Biology and Society

Unit Four: Genetics and Medicine

Topic One: A Primer on Human Genetics

• The human genome is the biological instruction for how an individual is formed and how the cells in the body function.

• There are between 20,000 to 25,000 genes in the human genome.

- Except for identical twins, the gene structure is unique in each individual.
- Half the DNA in the nucleus of each cell comes from each parent.

• Genes direct the formation, or expression, of proteins that a cell uses to function, repair or defend itself, and to divide.

• Genes are contained in the chromosomes in the nucleus of each cell.

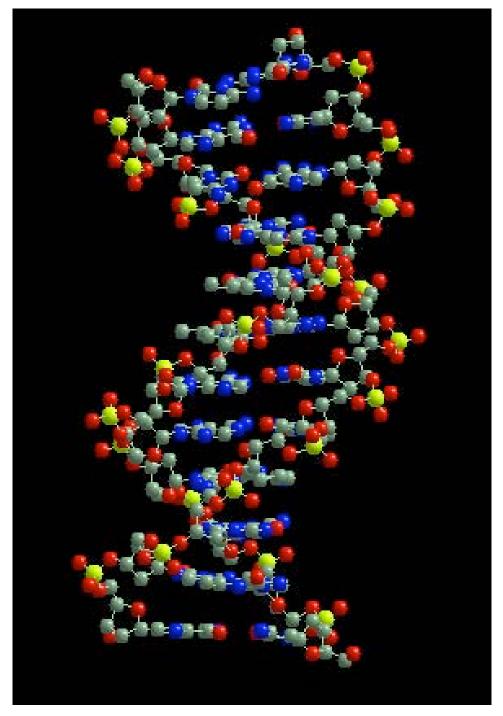
• There are 22 numbered chromosomes, plus two that determine gender, X and Y. A female has two X chromosomes, while a male has an X and a Y.

• A human normally has 23 pairs of chromosomes (46 total) in each cell.

• About 3 billion DNA subunits, called base pairs, make up the human genome.

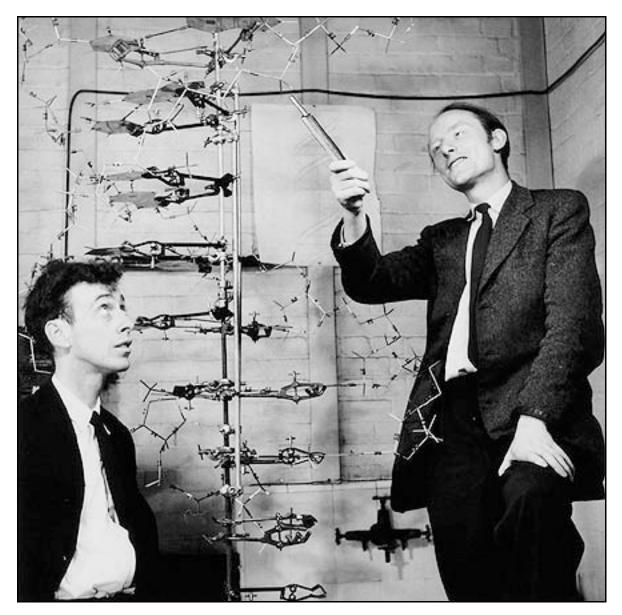
• Genes can have thousands of base pairs. The sequence and arrangement of these base pairs create a genetic code.

• Genes give coded instructions to the cell on how to assemble proteins. Making of a protein from this code is called "gene expression."

