BHARATHIDASAN UNIVERSITY

Tiruchirappalli- 620024, Tamil Nadu, India

Programme: M.Sc., Biomedical Science
(5 Year Integrated Program)

Course Title : Genomics

Course Code : BM47C12

Unit-I
Introduction to Genome

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INTRODUCTION TO GENOME

What is Genome?

Full set of genes in each cell of an organism

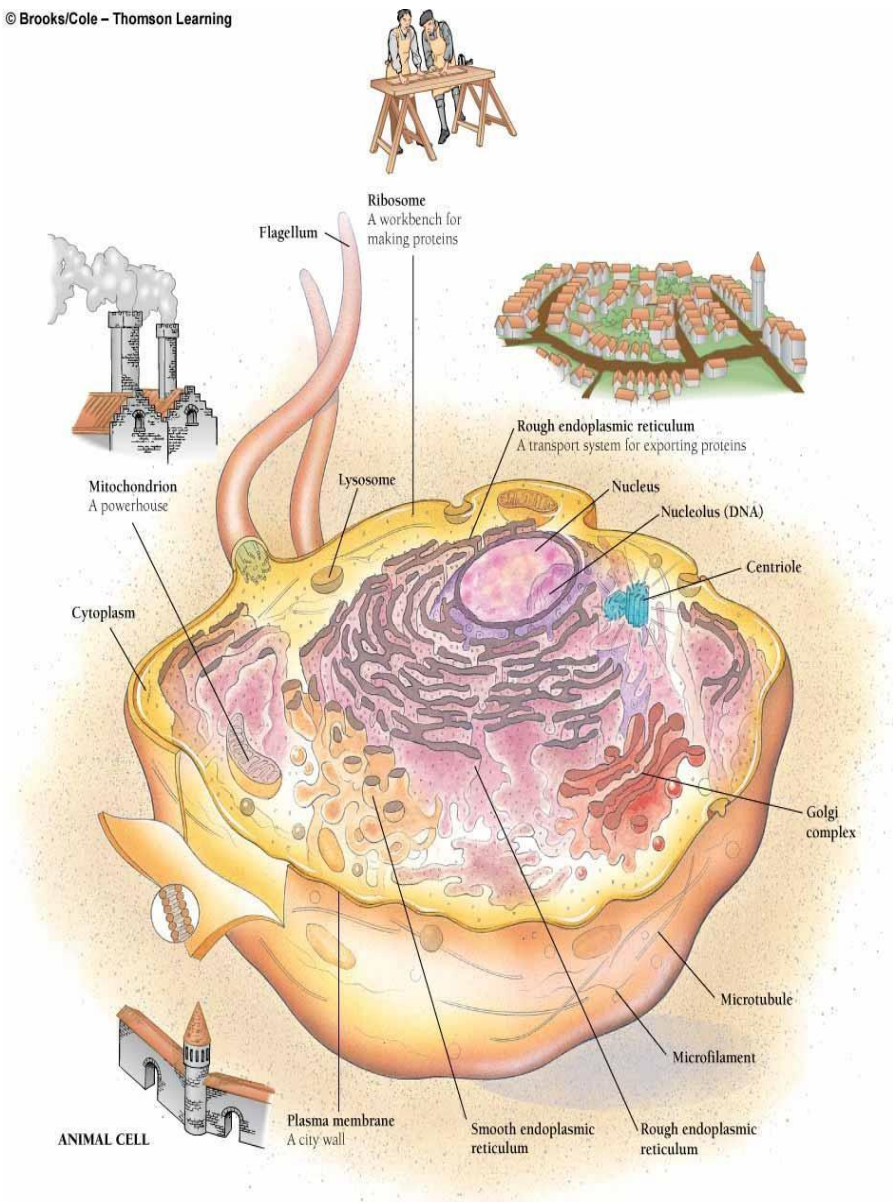
Who We Are on the inside!

