GENETIC CHARACTRISTIC OF HUMAN POPULATION

MEDICAL ACADEMY NAMED AFTER S.I. GEORGIEVSKY OF VERNADSKY CFU

DEPARTMENT OF MEDICAL BIOLOGY

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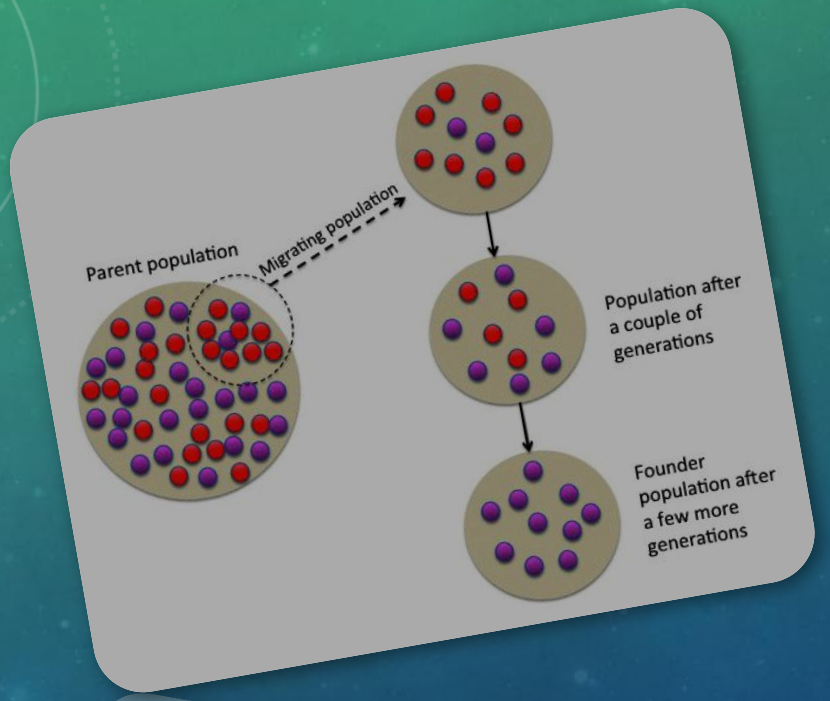
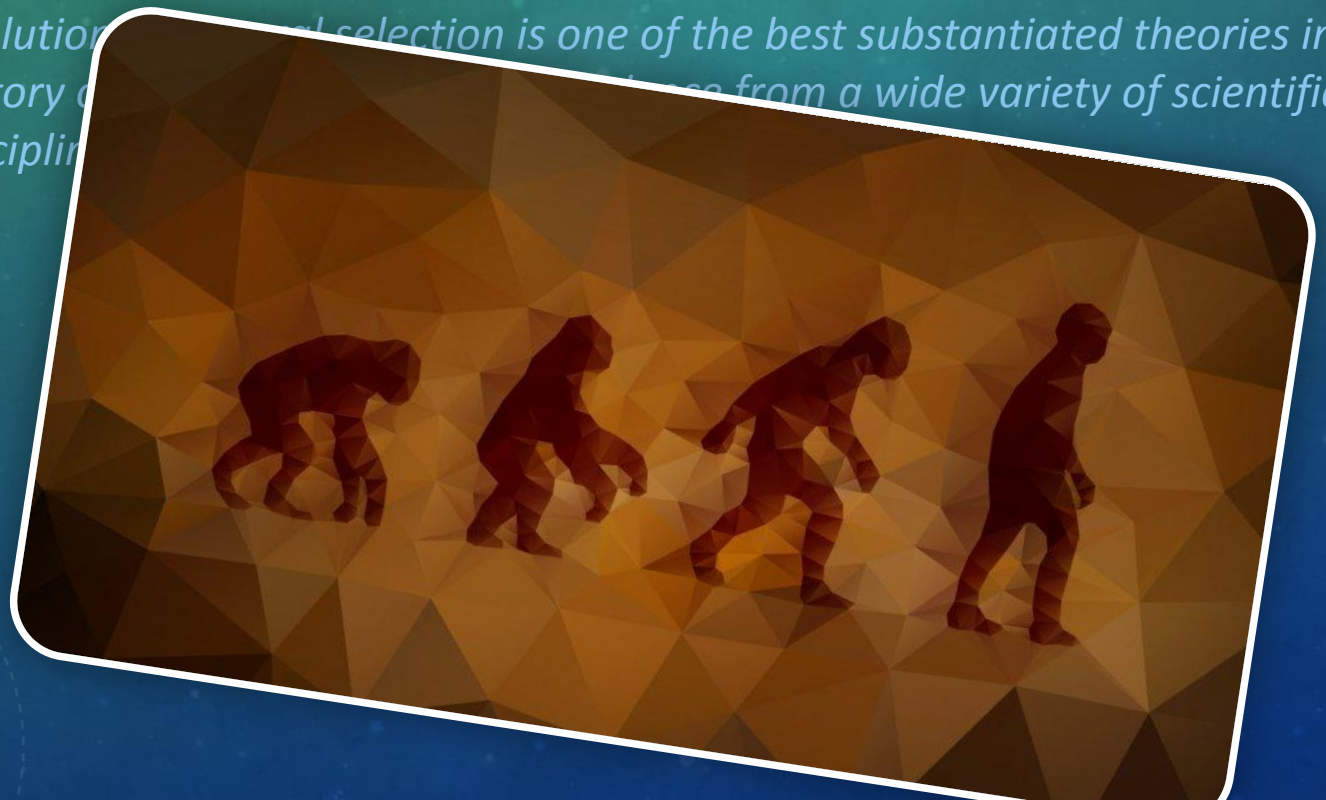
POPULATION

- POPULATION IS A GROUP OF ORGANISMS OF THE SAME SPECIES THAT LIVE IN THE SAME AREA
- HOW LARGE A POPULATION IS AND HOW FAST IT IS GROWING ARE OFTEN USED AS MEASURES OF ITS HEALTH
- A SINGLE INDIVIDUAL CANNOT EVOLVE ALONE; EVOLUTION IS THE PROCESS OF CHANGING THE GENE FREQUENCIES WITHIN A GENE POOL.
- THE PROCESS OF EVOLUTION OCCURS ONLY IN POPULATIONS AND NOT IN INDIVIDUALS

