Cribado neonatal de la galactosemia clásica. Revisión Sistemática [Neonatal screening for classic galactosemia. Systematic review]
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Record Status
This is a bibliographic record of a published health technology assessment from a member of INAHTA. No evaluation of the quality of this assessment has been made for the HTA database.

Citation

Authors' objectives
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. Its overall objective was to analyse existing evidence on neonatal galactosaemia screening to help decide on its introduction into the National Health System service portfolio. A specific objective was to assess different aspects of the disease, treatment and screening test which might serve to support decision-making. Secondary objectives were to recover information on the different screening strategies used world-wide (diagnostic tests, confirmatory tests, cut points).

Authors' conclusions
- As classical galactosaemia progresses, severe neonatal complications can develop. With early detection and treatment, it is estimated that most of the severe acute complications and deaths due to sepsicaemia which occur in the first weeks of life could be prevented. - Existing information highlights that fact that, despite early dietary treatment, patients with classical galactosaemia may present with important complications in the medium-long term (cognitive deficiency, speech/language disorders, motor disorders, etc.). - Based on data drawn from four descriptive studies, the screening programmes would be estimated to have a sensitivity and specificity of around 100%, though these data should be interpreted with caution since there are no studies that confirm negative cases. - Screening tests generally display a high FP rate and a low PPV. The only studies that obtained a PPV of over 20% were those that reported data from the Swedish screening programme and the Galician Regional programme, which uses four steps. - Screening protocols differ considerably in terms of screening strategy, analytical methods and cut points (GALT activity ≤15% to ≤30%). Depending on the cut point, there is a different probability of having classified mild heterozygous forms or benign Duarte variants as positive. In the long term, the lack of differentiation of certain mild heterozygous forms and the Duarte variant could be regarded as an adverse screening effect. - There is not enough information to establish whether screening is really effective in reducing the frequency of severe adverse effects or neonate mortality. On the basis of indirect assumptions and descriptive data from the Swedish and West German programmes, screening could be assumed to be capable of reducing the risk of mortality/morbidity provided that the screening results are obtained during the first week of life, though this assumption should be confirmed by suitably designed studies. - Currently, there are no comparative studies that would make it possible to establish whether neonatal galactosaemia screening is really superior to other secondary prevention measures aimed at preventing severe acute complications and mortality in the neonatal period (alert protocols, opportunistic screening). - The programme's cost-effectiveness has still to be assessed and its potential impact on the national health system is unknown.

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