

# **BIOINFORMATICS**

# **HUMAN GENOME PROJECT**

**Mrs. AROCKIAM THADDEUS**

**Associate Professor**

**PG & Research Centre of Zoology**

**III UG Zoology (R)**

# Bioinformatics

Human Genome Project

# About the project

- ❖ Multidisciplinary research project to determine the genomic structure of man
- ❖ Aiming... sequencing all DNAs of man, location of various DNAs in genes
- ❖ Under HUGO
- ❖ Funded by DoE, NIH (USA)
- ❖ 6 Laboratories involved (both silica and wet lab.)

# Methodology

- Human somatic cell – 23 pairs of chromosomes
- Long double stranded DNA and histone protein

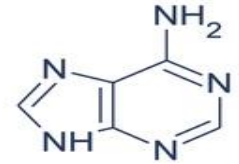
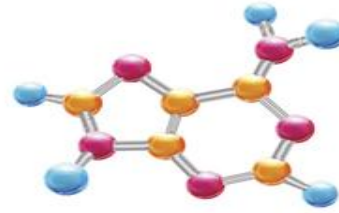
## **How to obtain DNA**

- ❖ **Human cell line – homogenized using sucrose solution**
- ❖ **Homogenate – differential centrifugation**
- ❖ **Chromosomal fraction – add Lysis buffer – ultracentrifugation**
- ❖ **Separation of chromosomes – separated and numbered**
- ❖ **Suspended in solution of Tris-HCL, EDTA, NaCl**
- ❖ **Release of DNA from histones**
- ❖ **Isolation of DNA by Cesium chloride density gradient centrifugation**
- ❖ **Stored in refrigerator at 20 °C**

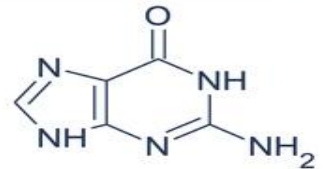
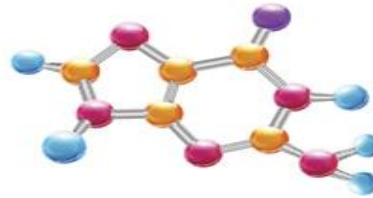
# DNA



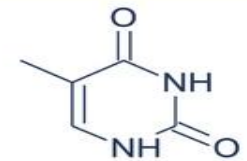
## NITROGENOUS BASES



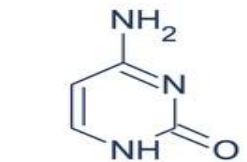
Adenine



Guanine



Thymine



Cytosine

# Preparation of DNA for study

- Long DNA –cut into pieces – 5000- 10000 nucleotides – restriction enzymes
- Separation of pieces – agarose gel electrophoresis – on the basis of restriction fragment length polymorphism (RFLP)
- Stored in vials for further use at 20 ° C
- One DNA fragment –inserted to plasmid DNA –**rDNA**
- **Introduce to E.Coli – in vivo amplification of DNA fragment – millions of amplification of DNA**

# Sequencing & Analysis

- Amplified DNA- target DNA - from bacterial cell – separated
- Add Fluorescent dye visible under laser light – to terminal end of nucleotides
- Resulting DNA solution in 96 tubes in the **DNA sequencing machine**
- Sequence can be observed in **fluorescence recorder**

## Result / Applications

- Biotechnology based companies – use information- manufacture human proteins- use in disease treatments.
- To detect genetic disorders in man
- 289 genetic diseases known.
- Chromosome – 1            -Alzheimer's disease
- Ch -----2            - loss of memory
- Ch----- 6            -Epilepsy
- Ch-----7            -Obesity
- Ch -----10            -Gyrate atrophy (progressive loss of vision)
- Ch-----17            - Breast cancer
- Ch -----20            -Severe combined immunodeficiency
- Ch-----Y            -Testis differentiation factor



# Result .....

- Matching human genome with Drosophila
  - This has remedial genes for 177 genetic diseases in man.
  - Information about genome – utilized to design babies with many superior characters - **skill, strength and free of genetic disorders.**
- 
- **Thanks to the scientists of Human Genome Project**