Title	*17.* ***A phase 3 extension study of Ataluren (PTC124) in patients with nonsense mutation cystic fibrosis (PTC124-GD-021e-CF)***
Project Coordinator	C. Braggion, MD (c.braggion@meyer.it), PI as partner of CTN
Internal Collaborators	Anna Silvia Neri, MD Michela Francalanci, Biologist, study coordinator
Study design	A phase 3 extension, open study, in which all patients which completed the previous study (PTC124-GD-021-CF) receive the study drug Ataluren for 96 weeks. EudraCT Number 2014-005355-83
Grant by	PTC Therapeutics, Inc.
Background and aims	In ~10% of patients with CF, the causative defect in the CFTR gene is a nonsense mutation that truncates CFTR protein production by introducing a premature stop codon into the CFTR messenger ribonucleic acid (mRNA). Ataluren (PTC124) was shown to promote ribosomal readthrough of mRNA containing a premature stop codon and so to overcome the genetic defect in patients for whom a nonsense mutation causes CF. A previous phase 3 study showed that Ataluren improved lung function (FEV1), when patients did not inhale tobramycin, and was generally well tolerated. The primary objective of this study is to evaluate the ability of ataluren to improve pulmonary function relative to placebo as assessed by FEV1. Secondary objectives are the effect of Ataluren on number of pulmonary exacerbations, quality of life and body weight, and the safety of study drug.
Inclusion criteria	Completion of study treatment in the previous Phase 3, double blind study protocol (protocol PTC124-GD-021-CF).
Exclusion criteria	Known hypersensitivity to any of the ingredients or excipients of the study drug.
Methods	Oral Ataluren will be administered orally 3 times a day to all included patients. Study visits include physical exam, spirometry, blood and urine samples, administration of quality of life questionnaire and will be performed every 12 weeks for 2 years. Adverse events and concomitant medications willl be recorded from screening to the end of study. ECG and renal ultrasound will be performed at the beginning and at the end of study.
Expected results and anticipated output	The objective of the study is to evaluate the long term safety and efficacy of Ataluren in patients with nonsense mutations, who previously participated in the placebo-controlled study PTC124-GD-021-CF.
Start of recruitment	February 2016
End of experimental plan	After 96 weeks of study drug treatment.
Pubblication on medical Journal	