

POSITION PAPER ON EXPANDED NEWBORN SCREENING

BY THE ITALIAN ASSOCIATIONS OF PEOPLE LIVING WITH RARE DISEASES

UNIAMO
FEDERAZIONE ITALIANA
MALATTIE RARE
ONLUS
Rare Diseases Italy

Authors: Annalisa Scopinaro* (Uniamo F.I.M.R.), Romano Astolfo (Sinodé Srl), Paola Bragagnolo (Sinodé Srl)

PATHWAY :

UNIAMO, the Italian Federation of Rare Diseases, and many POs have organised follow-up courses and meetings in different Regions both to raise awareness on the importance of the test and to outline the needs of pre and post blood sample collection. **New-born screening is more than just a test, it is a "system" and all aspects of it should be taken into account.**

INTRODUCTION:

In Italy new-born screening is a preventive, free and mandatory health activity since 1992, when Law no. 104 has provided mandatory screening for three diseases: phenylketonuria, congenital hypothyroidism and cystic fibrosis. Since 2016, Law no. 167 has extended mandatory new-born screening to about 40 other hereditary metabolic diseases (expanded new-born screening ESN), hitherto applied heterogeneously and not in all regions. The Budget Law 2019 (Law 30 December 2018, n. 145. "State budget for the financial year 2019 and multi-year budget for the three-year period 2019-2021"), amended the Law 167/2016 by expanding the ENS to neuromuscular diseases of genetic origin, severe congenital immunodeficiencies and lysosomal storage diseases including them in the panel.

OUR 10 RECOMMENDATIONS :

1. Ensure **the protection of human rights in the processes of carrying out expanded neonatal screening**. For this reason, screening tests for rare diseases must be: **timely, specific, sensitive, predictive, acceptable**.
2. Ensure **the timely inclusion**, in the panel of expanded new-born screening, of all hereditary metabolic pathologies, of neuromuscular diseases of genetic origin, of severe congenital immunodeficiencies and of lysosomal storage diseases or in any case **of all those pathologies for which there is scientific evidence of therapeutic efficacy for the pharmacological or dietetic therapy or for which there is scientific evidence that an early diagnosis, in neonatal age, entails an advantage in terms of access to therapies in an advanced state of trial, also of dietary type**.
3. Ensure **full implementation of new-born screening path in all the Regions/Autonomous Provinces** also through inter-regional agreement.
4. Provide for and guarantee **uniformity in the implementation of expanded new-born screening throughout the national territory** through the issue of **recommendations**.
5. Provide for and guarantee **training on expanded new-born screening of all health professionals involved in the birth path** including paediatricians and General Practitioners, **as well as the professionals and operators involved in the screening pathway**.
6. Provide for and ensure **adequate information on expanded new-born screening to citizenry** and in particular to families, from the pre-conception period to the time of the test.
7. Provide for and ensure **adequate communication of the results of new-born screening to parents**.
8. **If** the outcome of the expanded new-born screening is **positive ensure a timely recall and in the event of subsequent diagnostic confirmation, ensure adequate and timely take in charge**.
9. Provide a **communication to the parents also in case of negative test result**.
10. Provide for the possibility of **consciously opting for the retention, distribution and use of the residual biological material** of the screening and personal data **for research purposes**, also in an epidemiological perspective, and for further diagnosis, even over 2 years from execution of the test, **after acquiring informed consent as required by European legislation**.

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