Genetics & Genetic Engineering

(Autumn semester 2023)

- **Evaluation Process in this Semester Autumn 2023**
- **Plan of lectures/ Course structure (MKM's part)**
- **Text /Reference Books to study Genetics**

How we arrived in this planet?

Half of our genetic material is from mother and half from father

What about other organisms- single cellular & multicellular ?

- Propagation/ Reproduction (transmission of traits)
- Survivability and Continuity of species specificity
- Variation and Adaptation
- Evolution/ emergence of new species

How the genetic material of organism is varied/ modified ?

- DNA mutation and recombination (natural processes)
- DNA manipulation in laboratory (Genetic Engineering/ recombinant DNA technology)

Subject Genetics: 1) Classical definition and 2) modern definition

▪ Plan of lectures/ contents by MKM

Sl. No.	Genetics course topics (by MKM)
1	Introduction to the subject "Genetics"
2	Definitions and concepts of some terms
3	Inheritance pattern of single trait/gene is governed by Mendel's 1st Law or Principle of Segregation, as established by monohybrid cross
4	Inheritance pattern of two (or more) traits/genes is governed by Mendel's 2nd law or Principle of Independent Assortment, as established by dihybrid (or multiple-hybrid) cross
5	Rules of probability in inheritance of traits or genetic analyses
6	Chi-square (χ^2) test: statistical analyses of the Mendelian monohybrid and dihybrid ratios
7	Chromosomal basis or theory of inheritance
8	Concept of Pedigree Analysis
9	Extension and deviations from Mendelian Inheritance pattern
10	Linkage, Crossing over & Recombination; Genetic Mapping of Chromosome, Construction of Linkage Map
11	Chromosomal Variation/Mutation- Numerical and Structural
12	Concept of Quantitative Genetics
13	Concept of Population Genetics

Assignments on selected topics:

You need to present @ 10 min on each topic group-wise

▪Text /Reference Books to study Genetics

- ***Introduction to Genetic Analysis*** by Griffiths et al. (W. H. Freeman and Company)
- ***iGenetics: A Molecular Approach*** by P. J. Russell (Pearson Education Inc./ Benjamin Cummings)
- ***Genetics: From Genes To Genome*** by Hartwell et al. (McGraw-Hill)
- ***Principles of Population Genetics*** by Daniel L. Hartl and Andrew G. Clark (Sinauer Associates)

Introduction to the subject “Genetics”

- What is the subject Genetics? Classical definition and modern definition;
- Why we want to study Genetics?
- Genetics is the core of bioscience and genes are fundamental to biological information;
- Relationship among particulate factor, gene, biological information, chromosome and cell;
- Different sub-disciplines of Genetics

❖ What is the subject Genetics? 1) Classical definition and 2) modern definition;

1) Scientific study that deals with heredity and variation in living organisms.

heredity = transmission of traits (characteristic features) from parents to offspring or from one generation to the next;

variation = the differences among individual members of a species in population

Half of your genetic material is from mother and half from father

2) Scientific study of genes or biological information.

gene = the particulate factor determining the trait of inheritance or the physical/structural and functional unit of heredity or the molecular unit of heredity or the fundamental unit of information for life, that determines the biological properties or traits that are inherited or transmitted from parents to offspring.

In molecular biology term, a segment of DNA that encodes a functional product (RNA or protein), and essentially consists of a promoter, coding sequence and transcriptional terminator.

The year 2009 is the centenary year of the term ‘gene’.

