Introduction

Cystic fibrosis is an autosomal recessive disease caused by mutations in the CFTR gene (Grasemann and Ratjen, 2023). CFTR, also known as cystic fibrosis transmembrane conductance regulator, encodes a chloride channel protein that regulates the water-electrolyte balance on the surface of multiple organs. The most common mutation (approximately 85%) is the deletion of three base pairs in CFTR at position 508 (F508del), resulting in the loss of phenylalanine in the channel protein. Other variants of CFTR include premature stop codons, abnormal channel gating or conductance, transcription-related reduction, and stability deficiency.

CFTR dysfunction typically results in impaired ion transport and absorption in epithelial cells. People with CFTR dysfunction probably suffer reduced chloride absorption in the sweat-gland ducts, which consequently increases sweat chloride concentrations