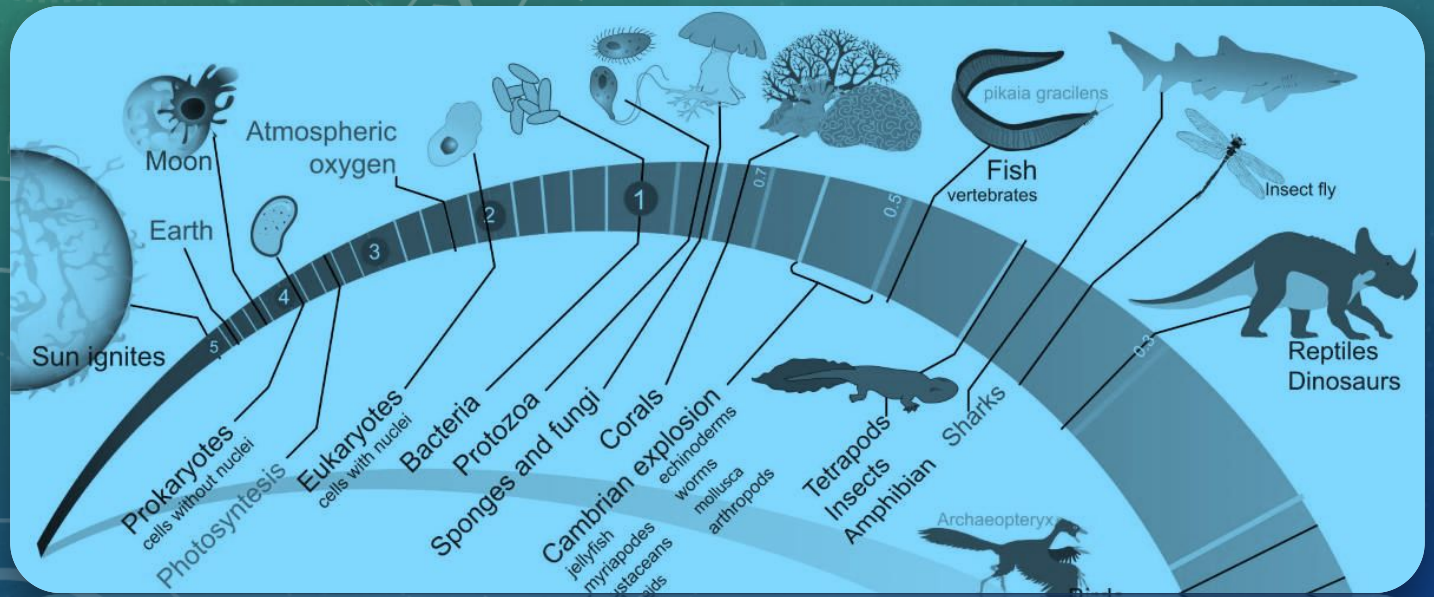


EVOLUTION

- *evolution is the change in the characteristics of a species over several generations and relies on the process of natural selection.*
- *The theory of evolution is based on the idea that all species are related and gradually change over time.*
- *Evolution[illegible] selection is one of the best substantiated theories in the history [illegible] from a wide variety of scientific disciplin[illegible]*

