primary studies - published, non RCT

Prospective and parallel assessments of cystic fibrosis newborn screening protocols in the Czech Republic: IRT/DNA/IRT versus IRT/PAP and IRT/PAP/DNA.

Code: PM22581207 **Year:** 2012 **Date:** 2012 **Author:** Krulisova V

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

Prospective assessment nationwide survey.

Participants

106,522 Czech newborns

Interventions

Immunoreactive trypsinogen (IRT)/DNA/IRT protocol currently in use nationwide, versus the IRT/pancreatitis-associated protein (PAP) and IRT/PAP/DNA CF NBS protocols. Dried blood spots (DBS) from 106,522 Czech newborns were examined for IRT concentrations. In the IRT/DNA/IRT protocol, DNA-testing was performed for IRTâ€‰â‰¥â€‰65 ng/mL.

Outcome measures

Dried blood spots (DBS) from 106,522 Czech newborns were examined for IRT concentrations.

Main results

In the IRT/DNA/IRT protocol, DNA-testing was performed for IRTâ€‰â‰¥â€‰65 ng/mL. Newborns with IRTâ€‰â‰¥â€‰200 ng/mL and no detected cystic fibrosis transmembrane conductance regulator gene (CFTR) mutations were recalled for a repeat IRT. In the same group of newborns, for both parallel protocols, PAP was measured in DBS with IRTâ€‰â‰¥â€‰50 ng/mL. In PAP-positive newborns (i.e., â‰¥1.8 if IRT 50-99.9 or â‰¥1.0 if IRTâ€‰â‰¥â€‰100, all in ng/mL), DNA-testing followed as part of the IRT/PAP/DNA protocol. Newborns with at least one CFTR mutation in the IRT/DNA/IRT and IRT/PAP/DNA protocols; a positive PAP in IRT/PAP; or a high repeat IRT in IRT/DNA/IRT were referred for sweat testing.

Authors' conclusions

CONCLUSION:the combined results of the utilized protocols led to the detection of 21 CF patients, 19 of which were identified using the IRT/DNA/IRT protocol, 16 using IRT/PAP, and 15 using IRT/PAP/DNA. Decreased cut-offs for PAP within the IRT/PAP protocol would lead to higher sensitivity but would increase false positives. Within the IRT/PAP/DNA protocol, decreased PAP cut-offs would result in high sensitivity, an acceptable number of false positives, and would reduce the number of DNA analyses. Thus, we concluded that the IRT/PAP/DNA protocol would represent the most suitable protocol in our conditions.

http://link.springer.com/article/10.1007%2Fs00431-012-1747-z

See also

European Journal of Pediatrics. 2012 Aug;171(8):1223-9

Keywords

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