primary studies - published RCT

Incorrect recall of residual risk three years after carrier screening for cystic fibrosis: a comparison of two-step and couple screening.

Code: PM10411814 **Year:** 1999 **Date:** 2003 **Author:** Marteau TM

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

double-blind, placebo-controlled, crossover trial

Participants

patients with stop mutations in CFTR or patients homozygous for the DeltaF508 mutation

Interventions

two drops containing gentamicin (0.3 percent, or 3 mg per milliliter) or placebo in each nostril three times daily for two consecutive periods of 14 days.

Outcome measures

Nasal potential difference was measured at base line and after each treatment period. Nasal epithelial cells were obtained before and after gentamicin treatment from patients carrying stop mutations, and the C-terminal of surface CFTR was stained.

Main results

Gentamicin treatment caused a significant reduction in basal potential difference in the 19 patients carrying stop mutations (from -45+/-8 to -34+/-11 mV, P=0.005) and a significant response to chloride-free isoproterenol solution (from 0+/-3.6 to -5+/-2.7 mV, P

Authors' conclusions

In patients with cystic fibrosis who have premature stop codons, gentamicin can cause translational "read through," resulting in the expression of full-length CFTR protein at the apical cell membrane, and thus can correct the typical electrophysiological abnormalities caused by CFTR dysfunction.

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See also

Am J Obstet Gynecol. 1999 Jul;181(1):165-9.

Keywords

Adolescent; Adult; Anti-Bacterial Agents; Child; CFTR Modulators; Gentamicin; Intranasal; pharmacological_intervention; Bacterial Infections; Respiratory Tract Infections; Respiratory Tract Diseases; Infection; Aminoglycosides;