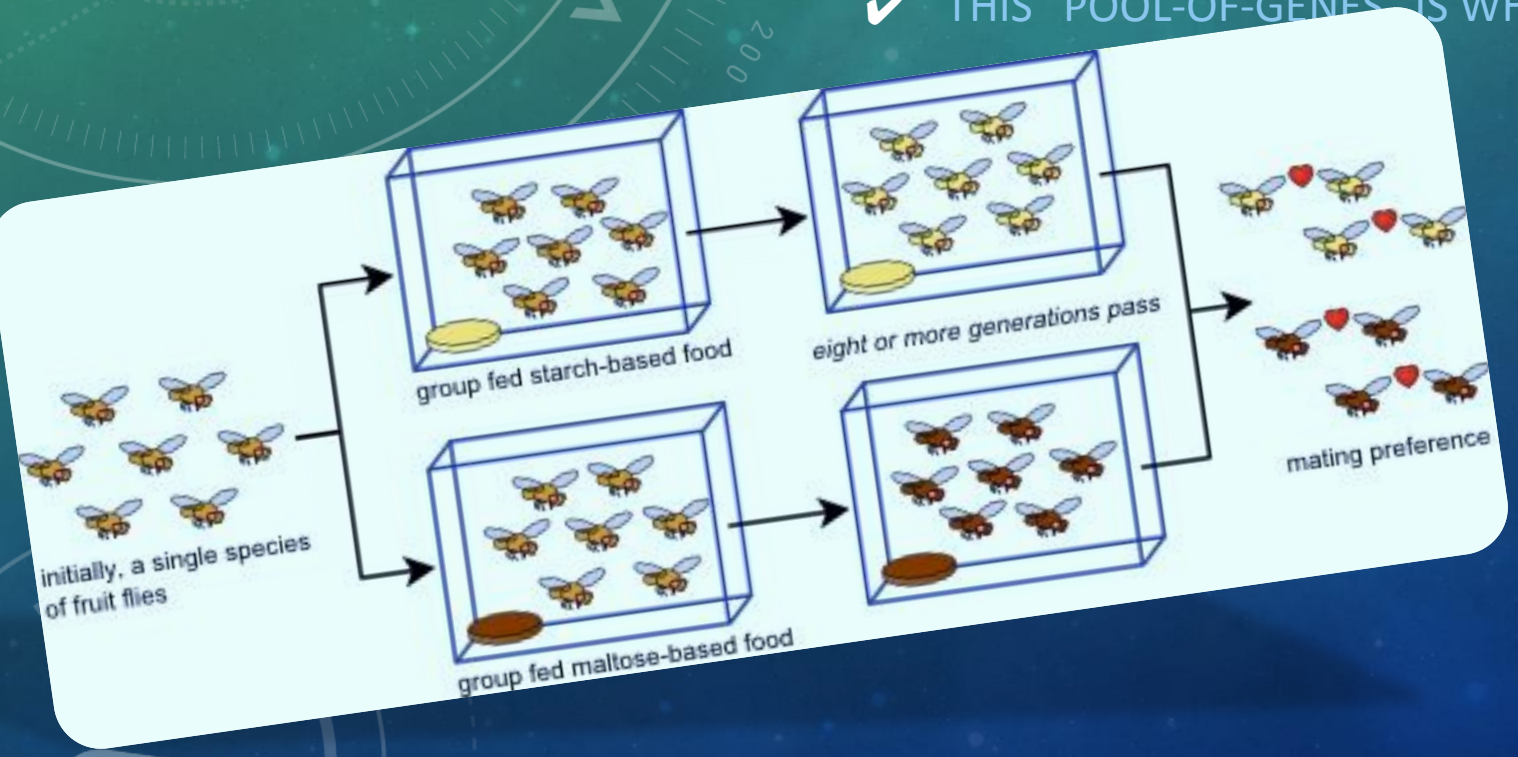


- DIFFERENT CHARACTERISTICS TEND TO EXIST WITHIN ANY GIVEN POPULATION AS A RESULT OF MUTATION, GENETIC RECOMBINATION AND OTHER SOURCES OF GENETIC VARIATION.
- EVOLUTION OCCURS WHEN EVOLUTIONARY PROCESSES SUCH AS NATURAL SELECTION (INCLUDING SEXUAL SELECTION) AND GENETIC DRIFT ACT ON THIS VARIATION, RESULTING IN CERTAIN CHARACTERISTICS BECOMING MORE COMMON OR RARE WITHIN A POPULATION.



POPULATION AS THE BASIC UNIT OF EVOLUTION.

- ✔ A **POPULATION** IS THE SMALLEST **UNIT** OF LIVING ORGANISMS THAT CAN UNDERGO **EVOLUTION**. ...
- ✔ A PECCARY **POPULATION**, THEREFORE, CAN BE CONSIDERED AS A "POOL" OF GENES AND GENE TYPES THAT REFLECT THE GENETIC DIVERSITY OF ALL THE INDIVIDUALS WITHIN THE GROUP.
- ✔ THIS "POOL-OF-GENES" IS WHAT CHANGES WHEN **EVOLUTION** TAKES PLACE.

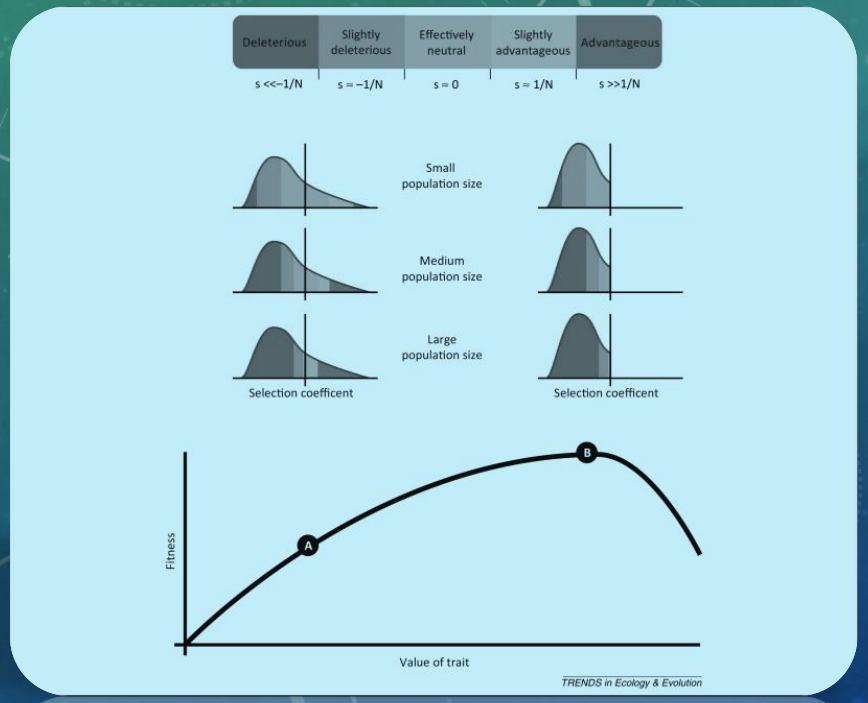


FACTORS OF EVOLUTION

- **NON-DIRECTED FACTORS OF EVOLUTION**
  - - GENETIC VARIATION IN POPULATION
    - GENE MUTATION
    - RECOMBINATION OF GENES
    - HYBRIDIZATION
  - - POPULATION SIZE
  - - REPRODUCTIVE ISOLATION
- **DIRECTED FACTORS OF EVOLUTION**
  - - NATURAL SELECTION

POPULATION SIZE

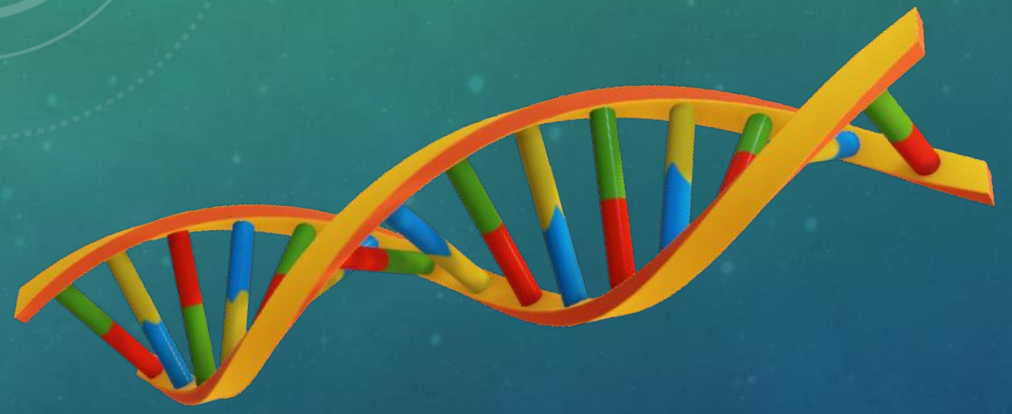
IN NATURE ALL POPULATIONS HAVE A TENDENCY AND A TREMENDOUS POTENTIAL FOR GROWTH, BUT REMAIN LIMITED TO A RELATIVELY CONSTANT SIZE DUE TO LIMITATION OF RESOURCES. IT MEANS ONLY A SMALL SIZE OF POPULATION ACTUALLY SERVE AS PARENTS FOR NEXT GENERATION. NORMALLY THE INDIVIDUAL BETTER ADAPTED TO THEIR ENVIRONMENT ARE MORE LIKELY TO SURVIVE. BUT CHANCE IS ALSO VERY IMPORTANT



