Genome and Genetic Code

What is Genome?

- Genetic material of an organism, essentially the instructions on making proteins and RNAs.
- Inscribed in DNA: complete DNA sequence.
- Includes both the genes and the non-coding regions.

What is Genetic Code?

- The set of rules by which information encoded within DNA or RNA is translated into proteins.
- In general, the genetic code specifies 20 standard amino acids by means of triple nucleotide codons and is <u>basically the</u> same for all organisms on Earth.

What is Gene?

- The portion of the genome that codes for a <u>single</u> protein or an RNA.
- The molecular unit of heredity of a living organism.
- The size of a single gene may vary greatly, ranging from ~1,000 bases to ~1 million bases in humans.



Human DNA

- The Human Genome Project (1990-2003) produced the first complete sequences of individual human genomes.
- <u>Human genome</u> 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 2012, 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.

DNA Damage

DNA is damaged up to 1 million times per cell per day.



- The cells have an elaborate type-of-damage-specific system of DNA repair that is constantly active.
- A cell that has accumulated a large amount of DNA damage, or one that no longer effectively repairs damage incurred to its DNA, can enter one of three possible states:
 - 1. an <u>irreversible state of dormancy</u>, known as *senescence*
 - 2. <u>cell suicide</u> (apoptosis) or programmed cell death
 - 3. <u>unregulated cell division</u>, which can lead to cancer

DNA Mutations

A <u>mutation</u> 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:
 - <u>external</u> environmental factors such as sunlight, radiation, and smoking
 - <u>native</u> errors during DNA replication
- Mutations can lead to:
 - an evolutionary advantage of a certain genotype
 - <u>disease</u>, developmental delays, <u>structural abnormalities</u>, or other negative effects.



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

DNA Half-Life

In 2012, researchers have calculated that DNA from bones has a <u>521 year half-life</u>, which means that the oldest clone-able samples of DNA could be no more than 2 million years old.



This result rules out any possibility of ever replicating dinosaurs, as the youngest dinosaurs were around more than 65 million years ago...

Basic Cell Types

<u>All cells</u> consist of a cytoplasm enclosed within a membrane.



Cells are typically categorized by <u>how</u> <u>their genetic material</u> <u>is packaged</u>:



<u>Eukaryote</u> - the DNA is partitioned off in its own membrane-bound room called the nucleus.

<u>Prokaryote</u> - the DNA within a cell is not separated from the cytoplasm.



Eukaryotic cells have specialized interior compartments, called organelles ("little organs"), that have specific functions.

