RNA Viruses and Transposable Elements or The Human Genome Project and Functional Genomics

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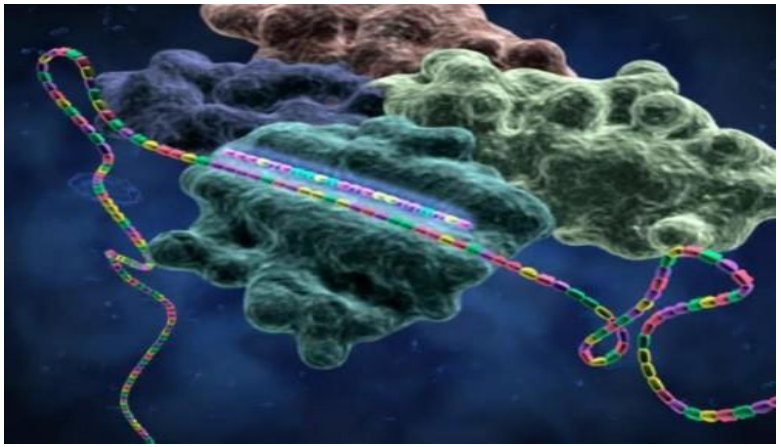
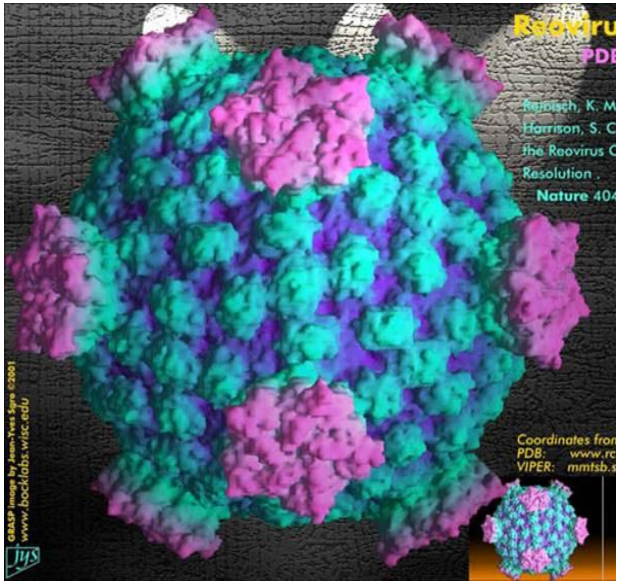
Plan

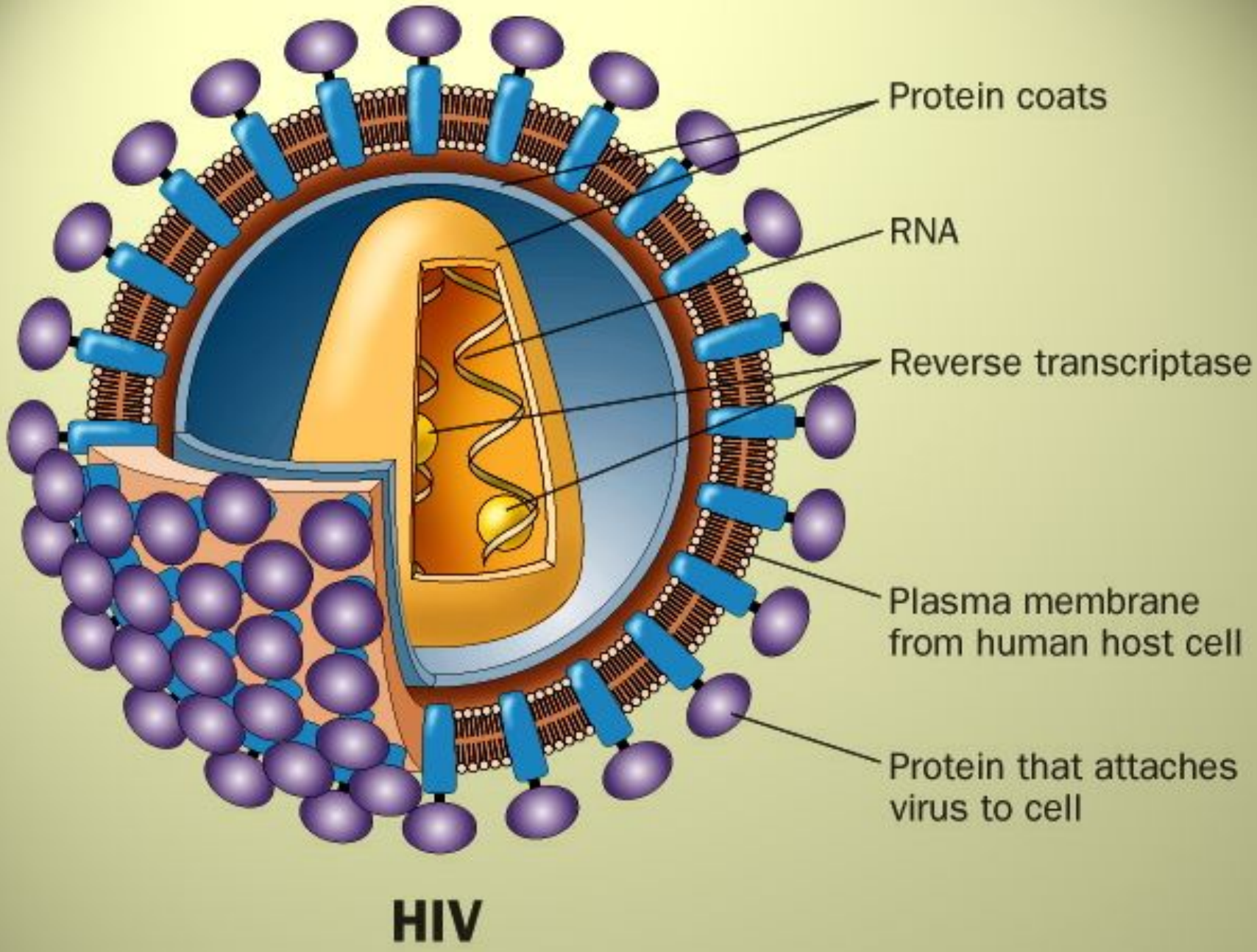
- 1. RNA containing viruses (HIV)
- 2. The human genome project
- 3. Genomes of viruses
- 4. Structure and Genome of viruses
- 5. Functional genomics