GENETIC CHARACTERISTICS OF POPULATION

I. HETEROGENEITY OF POPULATION

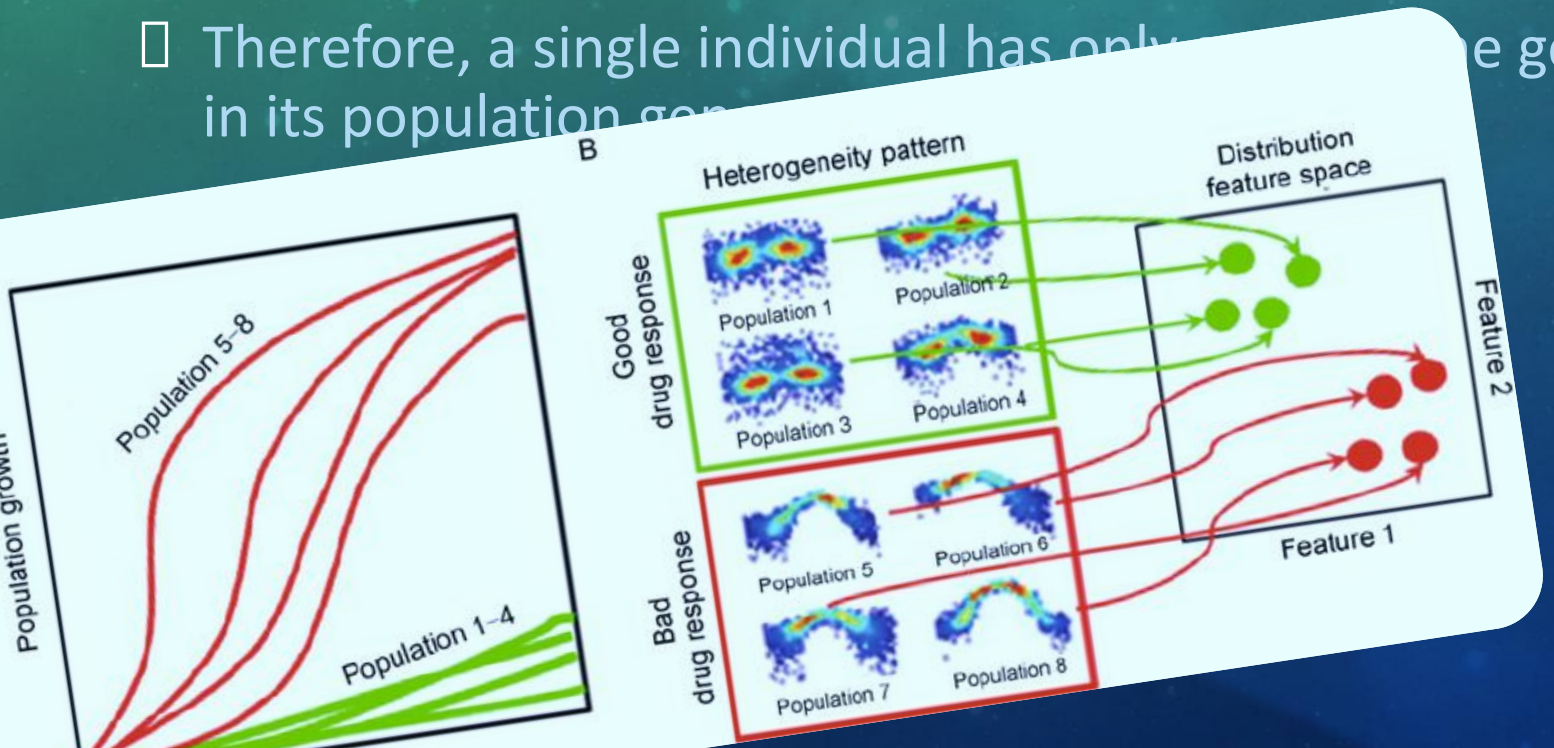
II. GENETICAL UNITY OF POPULATION



- IF A POPULATION IS NOT EVOLVING, THE FREQUENCIES OF EACH ALLELE REMAIN CONSTANT FROM ONE GENERATION TO THE NEXT GENERATION

