primary studies - published, non RCT

Cystic Fibrosis: A Simple and Customized Strategy for Genetic Screening Able to Detect Over 90% of Identified Mutated Alleles in Brazilian Newborns.

Code: PM32185651 **Year:** 2020 **Date:** 2020 **Author:** Rispoli T

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

Non randomized trial

Participants

CF patients

Interventions

The incorporation of molecular genetic testing into cystic fibrosis (CF) screening programs. A mini-sequencing assay was standardized using single-base extension in a previously genotyped control sample.

Outcome measures

Specificity of the diagnostic strategy

Main results

The inclusion of the 25 variants in the current newborn screening program increased the identification rates of two alleles from 33 to 52.43% in CF patients. This new approach was able to detect a total of 37 variants, which represents 93.01% of all mutated alleles described in the last CF Brazilian Register.

Authors' conclusions

Mini-sequencing for the simultaneous detection of 25 CFTR gene variants improves the screening of Brazilian newborns and decreases the number of inconclusive cases. This method uses minimal hands-on time and is suited for rapid screening, which reduces sample processing costs.

http://dx.doi.org/10.1007/s40291-020-00456-9

See also

Mol Diagn Ther. 2020 Jun;24(3):315-325. doi: 10.1007/s40291-020-00456-9.

Keywords

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