Abstract

Background: ERp29 was previously found to rescue the Cystic Fibrosis Transmembrane Conductance Regulator (CFTR) trafficking in both wild type and mutation F508-CFTR, the most common Cystic Fibrosis-causing mutation. Mutations Y1014C, F1016C and F1016S were researched as they were hypothesized to be in the same location as ERp29. It is expected that rescued trafficking would provide genetic therapy for Cystic Fibrosis patients.

Methods: Cell culture, PCR, immunohistochemisty and immunoblots were performed to determine protein movement and location within a cell.

Results: Evidence supported that these mutations are located in between the nucleus and the endoplasmic reticulum, directly after the CFTR is synthesized.

Conclusion: Since these mutations still do not traffick properly to the epithelial membrane in order to be functional, ERp29 may not have the same effect on this CFTR as the WT and mutated CFTR previously researched. Future research will include determining that barriers keep ERp29 from rescuing trafficking in these mutations.