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Title **Early Detection of Mucopolysaccharidosis and Oligosaccharidosis by Population Screening in the Newborn Period. Systematic Review**

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Aim

To systematically review the efficacy and effectiveness of neonatal mucopolysaccharidosis (MPS) and oligosaccharidosis screening, using tandem mass spectrometry or other analytical techniques.

Conclusions and results

Conclusion: The lack of quality studies that analyze the different aspects of neonatal mucopolysaccharidosis and oligosaccharidosis screening means that their inclusion in neonatal screening programs of congenital errors of metabolism cannot be recommended. Selective performance of diagnostic tests for MPS and oligosaccharidosis would, however, appear wise among patients judged to be at risk, specifically, those who present with metabolic disorders or scant weight gain at birth or in the first weeks of life. To enable active and regular follow-up in such cases, it is advisable to establish a case registry, which for healthcare, teaching, and research purposes would pool information on incidence, trends, survival, and other aspects linked to neonatal screening of these diseases.

Results: Of the papers retrieved in the bibliographic search, 35 were selected (4 clinical trials, 24 case series, 7 single cases). Diagnosis (10 papers) was performed by enzymatic activity assay of deficient blood enzyme (Delfia, tandem mass spectrometry, multiplex or fluorescence) or detection of urinary glycosaminoglycans (dimethyme thylene blue, Alcian blue or bidimensional electrophoresis). Treatment (25 papers) consisted of: enzyme replacement therapy with laronidase (MPS I), rhASB (MPS VI) and idursulfase (MPS II); hematopoietic stem cell transplantation (MPS I, MPS VII, aspartylglucosaminuria, fucosidosis); umbilical cord blood transplantation (MPS I); bone marrow transplantation (eg, MPS I, MPS VI, MPS VII); and peripheral-blood stem cell transplantation.

Recommendations

Epidemiological studies should be conducted to enable us to ascertain the distribution and frequency of these diseases, the validity of diagnoses based on enzymatic determination with tandem mass spectrometry, and the cost effectiveness of the new therapies. We should take advantage of the experience of existing screening teams and routine collection of specimens on paper strips to design and undertake parallel studies (aimed at long-term assessment of results and enabling a definitive conclusion to be arrived at regarding the use of these techniques and implementation of this type of screening in the context of a neonatal program).

Methods

Systematic review of literature from January 1996 to December 2006. Papers were selected using inclusion and exclusion criteria based on study design, patient characteristics, and outcome variables analyzed. Two reviewers, independently, carried out the selection of the studies, critical reading, data extraction, and evaluation of the methodological quality.

Further research/reviews required

The main biomedical databases used were MEDLINE, EMBASE, NHS Centre for Reviews and Dissemination, Health Technology Assessment (HTA), Database of Abstracts of Reviews of Effects (DARE) and Cochrane Library Plus.

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