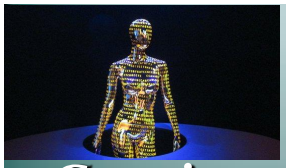
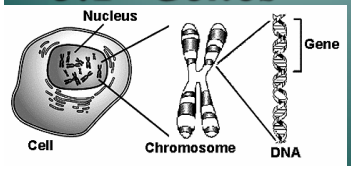
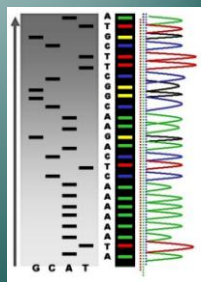


Genetics
3.1- Genes



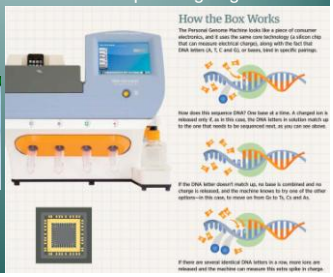


Essential idea:

- Every living organism inherits a blueprint for life from its parents.

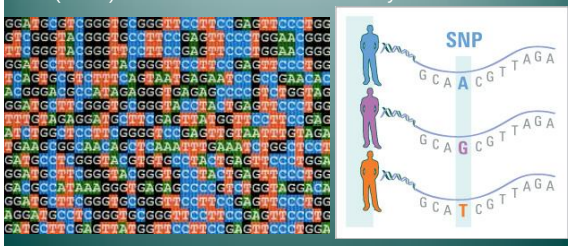
Nature of science:

- Developments in scientific research follow improvements in technology
  - Gene sequencers are used for the sequencing of genes. (1.8)



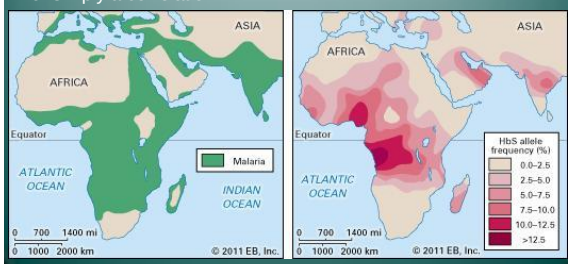
International-mindedness:

- Sequencing of the human genome shows that all humans share the vast majority of their base sequences but also that there are many single nucleotide polymorphisms (SNPs) that contribute to human diversity.



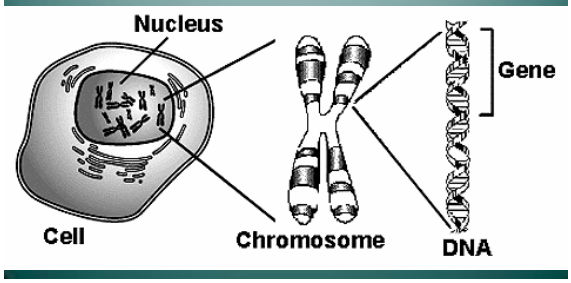
Theory of knowledge:

- There is a link between sickle cell anemia and prevalence of malaria.
- How can we know whether there is a causal link in such cases or simply a correlation?



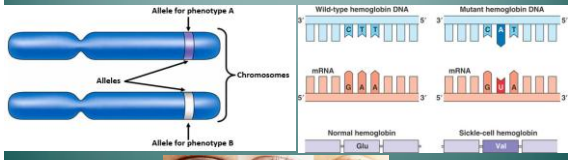
Genes

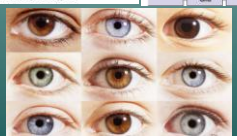
- A gene is a heritable factor that consists of a length of DNA and influences a specific characteristic.
- A gene occupies a specific position on a chromosome.



Alleles

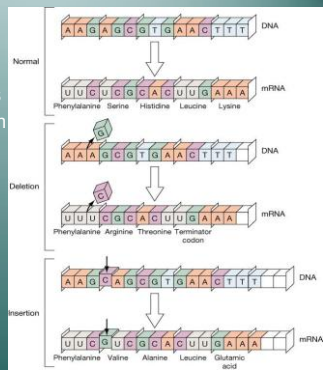
- The various specific forms of a gene are alleles.
- Alleles differ from each other by one or only a few bases.
- New alleles are formed by mutation.





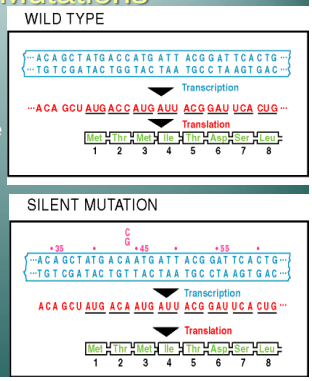
Gene Mutations

- Changes in DNA nucleotides
- Point Mutations
  - Frameshift Mutations
    - Base-pair insertion or deletion
    - The result is frequently a nonfunctional protein.



