



# Bioinformatics 1

## Principles of heredity Mutations, substitutions and polymorphisms

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office hours - see the web site

<http://www.bioinformatics.uni-muenster.de/teaching/courses-2011/bioinf1/index.hbi>

# Computer Lab B, Schlossplatz 2b

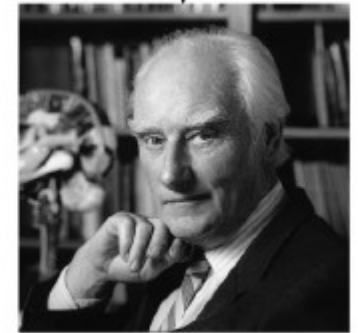
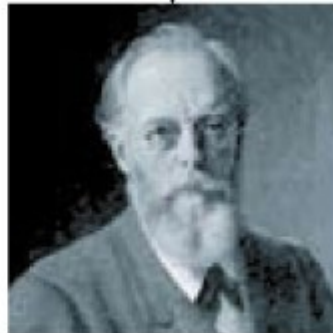
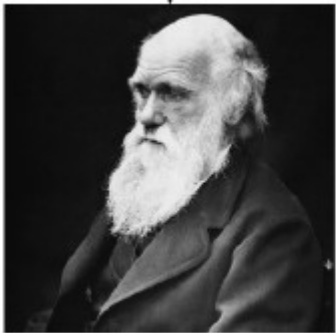
- Alignment and BLAST [November 14]
- Gene prediction [November 21]
- Phylogenetic inference [November 28]



Registration <http://www.bioinformatics.unimuenster.de/cgi-bin/teaching/coursereg.cgi>



# Understanding the principle of heredity following his historical development: from Darwin to Crick

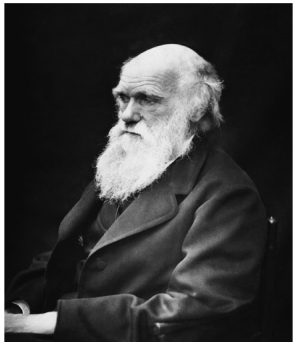


1859

1958



# All life on Earth as a common ancestor



Charles Darwin

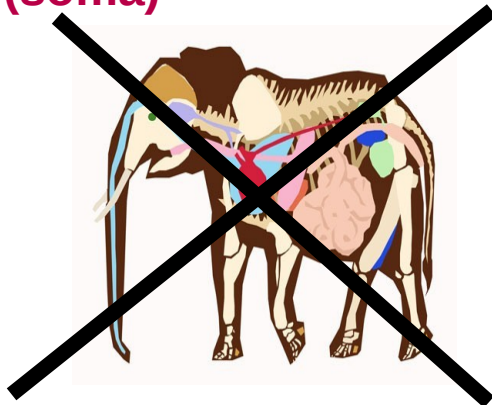
## Evolution by descent with modification



# How does heredity work?

What is the material basis of genetic continuity?

**Pangeneses**  
(soma)



vs.