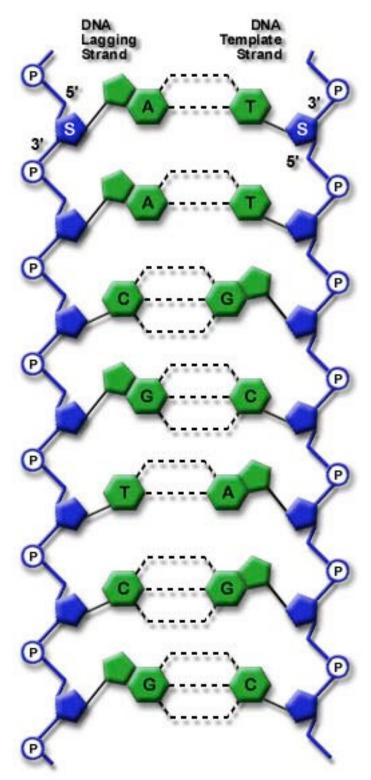


Nucleic Acids—DNA and RNA

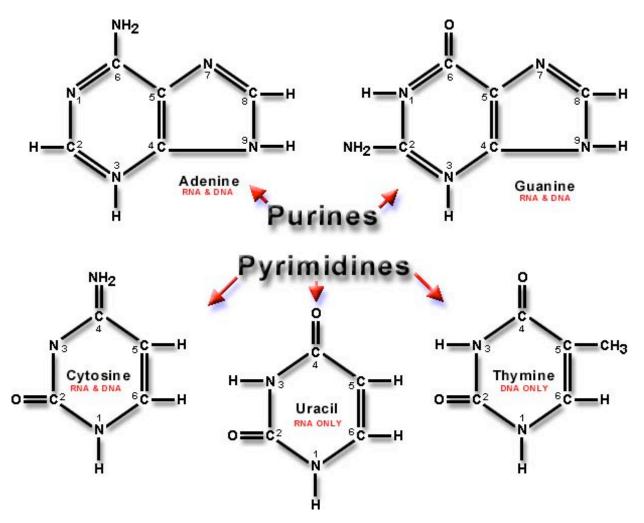
For more on DNA and RNA go to: <u>http://users.rcn.com/jkimball.ma.ultranet/BiologyPages/D/DoubleHelix.html</u>



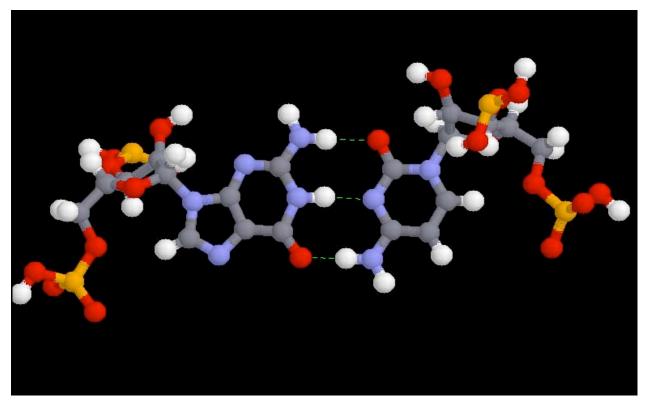
James Watson (left) and Francis Crick in 1953, were the first to discovered the structure of DNA .



The structure of DNA, blue is the sugar/phosphate backbone, the sides of the ladder, green are the nitrogen bases, the rungs of the ladder.



There are five different nitrogen bases that make DNA and RNA. Uracil is found only in RNA and Thymine only in DNA.

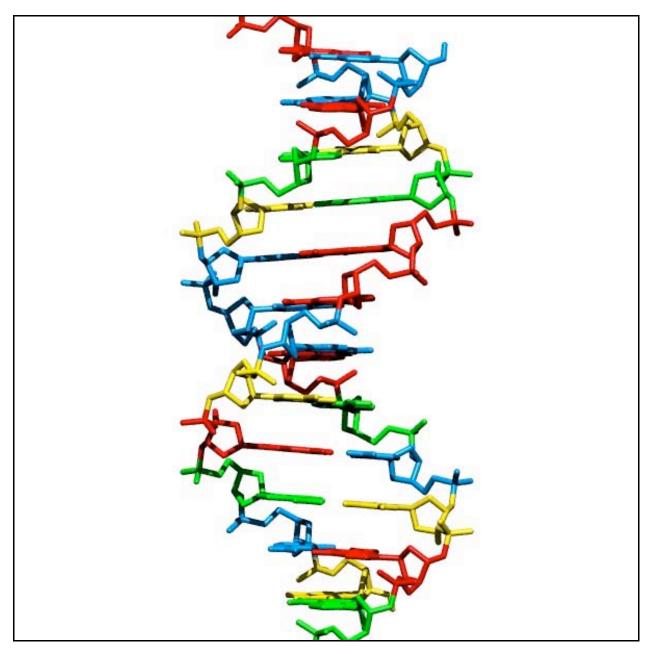


Structure of a single DNA base pair (Guanine to Cytosine)

• In DNA base pairs are composed of four different nitrogen bases, called nucleotides, that weakly bond in pairs to link the sides of the DNA double helix.

