

Bioinformatics 1

Principles of heredity Mutations, substitutions and polymorphisms

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<http://www.bioinformatics.uni-muenster.de/teaching/courses-2012/bioinf1/index.hbi>

Computer Lab B, Schlossplatz 2b

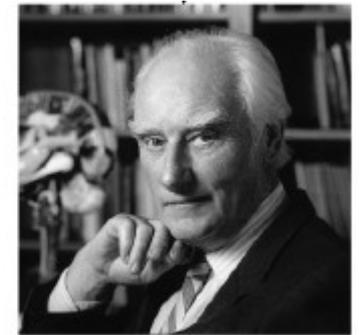
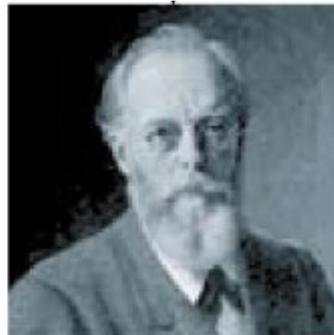
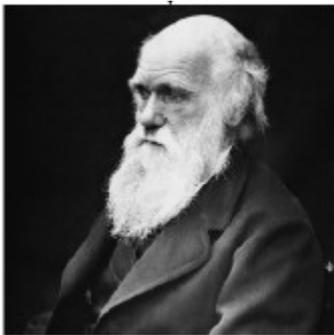
- Alignment and BLAST [November 12]
- Gene prediction [November 19]
- Phylogenetic inference [November 26]



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



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

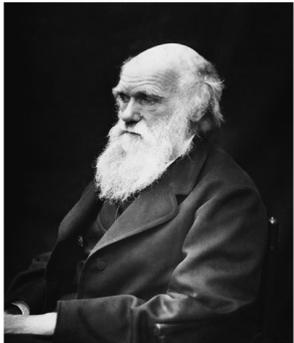


1859

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

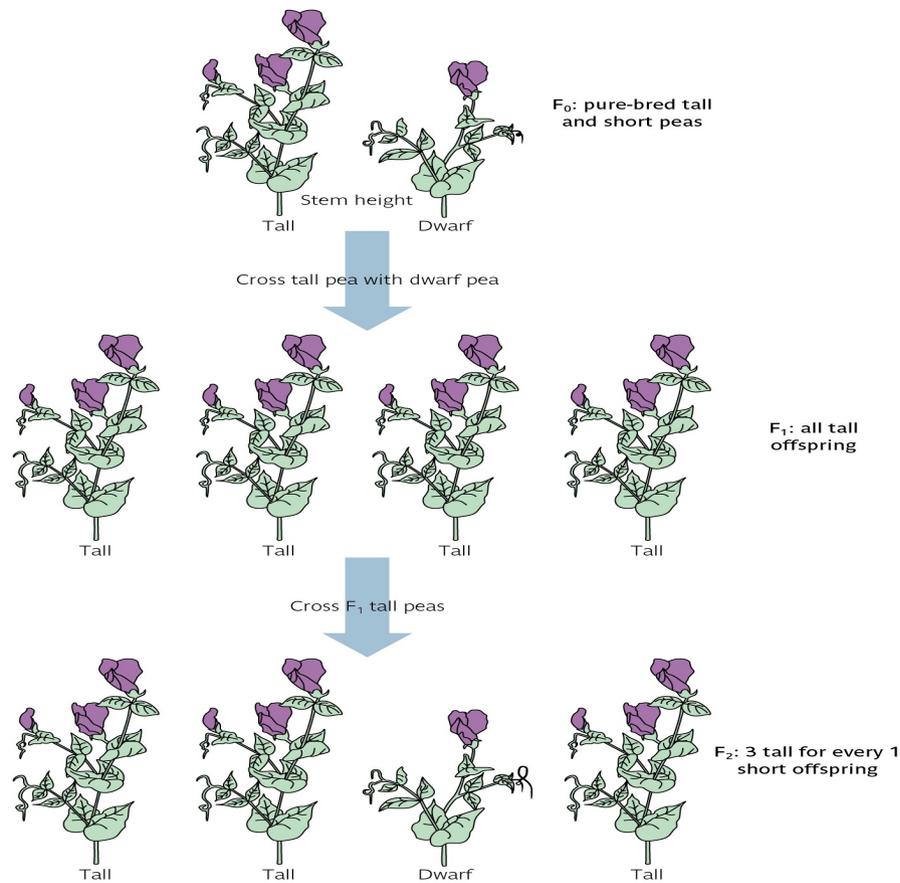
- **Germline separated from soma**
- **Immortal germline passes genetic information from one generation to the next**