Note that **“gene” is not the fundamental unit of “life”**, rather **fundamental unit of information for “life”.**

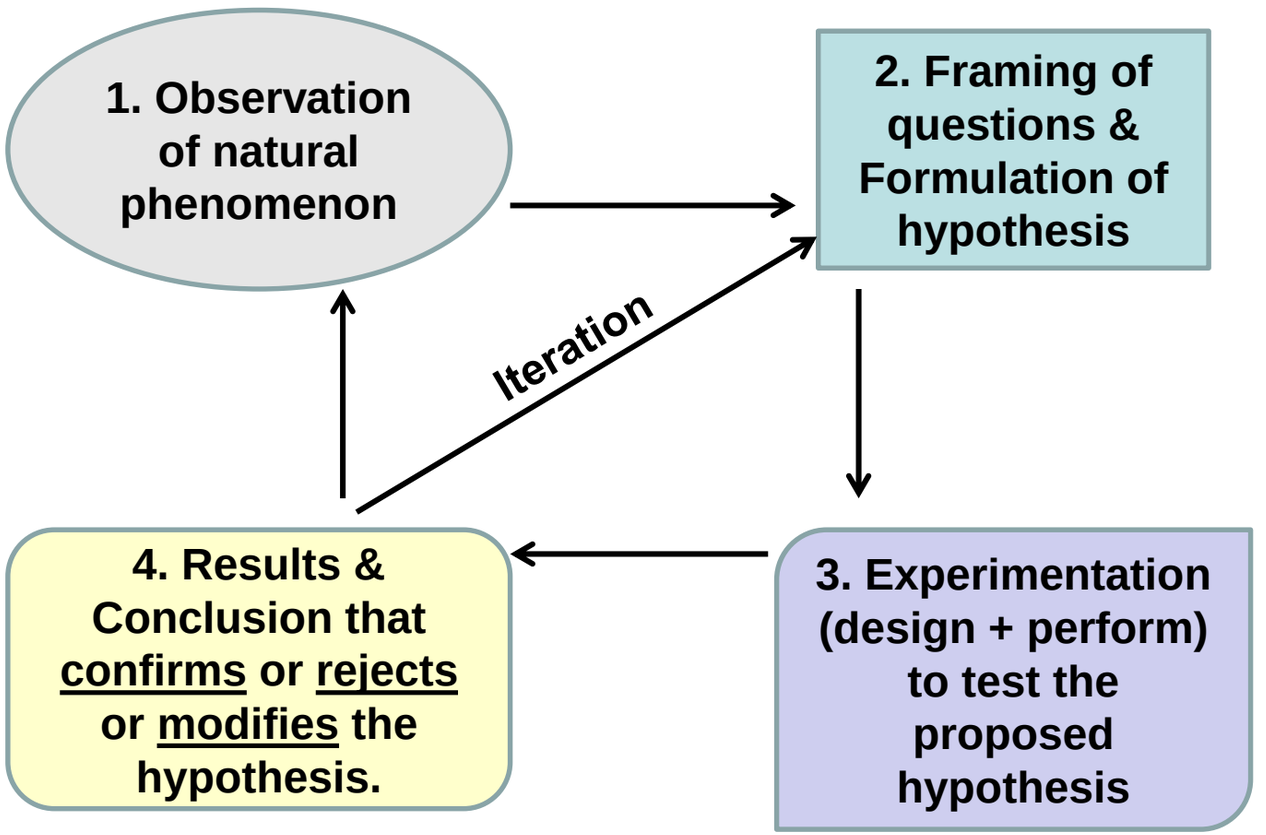
Hence, Life is beyond information; Life = information + something (?)

Other point to note-

“scientific study” What is it?

“Scientific study” always starts with a ‘well-posed problem’: a question that can be stated with enough clarity and precision that it is guaranteed an answer. It may take researchers decades or lifetimes to find clear and definite answer to a particular question.”

[*The Accidental Universe* — Alan Lightman, Physicist, served as faculties of both Harvard University and Massachusetts Institute of Technology]



I will explain this further when we study Mendelian Genetics.

Up to this slide, Lecture # 1 delivered on 08.08.2023

❖ Why we want to study Genetics?

as a common human being and as a biotechnologist?

(a) As a common human being:

we want to know ourselves- our past, present and future; our characters or behaviors; we want to know our surroundings and biodiversity- all living organisms and their activities ≡ we want to understand life holistically.