AATTCATGAAAATCGTATACTGGTCTGGTACCGGCAACACTGAGAAAATGGCAGAGCTCATCGCTA
AAGGTATCATCGAATCTGGTAAAGACGTCAACACCATCAACGTGTCTGACGTTAACATCGATGAACT
GCTGAACCGAAGAATCTTCAACCTGGCTTGCTGCCATGGCAATGAAGTACTGAAGGAAGCG
AATTTGAACCGTTCATCGAAGAGATCTCTACCAAAATCTCTGGTAAGAAGGTTGCGCTGTTCGGTTC
TTACGGTTGGGGCGACGGTAAGTGGATGCGTGACTTCGAAGAACGTATGAACGGCTACGGTTGCG
TTGTTGTTGAGACCCCGCTGATCGTTCAGAACGAGCCGGACGAAGCTGAGCAGGACTGCATCGAA
TTTGGTAAGAAGATCGCGAACATCTAGTAGACGTGTCTGACGTTAAGACGTTAAGACGTTA

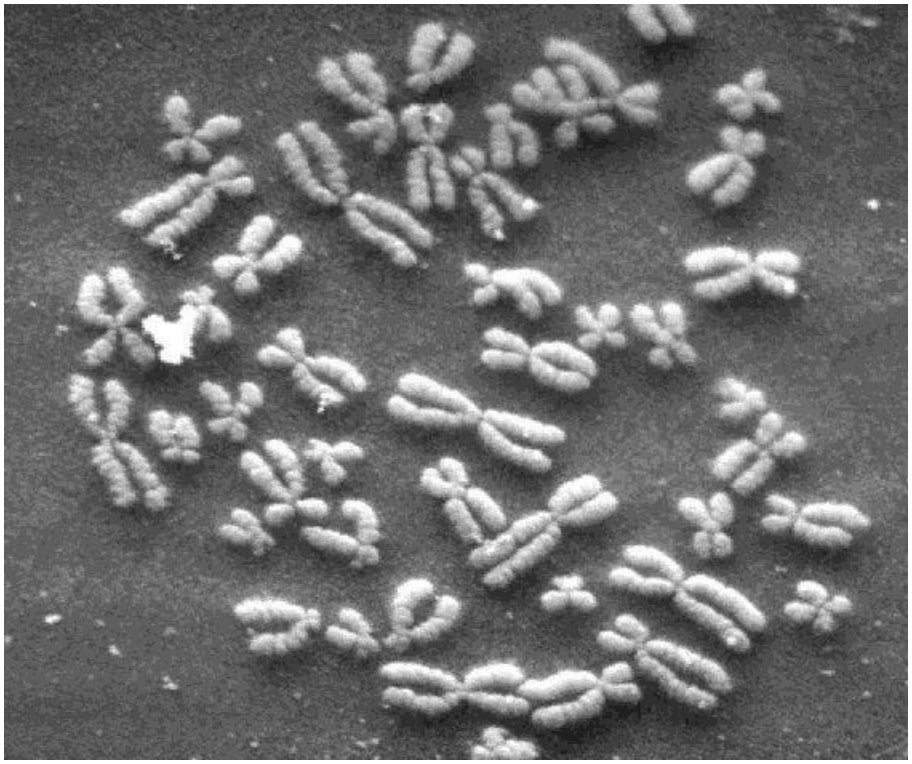
The Genome is our Genetic Blueprint

Genes

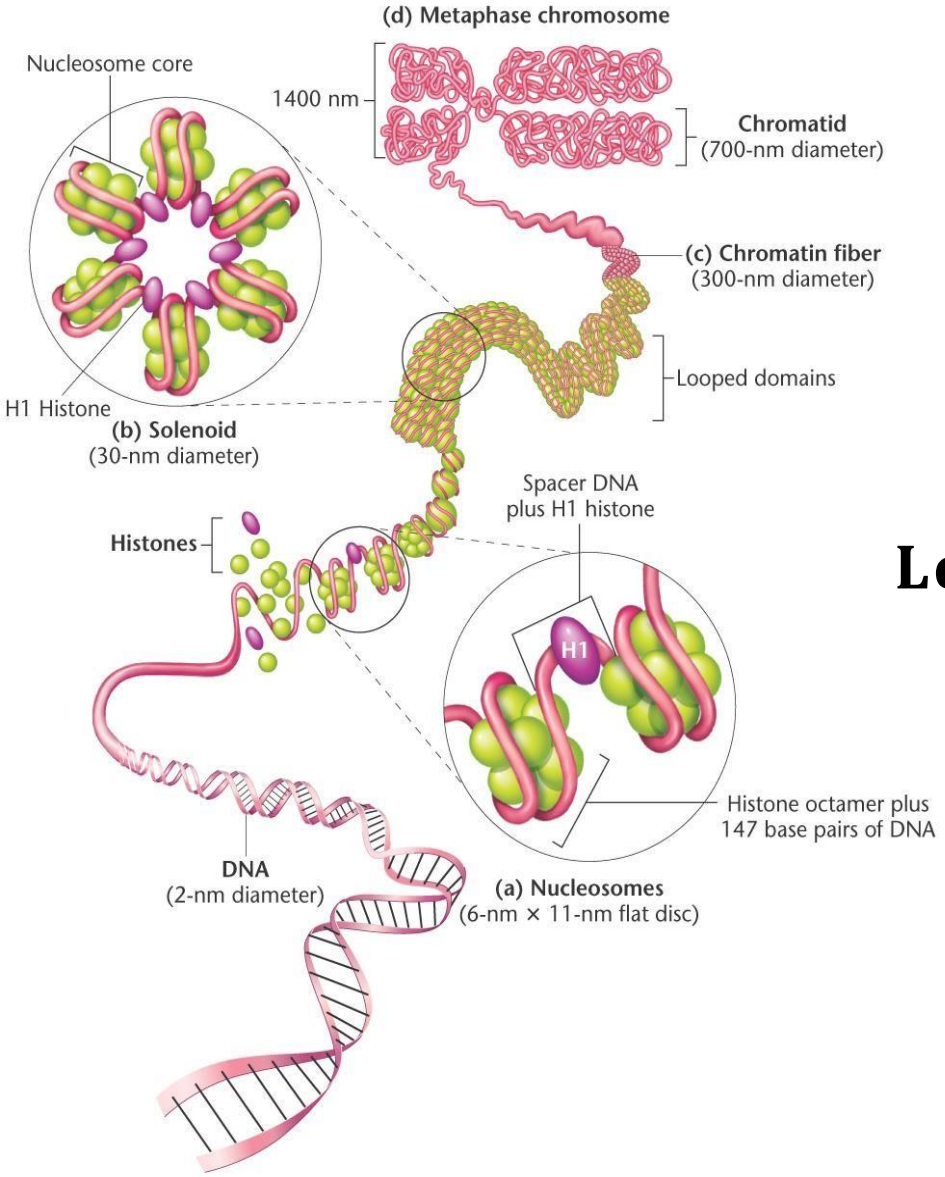
- Basic structural and functional unit of genome
- Located on chromosomes
- Genetic information is carried in DNA and RNA
- Comes in multiple forms called alleles
- Control cells by directing protein synthesis.
- Human cells posses 46 chromosomes



Chromosomes



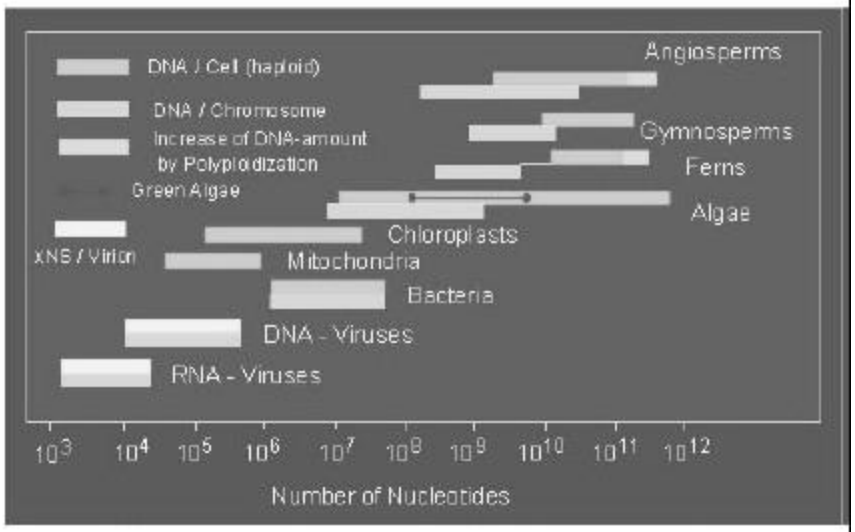
How Is it organized ?



