



Poster number: 279  
Poster Theme 9

# extended newborn screening

Author: Manuela Vaccarotto, AISMME / Co-Author: Giuliana Valerio

**The Extended Newborn Screening has finally become a compulsory national public service in Italy: no newborn will be left behind again**

## After Italy, it is up to Europe!

In 2018 every newborn baby in Italy is subjected to Extended Newborn Screening (ENS). This is a test now free and a “must” that in a few hours after birth can give an early diagnosis of nearly 40 rare metabolic hereditary diseases – illnesses that are hard to discover and to identify, for which there is treatment. This test is an important preventative action, even if secondary: it allows the early identification of rare metabolic hereditary diseases that, if caught on time, can be treated appropriately, avoiding or limiting disability or early death. The delay of diagnosis produces high social and human costs.

The Italian Parliament passed a bill in late 2016 (Law n.167) stating the requirement, finding the necessary funds and activating important tools to provide a support network for diseases and a permanent table that involves the Ministry of Health, the medical world and patient associations.

The table will provide the “screening path” from pre-conception to the baby’s birth that closely concerns information and sensitization of the screened disease’s path. Further, it provides a periodic and constant update of the screened rare disease panel.

This is an important step forward if we think that in Italy in 2016, only 50% of newborn babies were screened and that after the new bill, compulsory testing allowed the identification of 1 sick child in 1,250 babies born.

### What about Europe?

Law n.167 is the result of a large path of harmonization between institutions, the medical world and also associations like AISMME, which since 2005 has been working to support metabolic patients, to raise public awareness about this topic and to lobby in the political arena. The patient’s voice can be heard, if its sound is loud enough! This can be an example and an incentive for all European States and for all parliamentary representatives in Brussels, a call for hard work on the application of ENS for all newborn babies in the EU and on the extension of the panel of the screened rare diseases as unknown and extremely rare.

Today this goal is much more realistic than before, thanks to the establishment and growth of MetabERN, the voice of the patient with a rare metabolic disease in the European net ERN.

### The future of patients with rare metabolic diseases in Italy

Having achieved the goal of applying ENS on all the newborn babies in Italy, now AISMME is working on a media campaign to inform parents about the importance of this new test. This will help avoid uncertainty about subjecting people’s children to it.

During decisional discussions, AISMME continues working to increase the screening panel to include additional diseases, like lysosomal storage diseases, and to expand and improve the treating centres that have to take care of the increasing number of patients found by the screening.



## The test that can save your child’s life

### IT IS A LIFESAVER

It allows the identification of more than **40 hereditary metabolic diseases** and to intervene promptly, before the symptoms are evident, so that disability or death can be avoided

### IT IS SAFE AND SIMPLE

It is not invasive and ‘risk free’ for the baby. It takes just a few drops of blood, taken from the heel of the baby

### IT IS FREE AND COMPULSORY

It is required by the Law 167/2016 and it has to be done everywhere in Italy

### NEGATIVE RESULT

Everything is ok!

### POSITIVE RESULT

The baby is called back to do the test again for the genetic confirmation of the disease

Parents are given written information and could be asked to sign a form of ‘informed consent’.

For more information

[www.aismme.org/Infoscreening](http://www.aismme.org/Infoscreening)



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