(b) As a biotechnologist:

we want to utilize these knowledge banks for commercial applications to earn money for our sustenance

Why we want to study Genetics? Because------

- **Genetics is the core of bioscience and genes are fundamental to biological information:**

(1) Genetic material or DNA encodes biological information in the form of "gene"

(2) Biological functions or traits come from gene products (RNA or protein)

(3) Complex biosystem comes from the regulatory networks of gene-gene and gene-environment interactions,

(4) All living organisms are closely related in terms of DNA structure & function

(5) Genomes (complete set of genes) are modular, allowing rapid evolution

(6) Understanding the subject Genetics and devising tools (e.g., Genetic Engineering) permit the dissection of biological complexity

Why we want to study Genetics? Because------

- **Genetics is the core of bioscience and genes are fundamental to biological information:**

Genetics is at the heart/central to the biological science;

as the storage, distributions, variations, interactions, behaviors and activities of genes or biological information control the life processes/functions/features within an individual organism and in population for survival of that species in the community or ecosystem.

The processes/functions/features of life in different organisms may varies, but essentially include cellular structures, metabolisms (conversion of matter and energy), development (formation of tissues and differentiation of organs in higher/complex life forms), growth, maturation, response to external stimulus, reproduction to form new life, and death. These features or traits of life are determined and controlled by genes or unit of biological information.

The genes have the capacity to **store**, **express**, **replicate**, **diversify** and **control** the biological information within a living organism. Genes are capable of **transmitting the information** to next generation, **bringing variations** among individual members in a population of same species and also **causing evolution** for new organisms from existing ones.

❖ Relationship among Mendel's particulate factor, gene, biological information, chromosome and cell

Mendel's particulate factor: discrete unit of hereditary material, determining the trait of inheritance. The Mendel's particulate factor determining the trait of inheritance was named as "gene" by Wilhelm Johannsen in 1909, and the chemical nature of the hereditary material was only established as "DNA" by Avery, MacLeod and McCarty in 1944.

Gene: exactly identical to Mendel's particulate factor, a segment of DNA that encodes a functional product (RNA or protein) contributing to trait, and it essentially consists of a promoter, coding sequence and transcriptional terminator.

❖ Relationship among Mendel's particulate factor, gene, biological information, chromosome and cell

Biological information: the code or driving force behind the life. Mendel's particulate factor or gene is the fundamental unit of information for life, and determines the biological properties or traits that are inherited or transmitted from parents to offspring

- Think about the mystery/magic of information for life: (1) why only 4 nucleotides are enough to code for life, not 3 or 5 nucleotides? (2) Can you design new life by synthetic novel nucleotides?
- Note that the flow of biological information from 1-dimension (1-D, sequence in single stranded nucleic acid- DNA/RNA) to 3-dimension (3-D structure of protein)

❖ Relationship among Mendel's particulate factor, gene, biological information, chromosome and cell

Chromosome: a specially organized thread-like structure of genetic material, which is involved in storage and transmission of the biological information or inheritance of traits from parents to offspring (progenies). Thus, chromosome carries the Mendel's particulate factors/genes (fundamental units of biological information that determine the traits). *[Progeny (singular) > progenies (plural); but Offspring (both singular and plural)]*

Cell: structural and functional unit of life, fundamental unit of living system.