RNA-containing viruses:

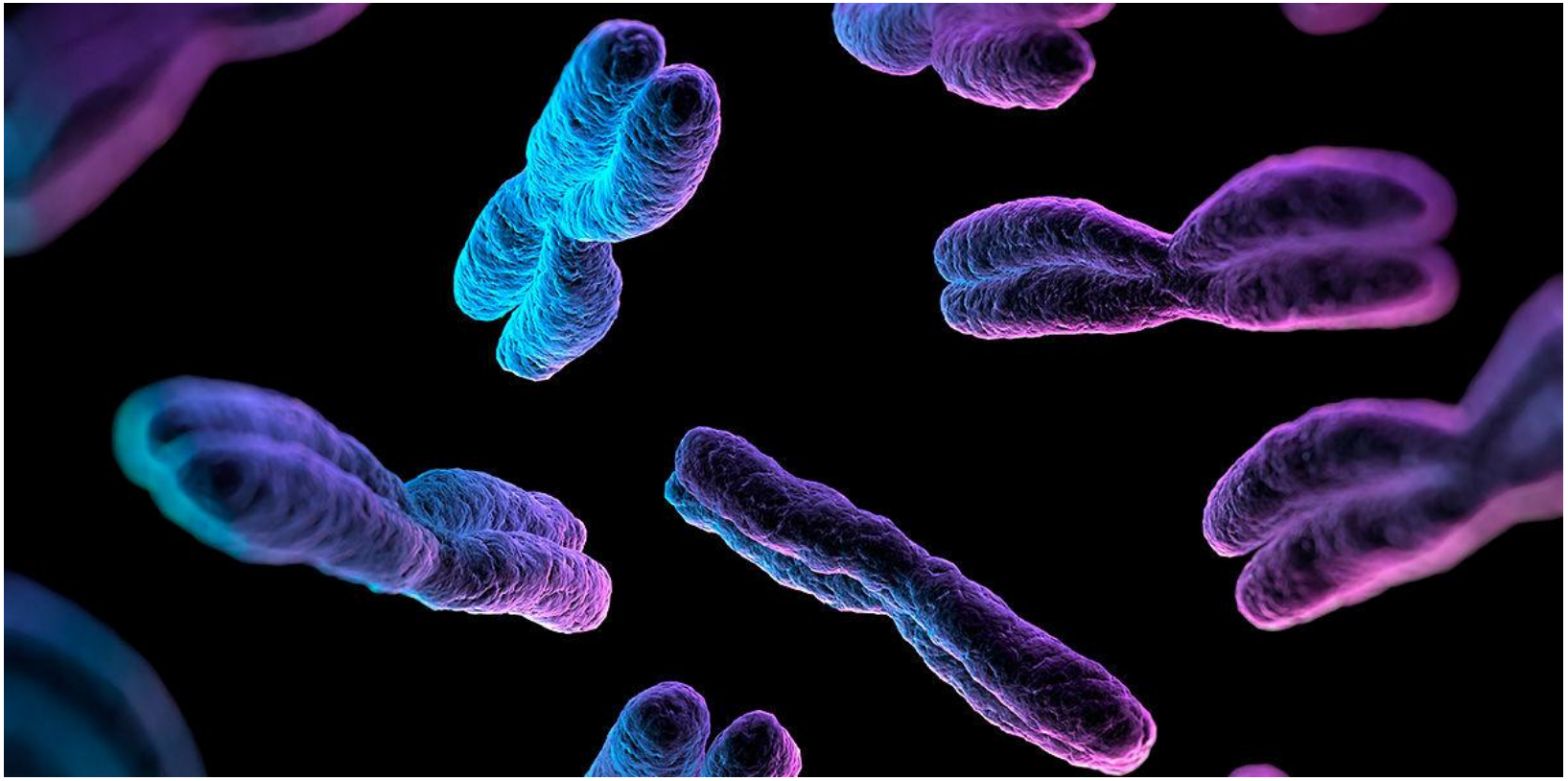
- 1 - paramyxoviruses;
- 2 - influenza viruses;
- 3 - coronaviruses;
- 4 - arenaviruses;
- 5 - retroviruses;
- 6 - reoviruses;
- 7 - picornaviruses;
- 8 - kapitsivirusy;
- 9 - rabdoviruses;
- 10 - togaviruses, flaviviruses;
- 11 - Bunyavirus




