ACTIVITY OF NEONATAL SCREENING PROGRAM IN SPAIN

A review since its inception from 2016

Comité de la Calidad de la Asociación Española de Cribado Neonatal

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SUMMARY

The neonatal screening program (NSP) began in Spain in 1968, when Professor Mayor Zaragoza set up the first laboratory for the early detection of congenital metabolic disorders in newborns. In 1979, there were 10 laboratories in Spain covering 2.45% of newborns. With the aim of expanding coverage, new laboratories were promoted, reaching 21 laboratories. The Autonomous Communities (CC. AA.), as competent authorities in public health matters, have carried out in recent years restructuring and collaboration agreements to improve efficiency in the provision of screening laboratory services. There are currently 15 laboratories that cover 100% of newborns in Spain.

The scientific societies related to the neonatal screening programs have collected activity data from the different laboratories from the beginning of the program until 2016. For this purpose, specific questionnaires were designed and filled out by the laboratory managers.

Phenylketonuria and congenital hypothyroidism are the two diseases included in neonatal screening programs since their inception. Consequently, 15,156,785 newborns and 14,595,780 newborns have been analyzed respectively.

The sample extraction strategy has changed over the years. In the beginning, double extraction and single extraction coexisted. Since the incorporation of tandem mass spectrometry (MS/MS) technology, only one sample is taken 24-72 hours of life of the newborn.

Over the years, the detection of new diseases in some autonomous communities has been incorporated into neonatal screening programs which highlight the heterogeneity of these programs.

In 2014, the Ministry of Health established the set of diseases that are part of the neonatal screening program for endocrine-metabolic diseases of the NHS from the common portfolio of services of the National Health System (NHS). Seven diseases (phenylketonuria, congenital hypothyroidism, cystic fibrosis, sickle cell disease, medium-chain acyl-CoA dehydrogenase deficiency, long-chain 3-hydroxyacyl-CoA dehydrogenase deficiency and glutaric aciduria type 1) whose detection should be incorporated in all the neonatal screening programs of the national territory. Likewise, the Information System of the NSP of the NHS (NSIS) is created.

This report collects screening activity data from the beginning of the program in Spain until 2016. The results obtained show the improvement in detection over time as well as the points for improvement in some of the key processes of the screening program.

From 2016 the Ministry of Health, in coordination with the CC. AA., collects activity data and issues the annual technical evaluation report of the NSP-NHS.