Lecture 1.
Introduction to Cytogenetics

Lovinskaya Anna Vladimirovna,

PhD, Departure of Molecular Biology and Genetics

- **Genetics is the science of the laws of heredity and variability**
- **CYTOGENETICS is the area of genetics, studying the cytological basis of heredity and variability**
- **CELLULAR GENETICS is the field of genetics that studies extrachromosomal (cytoplasmic) heredity**
- **The main subject of CYTOGENETICS is CHROMOSOME, its organization, functioning and inheritance**
- **Cytogenetics uses methods of genetics and cytology and is closely related to the sections of these sciences: molecular genetics, cytochemistry, karyology and others.**

Cytogenetics

General cytogenetics

- common cellular basis of heredity;
- Population cytogenetics;
- Radiation cytogenetics

on the level being studied:

- Microscopic cytogenetics;
- submicroscopic cytogenetics;
- molecular cytogenetics

Private cytogenetics

- plant cytogenetics;
- animal cytogenetics;
- human cytogenetics;
- medical (clinical) cytogenetics

The laws of inheritance

1) The law of uniformity
2) The law of segregation
3) The law of independent assortment
4) The chromosome theory of inheritance

Gregor Mendel,
1822-1884

The laws of inheritance

1) The ***law of uniformity*** refers to the fact that when two homozygotes with different alleles are crossed, all of the offspring in the F1 generation are identical and heterozygous.
2) The ***law of segregation*** refers to the observation that each individual possesses two genes for a particular characteristic, only one of which can be transmitted at any one time.
3) The ***law of independent assortment*** refers to the fact that members of different gene pair segregate to offspring independently of one another.

The laws of inheritance

The chromosomal basis of inheritance

Boveri and Sutton's chromosome theory of inheritance states that genes are found at specific locations on chromosomes, and that the behavior of chromosomes during meiosis can explain Mendel's laws of inheritance.

Thomas Hunt Morgan, who studied fruit flies, provided the first strong confirmation of the chromosome theory.

Walter Sutton, 1877-1916

Theodor Boveri, 1862-1915

Thomas Hunt Morgan, 1866-1945

The chromosome theory of inheritance

1) Genes are located on chromosomes.

2) Genes are located on the chromosome in a linear sequence.

3) Different chromosomes contain a different number of genes. In addition, the set of genes for each of the non-homologous chromosomes is unique.

4) Alleles of genes occupy identical loci in homologous chromosomes.