Protein coats
RNA
Reverse transcriptase
Plasma membrane from human host cell
Protein that attaches virus to cell
HIV

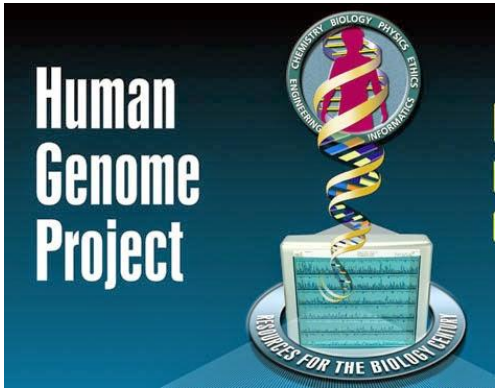
- **Genomics is the study of the genome of an organism - its entire genetic material in the form of RNA, DNA, genes and chromosomes. It concentrates on understanding the structure and function of an organism's genetic material from the molecular level upwards, including interactions between genes, interactions between genes and the proteins they produce, and interactions between genes and environmental factors. It has obvious links to the related science of proteomics, which focuses on understanding the structure and function of the proteins produced by the genome (the proteome).**

The human genome consists of 23 pairs of chromosomes present in the nucleus, as well as mitochondrial DNA. Twenty-two autosomal chromosomes, two sex chromosomes X and Y, as well as human mitochondrial DNA contain together approximately 3.1 billion base pairs.



Features of the human nuclear genome

1. The size of the genome is 3 × 10 9 bp
2. 3 0000 genes
3. Linear DNA molecules
4. Molecules of polyrep pleton DNA (the number of ori-sites depends on the length of the molecule)
5. There are no operatives
6. Mosaic (intron-exon) structure of genes
7. Greater (in comparison with prokaryotes) the number of regulatory elements
8. The presence of multi genic families and pseudogenes
9. The presence of unique and repetitive sequences.



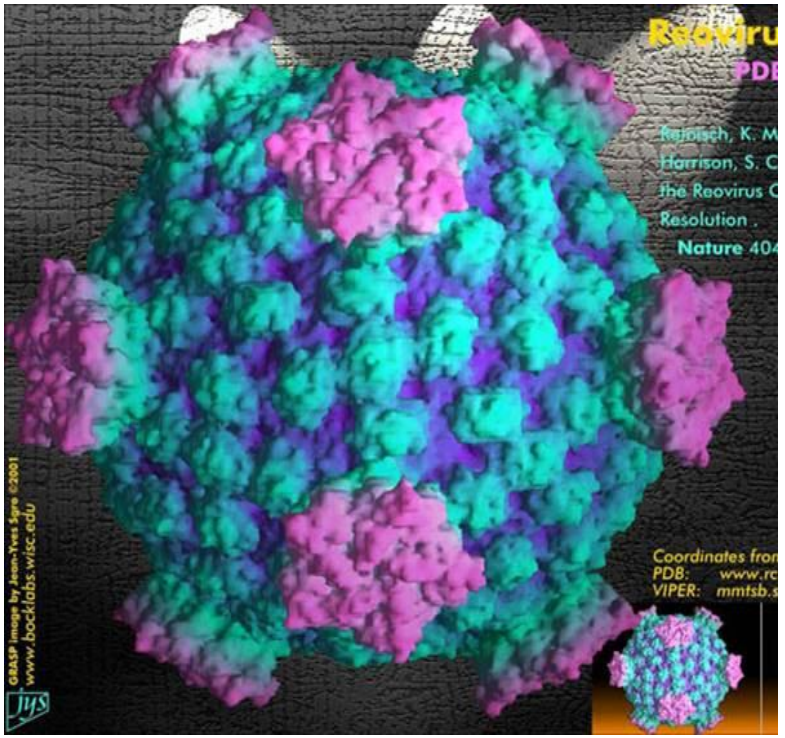
- **The genome (the Greek ending "ohm" is translated as "all, full")**
- **originally understood as a complete set of genes.**
- **The genome is a collection of hereditary**
- **cell or virus, stored and**
- **transmitted to the next generation in the form**
- **nucleotide sequence of DNA molecules**
- **(in the case of some viruses, RNA).**
- **That is, the genome is not only all genes, but all**
- **The remaining parts of the DNA from the yet unknown**
- **function.**