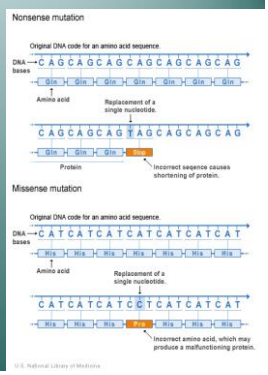
Gene Mutations

- **Base-pair substitutions**
  - **Silent mutations**
    - Changes to codons that have same effect
    - ACC to ACA both code threonine.



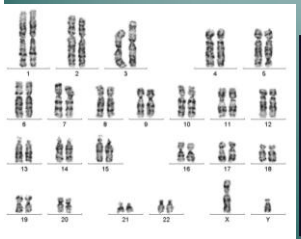
Gene Mutations

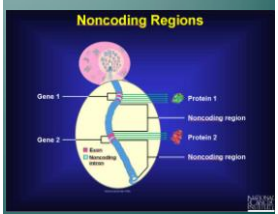
- Nonsense mutation (sickle cell)
  - A change that terminates the protein
  - CAG to TAG (a stop codon)
- Missense mutation (cystic fibrosis)
  - A change that changes the amino acid
  - CAT to CCT incorporates proline instead of histidine



Genomes

- The genome is the whole of the genetic information of an organism.
- It consists of DNA (or RNA in RNA viruses) and includes both the genes and the non-coding sequences





Human Genome Project

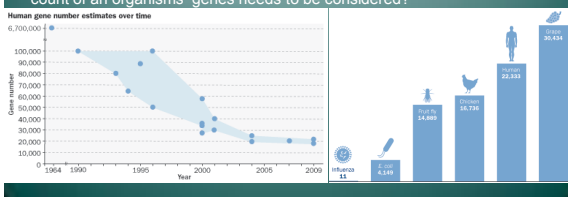
- The entire base sequence of human genes was sequenced in the Human Genome Project.
- The Human Genome Project (HGP) was an international 13-year effort, 1990 to 2003.
  http://www.genome.gov/10001772
- Primary goals were to discover the complete set of human genes and make them accessible for further biological study, and determine the complete sequence of DNA bases in the human genome.
- Chromosome Maps
  http://www.ncbi.nlm.nih.gov/genome/guide/human/

Comparing Gene Numbers

- Estimates of how many genes compose humans has declined (23000).
- The grapevine (30400) and water flea (31000) have a larger numbers of genes than humans.
- *E.coli* (4300) and chickens 16700) have less than humans.

End

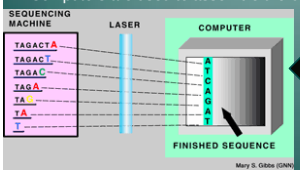
- Q - When analyzing an organisms' complexity, what other than the count of an organisms' genes needs to be considered?



Do Gene sequencing HW

Gene Sequencers

- The process of determining the precise order of nucleotides within a DNA molecule.
- PCR is used to prepare samples: the DNA needs to be copied to prepare a sufficiently large pure samples to sequence.
- Fluorescent labeling techniques enable all four nucleotides to be analyzed together.
- Lasers are used to fluoresce the dye markers.
- Digital camera technology reads the dye markers.
- Computers are used to assemble the base sequence.



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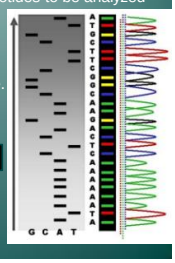
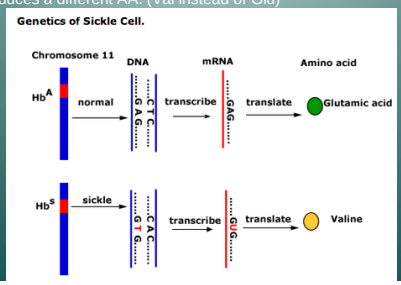


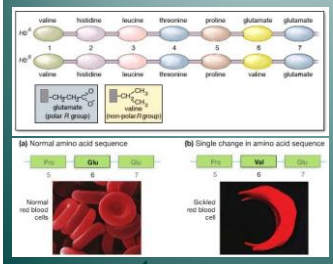
Sickle Cell Anemia (a SNP)

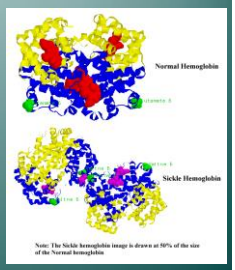
- Causes of sickle cell anemia
  - A base substitution mutation of DNA.
  - Changes the base sequence of mRNA transcribed from it.
  - Produces a different AA. (Val instead of Glu)



Sickle Cell Anemia (a SNP)

- Changes the 6th AA in the sequence of a polypeptide in hemoglobin.
- Different properties of functional group creates difference in folding.





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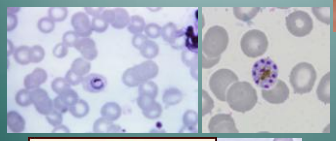
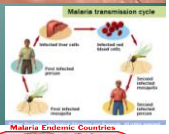
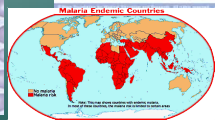
Malaria

- Caused by a *Plasmodium* protist.
- Carried by the female *Anopheles* mosquito
- Human is an intermediary in life cycle.







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