**Hard-inheritance**  
(germline)



August Weismann

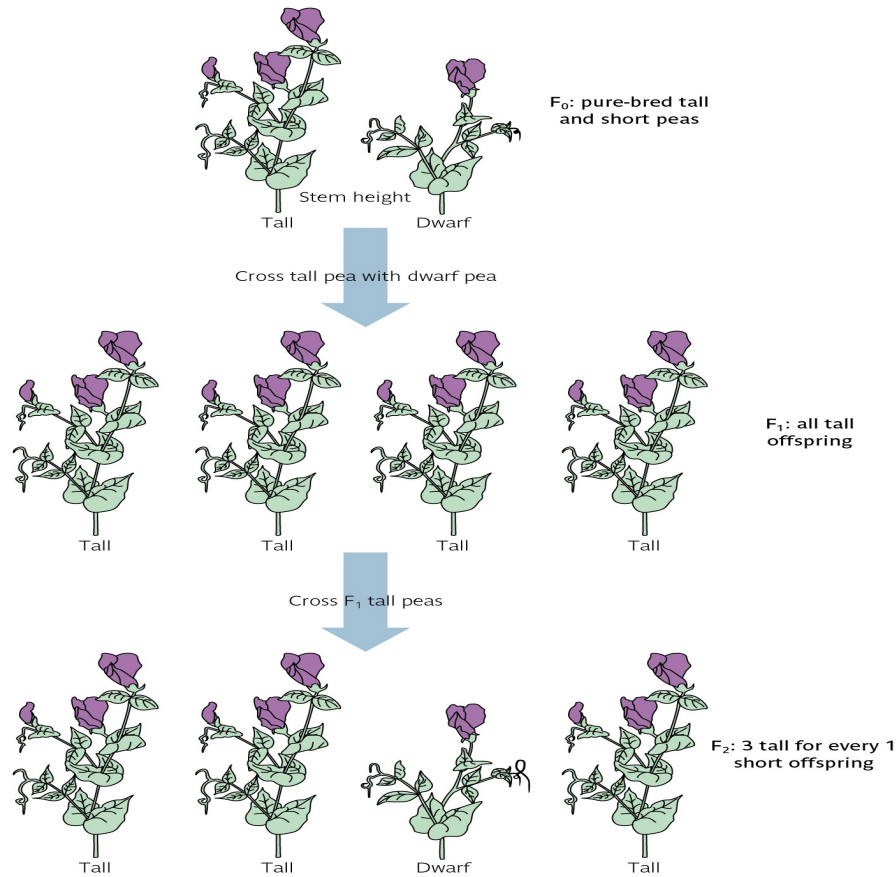
- **Germline separated from soma**
- **Immortal germline passes genetic information from one generation to the next**