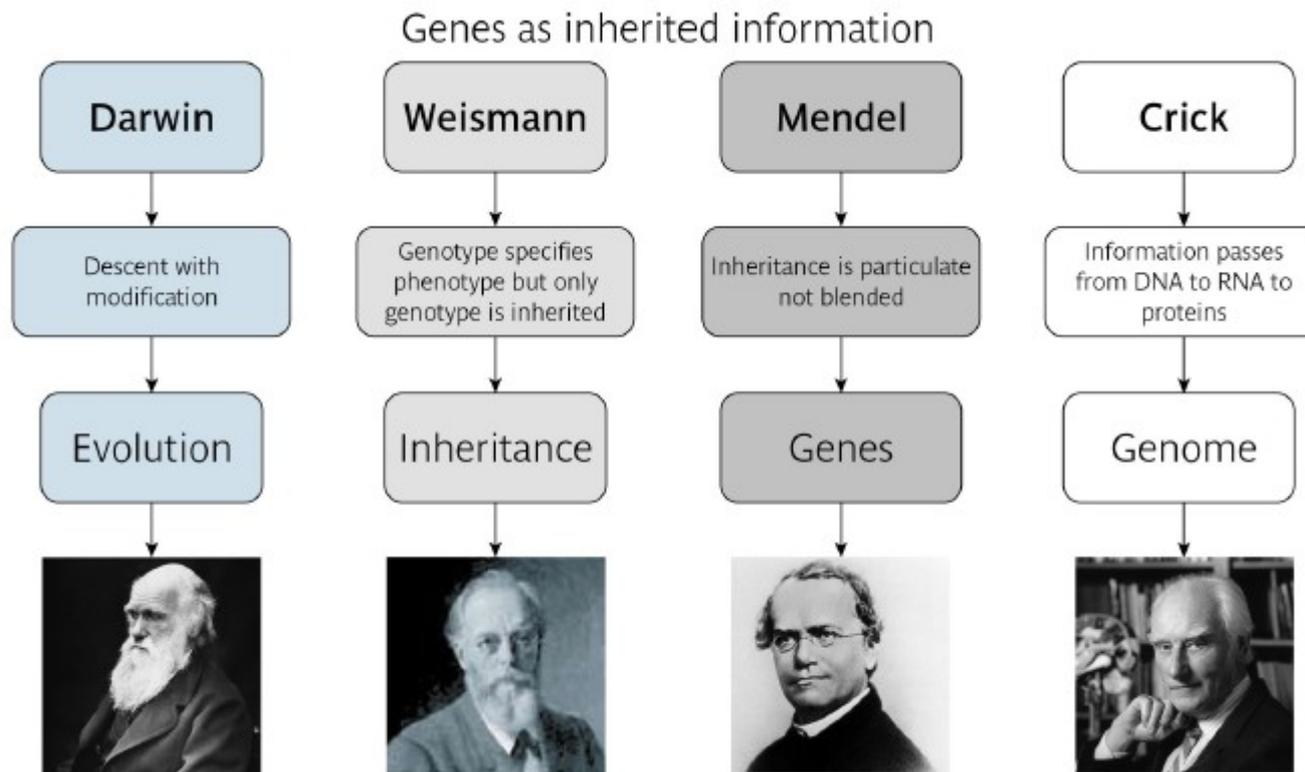


Discrete and discontinuous factors govern heritability: Genes



Gregor Mendel

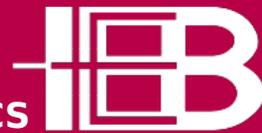




DNA → RNA → protein

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)