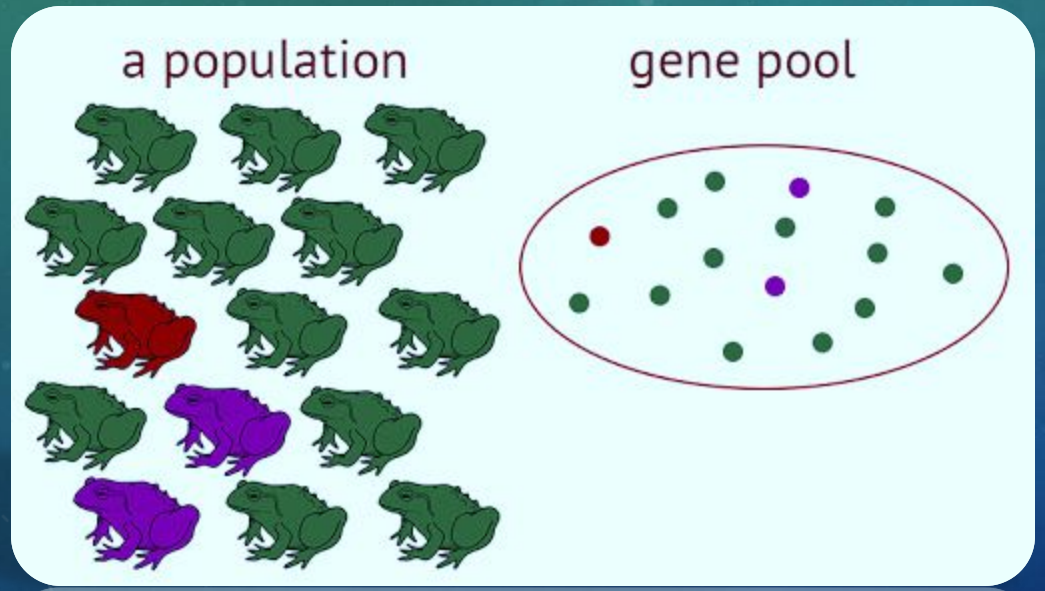
HETEROGENEITY OF POPULATIONS

- A given population (as illustrated by different phenotypes) indicates that each individual has a different portion of the genes that exist in the gene pool.
- Because most species are diploid, each individual member of a population contains only two alleles for each locus.
- Therefore, a single individual has only [illegible] genes found in its population [illegible]

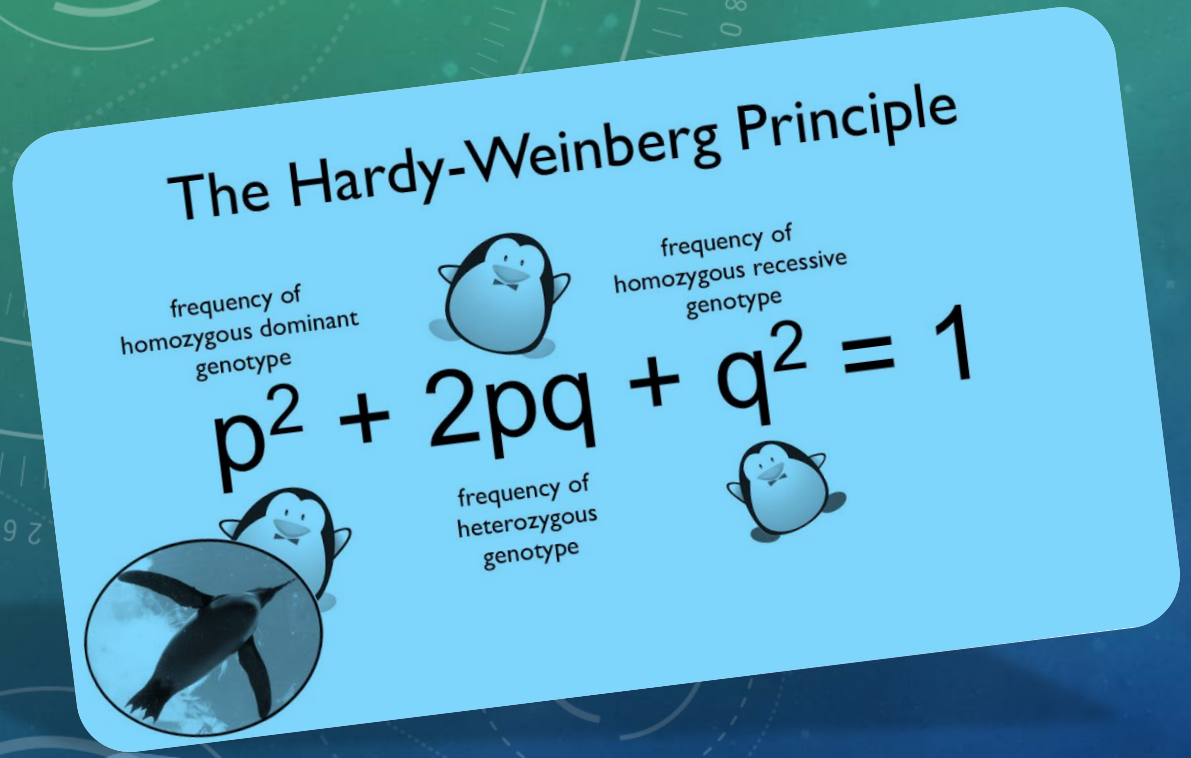


GENETICAL UNITY OF POPULATIONS.

- Each population possesses an iso- lated gene pool, which includes all possible alleles at each locus of each chromosome present in the breeding individuals of the population.



THE HARDY-WEINBERG LAW SHOWS THAT IN LARGE POPULATIONS, THE PROCESS OF INHERITANCE DOES NOT BY ITSELF CAUSE CHANGES IN ALLELE FREQUENCIES, WHICH REMAIN CONSTANT FROM GENERATION TO GENERATION.

