50 años del programa de cribado neonatal en Cataluña [

2020

text (article)

Analítica

The Catalonian Newborn Screening Program (CNSP) began in 1969, in Barcelona. It was promoted by Dr. Juan Sabater Tobella and supported by Barcelona Provincial Council and Juan March Foundation. That is how the Institute of Clinical Biochemistry was born, whose aims were diagnosis, research and teaching, along with the spirit of contributing to the prevention of mental retardation. The CNSP began with the detection of phenylketonuria (PKU), and, in 1982, the Program was expanded with the inclusion of congenital hypothyroidism detection. Towards 1990, the Program covered almost 100% of all newborns (NB) in Catalonia. In 1999, the CNSP was expanded with the incorporation of cystic fibrosis. It took fourteen years, until 2013, to make the largest expansion so far, with the incorporation of 19 metabolic diseases to the screening panel. The detection of sickle cell disease began in 2015 and in 2017 the detection of severe combined immunodeficiency was included. Currently, the CNSP includes 24 diseases in its main panel. Since 1969, 2,787,807 NBs have been screened, of whom 1,724 have been diagnosed with any of these diseases, and 252 of other disorders by differential diagnosis with those included in the main panel. The global prevalence is 1: 1,617 NBs affected by any of the diseases included in the CNSP and 1: 1,140 NBs if incidental findings diagnosed through the CNSP are included
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