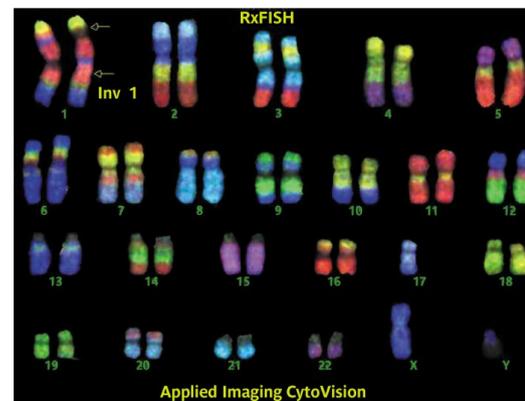




Chromosomal rearrangements

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

Inversions

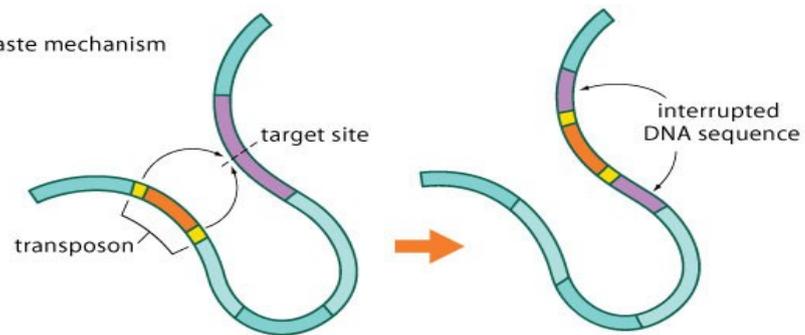


Transposable elements

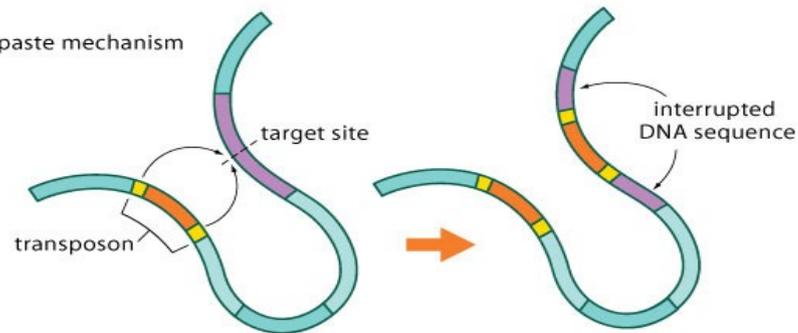
Jumping genes

Two methods of transposition:

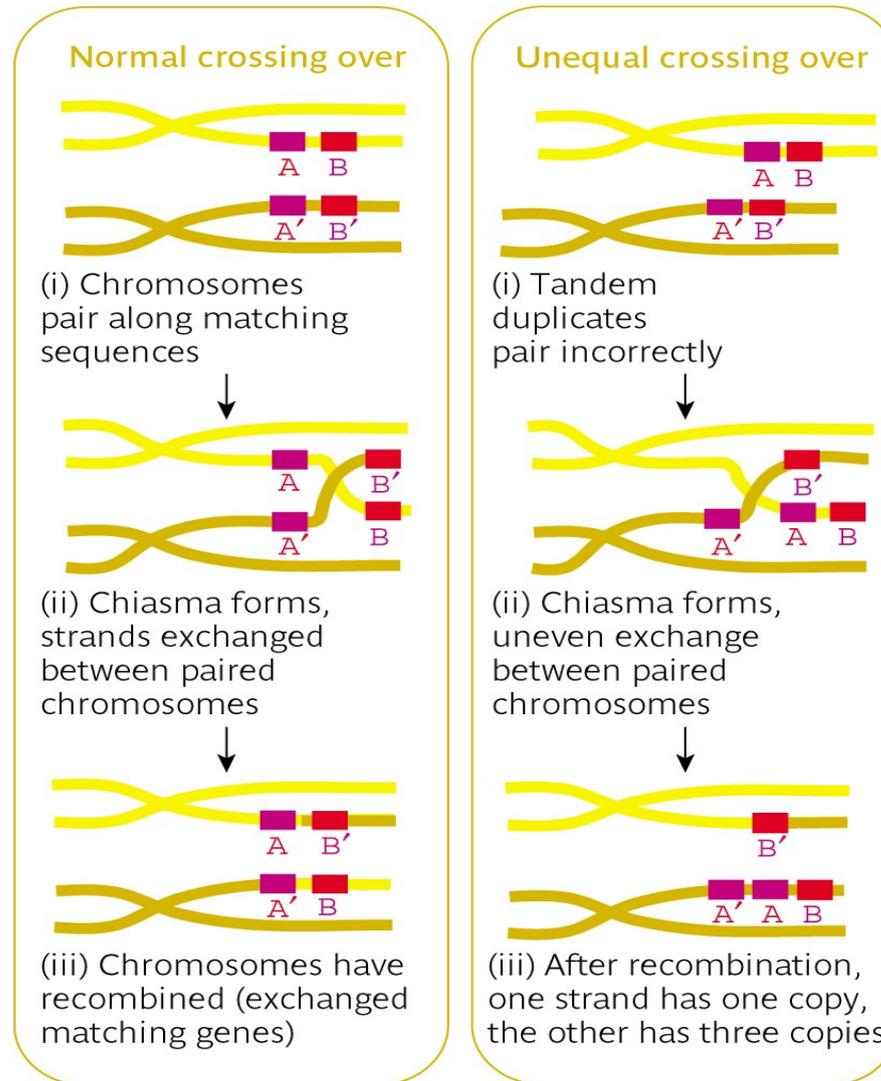
1. Cut-and-paste mechanism

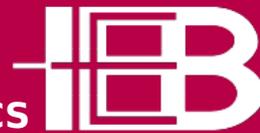


2. Copy-and-paste mechanism



Gene duplications





Point mutations

Substitutions

TAC TGG

AAC TGG

Deletions

TAC TGG

-AC TGG

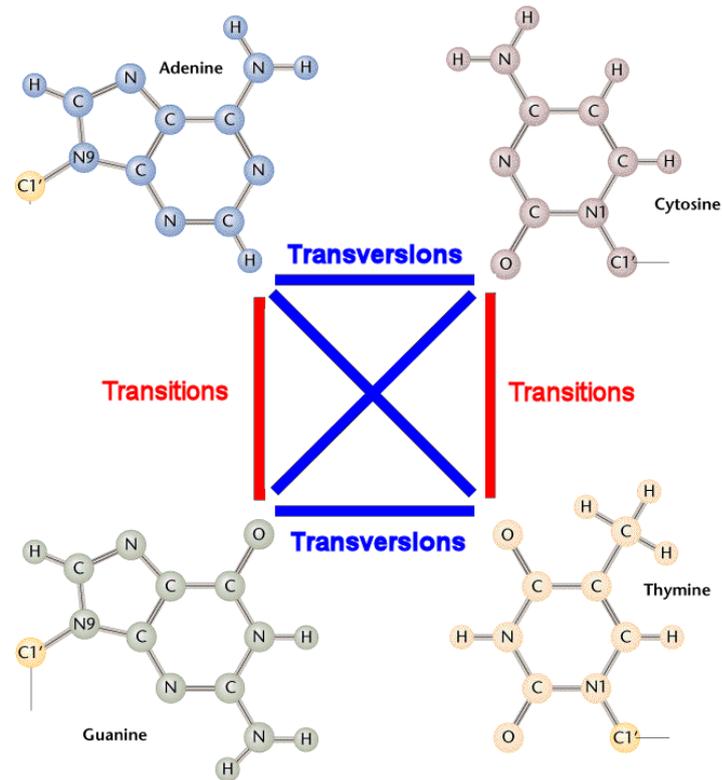
Insertions

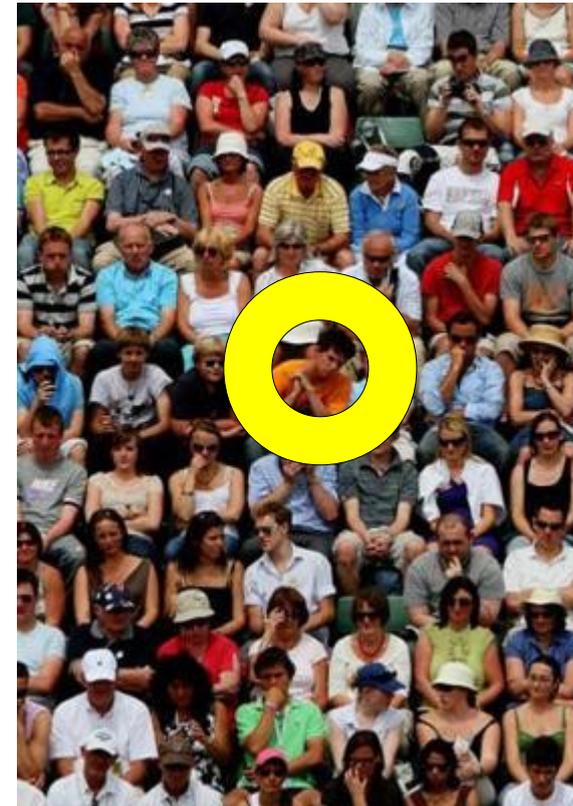
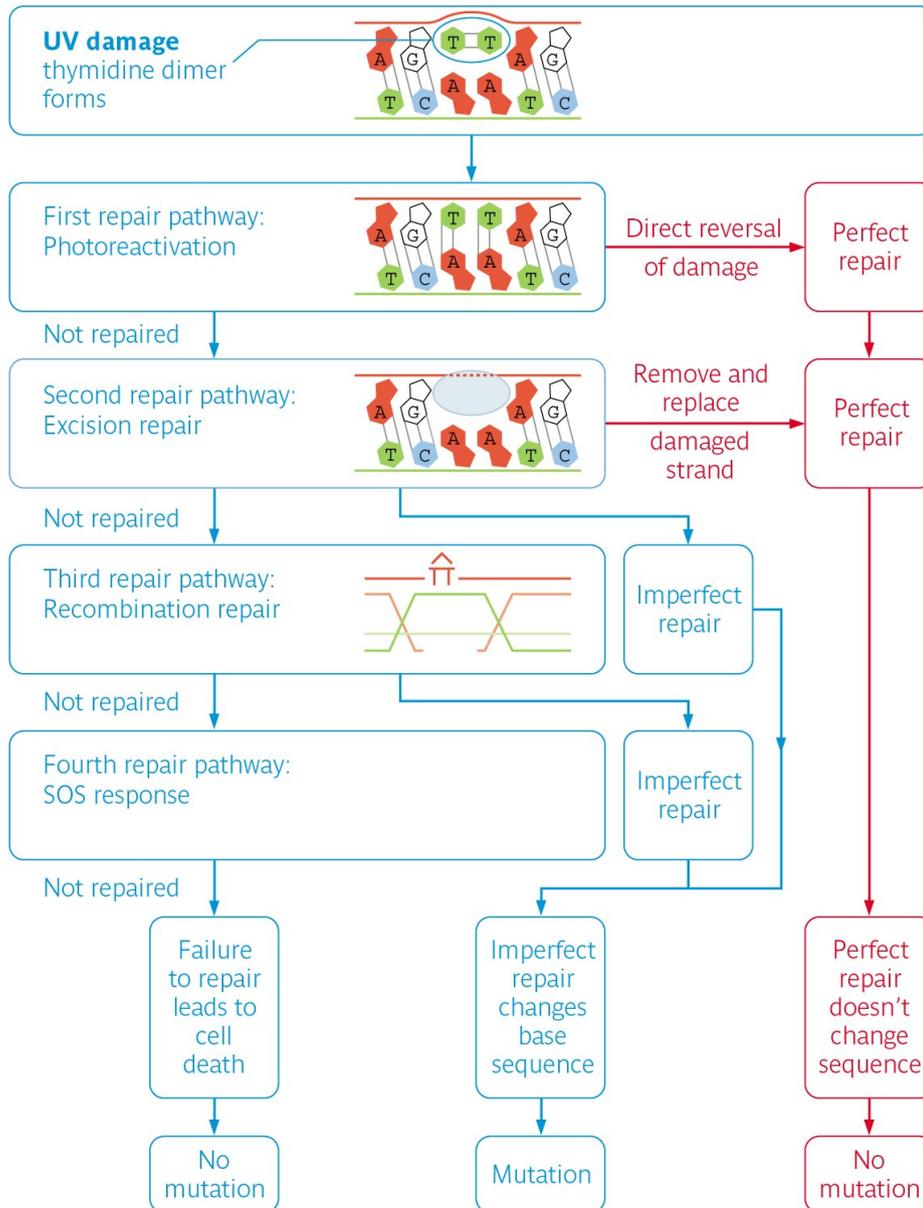
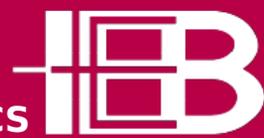
TAC TGG

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

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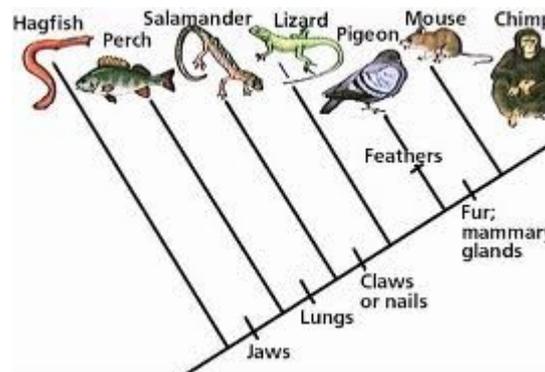




Mutation creates differences among individuals in a population POLYMORPHISMS



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



Evolution of taste receptors in vertebrates