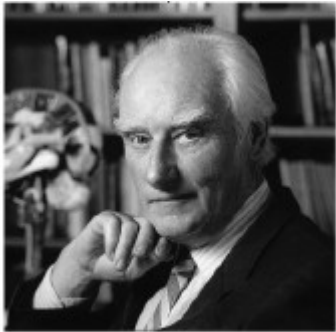


# Discrete and discontinuous factors govern heritability: Genes



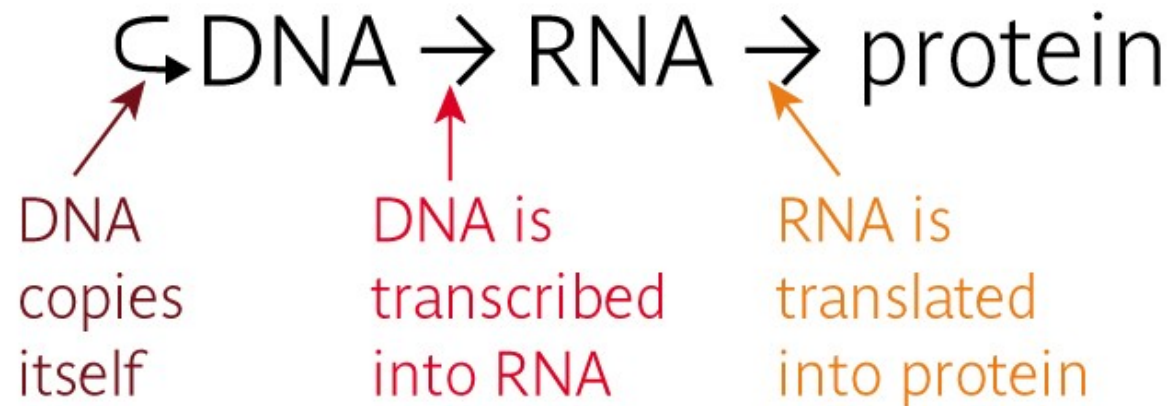
Gregor Mendel



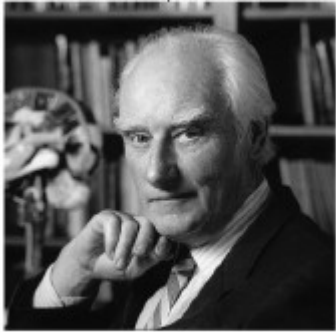


Francis Crick

# Central Dogma of Molecular Biology

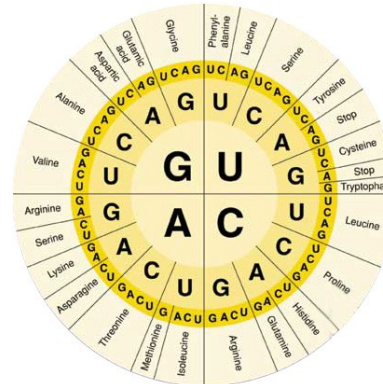
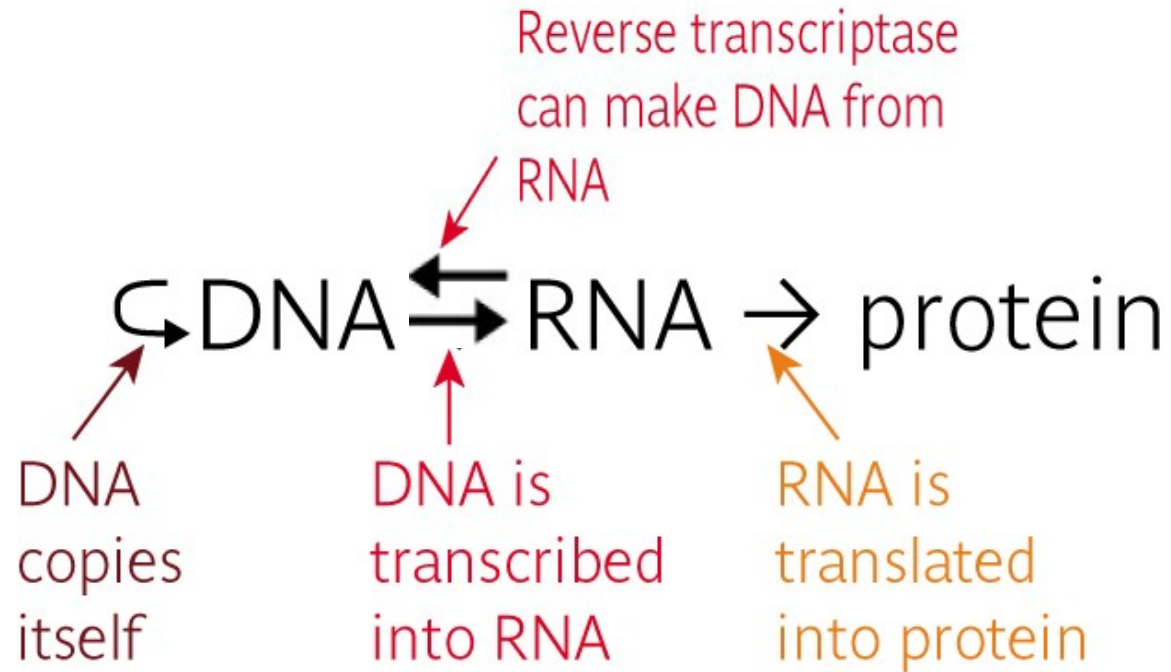


