primary studies - published, non RCT

Novel strategies in newborn screening for cystic fibrosis: A prospective controlled study.

Code: PM22271776 **Year:** 2012 **Date:** 2012 **Author:** Vernooij-Van Langen AMM

Study design (if review, criteria of inclusion for studies)

Prospective study

Participants

145,499 infants

Interventions

Two strategies were performed in all newborns. In the first strategy, concentrations of immunoreactive trypsinogen (IRT) and pancreatitis-associated protein (PAP) were measured. In the second method, samples with IRT â‰¥60 Î¼g/litre were analysed for 36 CFTR mutations, followed by sequencing when a single mutation was detected. Tests were positive only with two identified CFTR mutations.

Outcome measures

Sensitivity, specificity and positive predictive value (PPV) of both screening strategies.

Main results

145,499 infants were screened. The IRT/PAP approach showed a sensitivity of 95.0%, a specificity of 99.897% and a PPV of 12.3%. Test properties for the IRT/DNA/sequencing strategy were respectively 100%, 100% and 64.9%. Combining both strategies (IRT/PAP/DNA/sequencing) led to a sensitivity of 95.0%, a specificity of 100% and a PPV of 87.5%.

Authors' conclusions

In conclusion, all strategies performed well. Although there was no statistically significant difference in test performance, the IRT/DNA/sequencing strategy detected one infant that was missed by IRT/PAP (/DNA/sequencing). IRT/PAP may be the optimal choice if the use of DNA technology must be avoided. If identification of carriers and equivocal diagnosis is considered an important disadvantage, IRT/PAP/DNA/sequencing may be the best choice.

http://thorax.bmj.com/content/67/4/289.long

See also

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Keywords

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