- **Genomes of viruses**.

- The most simple

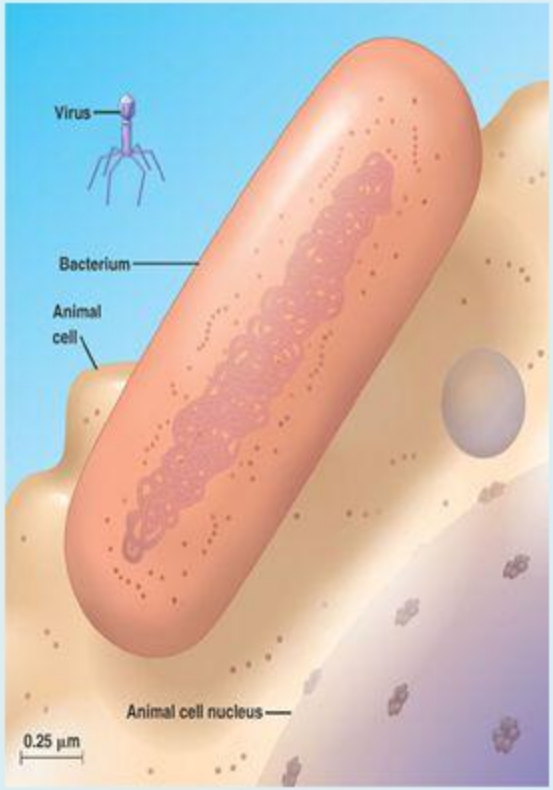
- Very compact - contain only genes closely adjacent to each other, sometimes overlapping

The number of genes ranges from 3 to 30 for simple viruses to 200 for the most complex ones.

