primary studies - published RCT

Effectiveness of PTC124 treatment of cystic fibrosis caused by nonsense mutations: a prospective phase II trial.

Code: PM18722008 **Year:** 2008 **Date:** 2008 **Author:** Kerem E

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

phase II prospective trial

Participants

adults with cystic fibrosis who had at least one nonsense mutation in the CFTR gene.

Interventions

Patients were assessed in two 28-day cycles. During the first cycle, patients received PTC124 at 16 mg/kg per day in three doses every day for 14 days, followed by 14 days without treatment; in the second cycle, patients received 40 mg/kg of PTC124 in three doses every day for 14 days, followed by 14 days without treatment.

Outcome measures

The primary outcome had three components: change in CFTR-mediated total chloride transport; proportion of patients who responded to treatment; and normalisation of chloride transport, as assessed by transepithelial nasal potential difference (PD) at baseline, at the end of each 14-day treatment course, and after 14 days without treatment.

Main results

Transepithelial nasal PD was evaluated in 23 patients in the first cycle and in 21 patients in the second cycle. Mean total chloride transport increased in the first treatment phase, with a change of -7.1 (SD 7.0) mV (p

Authors' conclusions

In patients with cystic fibrosis who have a premature stop codon in the CFTR gene, oral administration of PTC124 to suppress nonsense mutations reduces the epithelial electrophysiological abnormalities caused by CFTR dysfunction.

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See also

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Keywords

PTC124; Ataluren; CFTR Modulators; pharmacological_intervention;