Levels of DNA Condensation

Genome size

- Units: bp, pg/1C
- Range:
  - Small: < 50 Mbp
  - Large: 120,000 Mbp
  - Some sizes:
    - *Cardamine amara*: 54 Mbp
    - *Arabidopsis thaliana*: 125 Mbp
    - *Prunus persica*: 270 Mbp
    - *Medicago truncatula*: 466 Mbp
    - *Oryza sativa*: 490 Mbp
    - *Lycopersicon esculentum*: 1,005 Mbp
    - *Zea mays*: 2,671 Mbp
    - *Triticum aestivum*: 16,979 Mbp



Variation in genome size

- Plants:
  - Asterids: 0.3 – 24.8 pg
  - Rosids: 0.1 -- 16.5 pg
  - Ranunculales: 0.5 – 25.1 pg
  - Monocots: 0.3 – 127.4 pg
- Animals
  - Fruit fly 0.18 pg
  - Sea urchin 0.87
  - Chicken 1.13
  - Zebrafish 1.64
  - Mouse 3.01
  - Human 3.19
  - Octopus 4.56
  - Grasshopper 13.4
  - Salamander 38.3
  - Lungfish 140

Genome Size

Amoeba dubia
670000 Mb

Amoeba proteus
290000 Mb

Allium cepa
17000 Mb

Bufo bufo
6900 Mb

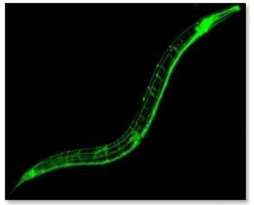
Homo sapiens
3000 Mb

Rhinolophus
1929 Mb

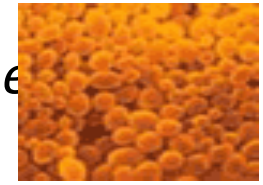
Arabidopsis
125 Mb

Drosophila melanogaste
120 Mb

