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TITLE: UPDATED SURVEY OF NEWBORN SCREENING FOR CYSTIC FIBROSIS IN EUROPE

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ABSTRACT BODY:

Abstract Body: Objectives: Newborn screening (NBS) for cystic fibrosis (CF) enables earlier pro-active treatment, which improves prognosis. The ECFS Neonatal Screening Working Group (NSWG) was established to track current practices in NBS, support implementation of NBS and establish consensus on strategies to improve this public health intervention. NSWG last conducted a survey on NBS in Europe in 2005. We aim to provide 1) an update on NBS in Europe, 2) discuss differences between protocols and 3) identify barriers to establishing NBS programmes.

Methods: Three questionnaires were sent to key workers in all European countries in 2015. The key workers completed the appropriate questionnaire depending on the situation in their country. If NBS was undertaken, this might be a national programme or regionally implemented. If no NBS was undertaken, we enquired about plans and barriers to implementation.

Results: 1) Compared to the 2005 survey, national NBS programmes in Europe have increased from 2 to 17 (including Russia & Turkey); 4 countries (Italy, Spain, Germany and Serbia) have regional programmes. In Spain from 2015, there is complete coverage of the population with these regional programmes. Germany will start a national NBS in 2016. 25 countries do not have NBS for CF. For 10 countries, a NBS programme is under consideration.**2)** For the national NBS programmes, the following protocols are used: IRT-DNA-IRT (7), IRT-IRT (4), IRT-DNA (3), IRT-PAP-DNA (1), and IRT-PAP-IRT (1). 56% (9/16) are using a fixed cut-off for IRT. There was a big variability in IRT cut-offs, age at testing and second tier strategies. Protocols that include DNA testing had better PPV (mean 0.50, range 0.12-0.91) compared to those without (mean 0.24, range 0.03-0.48), but at the cost of recognising more infants with an inconclusive diagnosis and carriers. **3)** Lack of financial support and cutting back health care budget are the main barriers to implementation of NBS, reported by 7 of 11, however 2 reported they expect to start a NBS programme within a year.

Conclusions: NBS programmes are now covering the majority of the population in Western Europe. There are numerous protocols, which to some degree reflect the different economic and health systems as well as the genetic diversity of the European population. However the degree of variability is not optimal. There is an urgent need to provide a more robust framework on which to evaluate the impact of NBS for CF and provide clearer information and

consensus on best practice.
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