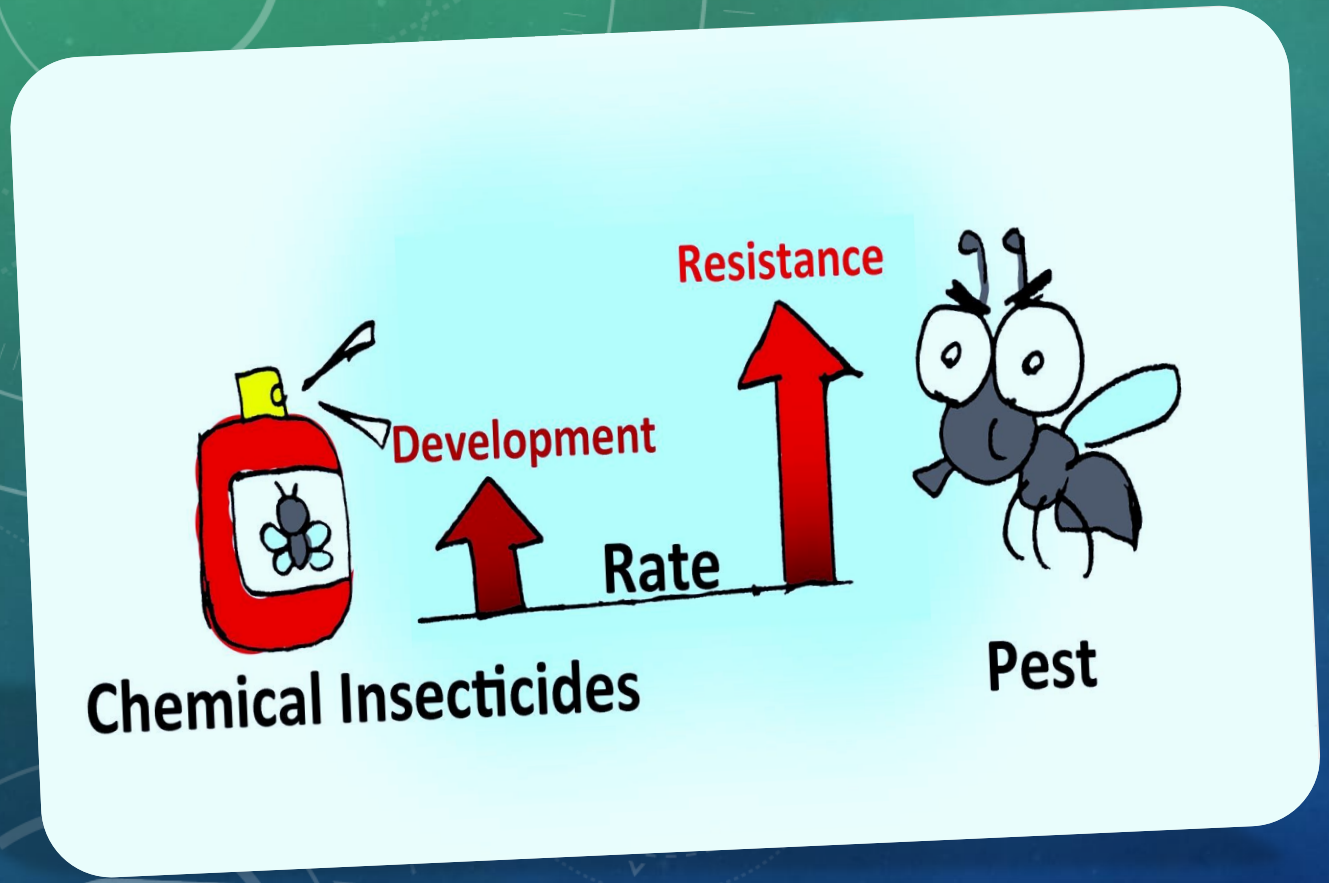


- *LARGE POPULATION SIZE*
- *ISOLATION*
- *NO MUTATION*
- *NO SELECTION*
- *RANDOM MATING*

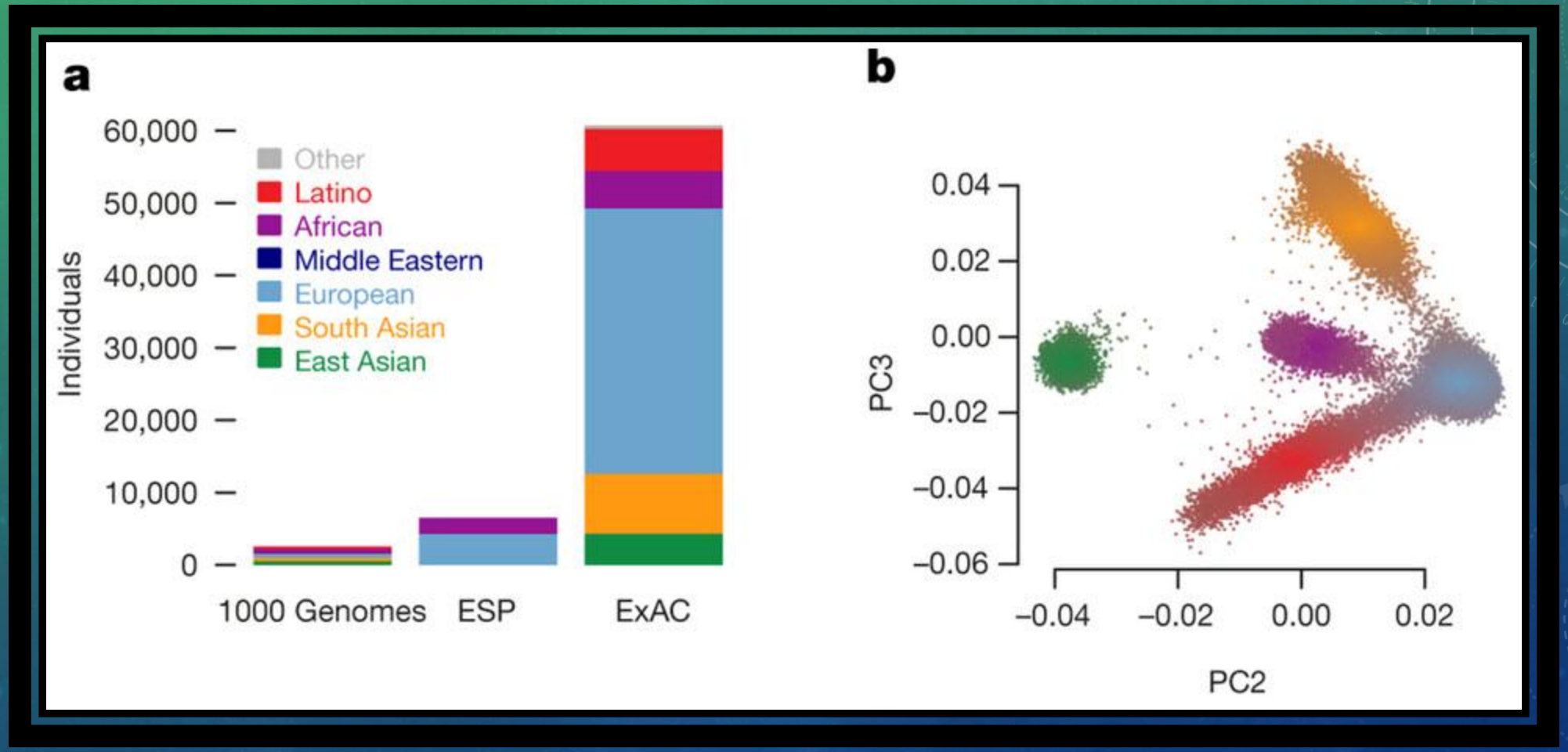
MOST POPULATIONS HAVE A LARGE RESERVOIR OF VARIABILITY

