Familial Idiopathic Pulmonary Fibrosis:

Mutations in TERT and TERC as Potential Risk Factors

Idiopathic Pulmonary Fibrosis is a progressive scarring lung disease, a subset of which is characterized as Familial Idiopathic Pulmonary Fibrosis. Shortened telomeres have been shown to have a correlation with both sporadic and familial cases. Telomerase, the enzyme responsible for adding repeating TTAGGG segments to the ends of our chromosomes, consists of a protein that controls polymerase activity (and is encoded for by TERT) and an RNA molecule (TERC) that serves as a template for the segments. In 2004, a paper noted that heterozygous TERC mutations were related to reduced telomerase activity and concluded a mechanism of haploinsufficiency. A 2007 study noted heterozygous TERT mutations and related their effects to pulmonary fibrosis. In 2008, a study revealed that a significant portion of individuals with IPF (25%) or FPF (37%) have telomere lengths in the lowest decile compared to control groups. Interestingly, the authors also found that 23% of IPF patients and 24% of FPF patients have short telomeres but no TERT or TERC coding mutations. A different paper published in 2008 suggests that telomere shortening, not mutations in TERT or TERC, cause pulmonary fibrosis.

Works Cited


