*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.

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### 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.

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### 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;