Each cell contains chromosomes, and chromosomes contain genes

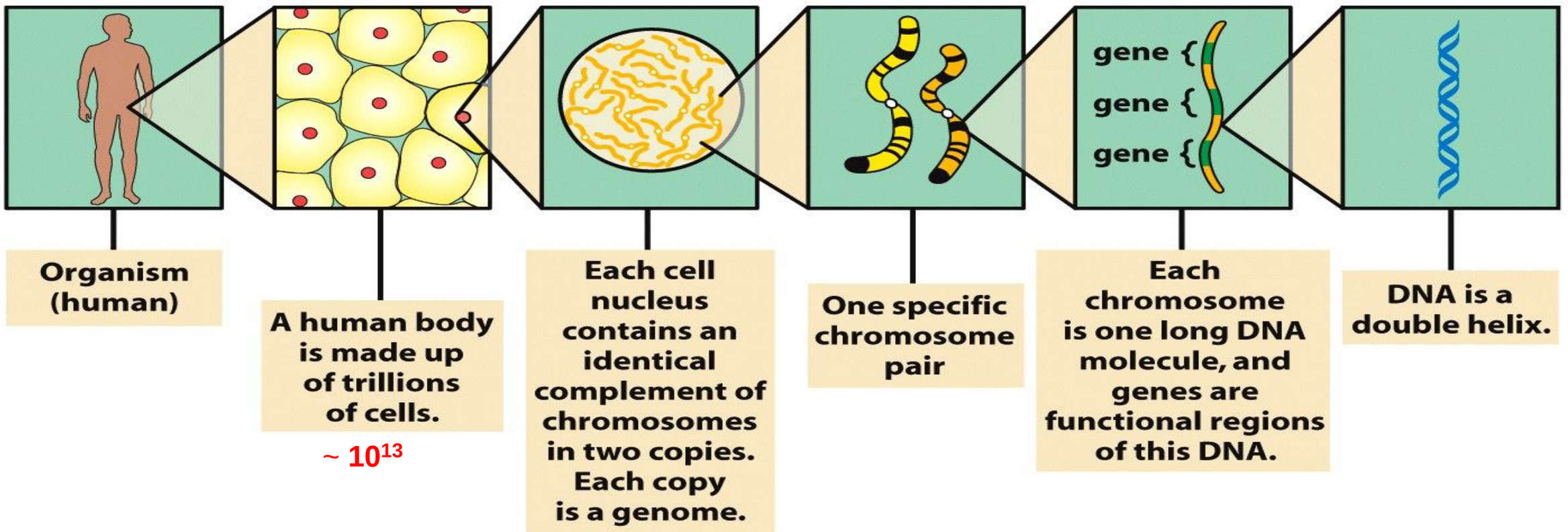


Figure 1-2
Introduction to Genetic Analysis, Ninth Edition

Sub-disciplines of Genetics

- Over the years, starting from the work of Gregor Johann Mendel (Mendelism) during mid-nineteenth century (1856-1863) till these days, the subject genetics have generated/expanded/evolved into several sub-disciplines.

The main subject genetics, broadly can be divided into four major sub-disciplines -

- ❖ Classical or transmission genetics
- ❖ Modern or molecular genetics (Genetic Engineering)
- ❖ Evolutionary genetics and
- ❖ Molecular breeding.

These, in turn, may be divided into several sub-subjects. Most importantly, there are no sharp boundaries between the sub-subjects and sub-disciplines.

Classical or Transmission Genetics

- Deals with the abstract (not concrete) and more intuitive nature (from feeling rather than fact = not able to touch) of genes (the particulate factors determining the traits are now know as genes) in heredity and variation among individuals in population (i.e., the role of particulate factors in inheritance of traits). [*Compare imagination vs. real*].

- Provides us answers on-
  - how traits are transmitted from parents to progenies generation after generation?
  - how traits are distributed in a population of individuals?
  - what determine or control the traits of inheritance ? (particulate factor or gene)
  - where genes are located ? (chromosomes in the nucleus)
  - how genes are arranged on chromosomes ? (linearly)
  - how genes recombine (exchange between chromosomes) to bring variations of traits in a population?
  - how chromosomes get separated during gamete formation?
  - whether a single gene determines or many genes determine the particular trait under consideration ?

Classical or Transmission Genetics

Contd…

The classical genetics sub-divided into:

- Mendelian Genetics
- Cytogenetics
- Population Genetics and
- Quantitative Genetics

Classical or Transmission Genetics

Contd…

Mendelian genetics:

- Study of the inheritance of traits or transmission of characters from parents to progeny **following two basic principles** known as Mendel's Law of Segregation and the Law of Independent Assortment.
- Further, it deals with the deviation from these two laws in studies on gene-gene interaction and association (linkage), and gene-environment interaction.

