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extended newborn screening

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Extended newborn screening a right of all european babies!

In Italy every newborn undergoes a test that identifies about forty rare metabolic disorders. It is the Expanded Newborn Screening (ENS), a free, mandatory test established by the 167/2016 bill and strongly sought by Aismme, Italian Association Inherited Metabolic Diseases Support. A full-fledged health path including not only the test and diagnosis, but also admittance to a continuity of care process for screened babies. The path has been developed by the Ministry of Health, patients associations, the Italian National Institute of Health and Scientific Medical Societies.

The ENS enables the early detection of about 350 affected children every year, hence preventing severe disabilities or death by means

of diets and therapies that halt the natural development of the pathology. The test provides families with serenity and a further certainty, with huge savings on social costs.



What is the ENS situation like in Europe?

- Newborns in other European countries are not as fortunate, as screened diseases are often too few.
- Equal access and availability of screening programmes are not uniform throughout Member States;
 - Screened diseases ranges from two to forty-two diseases, irrespective of the country's GDP;
 - Procedures and information parents receive on the disease and its treatment can differ.

The optimal screening path requires that:

- any introduction of a new screening path considers ethical, legal, social and privacy aspects;
- pathologies to be enlisted in the official panel undergo the Health Technology Assessment (HTA);
- decision boards involve not only experts on the disease, but also patients and/or caregivers to orientate health and political choices.



Every child in Europe has the right to the be screened for the highest possible number of diseases!

Aismme is currently working

- to make the ENS a right for all European newborns;
- to raise awareness of ENS among politicians, doctors and associations;
- to increase the quality of life of affected babies.



Associazione Italiana Sostegno Malattie Metaboliche Ereditarie

Italian Association Inherited Metabolic Diseases Support

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