Genetic Modulators of Pulmonary Function in Cystic Fibrosis & Genetic Susceptibility Factors of Emphysema

Jennifer S. Yarden

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This is a Ph.D. thesis. CF is one of the most common lethal autosomal recessive affecting the Caucasian population with an incidence of about 1 in 2500 births. Clinical manifestations are attributed to mutations in the CFTR gene, which encodes an epithelial chloride channel.

Specific mutations can influence the severity and progression of CF disease, but wide variations within genetically homogenous subgroups illustrate that other determinants of the clinical status do exist.

For example, certain CF phenotypes such as pancreatic insufficiency are clearly highly associated with the CFTR genotype. However, other phenotypes such as the severity of pulmonary disease are clearly influenced by other genes and environmental factors.

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