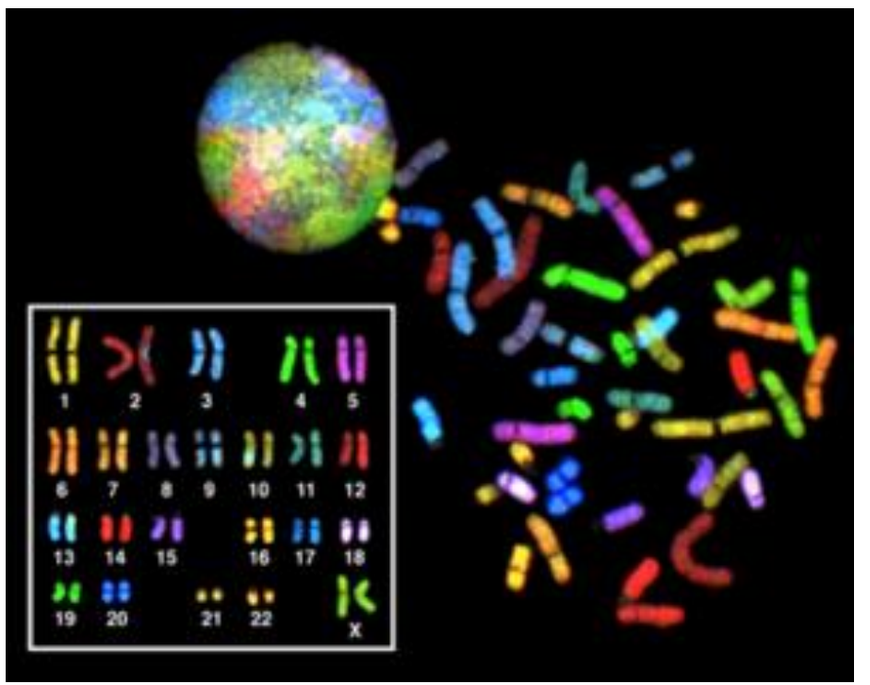
Classical or Transmission Genetics

Contd…

Cytogenetics:

- Cellular level study of genetics, mainly microscopic observation and analysis of chromosomes of the parental lines and progeny.

- Many improvements have been made during the years for staining, labeling, tagging, hybridization of DNA molecules of the chromosomes in generals and specific region, followed by simple to sophisticated detection techniques including imaging.



This is a spectral karyotype of a human female.
Credit: National Human Genome Research Institute, USA.

https://en.wikiversity.org/wiki/Cytogenetics

Classical or Transmission Genetics

Contd…

Population genetics:

- Study of heredity in groups of individuals of a single species for a particular trait (or a set of traits) that are determined by one or only a few genes.
- E.g., analyzing the frequency of disease causing gene in human population.
- Haemophilia B, a genetic mutation in the gene for blood clotting factor IX (British Queen Victoria was a carrier of this X-chromosome lined recessive trait) is rarer than Haemophilia A (defective in gene for blood clotting factor VIII (it is also X-chromosome lined recessive trait).

  [*Haemophilia = increased/easy bleeding upon bruising*].

Provides answer on-

- How the genetic structure/ variation of a population is maintained or modified generation after generation following **Hardy-Weinberg equilibrium / law**?
- How do the various biological features and forces like mating system, processes or sources of genetic variation, size of population, natural selection **influence or affect the genetic structure of the population**?

Classical or Transmission Genetics

Contd…

Quantitative genetics:

- Study of heredity for a particular trait (or a set of traits) that are determined by many genes referred as quantitative trait locus/loci (QTL/QTLs). E.g., analyzing the fruit weight and crop yield in plants and milk or meat yield in farm animals.
- **Quantitative traits vary over a range**