• The DNA nucleotides are adenine, thymine, cytosine and guanine. They are abbreviated A, T, C and G in the scientific description of the genome.

• The bases form specific nucleotide pairings, with "A" linking only with "T", and "C" only with "G".



The base pairs resemble rungs in a coiled ladder.

Nucleic Acids and Protein Synthesis

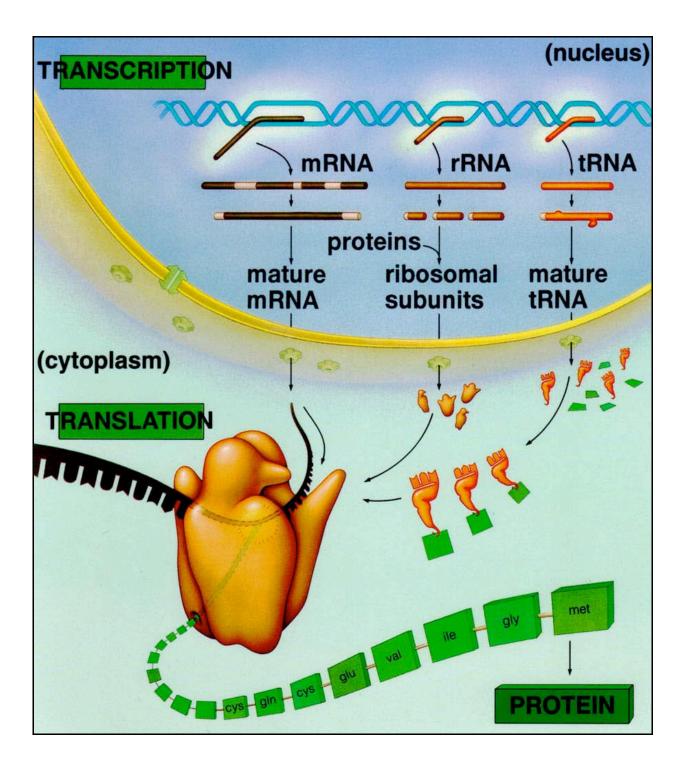
• Transcription

DNA (genes) -> to mRNA -> to ribosomes

• Translation

amino acids -> to tRNA -> to ribosomes -> makes proteins

• Genes code only for proteins and RNAs.



Genetics as Information – Terms and Definitions to Know

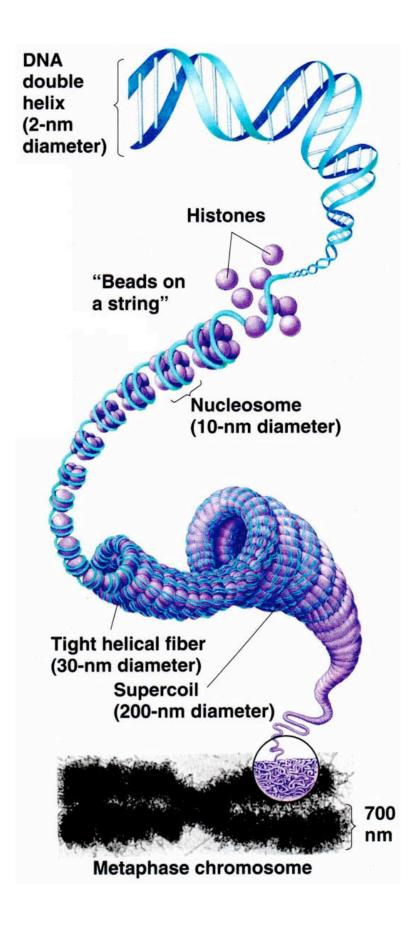
- genes \rightarrow genetic information
- genome \rightarrow genetic makeup of the individual
- gene pool → genetic makeup of a population
- biodiversity → genetic sum of all of life on earth
- genotype \rightarrow phenotype