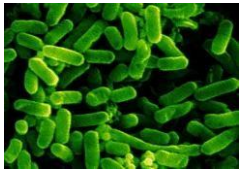
Caenorhabditis elegans
97 Mb



Saccharomyces cerevisia e
12 Mb



E. Coli 4.1 Mb

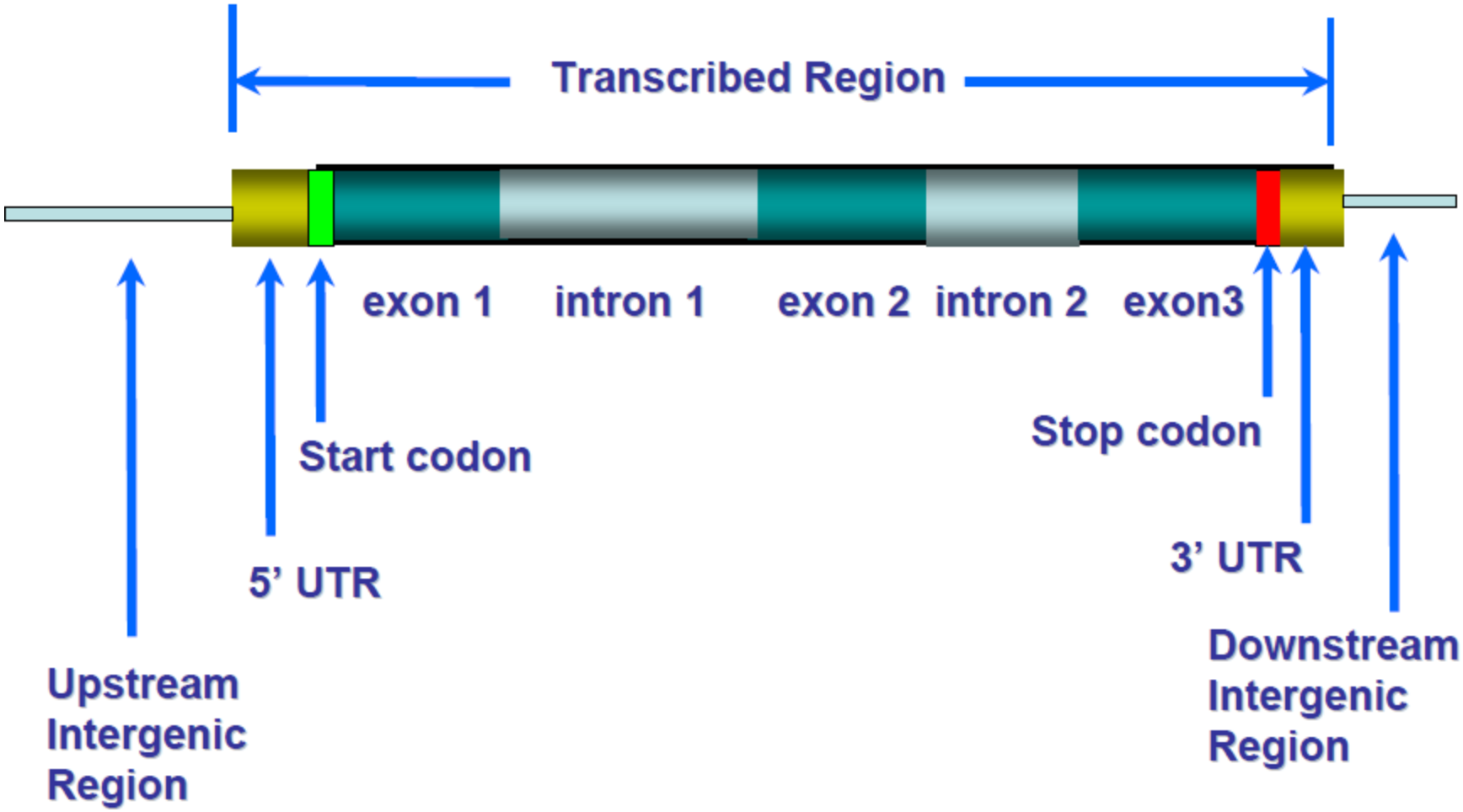


Human
immunodeficiency
virus type 1
0.19Mb

Eukaryotes

- Complex gene structure
- Large genomes (0.1 to 10 billion bp)
- Exons and Introns (interrupted)
- Low coding density (<30%)
  - 3% in humans, 25% in Fugu, 60% in yeast
- Alternate splicing (40-60% of all genes)
- High abundance of repeat sequence (50% in humans) and pseudo genes
- Nested genes: overlapping on same or opposite strand or inside an intron

Eukaryotic Gene Structure



Repetitive DNA

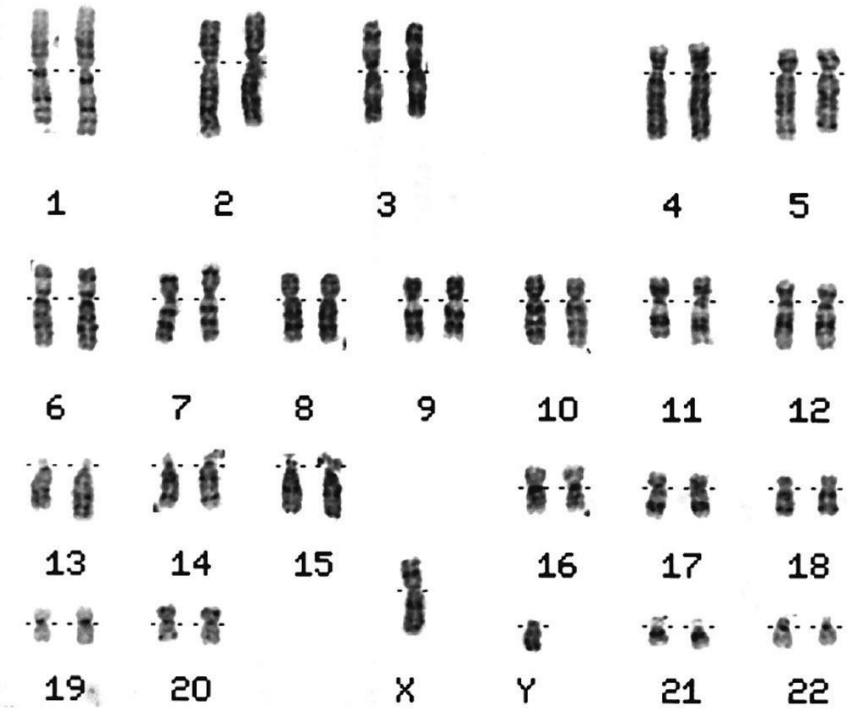
- **Moderately Repetitive DNA**
  - **Tandem gene families (250 copies of rRNA, 500-1000 tRNA gene copies)**
  - **Pseudogenes (dead genes)**
  - **Short interspersed elements (SINEs)**
    - **200-300 bp long, 100,000+ copies, scattered**
    - **Alu repeats are good examples**
  - **Long interspersed elements (LINEs)**
    - **1000-5000 bp long**
    - **10 - 10,000 copies per genome**

