

# 2014 - NEONATAL SCREENING FOR BIOTINIDASE DEFICIENCY.

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## Summary

**Introduction:** Neonatal screening programmes are a secondary prevention strategy that seeks to halt a disease's progression by ensuring early treatment in the presymptomatic stage and thereby improve its prognosis. Prior to implementing any neonatal screening programme, it is of the utmost importance to ascertain its efficacy, feasibility and cost-effectiveness by carrying out an assessment of the screening test, diagnostic confirmation tests, disease-management protocols and system used to evaluate the programme, which should, moreover, ensure adequate care at all screening stages. This assessment report was drawn up at the request of the National Health System Interterritorial Council's Services, Insurance & Finance Committee, in response to a proposal from the Galician Regional Health Authority.

**Objectives:** To assess existing evidence on the clinical effectiveness of neonatal biotinidase deficiency screening; and specifically, to analyse the incidence and/or prevalence, clinical characteristics and prognosis of the disease, as well as the screening test's analytical validity (sensitivity, specificity and predictive values) and benefits in terms of its effectiveness vis-à-vis morbidity and mortality.

**Methods:** Systematic review of the literature, covering the main biomedical databases (Medline, Embase, Cochrane Library Plus, HTA (Health Technology Assessment), DARE (Database of Abstracts of Reviews of Effectiveness), NHS EED (NHS Economic Evaluation Database), ISI Web of Science and Índice Médico Español (IME), among others). We used two search strategies -one centred on the epidemiology, clinical characteristics, morbidity, mortality, diagnosis and treatment of biotinidase deficiency, and the other centred on the screening of the disease- spanning the period 1 January 2009 to July 2013. After reading the abstracts of the papers retrieved, only those that met the pre-established inclusion/exclusion criteria were selected. The studies were then classified, and the data extracted and summarised in evidence tables.

**Results, discussion and conclusions:** See pdf summary below.

## DOCUMENTOS RELACIONADOS

[Summary \(pdf\)](#)

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