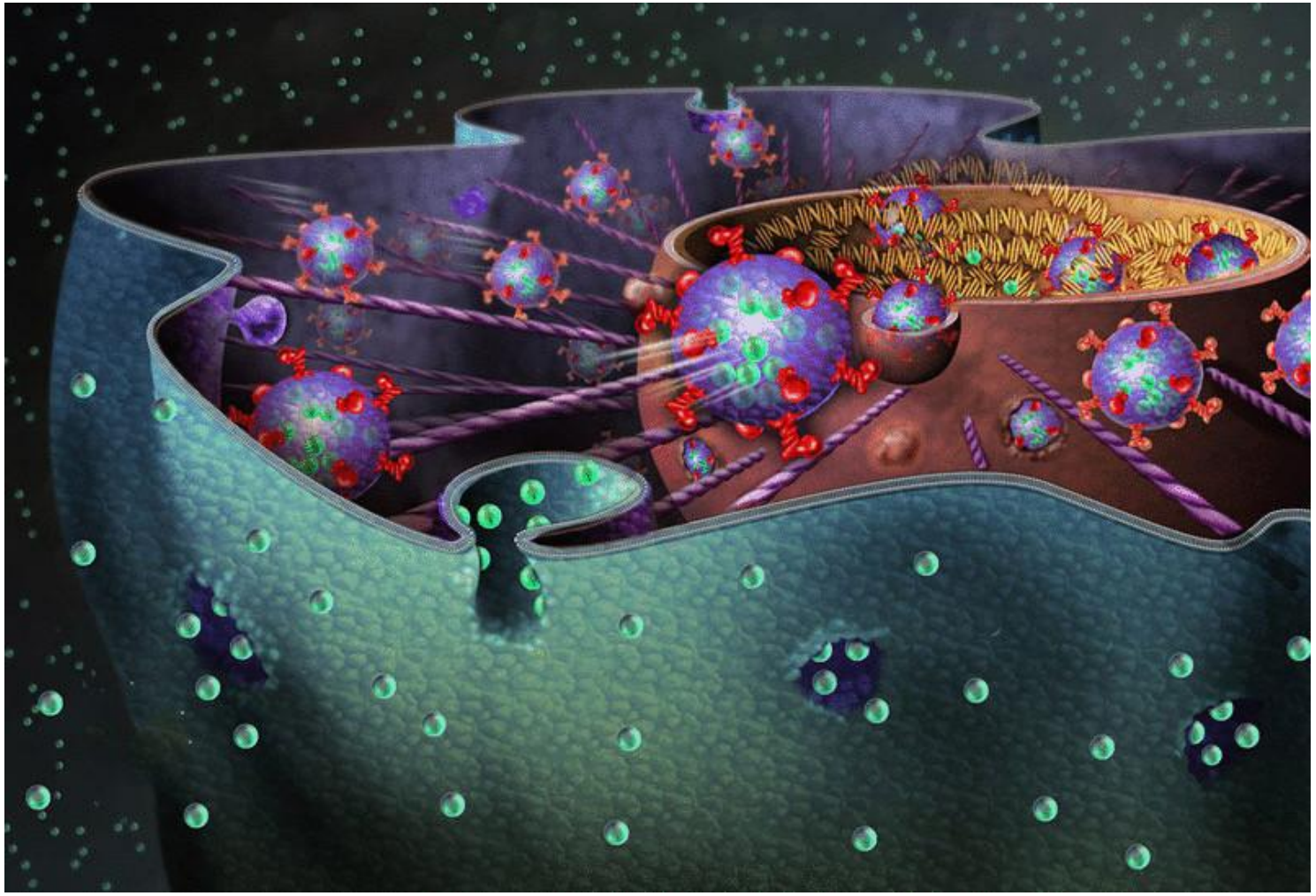


Structure and Genome of Viruses

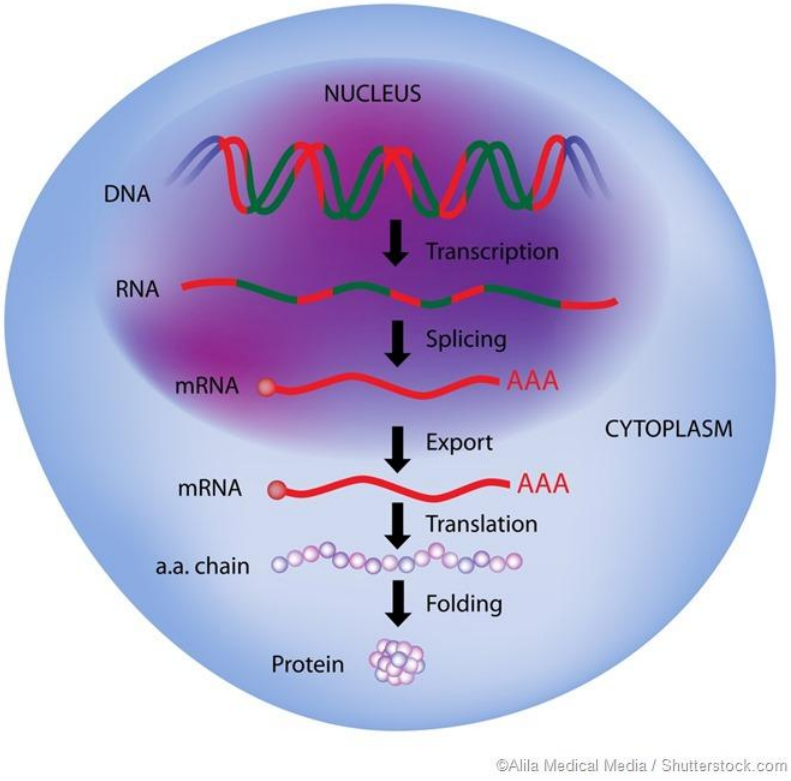
- Viruses are smaller and more simple that prokaryotes
- Lack structures and metabolic machinery found in most cells
- Most are a nucleic acid enclosed in a protein coat
- Genomes have many possibilities
  - -double stranded DNA
  - -single stranded DNA
  - -double stranded RNA
  - -single stranded RNA
- Most have genome in linear sequence or circular ranging from 4 genes to several hundred