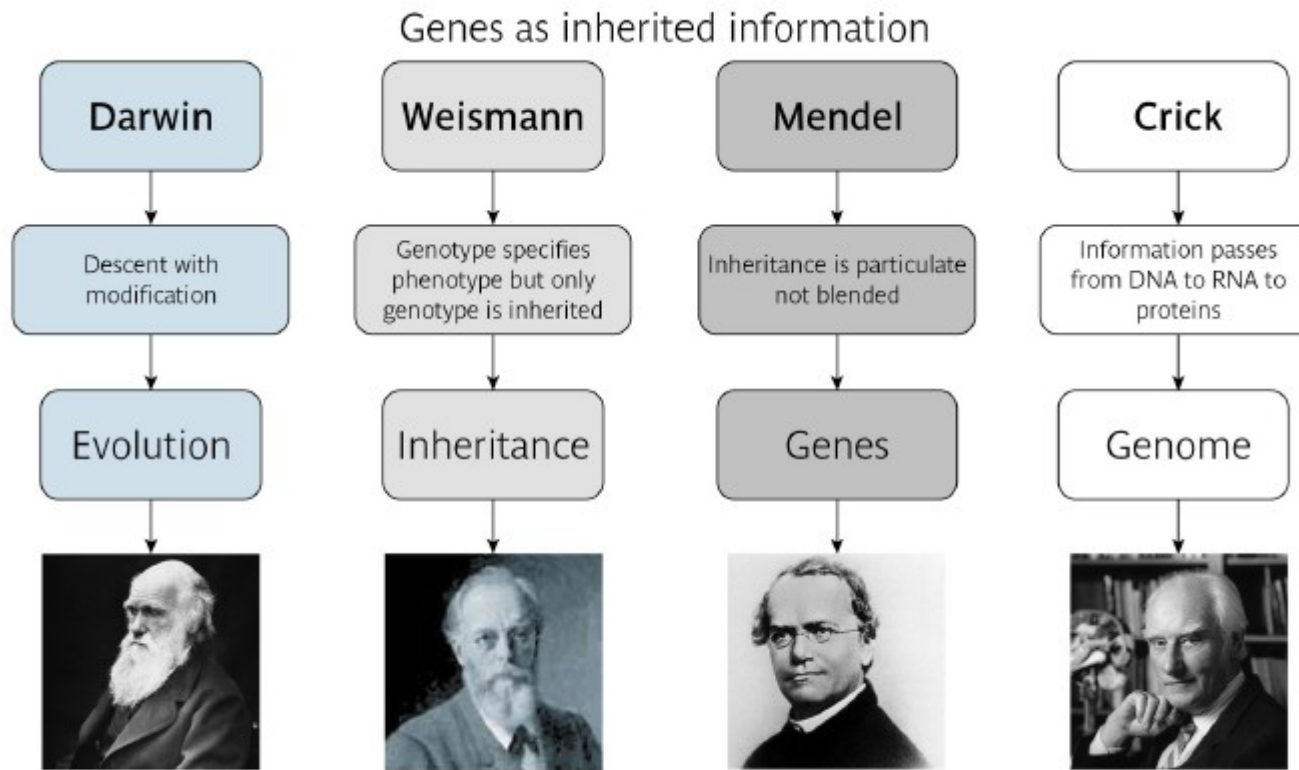




Francis Crick

# Central Dogma of Molecular Biology





DNA → RNA → protein

**Continuity of the germline:**  
hereditary information passed intact from parent to offspring

**Isolation of the germline:**  
changes to body cannot be coded into hereditary information



DNA damage

Copying errors



**MUTATIONS: heritable changes to the genome,  
essential for evolution.**

**LARGE SCALE** Chromosomal rearrangements  
Transposable elements



**SMALL SCALE** Gene duplications  
Single nucleotide changes (point mutations)

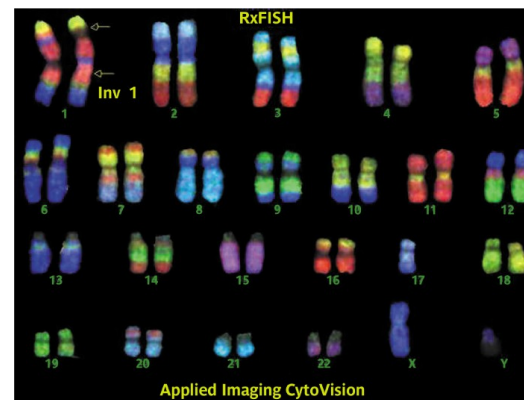




# Chromosomal rearrangements

Loss or duplication of full chromosomes  
(e.g., Trisomy 21)