Two extremes of the color range

Phenotypes of color range

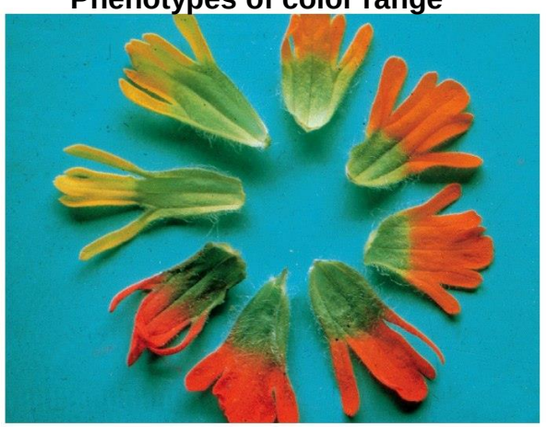
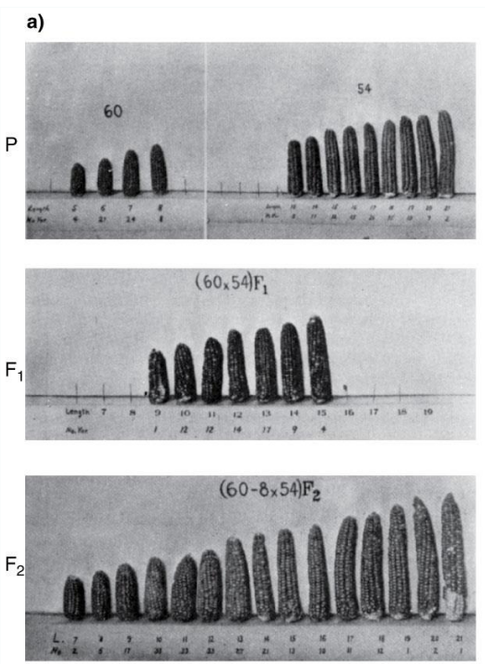


Figure 18-1
Introduction to Genetic Analysis, Ninth Edition

Bract (modified leaf) color of Indian paintbrush, *Castilleja hispida*

Inheritance of ear (cob) length (in cm) **in maize (corn)**



Modern or Molecular Genetics

- Deals with the genes at the molecular level.
- Provides answers on-
  - what is the chemical nature and structure of gene?
  - what about its expression or silencing, i.e. how, where, when and to what extent the particular gene is expressed or silenced?
  - what are the types of product (RNA or protein) formed by the gene and what is or are the function of each gene?
  - how gene change or mutate to cause variation in traits

Modern or Molecular Genetics

Contd…

- This domain of genetics also includes **recombinant DNA technology** and **genetic engineering** for improvement of quality and quantity of crop yield, farm animals and poultry birds; development of healthcare products, diagnosis of diseases in plants and animals including human, forensic applications and many other utility.
- Molecular genetics also spans **genomics** (genome analysis in terms of structure and function), i.e. knowing the sequences and positions of all the genes in the genome of an organism and their biological functions and application of this knowledge for improvement of the quality of human lives, our ecosystem and environment.

