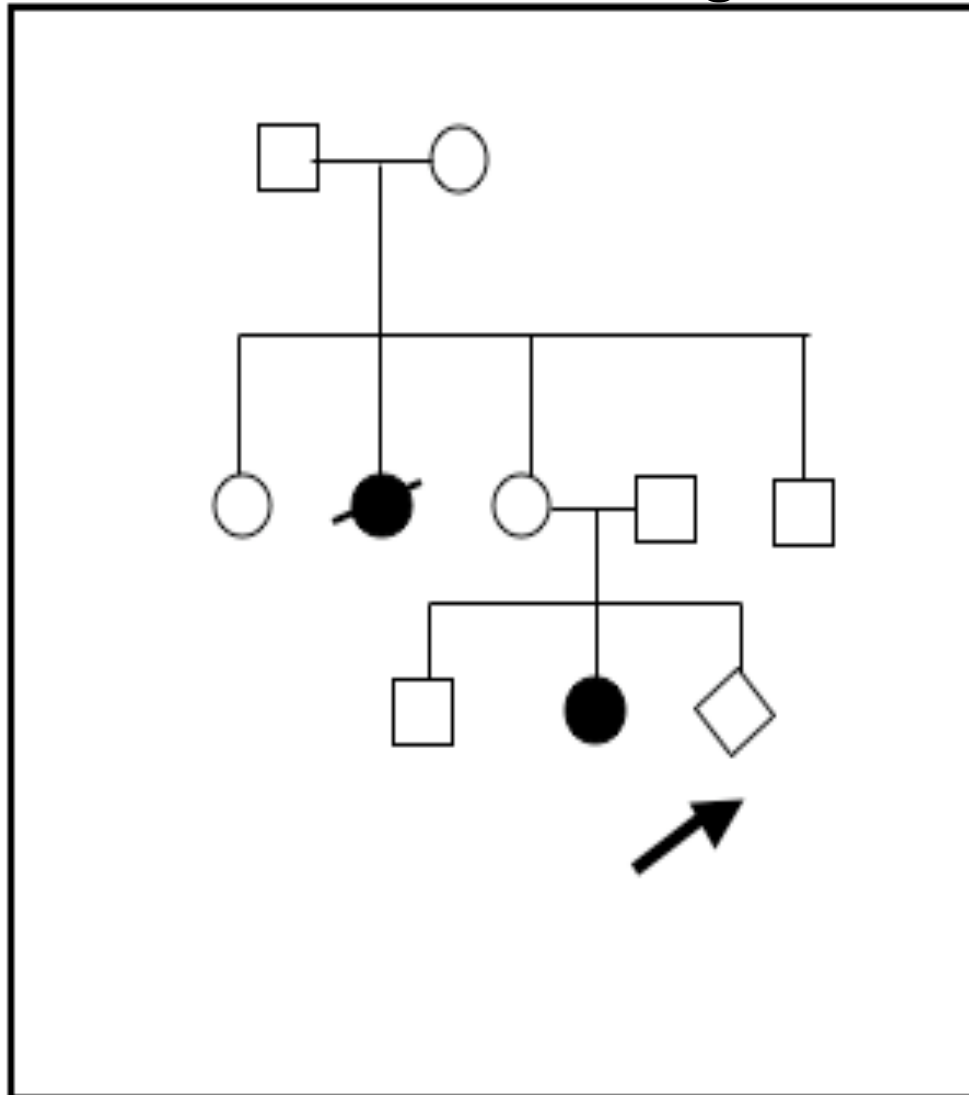


# Caccia al gene della fibrosi cistica

## Storia di una famiglia



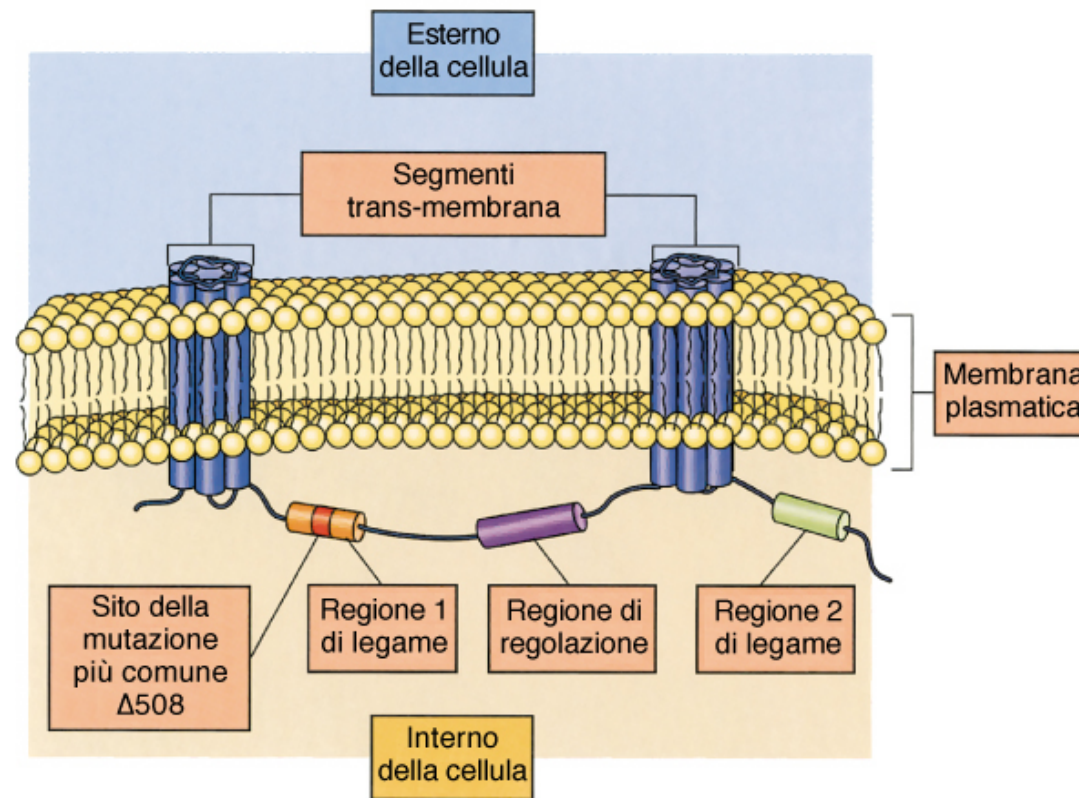
Mutazioni  
più frequenti

Mutazione	esone	%
p.F508del	10	81,0
p.Gly551Asp	11	3,5
p.Gly542X	11	1,1
c.621+1G>T	Introne 4	1,0
c.1898+1G>A	Introne 12	0,9

