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[Intervention Review]

Newborn screening for galactosaemia

Rohollah Lak¹, Bahareh Yazdizadeh², Majid Davari³, Mojtaba Nouhi⁴, Roya Kelishadi⁵

¹Vice-Chancellery for Health, Isfahan University of Medical Sciences, Isfahan, Iran. ²Knowledge Utilization Research Center, Tehran University of Medical Sciences, Tehran, Iran. ³Department of Pharmacoeconomics and Pharmaceutical Administration, Faculty of Pharmacy, Tehran University of Medical Sciences, Tehran, Iran. ⁴Health Equity Research Center, Tehran University of Medical Sciences, Tehran, Iran. ⁵Child Growth and Development Research Center, Research Institute for Primordial Prevention of Non-Communicable Diseases, Isfahan University of Medical Sciences, Isfahan, Iran

Contact address: Rohollah Lak, Vice-Chancellery for Health, Isfahan University of Medical Sciences, Isfahan, Iran. lak2346@yahoo.com.

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ABSTRACT

Background

Classical galactosaemia is an autosomal recessive inborn error of metabolism caused by a deficiency of the enzyme galactose-1-phosphate uridyltransferase. This is a rare and potentially lethal condition that classically presents in the first week of life once milk feeds have commenced. Affected babies may present with any or all of the following: cataracts; fulminant liver failure; prolonged jaundice; or *Escherichia coli* sepsis. Once the diagnosis is suspected, feeds containing galactose must be stopped immediately and replaced with a soyabased formula. The majority of babies will recover, however a number will not survive. There are long-term complications of galactosaemia, despite treatment, including learning disabilities and female infertility. It has been postulated that galactosaemia could be detected on newborn screening and this would prevent the immediate severe liver dysfunction and sepsis.

Objectives

To assess whether there is evidence that newborn screening for galactosaemia prevents or reduces mortality and morbidity and improves clinical outcomes in affected neonates and the quality of life in older children.

Search methods

We searched the Cochrane Cystic Fibrosis and Genetic Disorders Group Trials Register comprising references identified from electronic database searches, handsearches of relevant journals and conference abstract books. We also searched online trials registries and the reference lists of relevant articles and reviews.

Date of the most recent search of Cochrane Cystic Fibrosis Group's Trials Register: 18 December 2017.

Date of the most recent search of additional resources: 11 October 2017.

Selection criteria

Randomised controlled studies and controlled clinical studies, published or unpublished comparing the use of any newborn screening test to diagnose infants with galactosaemia and presenting a comparison between a screened population versus a non-screened population.

Data collection and analysis

No studies of newborn screening for galactosaemia were found.

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Main results

No studies were identified for inclusion in the review.

Authors' conclusions

We were unable to identify any eligible studies for inclusion in this review and hence it is not possible to draw any conclusions based on randomised controlled studies. However, we are aware of uncontrolled studies which support the efficacy of newborn screening for galactosaemia. There are a number of reviews and economic analyses of non-trial literature suggesting that screening is appropriate.

PLAIN LANGUAGE SUMMARY

Screening newborn babies for galactosaemia

Review question

We reviewed the evidence for screening newborn babies for galactosaemia in order to prevent or reduce death and illness, to improve clinical outcomes in affected babies and to improve the quality of life in affected older children.

Background

Galactosaemia is an inherited disease that affects the body's ability to breakdown the milk sugar galactose. Newborn babies with galactosaemia can have a variety of symptoms in the first weeks of life including poor feeding, cataracts, jaundice, an enlarged liver with liver failure or severe infection. Without treatment, babies with galactosaemia are often very unwell and highly likely to die from liver failure. Unfortunately, despite treatment, long-term complications for people with galactosaemia include learning difficulties and fertility problems (in females).

Search date

The evidence is current to: 11 October 2017.

Study characteristics

No studies were identified for inclusion in the review.

Key results

No suitable studies were found, but we are aware of some uncontrolled studies which suggest newborn screening for galactosaemia and early treatment can reduce death and illness. Future research is needed to provide robust evidence for or against screening.

Quality of the evidence

We have not identified any relevant studies for inclusion in this review.