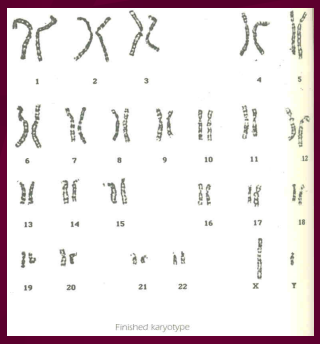
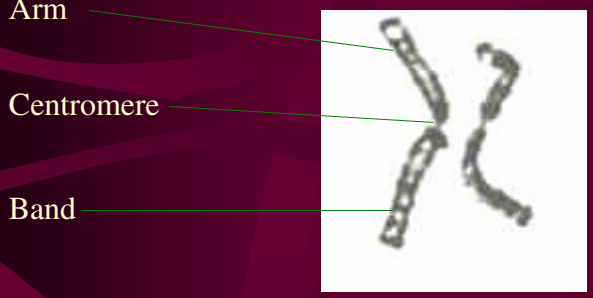
Karyotypes



What is karyotyping?

- A laboratory technique that allows scientists to visualize all the chromosome pairs of an organism in a glance to identify chromosome abnormalities.

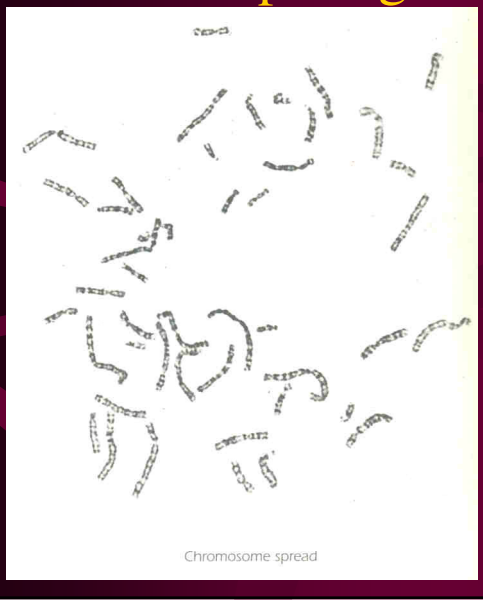
Chromosome Anatomy:



What is a karyotype?

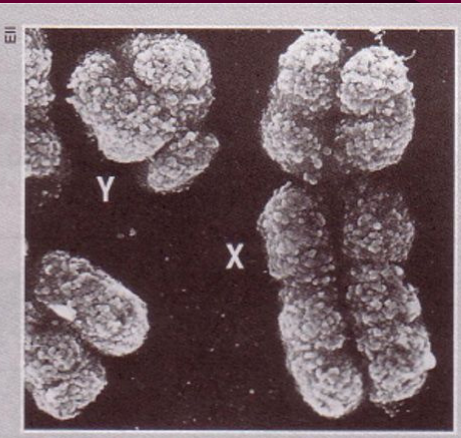
- A photograph of the chromosomes in a normal body cell organized by chromosome pairs.
- Chromosome pairs are determined based on arm size, banding patterns, and centromere location.

Preparing a Karyotype:

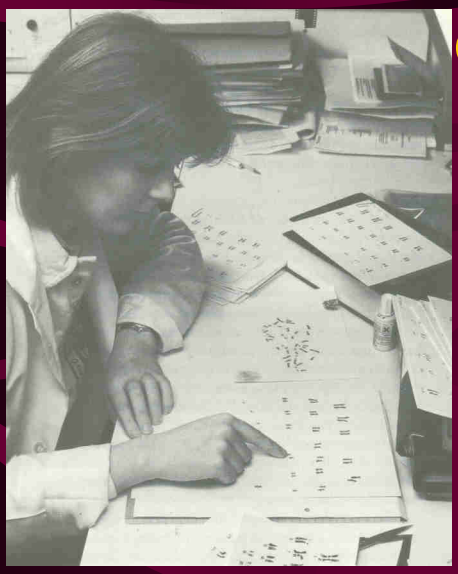


- Cells are removed from the organism.
- When the cells reach the very beginning of metaphase, a chemical called Colchicine is added to stop mitosis.
- Chromosomes are then placed on a microscope slide and photographed.
- The photograph is called a chromosome smear or chromosome spread.

