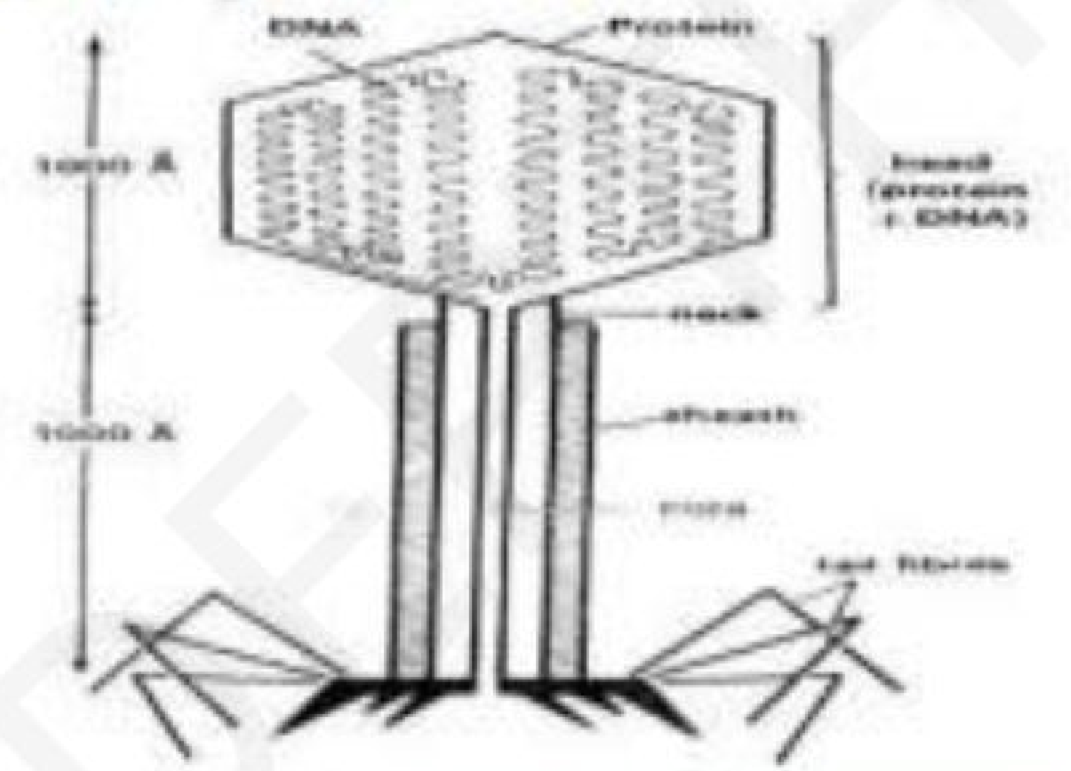
GENOMIC ORGANISATION IN MAMMALS

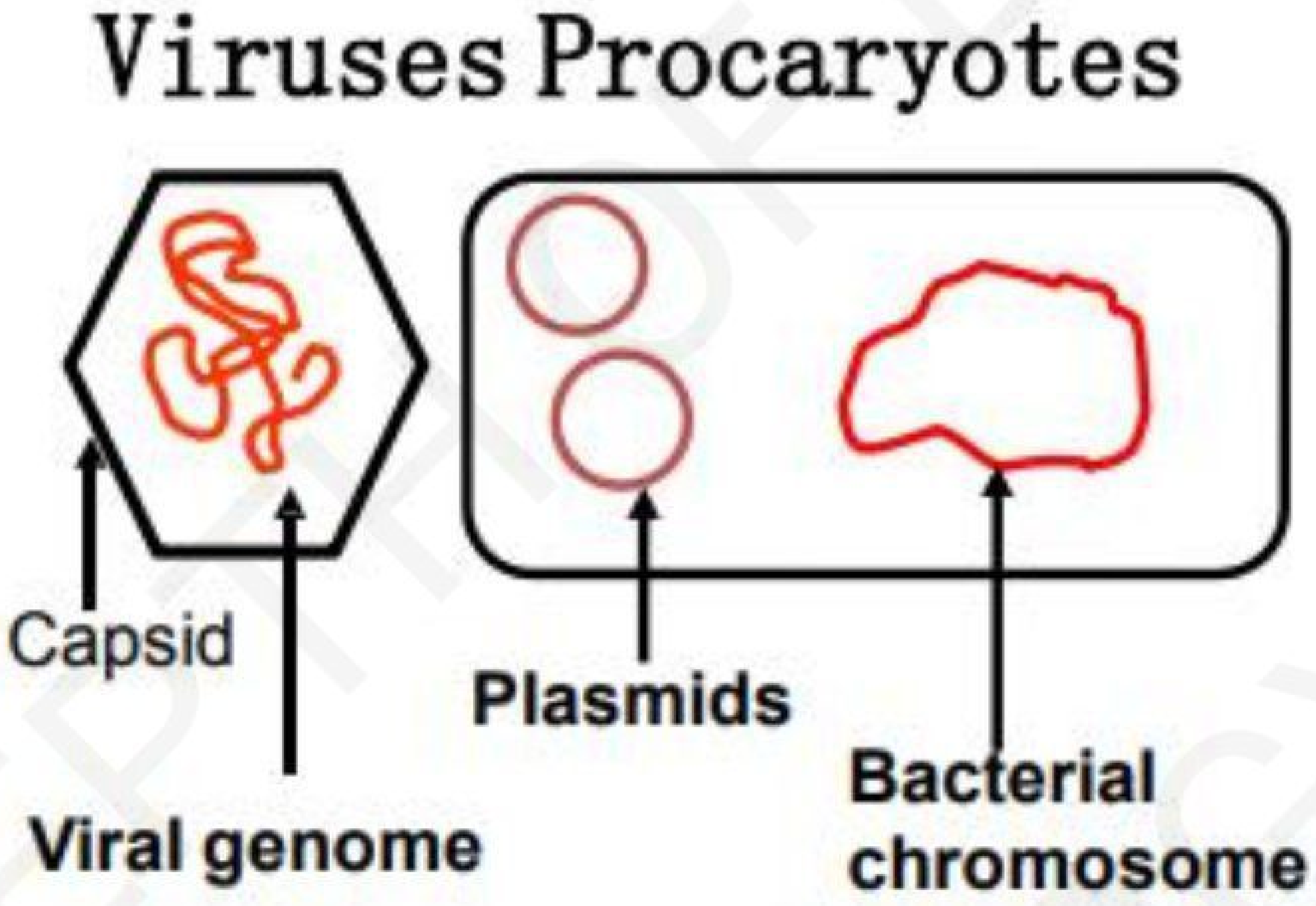
- The word “genome,” coined by German botanist Hans Winkler in 1920, was derived simply by combining ***gene*** and the final syllable of ***chromosome***.
- If not specified, “genome” usually refers to the nuclear genome!

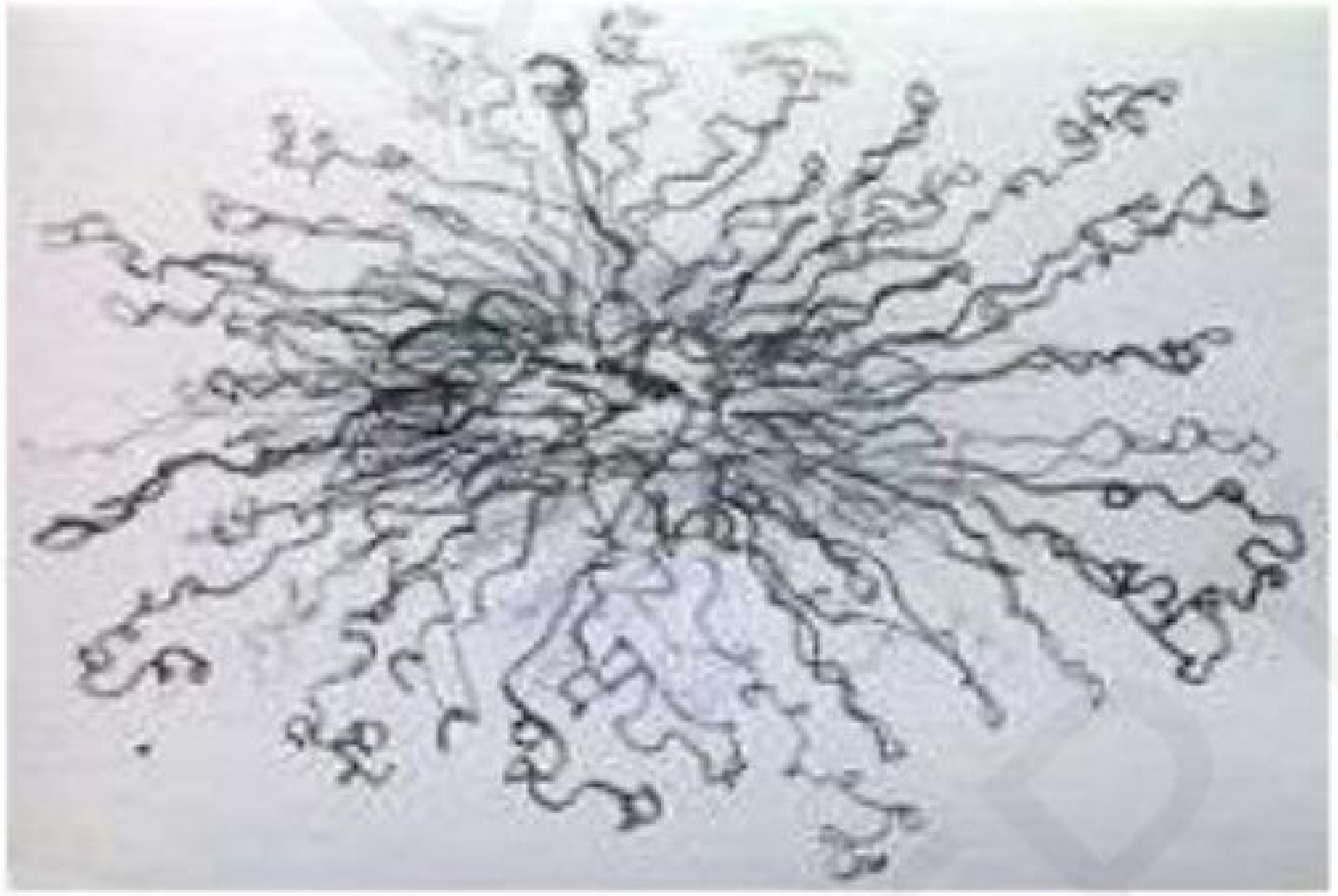
- An organism’s **genome** is defined as the complete haploid genetic complement of a typical cell.
- The genetic content of the organelles in the cell is not considered part of the nuclear genome.
- In diploid organisms, sequence variations exist between the two copies of each chromosome present in a cell.
- The genome is the ultimate source of information about an organism.