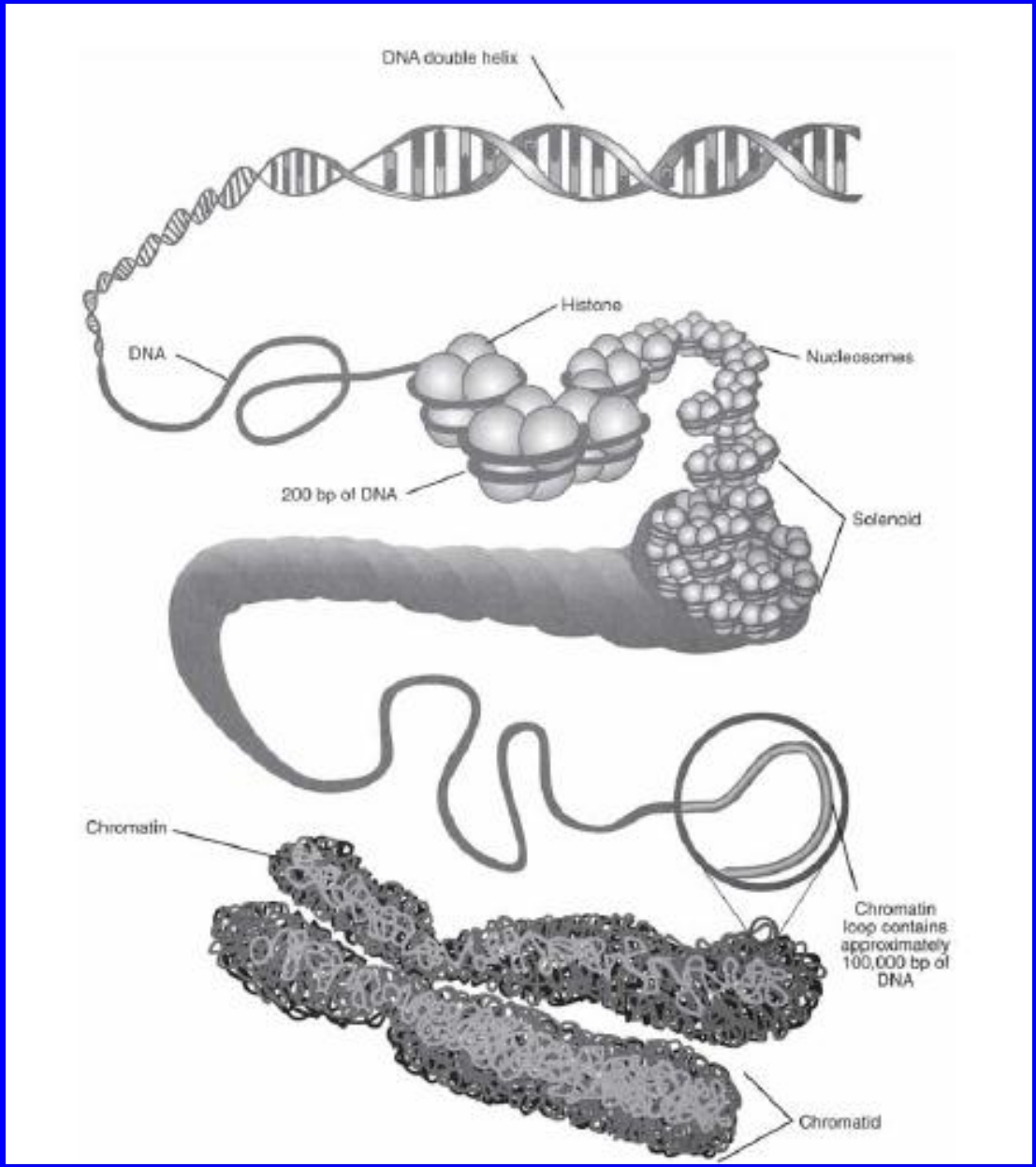
The chromosome theory of inheritance

5) The genes of one chromosome form a linkage group, that is, they are inherited mainly in a cohesive manner (due to which the linked inheritance of certain features occurs). The number of linkage groups is equal to the haploid number of chromosomes of this type (in the homogametic sex) or more by 1 (in heterogametic sex).