A scanning electron micrograph (SEM) of human chromosomes clearly showing their double chromatids.

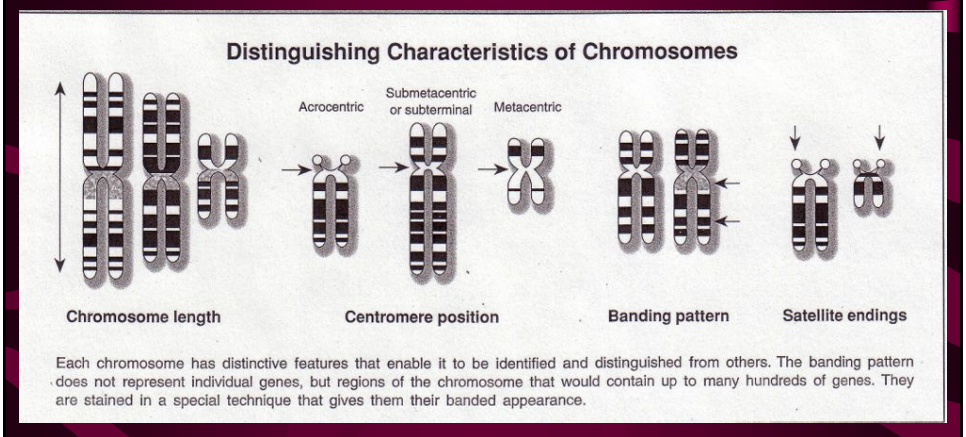


This SEM shows the human X and Y chromosomes. Although these two are the sex chromosomes, they are not homologous.

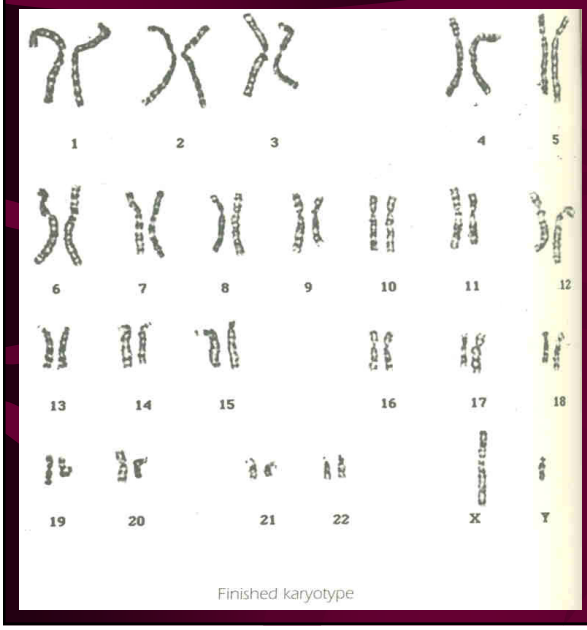


Cytogeneticists...

- Cytogeneticists are Scientists who study karyotypes.
- After the chromosome spread is made, the cytogeneticists enlarge and cut out the chromosomes.
- Then the chromosomes are organized by pairs.