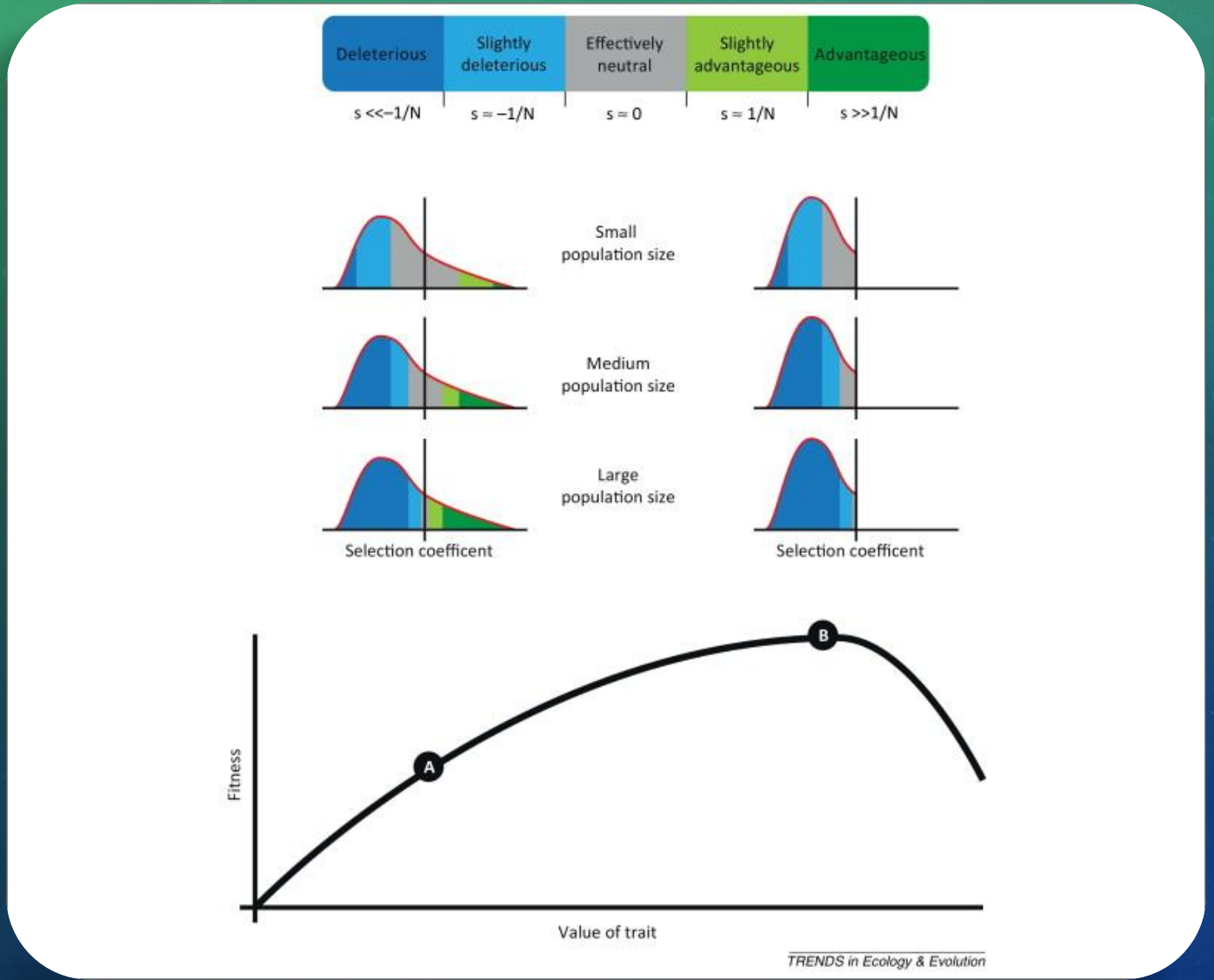
- ✔ HETEROZYGOTE ADVANTAGE OCCURS WHEN THE HETEROZYGOTE HAS A HIGHER DEGREE OF FITNESS THAN EITHER HOMOZYGOTE. BOTH ALLELES ARE MAINTAINED IN THE POPULATION.
- ✔ IN FREQUENCY-DEPENDENT SELECTION, A GENOTYPE'S SELECTIVE VALUE VARIES WITH ITS FREQUENCY OF OCCURRENCE.
- ✔ VARIATION THAT CONFERS NO DETECTABLE SELECTIVE ADVANTAGE IS CALLED NEUTRAL VARIATION.
- ✔ THE NEUTRALIST-SELECTIONIST CONTROVERSY IS A DEBATE AMONG BIOLOGISTS OVER THE RELATIVE IMPORTANCE OF SELECTION VERSUS GENETIC DRIFT IN MAINTAINING GENETIC VARIABILITY.

