HTA - - Health Technology Assessment Report

Neonatal screening for cystic fibrosis

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List of included studies (1)

CHOPIN study (it followed a Health Council's recommendation to undertake research into better screening methods)

Participants

72,874 neonates in Noord-Brabant, Utrecht, Gelderland and Limburg

Interventions

Heel prick blood was analysed in two ways: 1) by screening for IRT followed by analysis of PAP; and 2) by screening for IRT followed by mutation analysis (36 mutations possibly followed by EGA which consists of determining the sequence of exons and intron-exon boundaries of the CFTR gene). IRT-PAP method: IRT positive if > 50 Î¼g/l and PAP > 1.8 Î¼g/l or IRT positive if > 100 Î¼g/l and PAP > 1.0 Î¼g/l. IRT-DNA method: IRT positive if > 50 Î¼g/l and two mutations were found in the first mutation analysis; in the case of one mutation being found EGA was used to find a possible second mutation. Sweat tests were performed on all neonates with a positive test result.

Outcome measures

Evaluate different screening methods; cost-effectiveness of different screening methods and opinion of parents.

Main results

Using IRT 50 and PAP 1,8 or IRT 100 and PAP 1,0: 119 abonrmal results; 10 classical CF; 0 non-classical CF; 0 carriers. Using IRT 50 followed by DNA-EGA: 20 abonrmal results; 10 classical CF; 9 non-classical CF; 89 carriers. Using IRT-PAP-DNA-EGA (calculated result): 12 abnormal results; 10 classical CF; 2 non-classical CF; 5 carriers. Parents from ethnic minority groups experienced many more problems with the information on heel prick screening. Heredity appears to be a particularly difficult subject for them. Little use was made of the heel prick information provided on the RIVM website. Parents would like to be informed if they are carriers. Parents who receive positive test results are extremely shocked and worried. After false-positive results, many parents continue to be concerned even after a follow-up analysis has confirmed that the child does not have CF. Most parents feel that they have to wait too long (an average of four days) for the appointment for the follow-up analysis. The average period between carrying out the sweat test and hearing the results was 2-3 days, in a range of 0-28 days.

Authors' conclusions

Additional benefits of screening: the Committeeâ€™s conclusion on the grounds of the above is that neonatal screening for CF clearly provides additional benefits and that the benefits have been confirmed by the results of research conducted since 2005. Sensitivity and specificity: the Committee concludes that high specificity screening for CF can be performed using the IRT-PAP-DNA-EGA protocol. Bearing sensitivity considerations in mind, the Committee recommends that screening should be carried out using the IRT-PAP-DNA-EGA protocol, with the aforementioned failsafe procedure for the time being. Evaluations of mutation analysis may indicate that changes in the mutation panel and/or the failsafe procedure are advisable. Cost and savings: the Committee concludes that using these methods for neonatal screening does not appear to involve any exceptionally high costs and that doing so could lead to savings. There is little difference between the costs of the screening methods tested and the choice therefore has to be based on other more important factors, such as the stress screening causes for neonates and their parents. Carriers: no carriers are found when the IRT-PAP method is used, at least not by the screening process. Parents are generally of the opinion that they should be able to choose whether information is provided on whether or not their child is a carrier. Identification of carriers in neonatal screening for CF only occurs for a small number of neonates, the annual average being 12 neonates in the case of the IRTPAP-DNA-EGA strategy. Information: parents are well informed about screening and that they are positive about the information they receive on CF screening. 15% of obstetricians and screeners â€˜sometimesâ€™ provide information and 13% never do so. Half of the people who provided information did not point out that participation in CF research is voluntary. Moreover, 35% of those who provided the information never provided the brochure and 19% â€˜seldomâ€™ did so. Heredity and being a carrier appear to be problem areas in the information, especially in the case of immigrant parents and those with a low level of education. Information should be an important point for attention, especially with regard to the possibility of being a carrier. Improvements are also required in the field of refresher training courses for care providers. -

http://www.gezondheidsraad.nl/sites/default/files/201001E.pdf

See also

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Keywords

Neonatal Screening; non pharmacological intervention - diagn; screening; diagnostic procedures;