Completed Karyotype

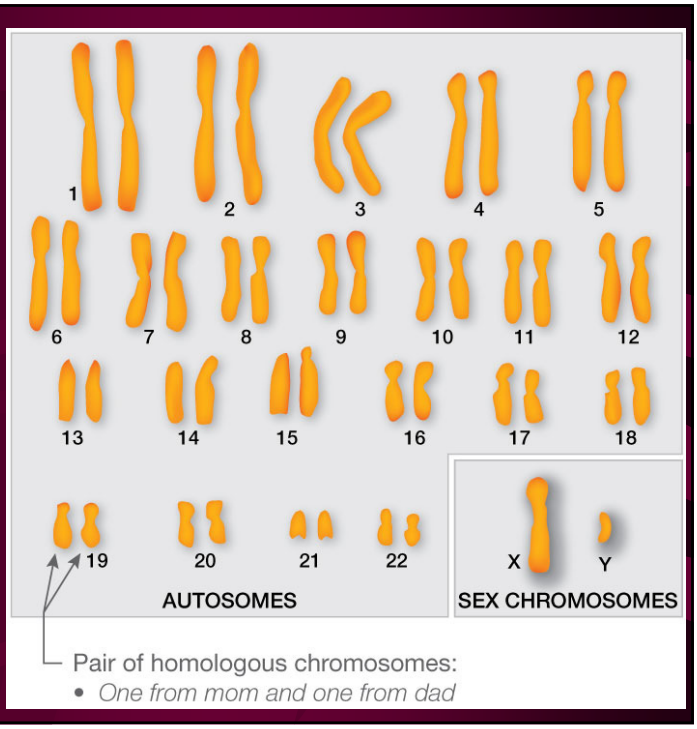


When the karyotype is complete, the cytogeneticist examines it for abnormalities (aneuploidies) and sex determination.

What is an aneuploidy?

- An abnormal number of chromosomes
- Normal is 2N
- Monosomy 2N -1
- Trisomy is 2N +1
- Tetrasomy 2N +2

- Sex Chromosomes:
  - X and Y chromosomes
  - 23rd pair
  - Used to determine sex
    - If there is a Y it is male
    - No Y it is female
- Autosomes:
  - All chromosomes except Sex Chromosomes
  - 1st -22nd pairs



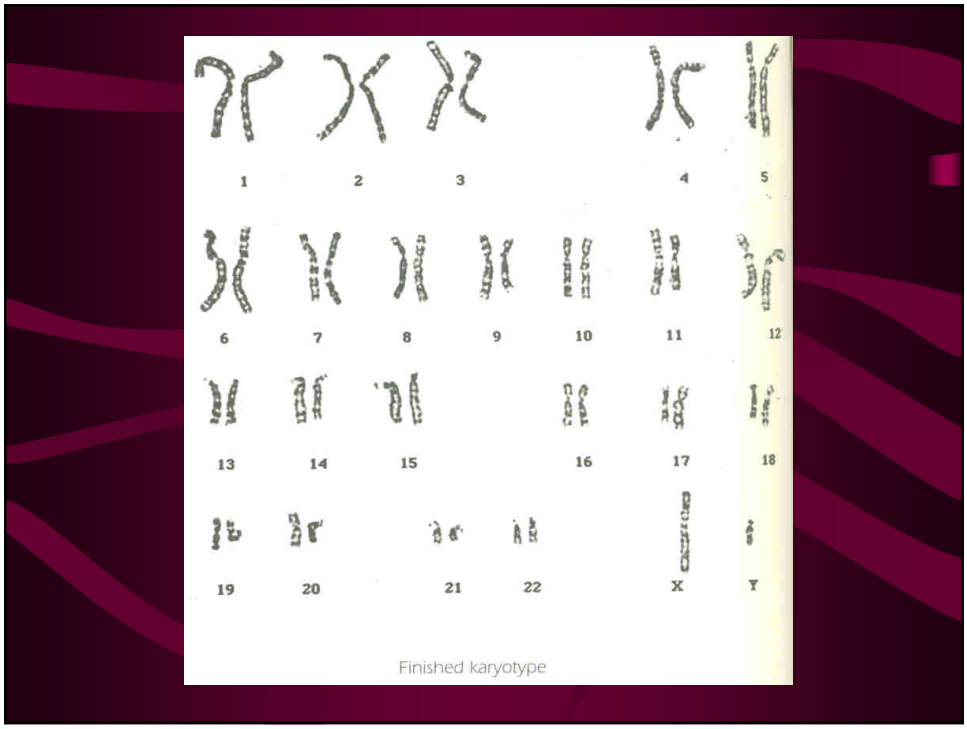
Notation

- Identifies the total number of chromosomes, sex chromosomes, and if there is a noticeable chromosome aneuploidy.
- Normal

  Female: 46, XX and Male: 46, XY
- Aneuploidy
  - Many kinds depending on the abnormality
    - i.e.:

