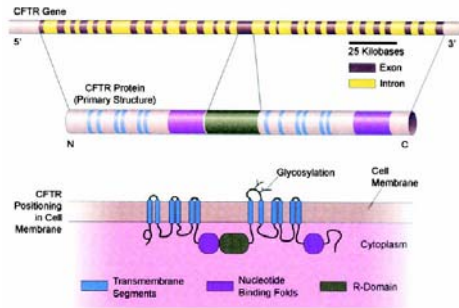




# Cystic Fibrosis



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# Cystic Fibrosis (CF)

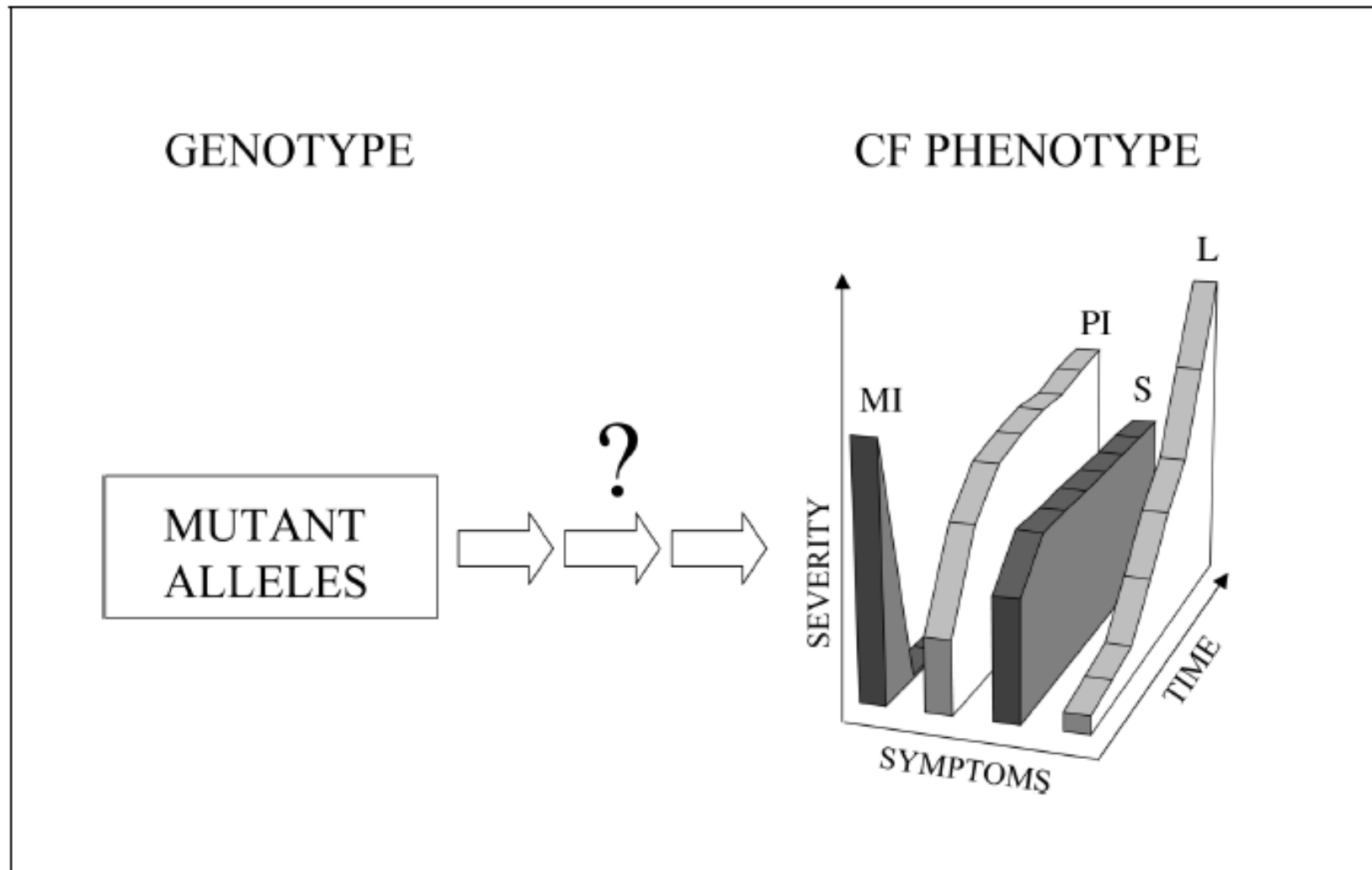
- the most common autosomal recessive genetic disorder of Caucasians
- 1:3,600 livebirth
- Cystic Fibrosis Transmembrane conductance Regulator (*CFTR*) gene on chromosome 7
- CFTR was first identified in 1989 at SickKids, Toronto by Dr. Lap-Chee Tsui



# CFTR mutations

- There are currently 1604 mutations
- $\Delta F508$  is the most common mutation, accounting for 70% of CF worldwide
- genetic variations influence severity of CF

CFTR mutation database:  
<http://www.genet.sickkids.on.ca/cftr/StatisticsPage.html>



MI= Meconium ileus, PI= Pancreatic insufficiency, S= Sweat Cl, L= Lungs