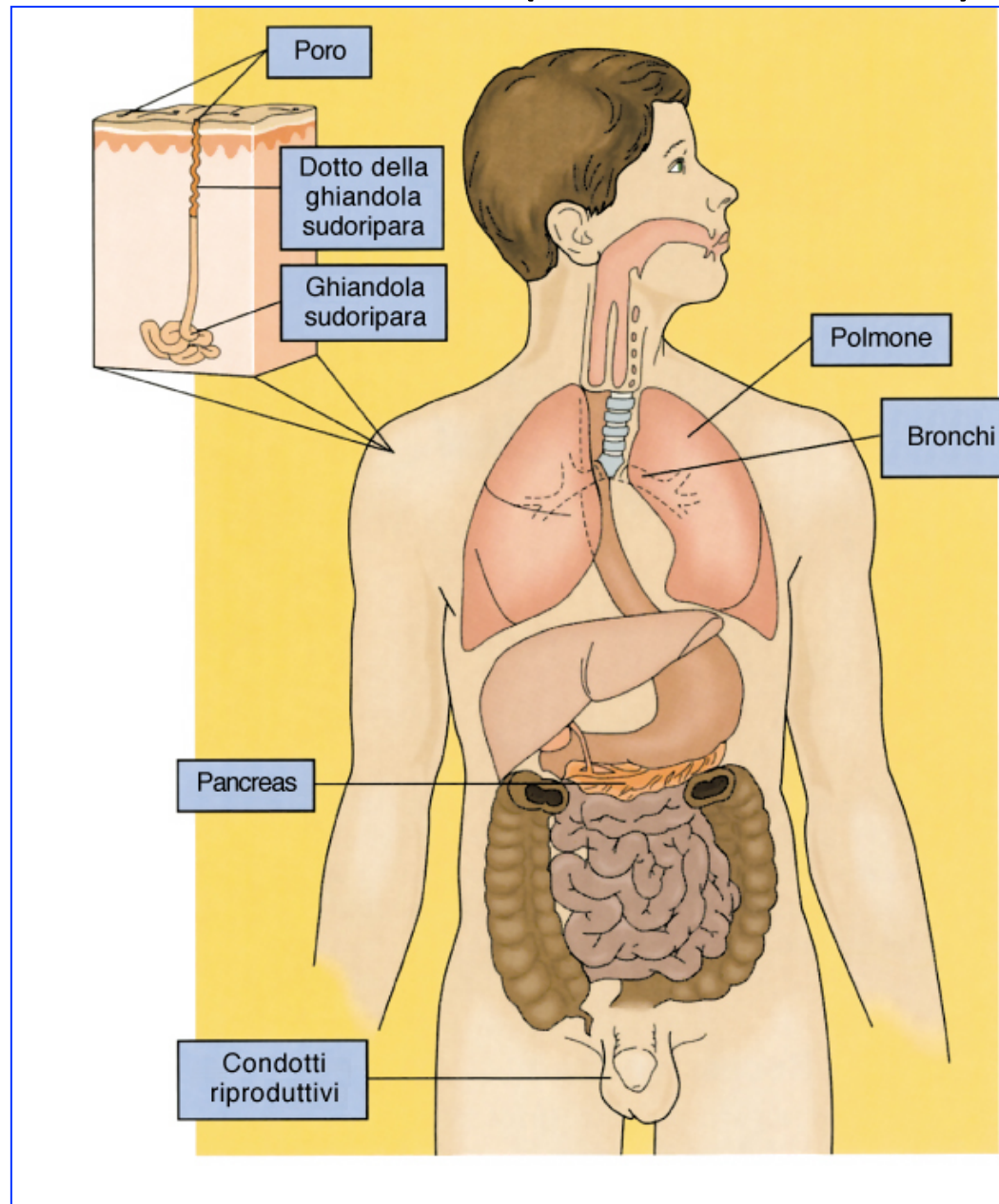
Dot Blot

	CF Probe	Control Probe
Father's DNA	●	●
Mother's DNA	●	●
Mary's DNA	●	○
Fetal DNA	●	●

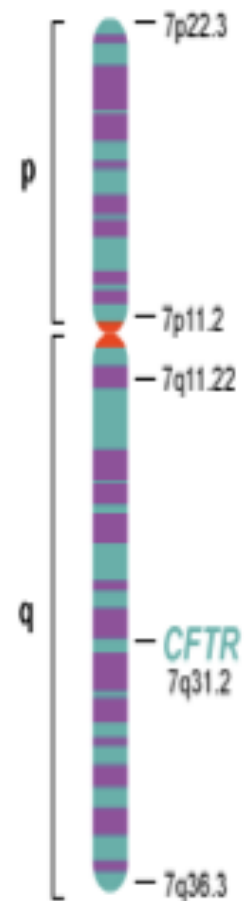
# La proteina Cystic Fibrosis Transmembrane Regulator (CFTR)



# La fibrosi cistica (OMIM 602421)



## Chromosome 7



### In Normal CFTR:

Nucleotide	AAT	ATC	ATC	TTT	GGT	GTT	TCC
Amino Acid	Asn	Ile	Ile	Phe	Gly	Val	Ser
	505			508			511

### In $\Delta$ F508 CFTR:

Nucleotide	AAT	ATC	ATC	GGT	GTT	TCC
Amino Acid	Asn	Ile	Ile	Gly	Val	Ser
	505					

## Alcune mutazioni e i loro effetti sulla funzionalità della proteina CFTR