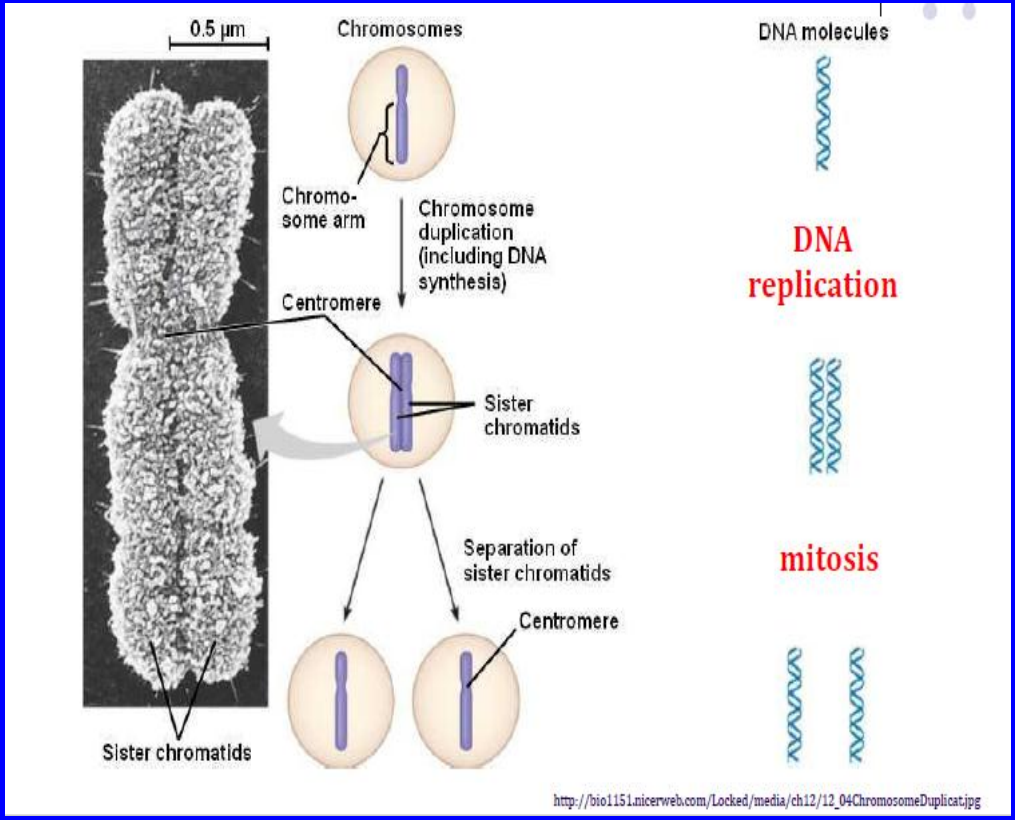
6) The clutch is broken as a result of crossing-over, the frequency of which is directly proportional to the distance between the genes in the chromosome (therefore, the adhesion force is inversely related to the distance between the genes).

7) Each biological species is characterized by a certain set of chromosomes - a karyotype.

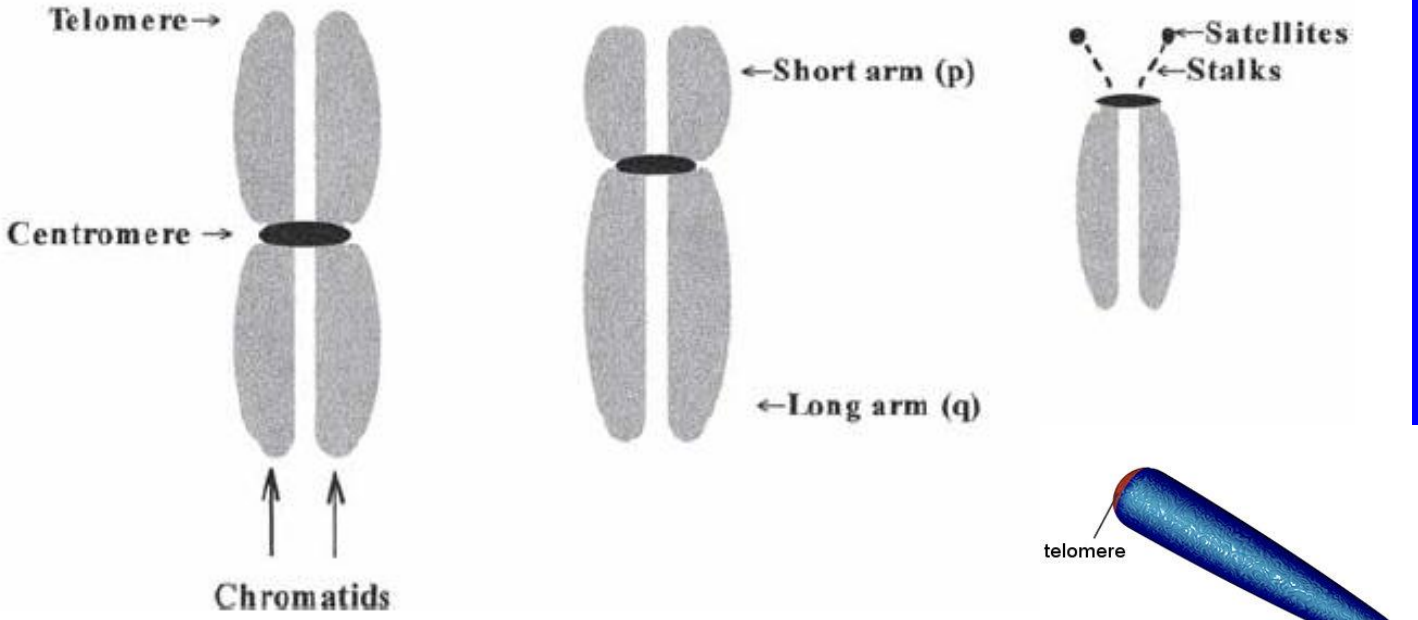
Chromosome. The levels of DNA organization

