The Basics of Genetics

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RBC vs WBC

- The most common lab source for DNA is blood.

- It is spun down to collect WBC, which contain a nucleus.

- Except for mature red blood cells, all human cells contain a complete genome because they contain a nucleus.
Mouth DNA:

DNA can also be obtained from mouth samples: saliva (high DNA yield) or buccal swabs (low DNA yield).

DNA

Deoxyribo-(Dee-oxee-rye-boe)

nucleic (New-klee-ick)

Acid

Deoxyribonucleic Acid

Contains a coded message: sequence

The Human Cell: inside the Nucleus

http://www.ornl.gov/hgmis
Adenine (A) pairs with thymine (T), while cytosine (C) pairs with guanine (G).
Almost all cells contain a complete copy of the human genome in their nucleus.
Chromosomes

We inherit one entire set of chromosomes from each parent. Each set has 23 single chromosomes: 22 autosomes and an X or Y sex chromosome for a total of 24 different chromosomes. The chromosomes range from 50 million base pairs to 250 million base pairs.

24 different chromosomes

DNA the molecule of life

Trillions of cells
Each cell:
- 46 human chromosomes
- 2 meters of DNA
- Trillions DNA segments (the bases: A, T, C, G)
- Approximately 20,000 genes, each for proteins that perform most life functions.

http://www.ornl.gov/hgmis
Definition of a gene:

A gene is a segment of a DNA molecule (ranging from fewer than 1 thousand bases to several million), located in a particular position on a specific chromosome, whose base sequence contains the information necessary for protein synthesis.

Proteins:

Required for the structure, function, and regulation of the body’s cells, tissues, and organs. Each protein has unique functions. Examples are hormones, enzymes, and antibodies.

The work of genetic research:

The search for a typographical error in 2 different editions of a 23 volume encyclopedia (the "genome"):
Mother’s vs. father’s chromosomes (affected vs. unaffected)
Pick one volume (Chromosome)
Choose a chapter (identify region of the chromosome)
Choose a subheading (identify the gene)
Compare affected to unaffected: uncover single typographical error (mutation causing disease).
   e.g. AACGTAG - CCTCAG (unaffected)
   AACGTAG - CCTCAG (affected)

Entire Human Genome
3,000,000,000 base pairs
Average Chromosome
120,000,000 base pairs
Average Gene
2,000-200,000 base pairs
Mutation That Can Cause Disease
1 base pair