IN PROKARYOTES

- Do not contain nucleus and membrane bound organelles
- Simple genome organization
- Genes usually do not have introns and histone protie
- circular double-stranded DNA molecule but may be a linear molecule in some major groups
- Supercoiling

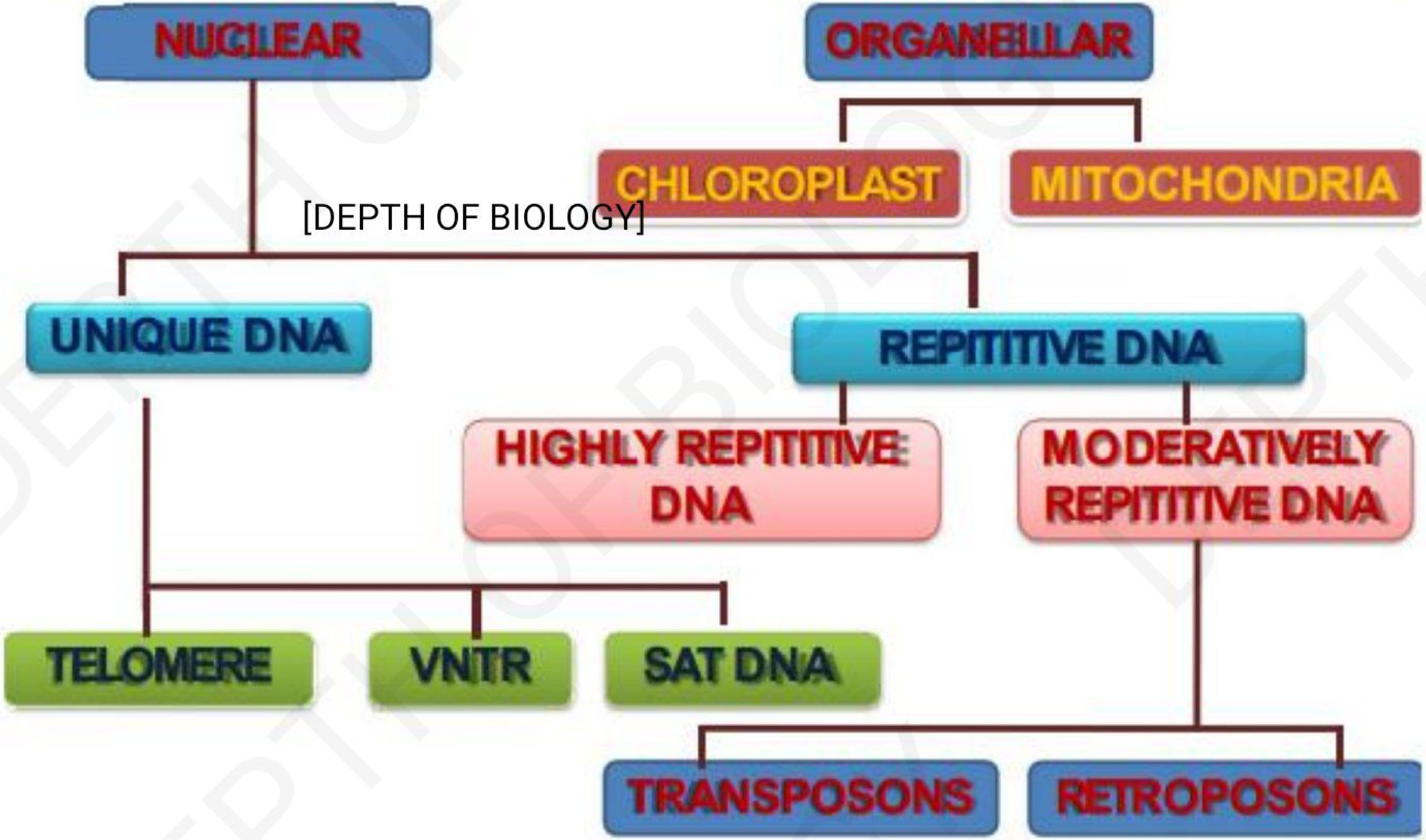






DEPTH OF BIOLOGY

GENOME IN EUKARYOTES



- NUCLEAR
  - UNIQUE DNA
    - TELOMERE
    - VNTR
    - SAT DNA
  - REPITITIVE DNA
    - HIGHLY REPITITIVE DNA
    - MODERATIVELY REPITITIVE DNA
      - TRANSPOSONS
      - RETROPOSONS
- ORGANELLAR
  - CHLOROPLAST
  - MITOCHONDRIA

HUMAN GENOME PROJECT

- Study of genome is called as ***genomics.*** [DEPTH OF BIOLOGY]
- HGP [1990-2003] was the 1st megaproject for sequencing of nucleotides and mapping of all the genes in human genome [DEPTH OF BIOLOGY]
- HGP was coordinated by **U.S. DEPARTMENT OF ENERGY & THE NATIONAL INSTITUTE OF HEALTH.**

GOALS OF HGP-

a) Identified all the estimated genes in human DNA.

b) Sequencing of 3 billion chemical base pairs of human DNA

c) Store this information in databases

d) Improve tools for data analysis

e) Transfer related technologies to other sector

f) Address the ethical, legal & social issues [ELSI] that may arise from the project

*In HGP, many non-human model organisms, such as bacteria ***(Haemophilus influenzae),*** yeast ***, Caenorhabditis elegans*** (nematode), Drosophila, plants [rice & arabidopsis] have also been sequenced

METHODOLOGIES

2 MAJOR APPROACHES

1. ***EXPRESSED SEQUENCE TAGS [EST`s]-*** focused on identifying all genes that are expressed as RNA.
2. ***SEQUENCE ANNOTATION-*** Sequencing whole Set of genomecontaining all the coding & non-coding sequence and later| assigning different regions in the sequence with functions

PROCEDURE

