# Modern Synthesis I

BIOL 4415: Evolution
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In 1843, a young lad named Johann Mendel took his vows as an Augustinian monk in the Abbey of St. Thomas, in the city of Brno. As part of his vows, he took a new name, and became Brother Gregor. . .

Brother Gregor studied at the University of Vienna, and came back to Brno to teach in the monastery school. In his spare time, he was an amateur scientist, who did research in physics and meteorology, but is most famous for his attack on the problem of *heredity*.

Between 1856 and 1863, Mendel crossbred and studied over 29,000 pea plants in the monastery garden, working out the basic rules by which traits were inherited.

Top: Purple-flowered and white-flowered peas
Bottom: Inflated and constricted pea pods.

## Mendel reasoned thus:

- There are "elements" inside the pea cells that determine what traits the peas have.
  - We now call these *genes*.
- A gene may exist in several forms called *alleles*.
  - In Mendel's classic experiments, each gene has two alleles: purple / white flowers, green / yellow peas, etc.
  - A gene may (and often does) have only one allele—or three, four, or more alleles. One human gene is known with 59 alleles.
  - What counts as an "allele" depends on the method of analysis: two gene copies might have different DNA sequences but produce identical phenotypes.

## New Alleles?

- Now known to come from *mutations*—heritable changes in genes
- Looks like I need to finish this slide. . .

The Russian scientist Sergei Chetverikov (1880-1959), studying wild populations of fruit flies, documented that populations contained far more alleles than was apparent to the eye. These must arise from mutation.

From year to year, from generation to generation, more and more new mutations arise, either similar to the preceding ones, or completely new. They are constantly absorbed into the basic species, which continually preserves its external homogeneity. This heterozygosity saturates the species in all directions, recombining and spreading in accordance with the laws of chance . . . .

## Point Mutation

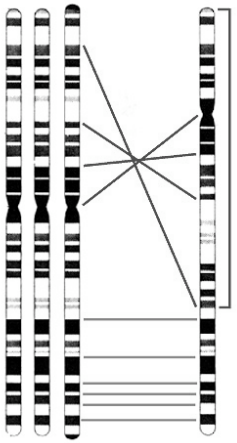
- Mis-sense mutations: one nucleotide replaced by another
  - Transition: replaces a purine with a purine (A to G or G to A) or a pyrimidine with a pyrimidine (T to C or C to T). Twice as frequent as a. . .
  - Transversion: mutation of a purine to pyrimidine (A or G to T or C, or vice versa)
- Frameshift mutations: addition or deletion of 1 or 2 nucleotides causes triplet codon reading frame to shift
- Nonsense or termination mutations: amino acid codon converts to stop codon, creating a shortened gene product

## Gene Effects

- Regulatory mutations: some genes "switch on" or "switch off" other genes—mutations in the control genes may have huge effects
- Mutations in genes for DNA repair enzymes can cause the overall mutation rate for the entire genome to increase or decrease

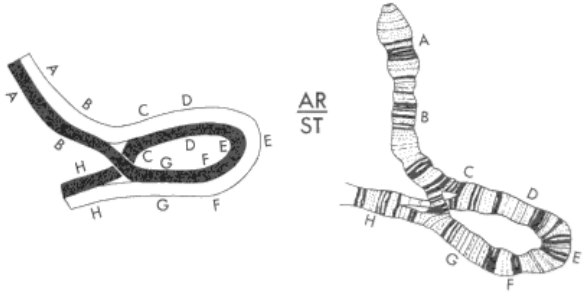
## Gene Effects

- Epistasis: one trait may be influenced by several genes, and a mutation in one might have unpredictable effects
- Pleiotropy: one gene might affect multiple traits, and again a mutation in one might have unpredictable effects

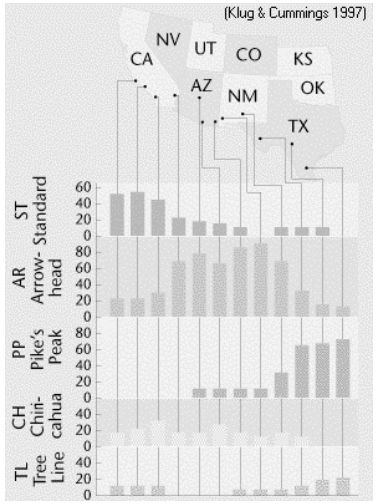


Inversion is another possible genetic change. The diagram shows chromosomes from four closely related animal species. The one on the right shows an inversion relative to the other three, as you can see if you try to match up the banding patterns.