Inversions

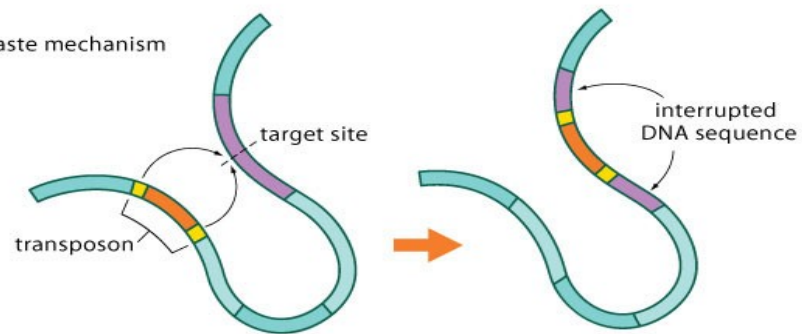


# Transposable elements

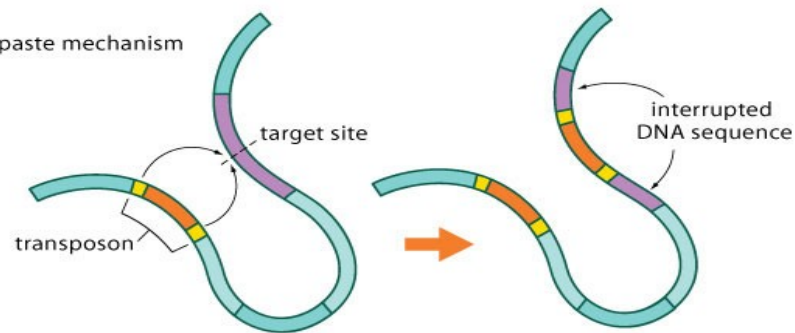
## Jumping genes

Two methods of transposition:

1. Cut-and-paste mechanism

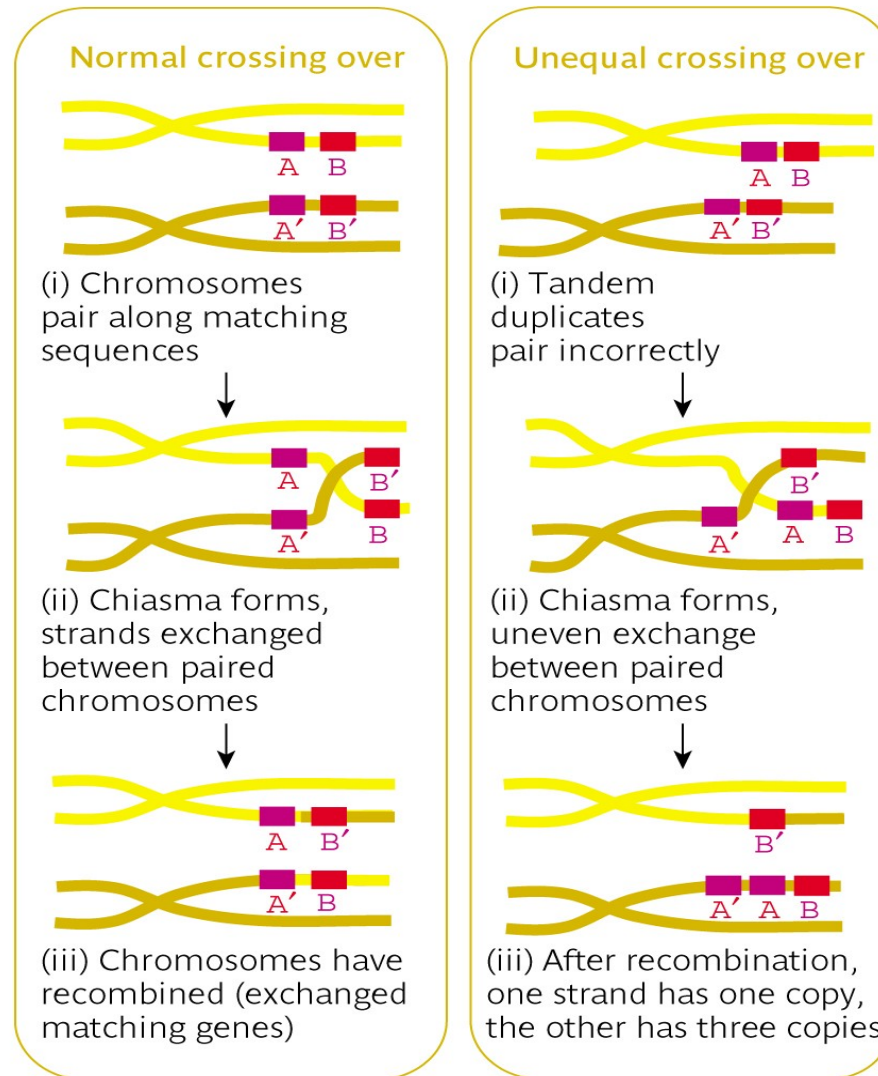


2. Copy-and-paste mechanism





# Gene duplications





# Point mutations

**Substitutions**

**TAC TGG**

**AAC TGG**

**Deletions**

**TAC TGG**

**ACT GG**

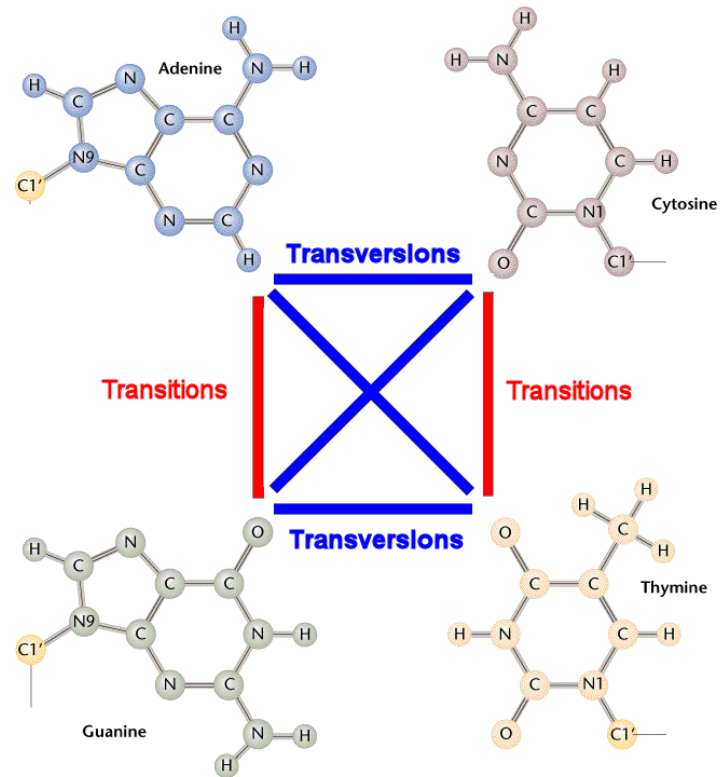
**Insertions**

**TAC TGG**

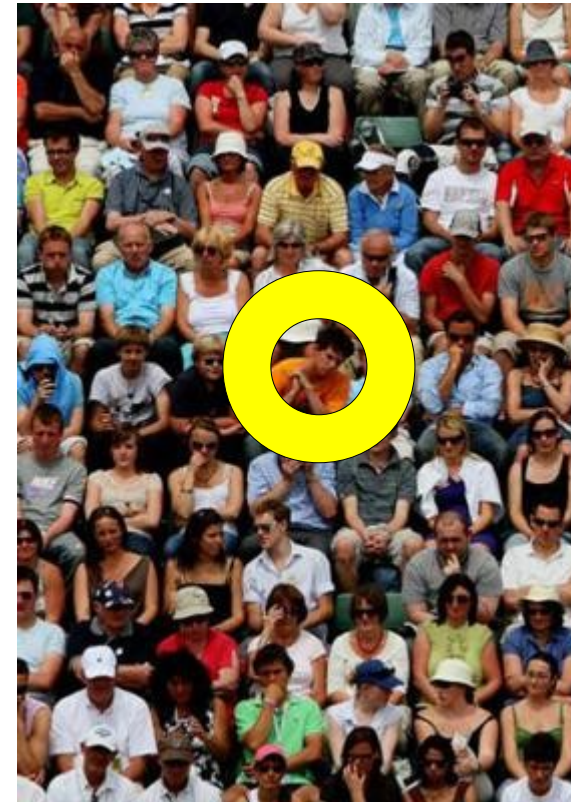
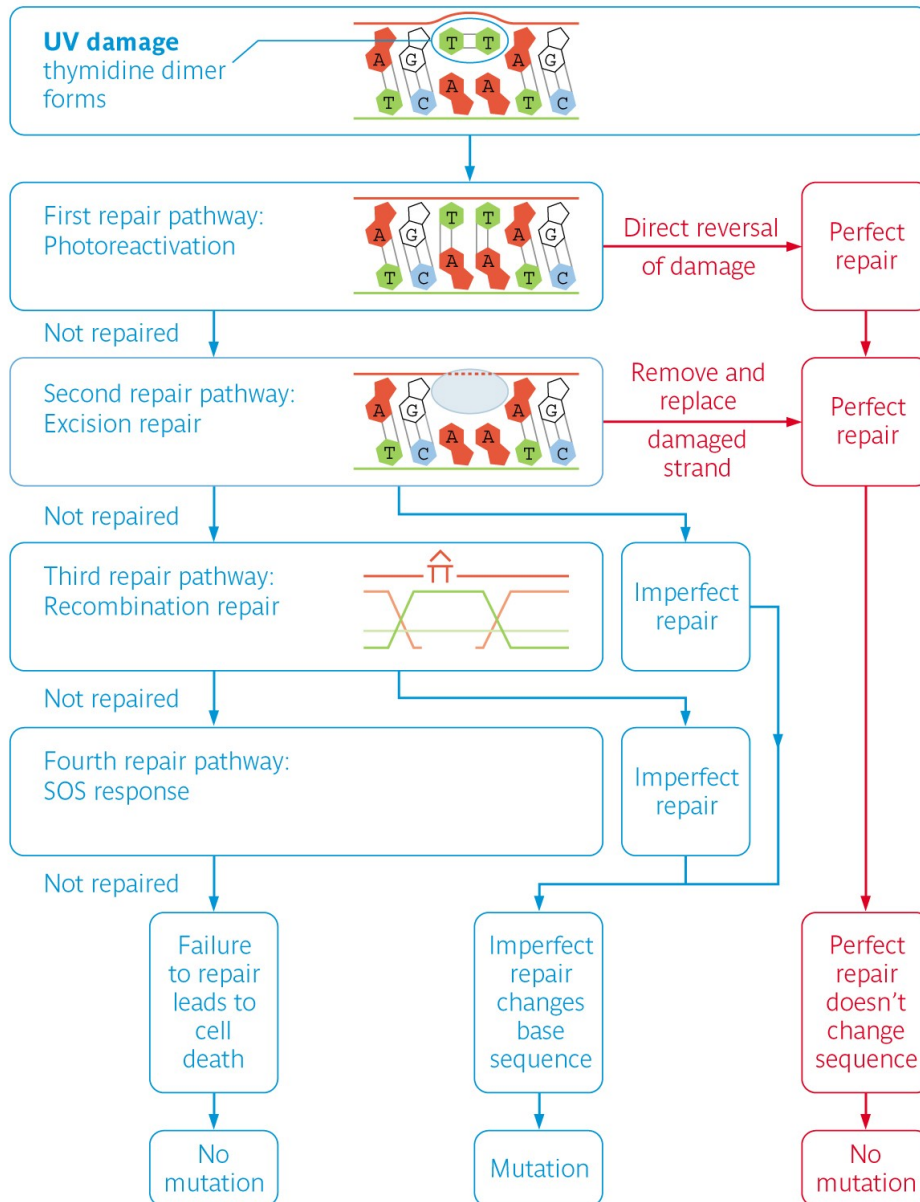
**TAA CTG G**



# Substitutions







# Single Nucleotide Polymorphism (SNPs)

# Genetic diversity: detecting and analyzing SNPs

**Biobanking: Collection of biological samples linked to informations about individuals (Etical issues!)**

**DeCODE: shedding light on schizophrenia**



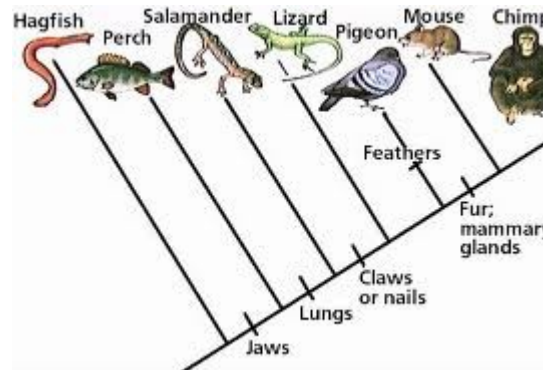
Genome-wide scan of 400 patients plus unaffected family members allowed to identify candidate genes associated with schizophrenia on chromosome 8



