Cribado neonatal de la anemia falciforme [Neonatal screening of sickle cell anemia]

Citation

Authors' objectives
To assess the efficacy/effectiveness and safety of neonatal sickle cell disease screening.

Authors' conclusions
The aim of a neonatal screening programme for sickle–cell disease is to detect the disease in a presymptomatic stage, to establish early treatment aimed at reducing morbidity and mortality caused by sickle cell disease in childhood. This proposed early treatment is based on two pillars: prevention of pneumococcal infection (with antibiotic prophylaxis and pneumococcal vaccination) and health education of parents in order to identify acute complications in its early stage.

There is no direct evidence, based on comparative studies, of the efficacy of neonatal screening programmes in reducing morbidity or mortality among children with sickle cell disease. There is evidence, based on good quality studies, showing that prophylactic penicillin reduces incidence of pneumococcal infections in children with homozygous sickle cell anaemia, which provides indirect evidence to indicate that children diagnosed during the presymptomatic stage are benefited by early administration of prophylactic antibiotics. The long-term effect of neonatal screening on chronic multiorganic complications of sickle-cell anaemia is not known. In population registers, a significant reduction over time in mortality among children with sickle-cell anaemia has been described but the contribution of newborn screening to this improvement in prognosis cannot be formally established. Current techniques for detecting haemoglobin disorders have demonstrated their validity, with a very high sensitivity and specificity, but they detect major drepanocytic syndromes, such as sickle cell trait carrier status, as well as other haemoglobinopathies of no clinical relevance or whose clinical relevance is unknown. Sickle cell disease is a hereditary disease, with a clear ethnic pattern, which mainly affects people originally from regions where malaria was endemic, though, as a result of migrations, incidence has risen in other regions, such as Europe and North America. In Spain there are no data on the real prevalence of sickle cell disease. There is an overall estimation, based on migratory flows, which classifies Spain in the group of countries with low risk of incidence of sickle-cell anaemia, but the distribution of immigrants is very variable throughout the country's regions (Autonomous Communities). The effectiveness and the cost-effective ratio of sickle cell disease screening depend on the prevalence of the disease.

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