**(Left to right: human, chimp, gorilla, and orangutan chromosome 3)**

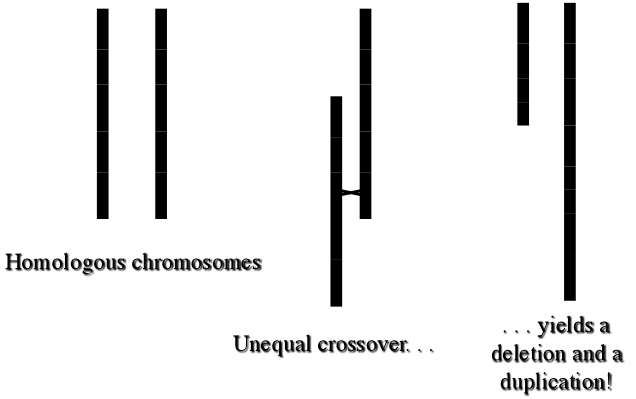



In the species of fruit fly *Drosophila pseudoobscura*, inversions are easily seen, because in an individual with both a "normal" and "inverted" chromosome, the chromosomes must form loops in order to pair up.

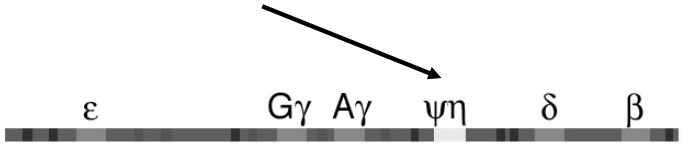



In *Drosophila pseudoobscura* there are several different inversions on chromosome 2 (which have various names, shown on the left). These have different frequencies in different parts of the fly's range.

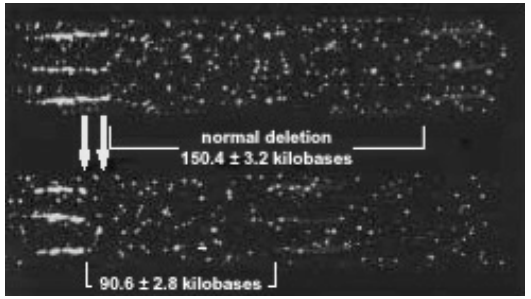
Gene duplication and deletion usually results from unequal crossover in meiosis.



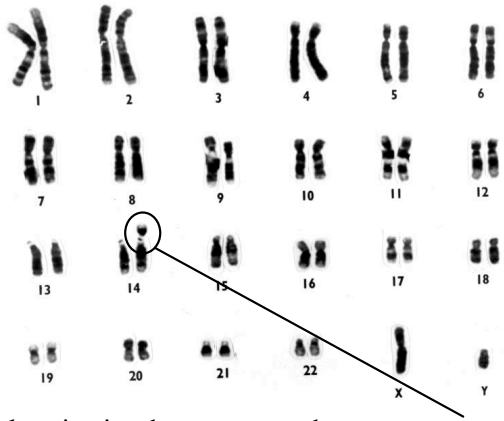

Duplication explains the existence of several closely related beta-globin genes (including a nonfunctional sequence, or *pseudogene*) on human chromosome 16



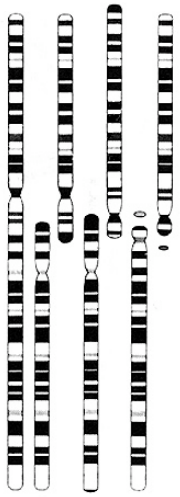

Deletion can be visualized clearly by the DNA combing technique.




Top: DNA strands from three normal people.
Bottom: DNA strands from three people with a 60,000-bp deletion.
(Image taken from Herrick and Bensimon 2001)



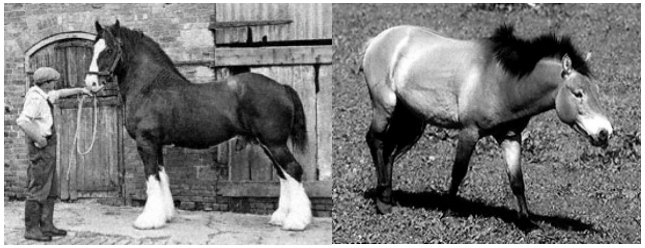

Translocation in a human—note that an extra copy of chromosome 21 has attached to chromosome 14.



Chromosome fusion or fission shown in four closely related animal species. The first one has a single chromosome that matches with the banding pattern of two separate chromosomes of each of the other three species. (In this case, fusion is the most likely reason why—we'll get to that later. . . . )

**(Left to right: human, chimp, gorilla, and orangutan chromosome 2 homologues)**

Chromosome fusion probably explains why domestic horses have 64 chromosomes. . .



. . . and the closest living wild species and probable ancestor of domestic horses, Przewalski's horse, has 66 chromosomes.

Duplication of a chromosome (polysomy) or a genome (polyploidy) can also occur as a result of a meiotic error.

Compare these two windflowers (genus *Anemone*) and their karyotypes—the one on the right is double the one on the left

Chromosome and karyotype changes can be associated with speciation, but not always. Spring beauty (*Claytonia virginica*) exists in over 50 karyotypes, ranging form 2n=12 to 2n>190. Different karyotypes coexist within the same population—and can apparently hybridize freely.

## So are mutations good or bad?

- We commonly think of mutations as a "bad" thing
  - Many human diseases are caused by mutant genes
- The majority of mutations are thought to be neutral, with no effect on phenotype
  - Japanese biologist Motoo Kimura developed this idea as the *neutral theory of molecular evolution*
  - Neutral mutations, however, create an unseen "reservoir" of genetic diversity within a population