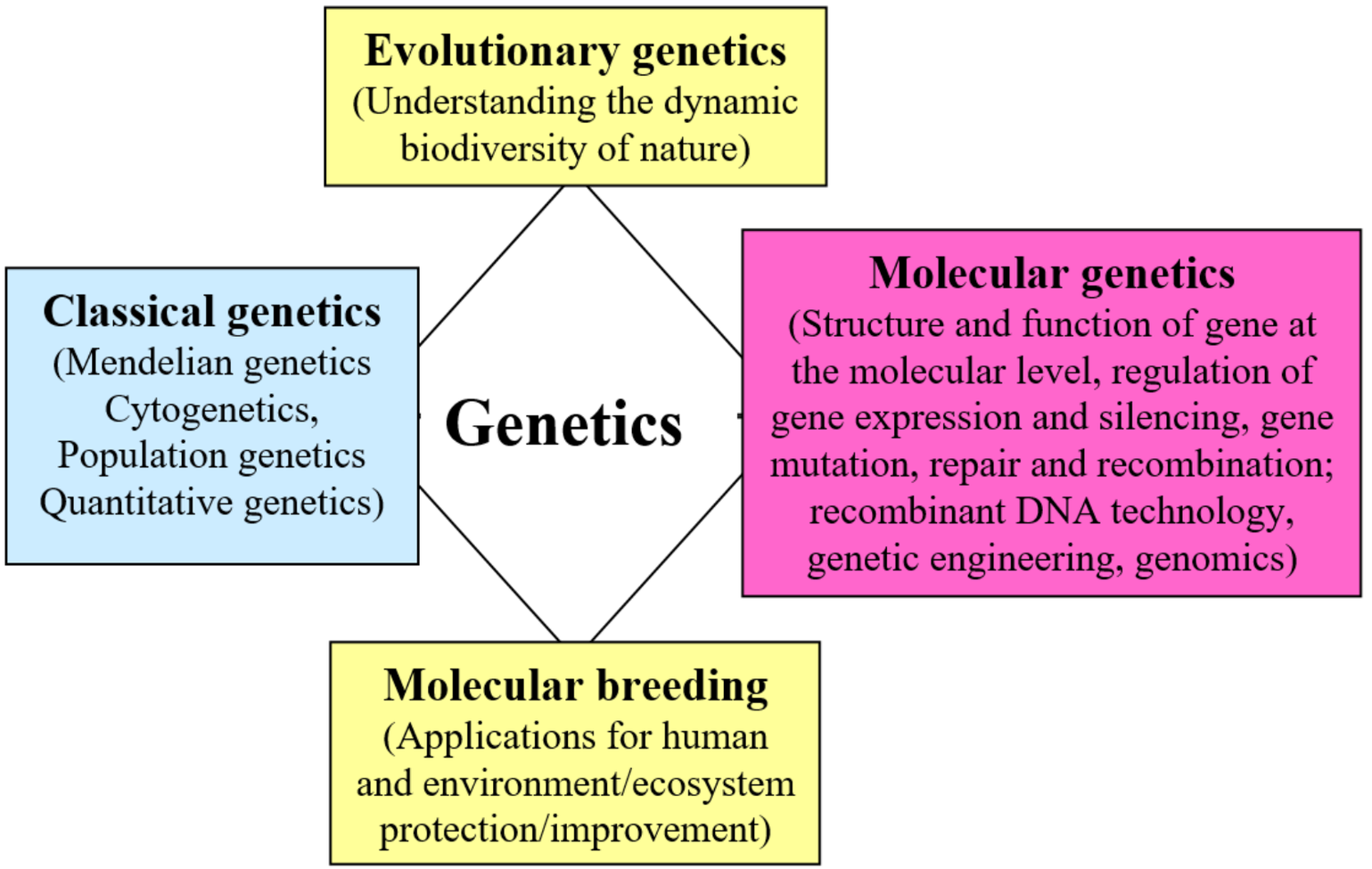
Evolutionary Genetics

- **Evolutionary Genetics** involves study of the genetic changes/ variations that take place **over the time within a group of organisms distributed in different regions** of the Earth.
- How the genetic changes/variations could arise and how they are distributed?
- What is the frequency of new variation?
- This branch of genetics helps us to understand how the evolutionary forces maintain the dynamic patterns of biodiversity observed in nature.
- How simple organisms evolve into complex organisms?
- It requires knowledge, techniques and tools of both transmission genetics and molecular genetics.
- This sub-discipline also involves in studying the DNA/gene and protein molecular evolution.

