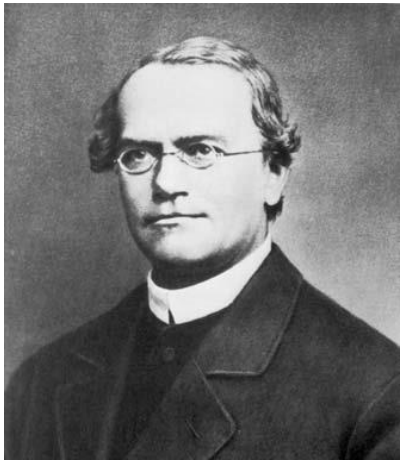
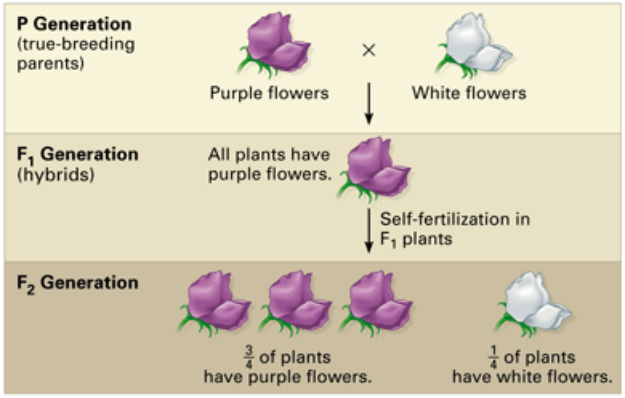
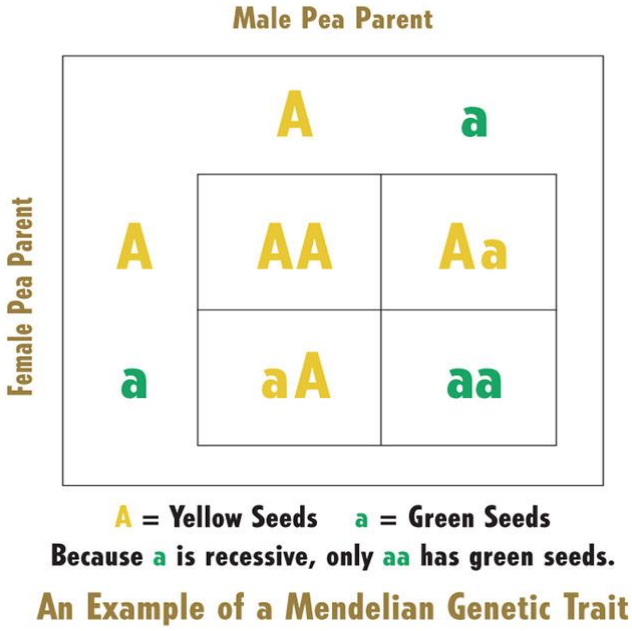
Laws of Mendelian Inheritance

Gregor Mendel, 1856-1863: pea plant experiments







- **Cultivated and tested some *29,000 pea plants* in the monastery's 2 hectares (4.9 acres) experimental garden.**
- **Worked with seven characteristics: plant height, pod shape and color, seed shape and color, and flower position and color.**
- **Law of Segregation: one random *allele* (gene variation) from each parent.**
- **Law of Independent Assortment: alleles for different traits are independent.**
- **Law of Dominance: some alleles are dominant while others are recessive; an organism with at least one dominant allele will display the effect of the dominant allele.**
- **"Father of modern genetics"**

DNA mutation: a change in sequence

THE CAT ATE THE RAT
THE KAT ATE THE RAT

THE CAT ATE THE RAT
THE HAT ATE THE RAT

THE CAT ATE THE RAT
THE CAA TET HER AT

THE CAT ATE THE RAT
THE ECA TAT ETH ERA T

- **Substitution:**
  - Silent – the same meaning.
  - Missense – the meaning is changed.
- **Deletion** – nonsense (the sentence makes no sense)
- **Insertion** – nonsense (the sentence makes no sense)

Substitutions are ***point*** **mutations, while insertions and deletions are *frameshift* mutations.**

DNA Mutations

A mutation is a permanent change in the DNA sequence.

- **Mutations can be:**
  - ***spontaneous*** **(by chance)**
  - ***induced*** **by mutagens (physical, chemical or biological agents)**

- **Factors that cause mutations:**
  - **external - environmental factors such as sunlight, radiation, and smoking**
  - **native - errors during DNA replication**

- **Mutations can lead to:**
  - **an *evolutionary advantage* of a certain genotype**
  - **disease, developmental delays, structural abnormalities, or other effects.**

Example: Sickle cell anemia is a disorder in which the body makes sickle-shaped red blood cells as a result of DNA mutation.

Human DNA

- The Human Genome Project (1990-2003) produced the first complete sequences of individual human genomes.
- Human genome contains ~3 billion bases and ~20,500 genes.
- Over 98% of the human DNA comprises *non-coding repetitive sequences* (the role, functions and descriptions of these sequences are currently being investigated by scientists).

- By 2020, tens of thousands of human genomes have been completely sequenced.
- All humans have the DNA that is 99.9% similar, however the rest 0.01% is enough to identify different individual DNA sequences (*i.e. tell apart which DNA belongs to whom*).
- Primary (and now standard routine!) applications include paternity testing as well as DNA profiling in criminal investigations.