      47, XX, +13

      47, XXY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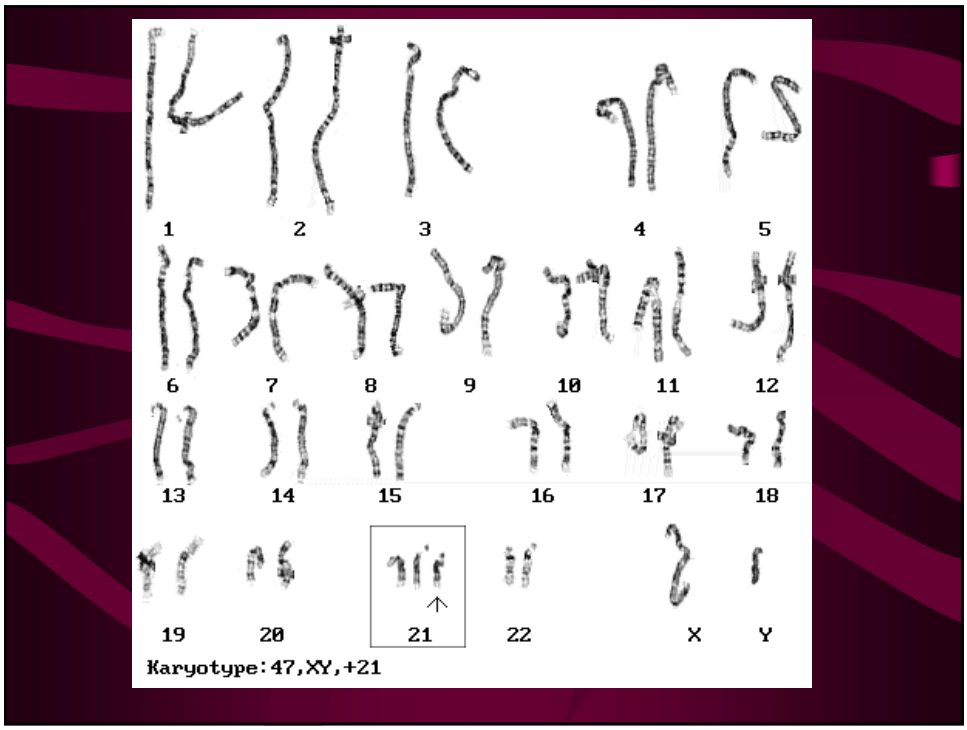
Finished karyotype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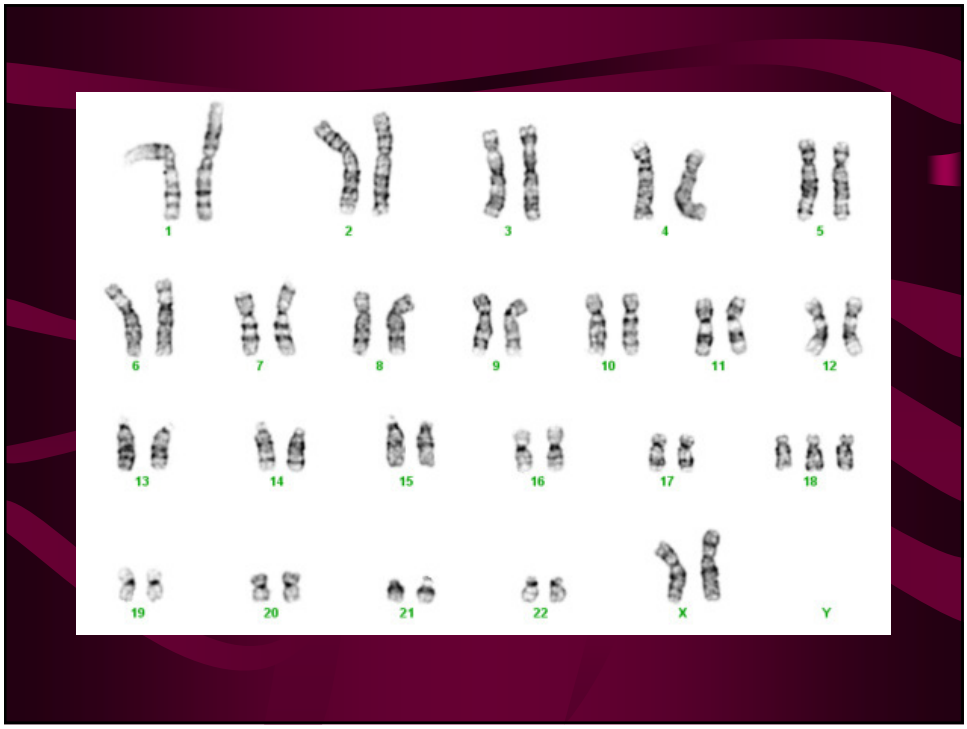