Repetitive DNA

- **Highly Repetitive DNA**
  - **Minisatellite DNA**
    - repeats of 14-500 bp stretching for ~2 kb
    - many different types scattered thru genome
  - **Microsatellite DNA**
    - repeats of 5-13 bp stretching for 100's of kb
    - mostly found around centromere
  - **Telomeres**
    - highly conserved 6 bp repeat (TTAGGG)
    - 250-1000 repeats at end of each chromosome

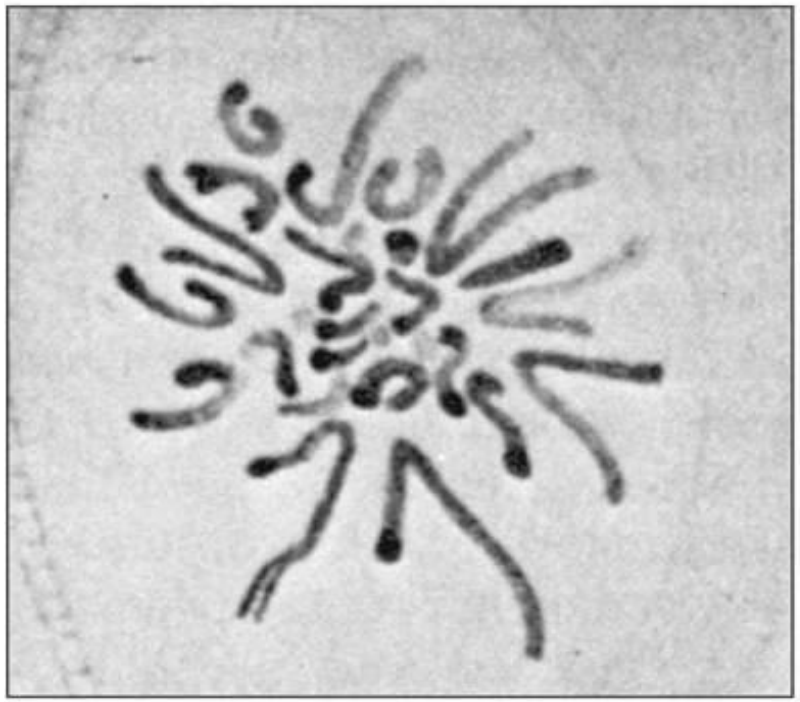
Cytogenetics

- Studies on chromosome number & structure



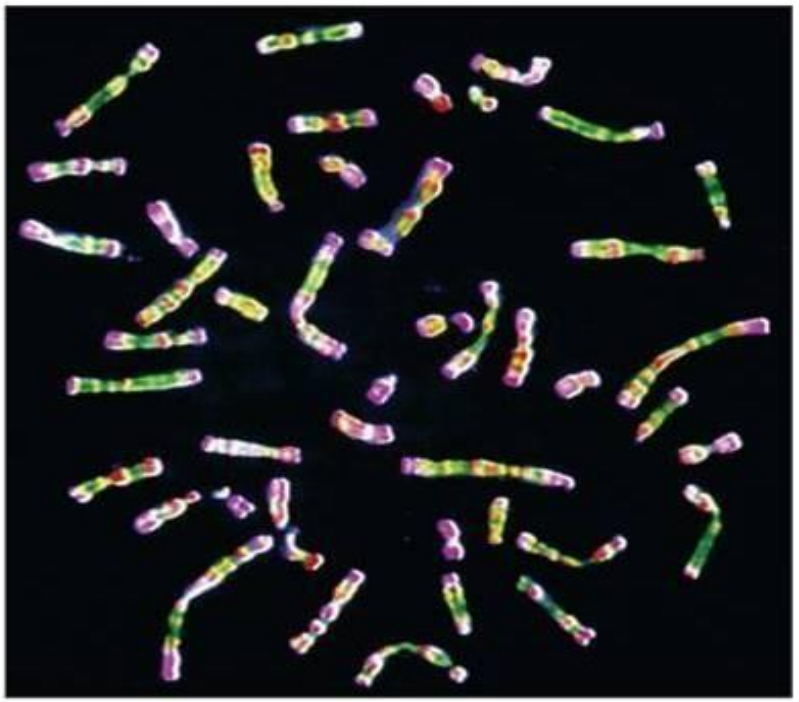
Cytogenetics: Then & Now

1882



Drawing by German biologist Walther Flemming

Now



Micrograph of actual stained human chromosomes

THANK YOU