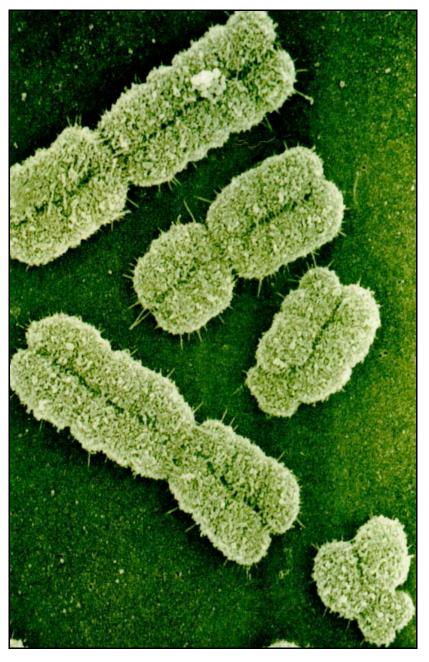
Genetics and Reproduction – Terms and Definitions to Know

• genetic mutation

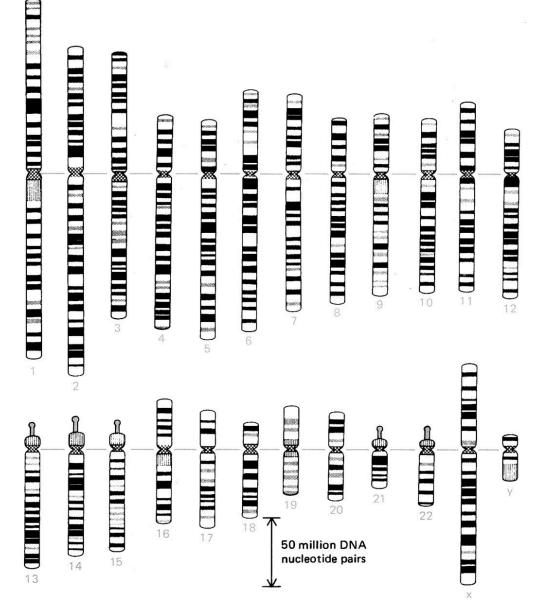
• allele

- variation in genes variation in alleles
 - dominant alleles recessive alleles
 - heterozygote
 homozygote
 - gene expression
- single gene traits polygenic traits pleiotropy
 - somatic cells germ cells
 - mitosis meiosis

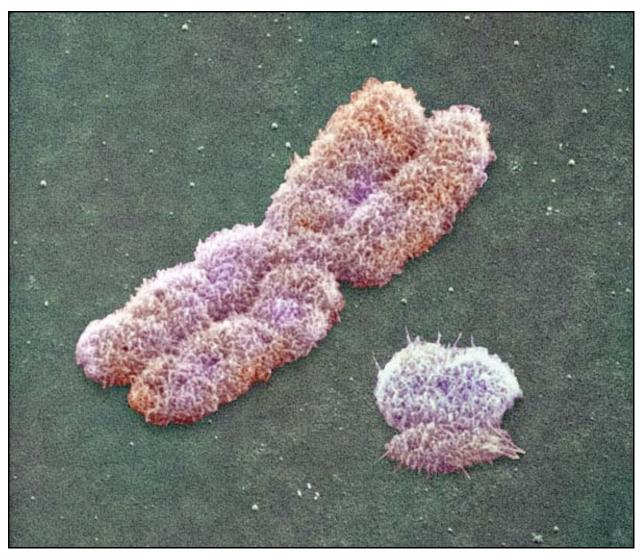




Electron Micrograph of Human Chromosomes



There are 24 different chromosomes in the human genome.



X (left) and Y Chromosomes Magnified 10,000 Times

Cell Division-Mitosis and Meiosis

For more on chromosomes and cell division go to: <u>http://www.biology.arizona.edu/cell_bio/cell_bio.html</u>

and

M&M.pdf

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