- Isolate total DNA from a cell —> Convert into random fragments —> Clone in suitable host (bacteria & yeast) using vectors (eg: BAC & YAC) for amplification —> Fragments are sequenced using Automated DNA sequencers (Frederick sanger method) —> Sequences are arranged based on overlapping regions —> Alignment of Sequences using computer programs.

- BAC= Bacterial Artificial Chromosomes
- YAC= Yeast Artificial Chromosomes

- DNA is converted to random fragments as there are technical limitations in sequencing very long pieces of DNA.
- HGP was closely associated with ***Bioinformatics***
- ***Bioinformatics:*** *Application of computer science and information technology to the field of biology & medicine.*
- Of the 24 chromosomes (22 autosomes and X & Y), the last sequenced one is chromosome 1 (May 2006).

SALIENT FEATURES

- Human genome contains 3164.7 million nucleotide bases.
- Total number of genes= about 30,000.

- Average gene consists of 3000 bases, but sizes vary.Largest known human gene (dystrophin on X-chromosome) contains 2.4 million bases.
- 99.9% nucleotide bases are same in all people. Only 0.1%($3x10^{\circ}$ bp) difference makes every individual unique.
- Functions of over 50% of discovered genes are unknown.
- Chromosome has most genes (2968) and Y has the fewest (231).Less than 2% of the genome codes for proteins

- Very large portion of human genome is made of Repeated(repetitive) sequences. These are stretches of DNA sequences that are repeated many times. They have no direct coding functions. They shed light on chromosomes structure, dynamics and evolution.

- About 1.4 million locations have single-base DNA differences. They are called SNPs (Single nucleotide polymorphism or ‘snips’). This helps to find chromosomal locations for disease-associated sequences and tracing human history