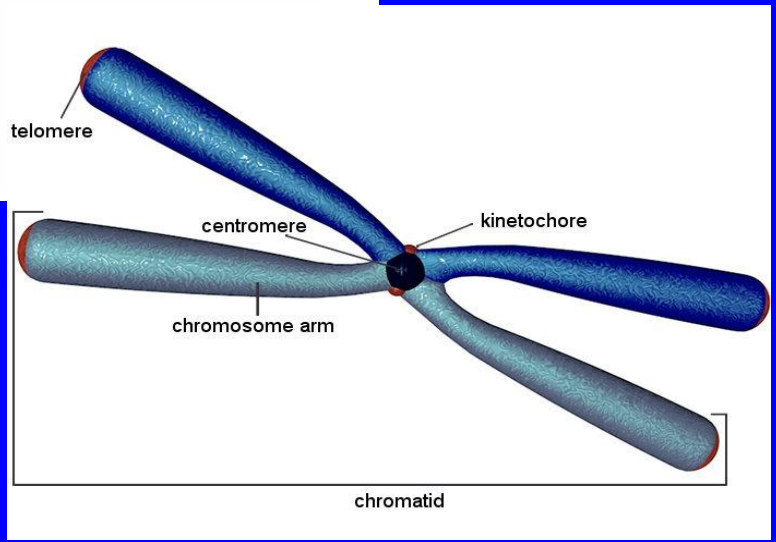


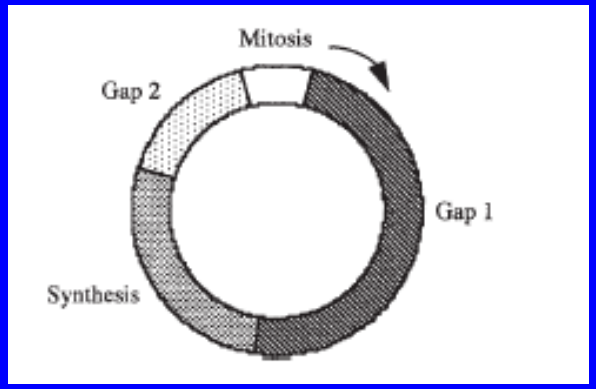
- cell division: equal distribution of DNA → chromosomes form
- 2 chromatids = chromosome → are identical
- 1 DNA molecule = chromatid