Karyotype:47,XY,+21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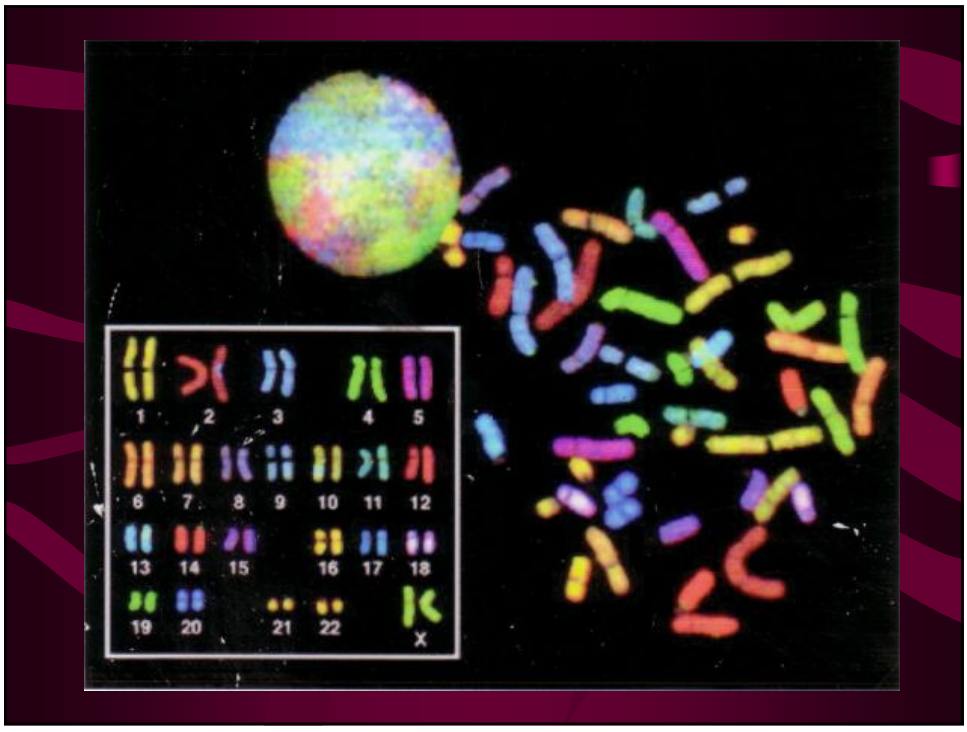
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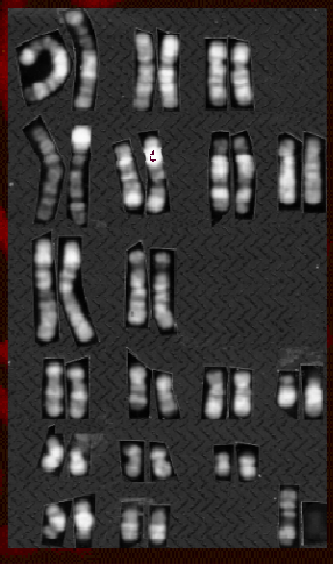
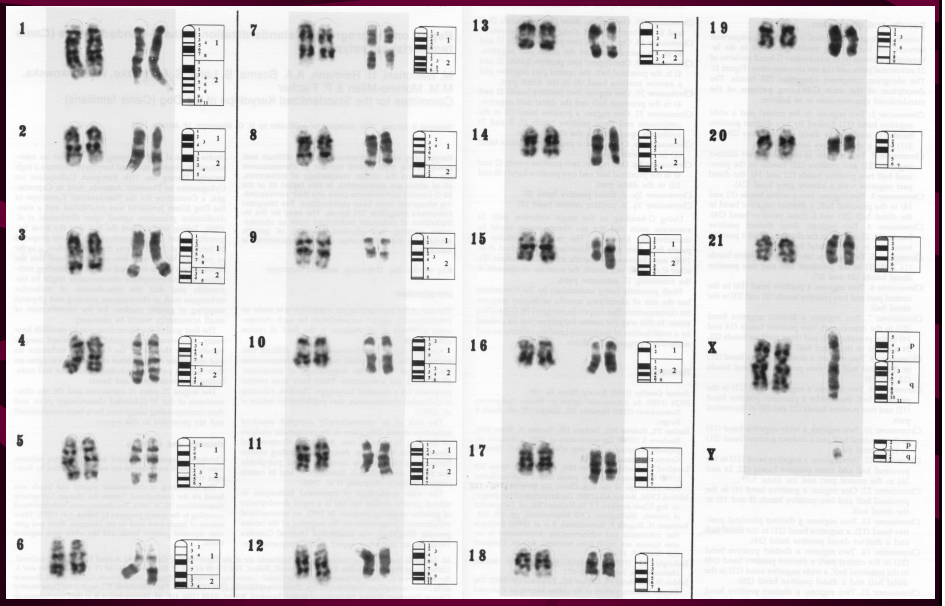
Karyotypes of other Animals:

Normal karyotype, cat

2N = 38
XY



Dog: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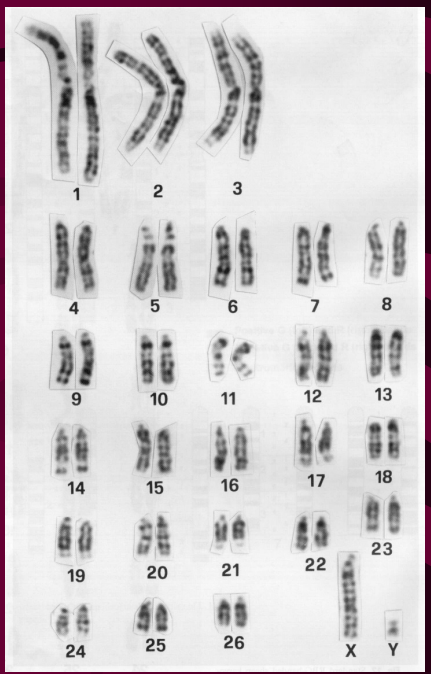
Sheep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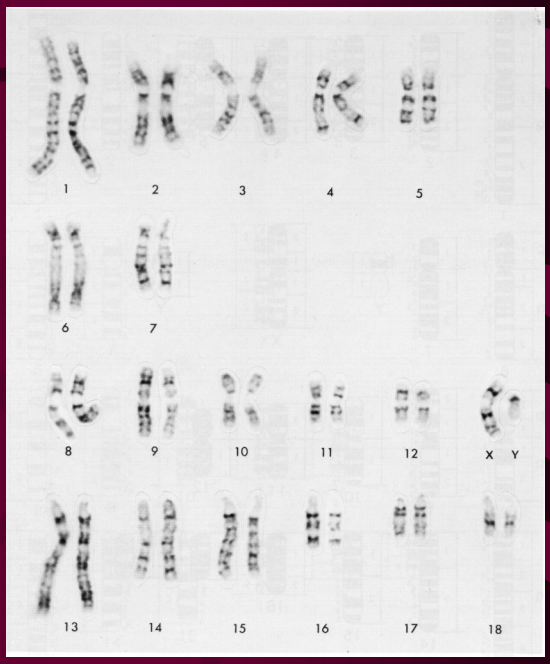
Pig: