

# NEWBORN SCREENING IN ITALY: IT IS TIME FOR A DEEP REFLECTION

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**KEYWORDS:** Newborn screening, Inherited metabolic disease, Health technology assessment.

Newborn screening (NBS) is currently the most challenging topic for the Italian Society for the Study of Hereditary Metabolic Diseases and Newborn Screening (SIMMESN). Many problematic aspects are relevant according to the different components involved in the process; we have chosen those that are particularly critical and need to be shared. For this purpose, we have selected some keywords that require careful consideration and thought. They represent the main starting points toward the quality assurance and review of delivered services, i.e., health technology assessment, within this successful preventive public healthcare program.

## EFFICIENCY

In the healthcare system, as known to most, efficiency is the ratio of resources used (inputs) to services delivered (outputs) and is measured by performance indicators; these allow monitoring of the productivity of resources used in relation to the activities carried out in the evaluated system, including comparisons to precise benchmarks. Efficiency can also be measured by the number of mistakes made before a given goal is achieved or by the time invested or resources needed to implement it.

Measuring and monitoring the efficiency indicators of a system, such as NBS, would enable shared measures to be employed, perhaps even prospectively, to maximize the services provided, according to the inputs used, and minimize production costs. The medical literature has evaluated the efficiency of many individual NBS programs for certain diseases; yet, coordinated action at the national level is still lacking. Nevertheless, such estimation, in terms of benchmarking by pathology or center, could be relevant to indicate corrective actions for improvement.

## EQUITY

The Italian National Health Service (NHS), established in 1978, is based on the inalienable principles of universality and equality, authentic pillars of the reform that guarantee the country's social cohesion and counteract the health consequences of social inequalities resulting from the different socioeconomic conditions of each territory. Over time, considering the increasing age of the general population and the pediatric age of most children with previously intractable chronic conditions, the traditional idea of equality that individuals with the same health status or the same needs should be treated equally (horizontal equity), has gradually been combined with the belief that individuals with worse health sta-

tus, or greater needs, should be treated with greater care (vertical equity). As a result of this evolution, three different areas have emerged in which the result of equity must find concrete realization: access to healthcare services, health outcomes in charge of individual subjects, and allocation of resources in proportion to needs.

We cannot remain silent about the horizontal and vertical inequities that the NHS, now fragmented by its very nature and poorly coordinated after the 2001 reform of Title V of the Italian Constitution, allows and does not seem able to reduce. In this regard, SIMMESN must first acknowledge these realities and, in compliance with statutory regulations, take action to improve the quality of care throughout the country. This will require action on the organizational development of the various regional NBS programs, without neglecting the promotion of the indispensable integration between the activities of screening and diagnostic confirmation laboratories, on the one hand, and clinical reference centers, on the other.

## SUSTAINABILITY

Sustainability is a concept whose complexity seems to be increasing exponentially. The most frequently cited definition comes from the UN World Commission on Environment and Development: “Sustainable development is a development that meets the needs of the present time without compromising the ability of future generations to meet their own needs”<sup>1</sup>. Accordingly, sustainability can be defined as the ability to meet current needs without compromising the ability of future generations to meet their own needs (vertical sustainability). However, most people seem to miss the issue of the current sustainability of complex systems, such as healthcare, which, regardless of the level from which it is managed, has finite resources that must be allocated according to priorities and the long-term consequences of the choices made. Thus, sustainability is the process of living within the limits of the available physical and natural resources and current social and economic resources (horizontal sustainability), sufficiently to enable all human organized systems to thrive perpetually. The global abstraction of the concept of sustainability seems to alienate the public, and even many healthcare professionals, from the relevance of prioritizing the inherent sustainability of the country’s system, especially with the possibility of maintaining a universal NHS despite the exponential increase in costs, primarily technological, required to maintain it.

Today, we are urged to question whether the current NBS program in Italy, recently expanded nationwide to 59 inherited metabolic diseases, is sustainable for our country to an equitable and universal extent without sacrificing its quality levels<sup>2</sup>. Furthermore, as a result of recent technological advances in the field of genomic sequencing, the potential for incorporating these technologies into NBS programs is very promising; however, it adds considerable costs and carries ethical implications that must be carefully considered not to compromise the satisfactory level of trust achieved in our country toward this secondary prevention intervention. We believe it is necessary to undertake a careful and systematic analysis of the services currently provided, attentively evaluating the many possibilities for increased efficiency that coordination and convergence of different local programs, from regional to supra-regional, could offer. Without our pragmatic and shrewd programmatic intervention, we may risk seeing the quality level of deliverable laboratory and clinical services increasingly reduced; in addition, compared with the current situation, there may not be the necessary resources to implement, for example, more preventive interventions (eight inherited metabolic diseases are on hold, having received conditional approval), and prospective evaluation actions (population registries to assess actual effectiveness including cost/benefit).

As a form of holistic medicine dedicated to subjects (children and adolescents) at key stages of growth and development, pediatrics is undoubtedly one of the most critical disciplines in determining the health and well-being of present and future generations.

Modern pediatrics can only be a 5-P medical science: predictive, preventive, precise, participatory, and proactive, and the practice of NBS is among the few activities in which all these aspects coincide.

We must always be proud of everything accomplished to date, remembering the merits of the Experts who have preceded us. Concurrently, we must become aware of what remains to be done with the utmost sense of responsibility and willingness to dialogue with institutions without neglecting the training of young people interested in the ever-widening and challenging field of inherited metabolic diseases.

**ACKNOWLEDGEMENTS:**

Editorial assistance was provided by Valentina Attanasio and Aashni Shah (Polistudium s.r.l).

**CONFLICT OF INTEREST:**

The author declares that he has no conflict of interest to disclose.

**ETHICS APPROVAL AND INFORMED CONSENT:**

Not required due to the nature of the study.

**FUNDING:**

Editorial assistance was supported by SIMMESN.

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