primary studies - published, non RCT

Newborn screening for cystic fibrosis in Wisconsin: comparison of biochemical and molecular methods.

Code: PM9164776 **Year:** 1997 **Date:** 1997 **Author:** Gregg RG

Participants

group I: 220 862 neonates; group II: 104 308 neonates

Interventions

groupm I: IRT; group II: IRT/DNA. For the IRT protocol, neonates with an IRT >/=180 ng/mL were considered positive, and the standard sweat chloride test was administered to determine CF status. For the IRT/DNA protocol, samples from the original dried-blood specimen on the Guthrie card of neonates with an IRT >/=110 ng/mL were tested for the presence of the DeltaF508 CFTR allele, and if the DNA test revealed one or two DeltaF508 alleles, a sweat test was obtained.

Outcome measures

specificity, sensitivity, positive predictive value, false-positive IRT/DNA population, type of CFTR mutations, cost of screening

Main results

Both screening procedures had very high specificity. The sensitivity tended to be higher with the IRT/DNA protocol, but the differences were not statistically significant. The positive predictive value of the IRT/DNA screening protocol was 15.2% compared with 6.4% if the same samples had been screened by the IRT method. Assessment of the false-positive IRT/DNA population revealed that the two-tier method eliminates the disproportionate number of infants with low Apgar scores and also the high prevalence of African-Americans identified in the previous study of newborns with high IRT levels. 55% of DNA-positive CF infants were homozygous for DeltaF508 and 40% had one DeltaF508 allele. Adding analyses for 10 more CFTR mutations has only a small effect on the sensitivity but is likely to add significantly to the cost of screening.

Authors' conclusions

Advantages of the IRT/DNA protocol over IRT analysis include improved positive predictive value, reduction of false-positive infants, and more rapid diagnosis with elimination of recall specimens.

http://www.mrw.interscience.wiley.com/cochrane/clcentral/articles/514/CN-00208514/frame.html

See also

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Keywords

Newborn; non pharmacological intervention - diagn; screening;