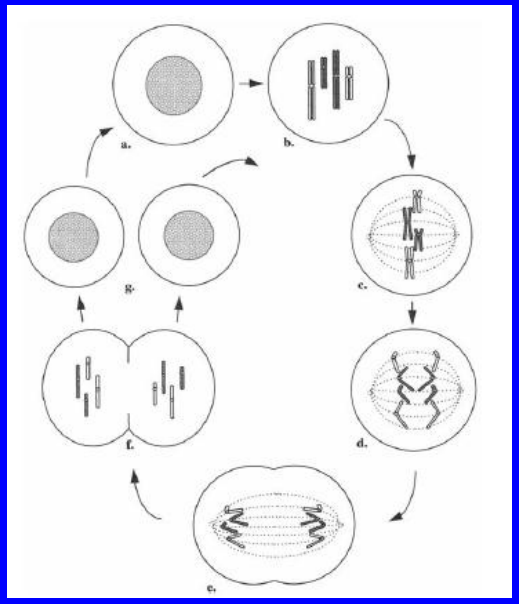
The functional and structural components of metaphase chromosome



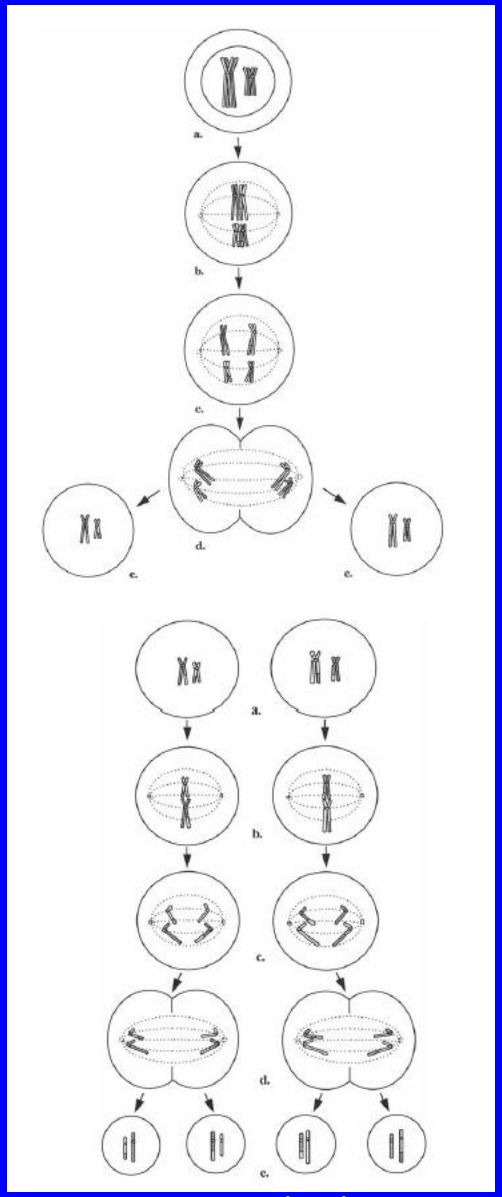




Cell cycle



Mitosis



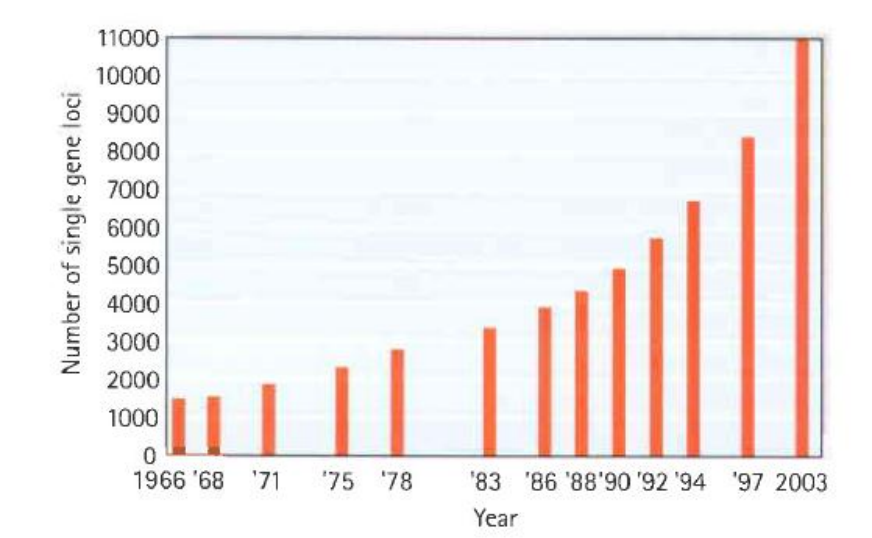
Meiosis

Chromosome number in various animal

Animal	Total number of chromosomes	Animal	Total number of chromosomes
Human	46	Toad	22
Cat	38	Chimpanzee	48
Dog	78	Mosquito	6
Pig	38	Earthworm	36
Goat	60	Chicken	78
Sheep	54	Fruit fly	8
Cattle	60	Crayfish	>100
Horse	64	Mallard	80
Donkey	62	Rat	42