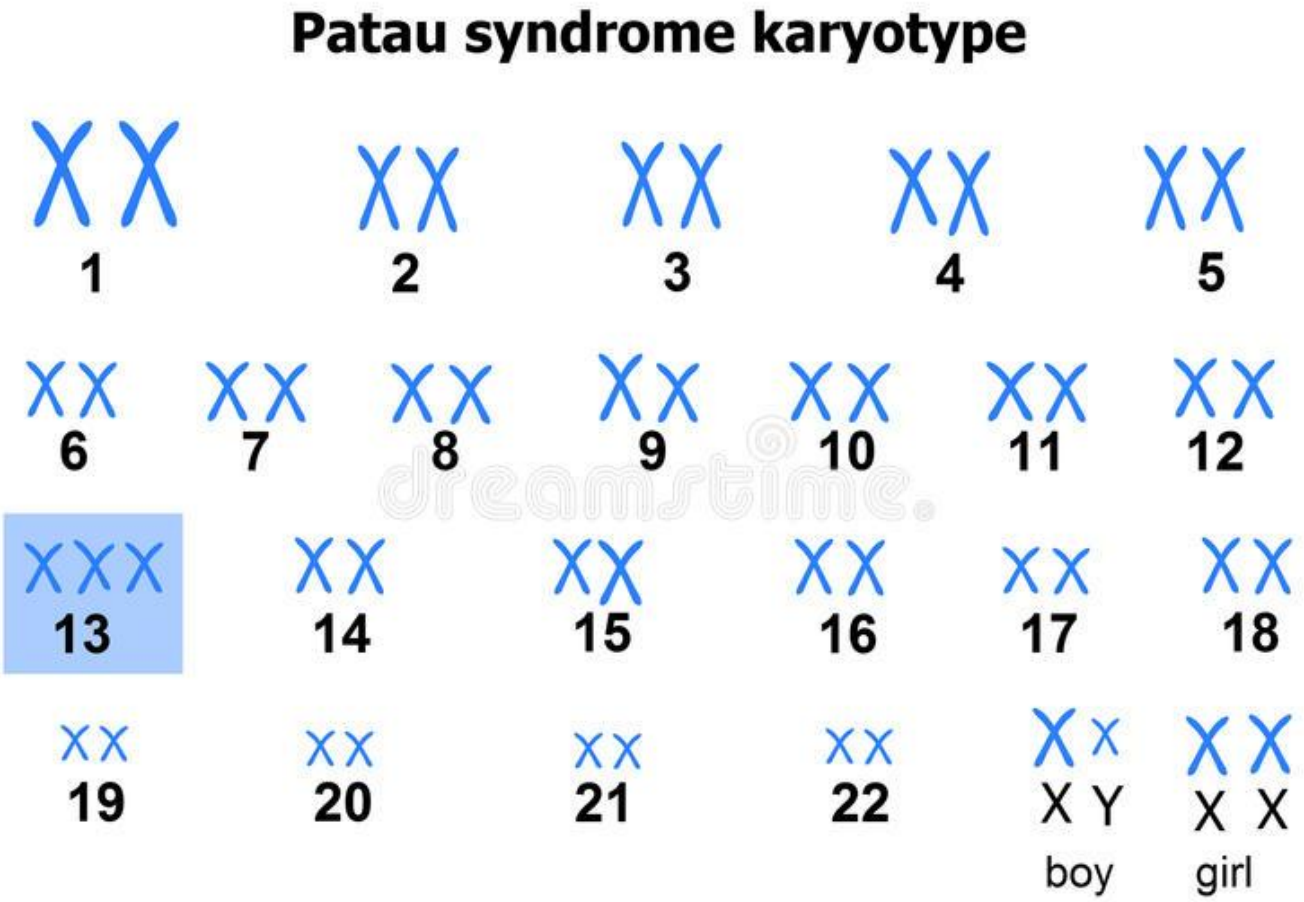
- **Viruses are an integral part of the human genome. Unfortunately this is the case.**



Functional genomics - the implementation of information recorded in the genome, from the gene - to the trait.