# Mutation creates differences among individuals in a population POLYMORPHISMS

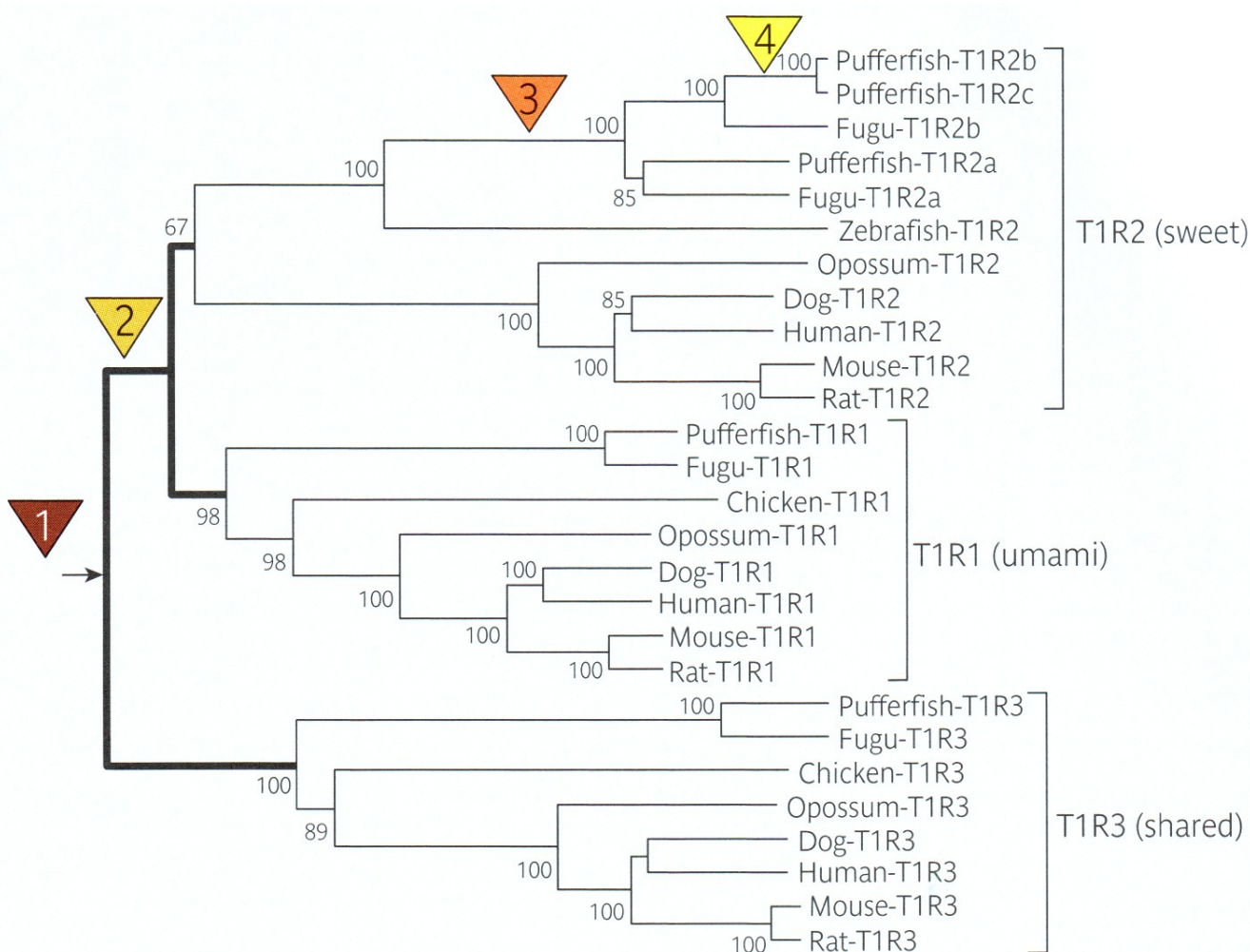


# What has this to do with differences between different species? SUBSTITUTIONS





# Evolution of taste receptors in vertebrates





# Evolution of taste receptors in vertebrates