Rabbit	44	Mouse	40

Hereditary conditions:

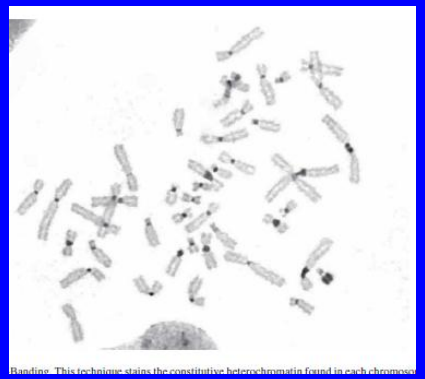
- Single gene disorders (albinism);
- Chromosome abnormalities (Down syndrome);
- Multifactorial disorders (diabetes mellitus);
- Acquired somatic genetic disease (cancer)

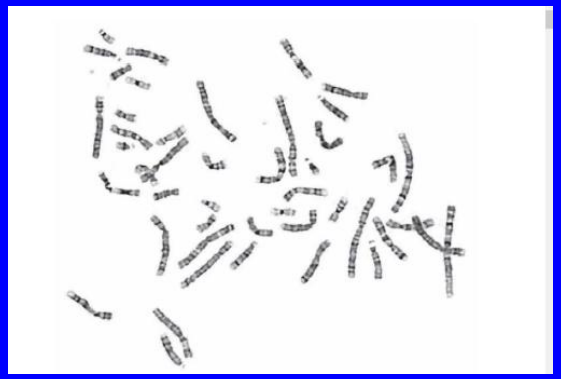


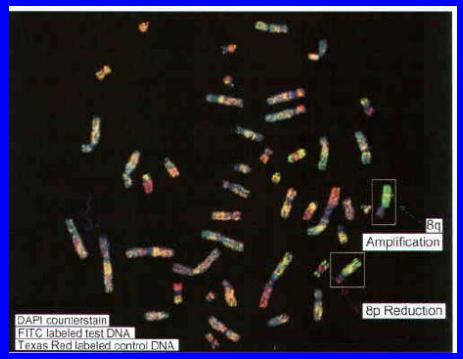
Histogram showing the rapid increase in recognition of conditions and characteristics (traits) showing single-gene inheritance (Adapted from McKusick 1998 and OMIM)