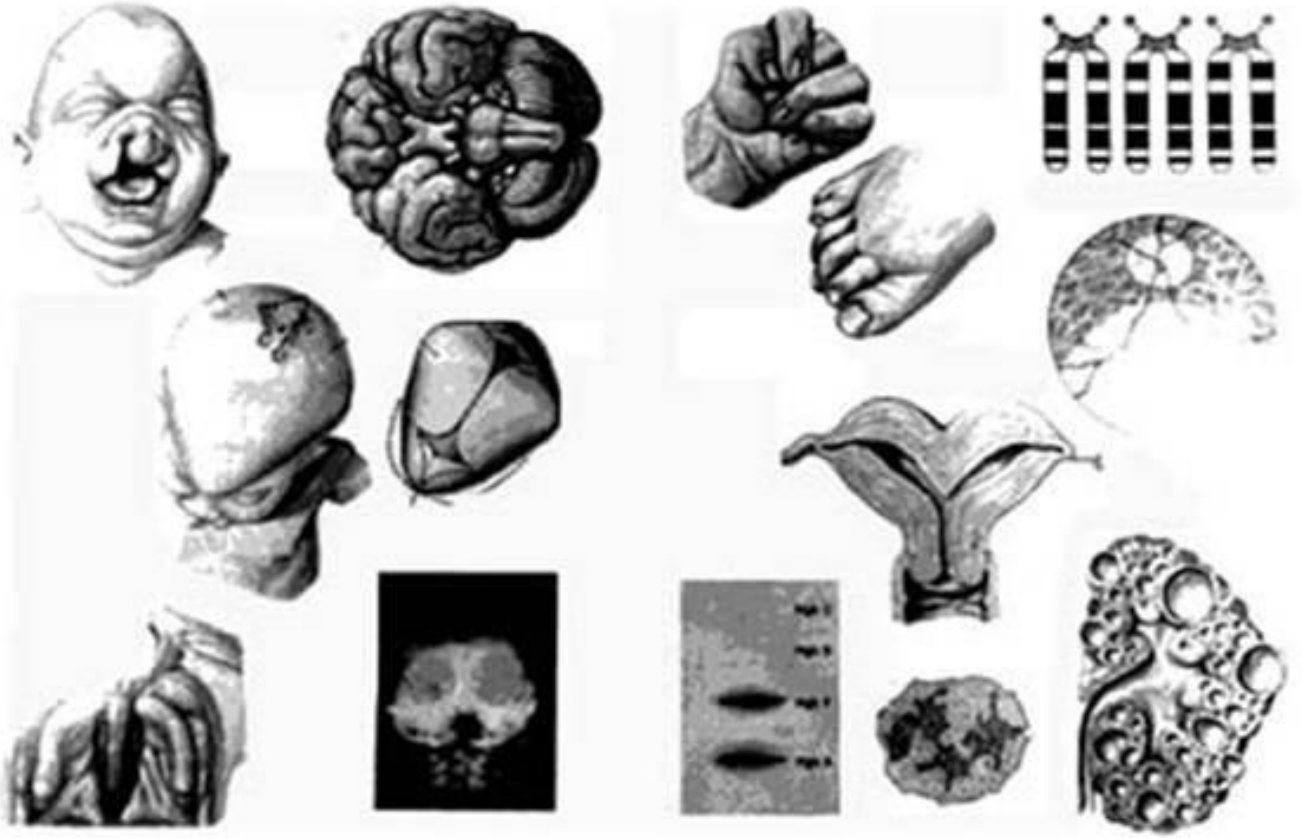
Tuberous Sclerosis or Patau's Syndrome (Trisomy 13)



PLAN

- The Patau Syndrome
- Congenital malformations
- The history of the discovery of the Patau Syndrome
- Defects of internal organs
- Cleft lip and palate
- Treatment of the Patau syndrome
- Clinical sings
- Forecast
- Polydactyly

The Patau syndrome is a chromosomal abnormality, a syndrome in which the patient has an additional 13 chromosomes, due to the non-dissociation of chromosomes during meiosis (also known as trisomy 13 and trisomy D). Some of them are caused by Robertson's translocation. Additional 13 chromosomes disrupt the normal course of the child's development, namely the occurrence of heart and kidney defects, in addition to other features characteristic of the Patau syndrome.



PATAU SYNDROME

- Generally not inherited, sometimes can be
- Genetic Changes:
  - Additional DNA from 13 (usually extra copy)
- Problems:
  - Polysomy due to nondisjunction
  - Translocation (partial trisomy of 13)
- Symptoms:
  - Sever retardation
  - Small eyes
  - Cleft lip/palate
  - Don't often live past infancy

Congenital malformations

• violation of the formation of the brain;
• eyeballs, brain and facial parts of the skull;
• the circumference of the skull is usually reduced,
• the forehead is beveled, low;
• the eye slits are narrow;
• sunken nose; auricles low situated and deformed;
• clefts of the upper lip and palate (usually bilateral).

The history of the discovery of the Patau syndrome

He was the first to describe in scientific medicine Patau syndrome - a chromosomal disease of a person, which is characterized by the presence in cells of an additional chromosome.