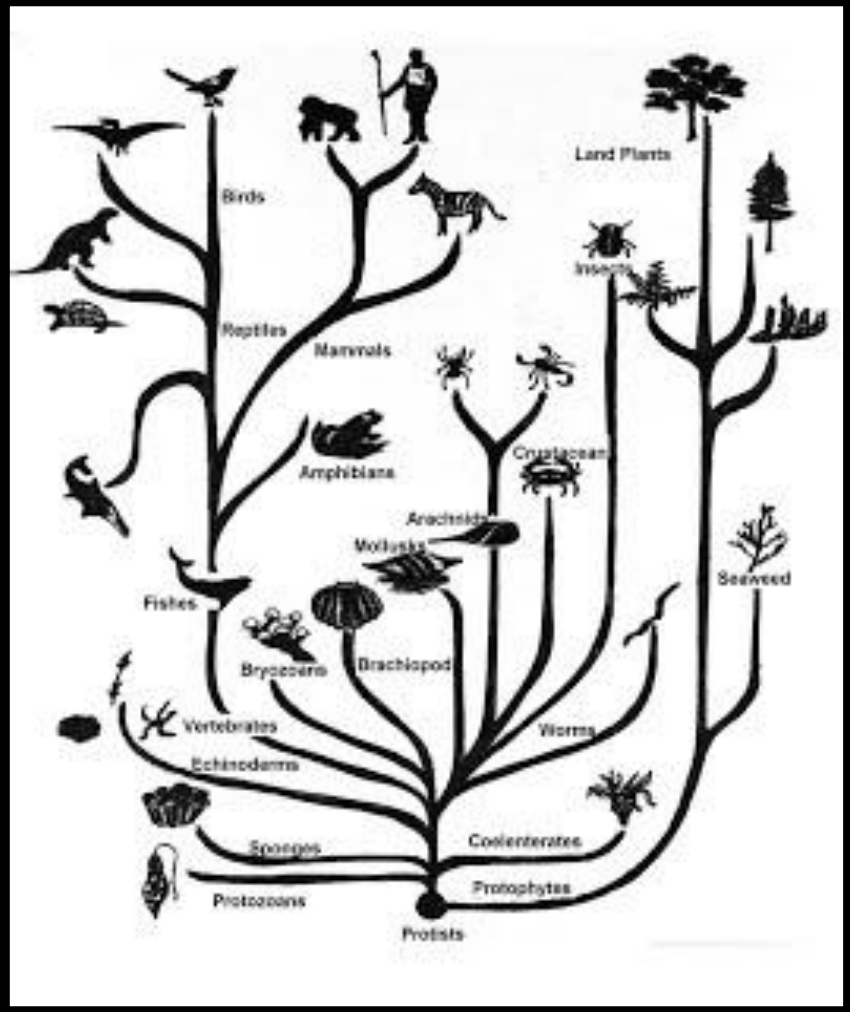
EVOLUTION → RESISTANCE



POPULATION GENETICS IS A SUBFIELD OF GENETICS THAT DEALS WITH GENETIC DIFFERENCES WITHIN AND ... HALDANE ALSO APPLIED STATISTICAL ANALYSIS TO REAL-WORLD EXAMPLES OF NATURAL SELECTION, SUCH AS PEPPERED MOTH ... MUTATIONS CAN EVENTUALLY CAUSE THE GENETIC ASSIMILATION OF TRAITS THAT WERE PREVIOUSLY INDUCED BY THE ENVIRONMENT.


a
60,000
50,000
40,000
30,000
20,000
10,000
0
Individuals
Other
Latino
African
Middle Eastern
European
South Asian
East Asian
1000 Genomes
ESP
ExAC
b
0.04
0.02
0.00
−0.02
−0.04
−0.06
PC3
−0.04
−0.02
0.00
0.02
PC2


Deleterious
Slightly deleterious
Effectively neutral
Slightly advantageous
Advantageous
s <<–1/N
s ≈ –1/N
s ≈ 0
s ≈ 1/N
s >>1/N
Small population size
Medium population size
Large population size
Selection coefficent
Selection coefficent
Fitness
A
B
Value of trait
TRENDS in Ecology & Evolution


Land Plants
Birds
Insects
Reptiles
Mammals
Amphibians
Crustacean
Arachnids
Molluscs
Seaweed
Fishes
Bryozoans
Brachiopod
Vertebrates
Worms
Echinoderms
Sponges
Coelenterates
Protophytes
Protozoans
Protists

POPULATION GENETICS BEGAN AS A RECONCILIATION OF MENDELIAN INHERITANCE AND BIOSTATISTICS MODELS. NATURAL SELECTION WILL ONLY CAUSE EVOLUTION IF THERE IS ENOUGH GENETIC VARIATION IN A POPULATION. BEFORE THE DISCOVERY OF MENDELIAN GENETICS, ONE COMMON HYPOTHESIS WAS BLENDING INHERITANCE. BUT WITH BLENDING INHERITANCE, GENETIC VARIANCE WOULD BE RAPIDLY LOST, MAKING EVOLUTION BY NATURAL OR SEXUAL SELECTION IMPLAUSIBLE.

THE HARDY–WEINBERG PRINCIPLE PROVIDES THE SOLUTION TO HOW VARIATION IS MAINTAINED IN A POPULATION WITH MENDELIAN INHERITANCE. ACCORDING TO THIS PRINCIPLE, THE FREQUENCIES OF ALLELES (VARIATIONS IN A GENE) WILL REMAIN CONSTANT IN THE ABSENCE OF SELECTION, MUTATION, MIGRATION AND GENETIC DRIFT.

FOR BETTER UNDERSTANDING

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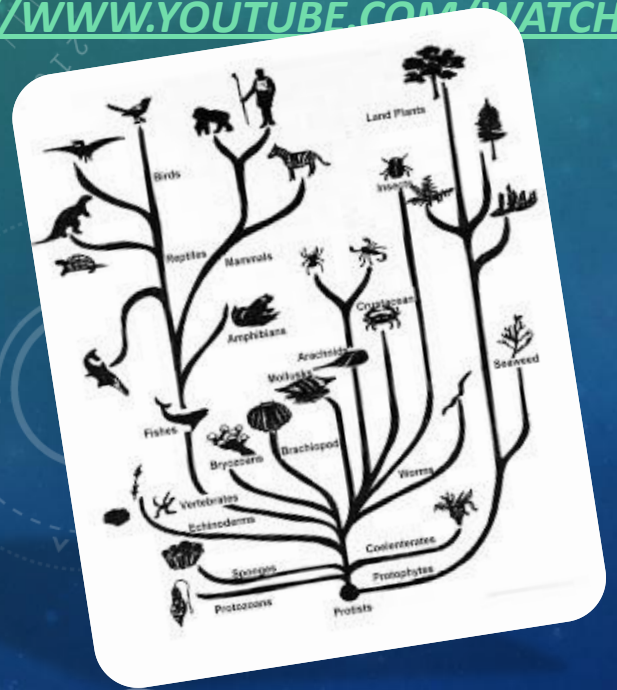


thank you