Ideally, this practice is part of a larger NBS programme that also includes confirmatory diagnosis, immediate care, treatment and follow-up. Recent and continued developments of screening techniques, as well as the increase of possibilities for treatment, have led to the expansion of NBS to include potentially detectable rare diseases (RDs). Broader benefits of NBS are also being considered, which include not only the newborn baby, but also the family.

As additional tests have been sporadically added to NBS programmes in some European countries, a growing discord in the field and health inequality among European populations has been observed. With support from the European Commission, the NBS expert community is now engaged in the analysis of existing NBS policies for RDs in Europe and in the preparation of recommendations for the evaluation and implementation of new NBS policies at the national level. The RD patient community hopes that this European action will lead to increased collaboration between Members States (MS) and subsequent harmonisation of criteria as well as greater convergence of practices, potentially translating into the introduction of new NBS programmes with the systematic screening for additional RDs in Europe.

EURORDIS is dedicated to representing the needs of RD patients and their families. Some of our actions for newborn screening are the following:

- Partnering with the European Union Tender "Evaluation of population newborn screening practices for rare disorders in MS of the EU" led by the Istituto Superiore di Sanità - Centro Nazionale Malattie Rare
- Hosting a workshop on NBS for RD during the EURORDIS Membership Meeting 2011 Amsterdam
- Producing a Decide topic on NBS to promote well informed citizens debates and citizens preferred policy scenarios
- Organising sessions on NBS in the European Conference on Rare Diseases & Orphan Products 2012 Brussels.
- Officially representing patient needs in the European Union Network of Experts on Newborn Screening (EUNENBS)
- Promoting this public health topic within the national plans on rare diseases (EUROPLAN - and national conferences on national plans for rare diseases)
- Contributing within the European Union Committee of Experts on Rare Diseases (EUCERD) to shape the EU policy on Newborn Screening.

WHY IS NEWBORN SCREENING FOR RARE DISEASES REQUIRED?

The Commission Communication, "Rare Diseases: Europe's Challenges"[1] proposes that Member States (MS) implement mechanisms to gather expertise on RDs at the national level in concert with European counterparts. The expected result is to strengthen the development of European guidelines on diagnostic tests or population screening, while respecting national decisions and competences.

The Council Recommendation on an Action in the Field of Rare Diseases[2] encourages cooperation to generate evidence for decisions to be made at MS' level.

A European Union Network of Experts on Newborn Screening (EUNENBS) supported by the European Commission, has been created to evaluate current practices in NBS. Patient representatives, through EURORDIS, are part of this network.

The recommendations of the European Project for Rare Diseases National Plans Development (EUROPLAN)[3] echo that cooperation among MS may be advantageous in carrying out screening programmes. More specifically, a future common framework is advisable for the development of NBS programmes in order to assure equal access to prevention, diagnosis and care, while recognising that geographic patterns of distribution of certain diseases and public health systems and social issues may be different in different countries.

1 - COM (2008) 679 Communication from the Commission to the European Parliament, the Council, the Economic and Social Committee and the Committee of the Regions on Rare Diseases: Europe's challenges.
2 - Council Recommendation of 8 June 2009 on an action in the field of rare diseases.
3 - Recommendations for the Development of National Plans for Rare Diseases - Guidance, www.europlanproject.eu.

WHY IS NEWBORN SCREENING IMPORTANT FOR RARE DISEASE PATIENTS?

RDs are often severe, chronic, and with a degenerative evolution. For approximately 50% of RDs the onset occurs during childhood but for many diseases, clinical signs of symptoms do not appear in the first days or months following birth. For this reason, some RDs fully qualify for inclusion in NBS programmes where early intervention can prevent or minimize the onset of the disease symptoms, improve individual health outcome together with family and social responses. This strategy to prevent late or incorrect diagnosis minimises undesired community level consequences such as unnecessary health expenditures or unadapted family behaviour. In addition, patient organisations highlight the benefit of NBS for family planning; indeed, when a child is diagnosed at birth with a very severe and life threatening disease, parents are able to shape their family planning accordingly (e.g.; adequate genetic counselling, pre-implantation diagnosis, pre-natal diagnosis when technology is available).

Until now, a few RDs such as phenylketonuria (PKU) and congenital hypothyroidism (CH) were screened for widely across Europe due to the undisputed significant benefit to the newborn population as a whole. But equal access and availability of screening programmes (up to 29 RDs are screened in some countries versus only one or two or even none in others), their mandatory nature and adequate care and treatment following diagnosis has not been established in all MS. This is unfair to families and their children in MS where programmes are inadequate.

HOW TO ADDRESS NEWBORN SCREENING FOR RARE DISEASES?

There are clear and valid principles that EURORDIS endorses and recommends to RD patient advocates and upon which any decision for introducing new NBS programmes should be based. Beyond these objective principles, patients and their families together with rare disease experts expect to be included in discussions on new potential NBS programmes for their conditions.

Expansion of NBS programmes requires each MS to assess the potential harms and demonstrate benefits commensurate with the financial commitment required. Tests used to detect RDs in NBS programmes must be:

- Specific - have few false positives that unnecessarily cause parental stress and anxiety
- Sensitive - have few false negatives that can cause potential delays in diagnosis
- Predictive – have a high probability that persons with a positive test result has, or will get, the disease
- Acceptable - low level of risk, discomfort, etc.

NBS programmes must also follow criteria based on the Wilson and Jungner Principles and Practice of Screening for Disease[4]:

- Eliminate or minimise any harm
- Address concerns about privacy and autonomy
- Evaluate ethical, legal and societal aspects
- Recognise that most screened conditions are inherited disorders and may have consequences for family members.

The decision for expanding NBS programmes should be not only driven by the evaluation of the potential benefit for the specific newborn population and individual patient, but by positive reported outcomes and additional benefits to the family:

- Shorter time to correct diagnosis
- Improvement in family dynamic and educated reproductive decisions
- Adjunctive and curative therapy and early intervention programmes
- The potential participation of the affected child in research on innovative therapies.

RD patients and patient groups should continue to be involved in the discussions that will ultimately influence clearly stated, transparent standards and good practices agreed upon at the EU level. The Decision Making Matrix developed under the EU Tender "Evaluation of population newborn screening practices for rare disorders in MS of the EU" may soon serve as a practical guide for national stakeholders to update newborn screening policies.

This should lead to harmonisation across MS of criteria for public health decision-making on NBS programmes for RDs for which there is strong clinical evidence of benefit to individual patients, their families and society as a whole.

New and expanded NBS programmes for RDs may:

- Include diseases that can now be screened using new technologies
- Expand the definition of benefit to include not only clinical evidence, but also patients' and their families' quality of life, benefit for society, etc.

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