Molecular breeding

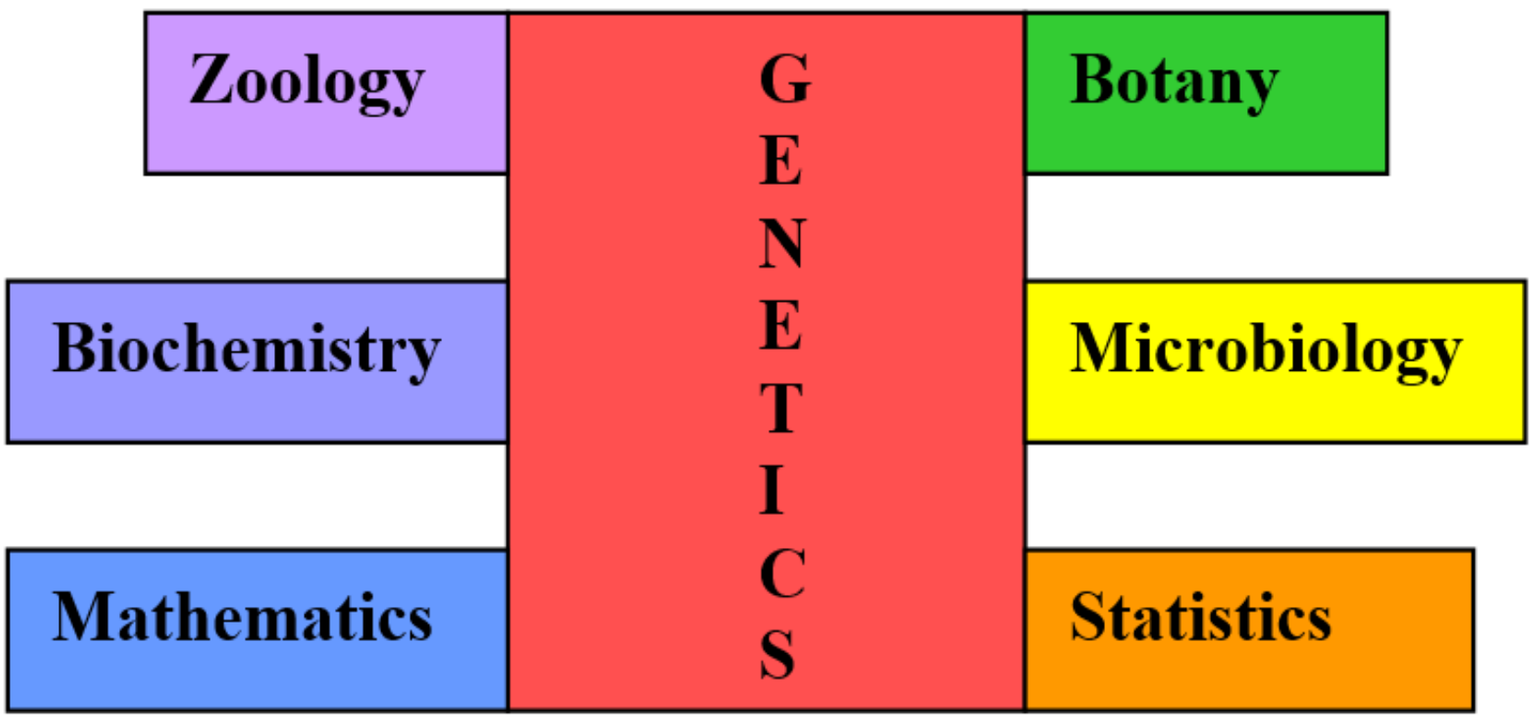
- In this process of **sexual reproduction between selected individuals** of animal or plant species, the desirable traits (many cases controlled by several genes referred as quantitative trait loci, QTLs) are introduced in the offspring and the **molecular genetic techniques are used for screening and selection of the final breed**.
- Through molecular breeding the time period involves to develop a suitable stock or cultivar is relatively shorter compared to the conventional breeding program that takes several years usually decades or so.
- This sub-discipline of genetics also requires knowledge, techniques and tools of both transmission genetics and molecular genetics.

Learning genetics needs a balanced approach that integrates knowledge of the classical genetics with the concept and tools of molecular genetics in order to understand the dynamic biodiversity of nature through evolutionary genetics and to get the practical benefit of molecular breeding for improvement of the stocks of domestic livestock or cultivars of crop plants for human requirement and improving the quality of ecosystem and environment.



Studying genetics requires knowledge on a few other disciplines:

Knowledge about other subjects, i.e. zoology, botany, microbiology, biochemistry, mathematics and statistics are important to study genetics. In this sense, genetics is an interdisciplinary subject.



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