CHROMOSOMES and KARYOTYPES

Chromosomes of higher organisms (eukaryotes) contain DNA and protein.

One long DNA double helix is present per chromatid

DNA is highly organized in association with histones (protein)

- 5 histones are highly conserved from 1 species to another
- 4 histones (2 copies of each) make a ball
- 1 histone links adjacent balls together
- 146 base pairs of DNA wrap around each ball (2 loops/ball)
- each ball is a nucleosome

Nucleosomes are coiled into solenoids (6 nucleosomes/solenoid)

- solenoids also coil, forming a fiber
- · fibers coil to give a visible (microscopic) chromosome

Chromosomes can best be seen when they are highly condensed at metaphase

Stains such as Giemsa that bind to DNA make condensed chromosomes visible

Adding colchicine to actively dividing cells destroys spindle fibers and traps the chromosomes at metaphase



Individual chromosomes can be cut from a picture and aligned by pairs, as shown from the work of Dr. T. C Shu



By convention, the largest chromosome pair is #1, the next # 2 etc.

The resulting picture shows the karyotype (chromosome composition) of the individual.

Humans have 23 "pairs" of chromosomes; 22 pairs of autosomes and one pair (XX or XY) of sex chromosomes

Special techniques produce banding patterns that permit chromosomes of similar size and shape to be distinguished

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Especially useful are fluorescent probes that identify individual chromosomes based on DNA hybridization (FISH or fluorescent in situ hybridization)

Dyes that result in different colors help in karyotyping:



Chromosome vary in relative length of the arms and position of the centromere;

- telocentric: the centromere is at the end (telomere) of the chromosome
- metacentric: the centromere is near the middle
- submetacentric: slightly off-center
- <u>acrocentric</u>: very off center

Centromeres provide the point of attachment (kinetochore) for pulling daughter chromosomes to opposite poles in mitosis and meiosis.

Telomeres are very special; they cannot replicate using DNA polymerase

- . Telomeres:
 - are short highly repeated sequences that protect the ends of chromosomes from exonucleases
 - grow shorter with age in most cells
 - are maintained by telomerase enzyme in germline cells, and in some cancer cells (required for "immortality")

-telomerase is a ribozyme, ie it has an RNA component -the RNA overlaps the repeat and serves as a template to re-exetend

The numbe	r of ch	iromosomes	varies	between	species:
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Humans	46 (23 pairs) per diploid cell		
Dogs	78 (39 pairs)		
Cats	38 (19 pairs)		
Horses	64 (32 pairs)		
Ducks	80		
Alligator	32		
Drosophila (fruitflies)	8		
Horsetail	216		
Sequoia	22		

A genome is one complete set of chromosomes

"ploidy" refers to **the number of sets** of chromosomes per cell

Gametes each contain one genome so are haploid

Most body cells contain 2 sets so are diploid

The amount of DNA (or RNA in some viruses) per genome varies among species

Some viruses have only around a thousand bases, enough for 3 genes

Humans have over 3 billion base pairs per genome

Amphibians and some primitive plants have 10-100 times as much DNA as mammals.

Crickets have 10 times as much DNA/cell as Fruitflies

Yeast, a eukaryotic fungus, and *E. coli*, a bacterium, both have about 6,000 genes even though yeast has more DNA/genome

Much of the eukaryotic genome consists of highly repeated sequences that do not function as genes.