Rasmus Bartholinus (latin Erasmus Bartholinus, August 13, 1625, Roskilde - November 4, 1698) - Danish scientist, physician, anatomist, physicist, mathematician, professor of the 17th century

Defects of internal organs

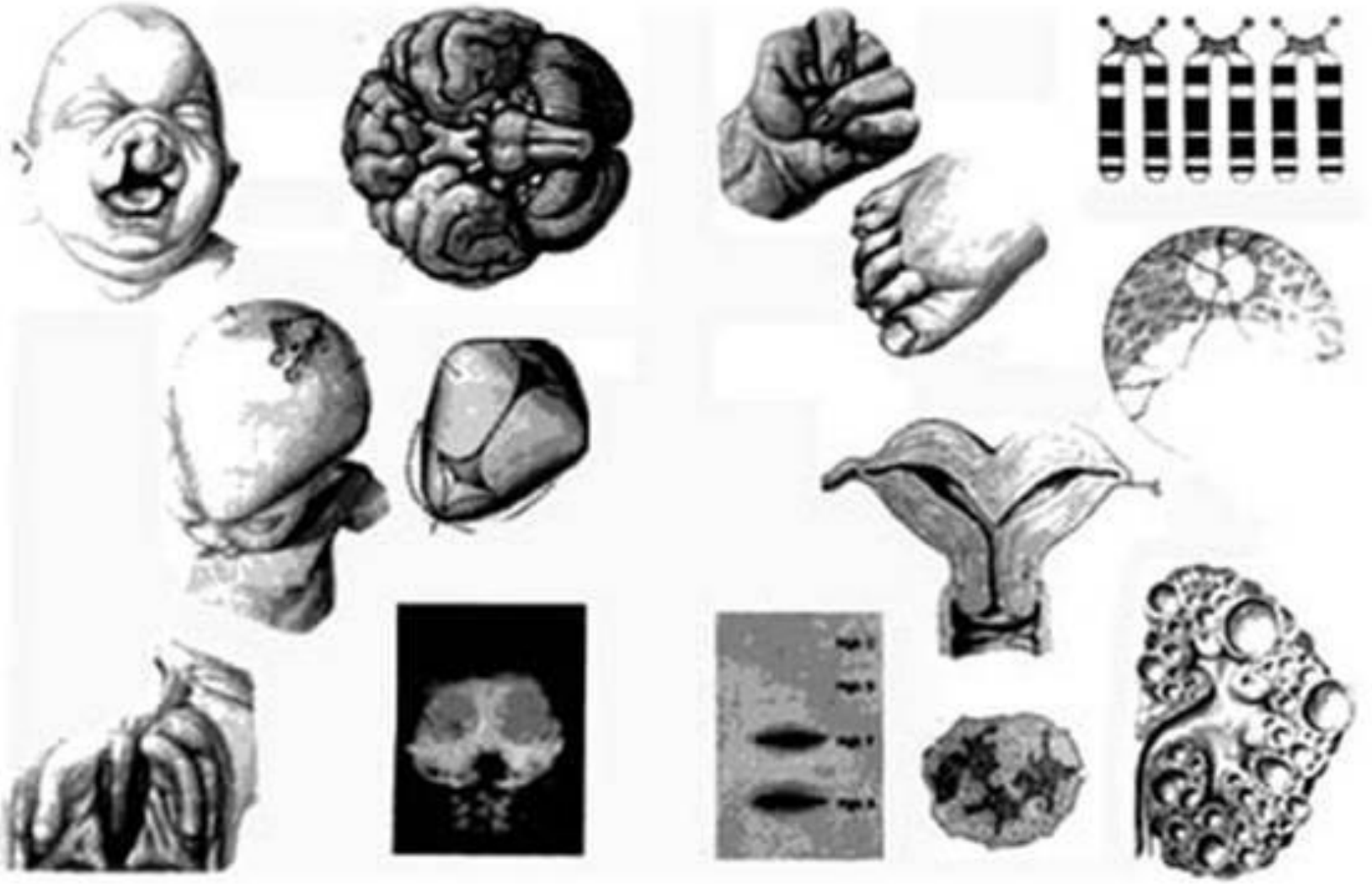
- Defects of the partitions of the heart,
- Incomplete turn of the intestine,
- Kidney cysts,
- Anomalies of internal genital organs,
- Pancreatic defects. polydactyly (more often bilateral and on hands) and flexory position of brushes are observed.

Cleft lip and palate

Treatment of the Patau syndrome

- Treatment of the Patau syndrome is non-specific, which includes carrying out operations on the fact of congenital malformations, mandatory prevention of infectious and catarrhal diseases, conduction of general restorative treatment. Most children born with Patau's syndrome, due to the presence of severe malformations, die before reaching one year (95% of cases), but cases were recorded when prolongation of life up to five and even up to ten years was provided by prompt elimination of malformations, full nutrition and careful care.

CLINICAL SIGNS



Forecast

- Children with the Patau's syndrome die in the first weeks or months (95% - up to 1 year). Some patients live for several years. There is a tendency to increase the life expectancy of patients with Patau's syndrome up to 5 years (about 15% of children)

Polydactyly

Thank you for attention!