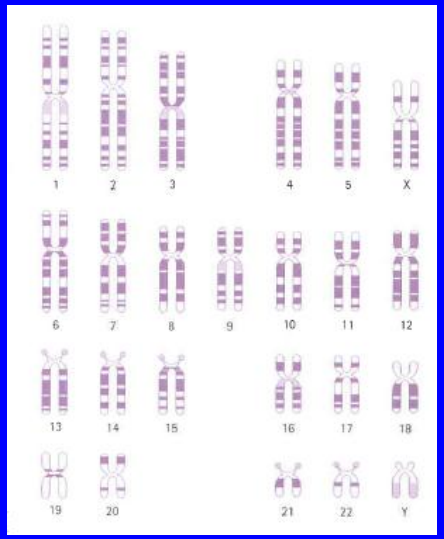
Methods of chromosomal analysis

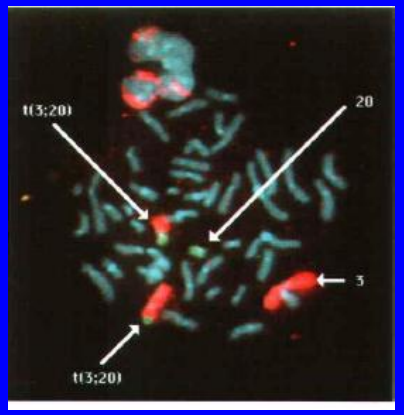
- Chromosome preparation and chromosome banding (G-, Q-, R-, C-banding, high-resolution banding);
- Karyotype analysis;
- Fluorescent *in-situ* hybridization (FISH);
- Comparative genomic hybridization (CGH)





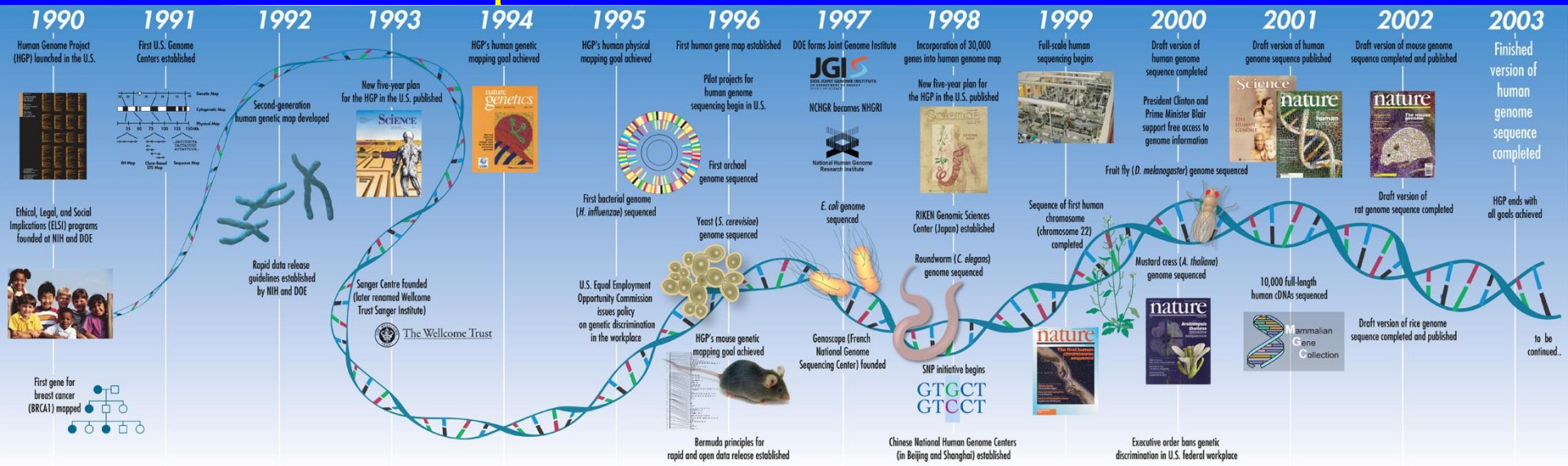






Major new developments

The ***Human Genome Project*** **(HGP)** was an international scientific research project with the goal of determining the sequence of nucleotide base pairs that make up human DNA, and of identifying and mapping all of the genes of the human genome from both a physical and a functional standpoint. The Human Genome Project was declared complete in 2003.



Major new developments

Genome 10K (G10K) is a project to sequence the genome of at least one individual from each vertebrate genus, approximately 10,000 genomes. This target number could expand in the future to consider all ~66,000 named species of vertebrates. The Genome10K project was established in 2009.

Thank you for attention!