Genetics 101: Fundamentals of Heredity and Inheritance

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The fields of genetics gained immense importance as a result of famous experiments by Frederic Griffith in the late 1920s and 1930s. These famous experiments jolted the very concept of considering proteins as genetic material and gave us clue that it is something else. It wasn't proven until 1952 when Alfred Hershey and Martha Chase did their famous experiment to prove DNA as the genetic material for all living organisms. After these landmark experiments and then the arrival of Watson and Crick model of DNA we saw the boom of advancement in genetics which continues to date. Going back to the basics is essential to develop an understanding of the structural and functional genomics in the modern era. The core terms related to the discipline of genetics which we frequently come across are as follows:

Genetics
Genetics is a scientific study of heredity and of mechanisms by which characteristics are transmitted from parents to their offspri.

Gene (Gr: genos = birth, race)
Genes are basic units of inheritance. Gene is a segment of DNA that codes for a protein chain (e.g. hemoglobin is a complex protein and made up of four protein chains so four genes will code for four protein chains of hemoglobin).

Genome
It is the total number of genes present in an individual at a given time (it is important to note that we develop from a zygote through the process of mitosis and mitosis does not change genetic makeup of cell. So technically we all have same DNA and genes which were present in our zygote except for our gametes).

Gene Pool
Population is group of members of same species living in a same area at same time. Total number of genes present in whole population at a given time is its gene pool.

Locus
Genes are segments of DNA and DNA resides on chromosomes. So each and every gene has a specific place on chromosome where it resides. That particular position of gene on chromosome is called locus.

Alleles (Gr: allelon = of one another)
We human inherit two sets of chromosomes each from our parent i.e. one from mother and one from father. As both our parents belong to same species and share somatic characters so we have at least two genes for all characters on our chromosomes. These genes which control same character are called alleles of each other.

Homozygous Alleles
If both parents share same gene for a character to their offspring then the particular character said to have homozygous alleles (if a mother and father both have blood group O and share same gene for blood group O to their child then child said to have homozygous allele for blood group O).

Heterozygous Alleles
If both parents share different gene for a character to their offspring then the character said to have heterozygous alleles (if a mother has blood group O and father has blood group A then they share gene for blood group O and A to their child then child said to have heterozygous allele for blood group).

If whole population is homozygous for a specific gene then that gene is said to be fixed in a population (it is a rare phenomenon).

Dominant Gene
When two alleles are brought together in an individual, only one expresses or appears in a subsequent generation. Gene which expresses itself is said to be the dominant gene (e.g. a child receive blood B gene from mother and O from father but have blood group B. In this case blood group B gene is dominant gene).

Recessive Gene
The alleles which are unable to express called recessive (e.g. allele for blood group O in above example)

Codominance
Sometimes when an individual receives two different genes from parents they both

express themselves in offspring and said to be codominant (e.g. if a mother has blood group homozygous A and father has blood group homozygous B then offspring will have blood group AB).

Allele Frequency
Relative frequencies of different alleles in a population constitute the phenomenon of frequency of an allele.

Haplotype
A combination of alleles at closely linked gene loci that are inherited together. By analyzing the haplotypes of individuals, researchers can identify regions of the genome that are associated with the trait or disease.

Phenotype
We are unable to observe genes as real material but we continuously observe different characters produced by gene as their expression. Expression of gene which can be observed is considered as phenotype (blue eyes, tall height, blonde hair, widow's peak, etc.).

Genotype
To study transmission of genes from one generation to other we need calculable representation of phenotype i.e. symbols that represent phenotypes. That symbols which represent phenotypes are labelled as genotypes (e.g. Blood group O is genotyped as ii, Blood group AB is genotyped as IAIB).

True Breeds / Pure Parents
True breeds or pure parents are individual which are homozygous for their genetic characters i.e. they will produce offspring with their own characters (if a women with blood group O marries with men with blood group O then all children of this couple will have blood group O).

Hybrid
An individual which is offspring of two parents have different homozygous alleles i.e. true breeds for different alleles (e.g. a women with blood group homozygous A marries with a man who is homozygous B then their children are hybrid i.e. AB).

Multiple Alleles
If a genetic character is controlled by single gene in an individual but have more than two alleles then it is said to be a multiple allele (e.g. blood grouping in man is controlled by three alleles i.e. IA, IB and i in human so it is an example of multiple allele).

Polygenic Characters
If a genetic character is controlled by more than one gene (may or may not have multiple alleles for all those genes) then it is said to be a polygenic character (e.g. Human height, skin color, etc.).

Multifactorial characters
These are characters in which presence of genes is not sufficient for desired outcome but also require some environmental support for genes to express themselves (e.g. a child with genes for being tall heighted can achieve more height then normal if have good diet and sporting facilities available).

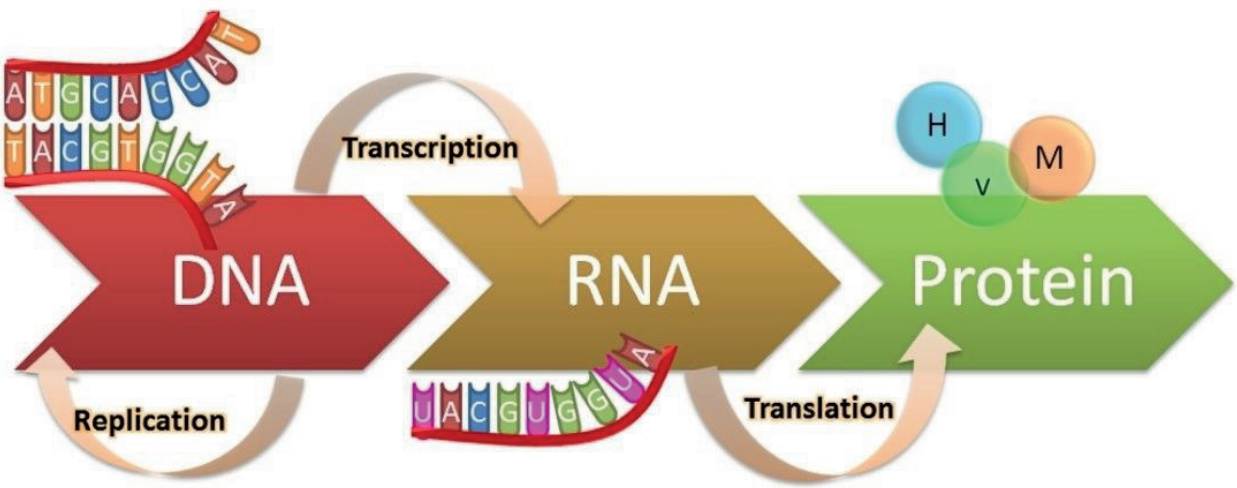
Genetic Crossing
It is a method use to study transmission of genetic character from parents to their offspring.

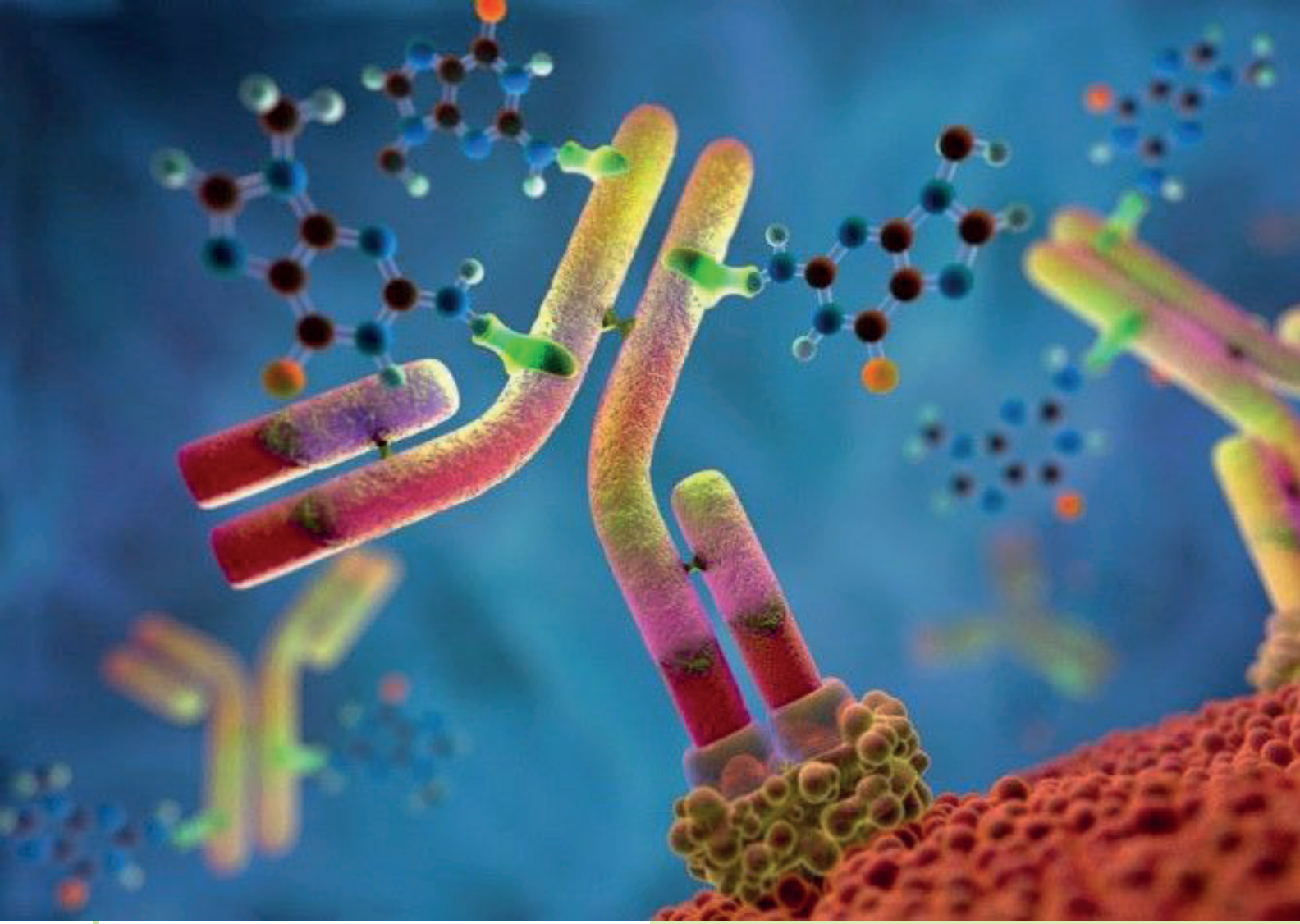
Mutation
At their locus on chromosomes or during their transmission from one generation to other genes can get changed, damaged, altered, etc. by different agents present in our environment. This change in gene is termed as mutation.

Mutagens
Mutagens are different agents which lead to mutation e.g. ionizing radiations, chemical mutagens, viral infections, etc.

Pleiotropy
Effects of gene at a locus on two or more characters are called pleiotropy (e.g. Marfan syndrome is caused due to the mutation in a single gene yet it affects various aspects of growth and development that include vision, height, and heart functioning).

Genetic variation
Genetic variation is the differences in DNA sequences among individuals or populations. This variation forms the basis for evolution and adaptation.





Genetic association
Genetic association is a statistical relationship between a particular genetic variant, or a set of variants, and the occurrence of a particular trait or disease.

Monogenic disorder
It is a disease that is caused by one or more mutations in a single gene, such as cystic fibrosis, which is caused by mutations in the CFTR gene. These disorders are also referred to as Mendelian diseases.

Polygenic Disorder
A polygenic disorder is a medical condition that is influenced by the interaction of multiple genes, each with a small effect on the disease risk. Examples include Type 2 diabetes caused by a combination of genetic and environmental factors, with multiple genes playing a role in the development of the disease.

Complex disease
A disorder in which the cause is considered to be a com-bination of genetic effects and environmental influences such as schizophrenia.

Methylation
Methylation of DNA is the process of adding a methyl group to DNA, which usually occurs at CpG sites (cytosine-phosphate-guanine sites) in the DNA sequence of vertebrates. This results in the conversion of cytosine to 5-methylcytosine.

Penetrance
It refers to the percentage frequency with which a dominant or homozygous recessive gene combination manifests itself in the phenotype of carriers.

Codon
A codon is a three-nucleotide sequence in DNA or RNA that codes for a specific amino acid or stop signal in protein synthesis.

Translation
Translation is the process by which mRNA is used to synthesize a protein.

Transcription
Transcription is the process by which DNA is used to produce a complementary RNA sequence.

Epigenetics
Epigenetics refers to the changes in gene expression that do not involve changes to the underlying DNA sequence. These changes can be influenced by environmental factors and can be passed down to future generations.

CRISPR
CRISPR (Clustered Regularly Interspaced Short Palindromic Repeats) is a gene editing technology that allows scientists to make precise changes to DNA sequences in living organisms.

Proteome
The proteome is the entire set of proteins that are expressed in a cell, tissue, or organism. It includes all of the proteins produced from an organism's genome, as well as those that arise due to post-translational modifications.

Exome
The exome is the portion of an organism's genome that codes for proteins. It represents only a small fraction of the total genome, but contains the majority of disease-causing mutations.

Genome assembly
Genome assembly is the process of piecing together the fragments of DNA sequence data obtained from high-throughput sequencing technologies into a contiguous sequence that represents the organism's genome.

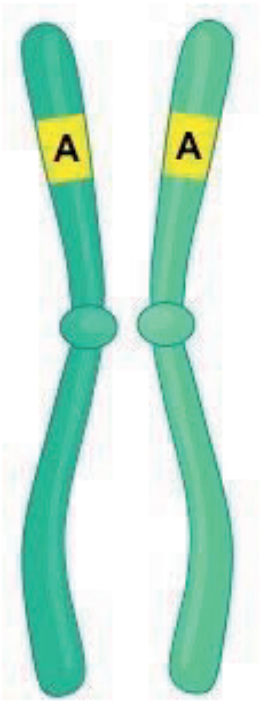
Annotation
Annotation is the process of identifying and labeling the functional elements within a genome sequence, such as genes, regulatory regions, and repetitive sequences.

Structural variation
Structural variation refers to differences in the genome structure that are larger than a single nucleotide, such as insertions, deletions, inversions, and duplications.

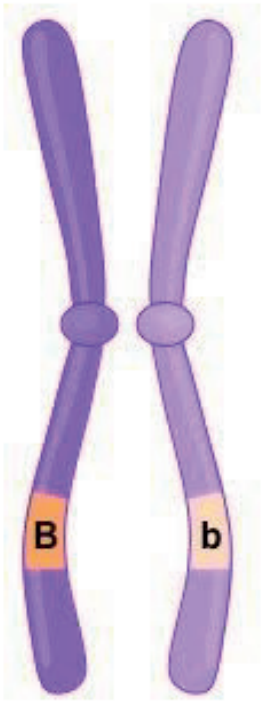
Single nucleotide polymorphism (SNP)
SNP is a variation in a single nucleotide that occurs at a specific position within a genome. SNPs are the most common type of genetic variation and can be used as markers to track inheritance and disease risk. For example an AG in place of an AT.

Copy number variation (CNV)
CNV is a type of genetic variation that involves changes in the number of copies (in comparison with single base pair in SNP) of a particular segment of DNA in the genome. CNVs can range in size from a few hundred base pairs to several megabases

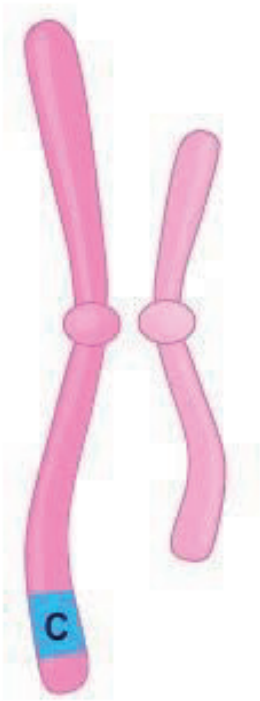
Comparative genomics



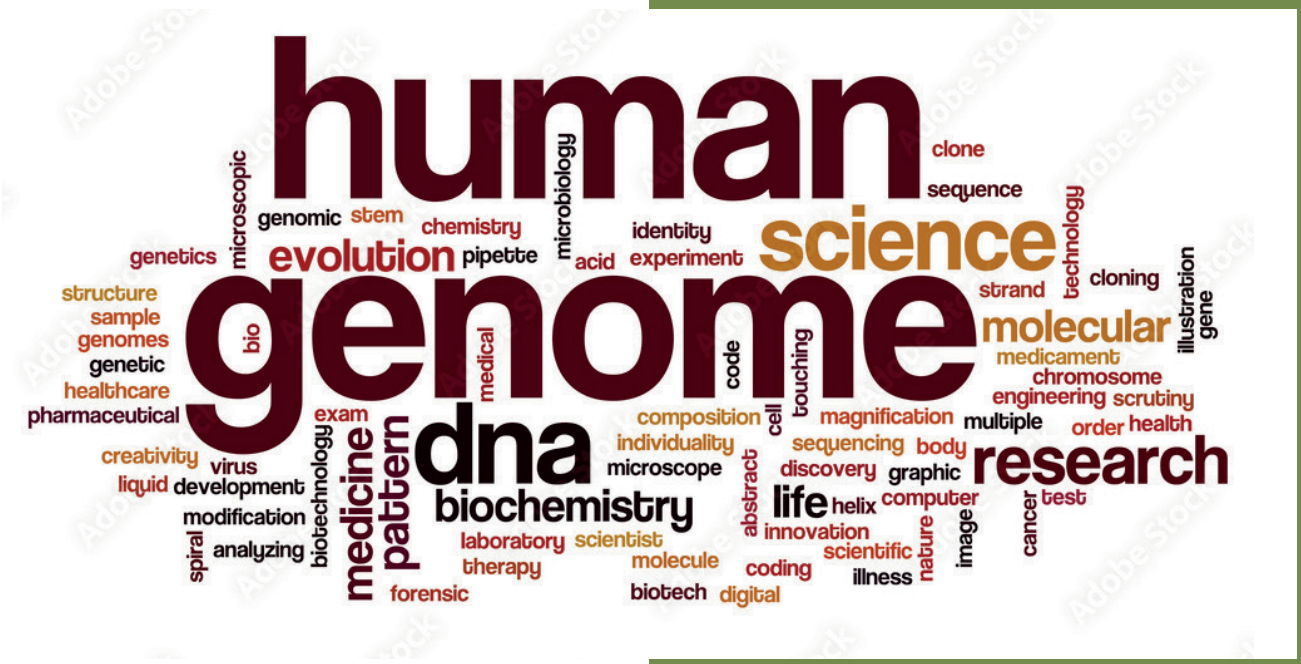
Homozygous
Alleles are same



Heterozygous
Alleles are different



Hemizygous
Only one allele
(e.g. XY)



Comparative genomics is the study of the similarities and differences in genome structure and function across different organisms.

Next-generation sequencing
Next-generation sequencing (NGS) refers to a group of high-throughput sequencing technologies that allow for the rapid and cost-effective sequencing of large amounts of DNA or RNA. NGS has revolutionized the field of genomics by enabling the sequencing of entire genomes and transcriptomes in a single experiment.

Polymerase chain reaction (PCR)
PCR is a laboratory technique used to amplify a specific region of DNA. It allows researchers to make multiple copies of a DNA fragment in a short period of time.

DNA sequencing
DNA sequencing is the process of determining the order of nucleotides (A, C, G, T) in a DNA molecule. This information can be used to identify mutations, study genetic variation, and determine the function of genes.

Genotyping
Genotyping is the process of determining an individual's genotype for a specific set of genetic markers, such as single nucleotide polymorphisms (SNPs).

Microarray analysis
Microarray analysis is a technique used to simultaneously analyze the expression of thousands of genes in a sample. It involves hybridizing a sample of DNA or RNA to a microarray chip that contains probes for specific genes.

RNA interference (RNAi)
RNAi is a technique used to silence gene expression by using small RNA molecules to target and degrade specific messenger RNA (mRNA) molecules.

Knockout mice
Knockout mice are mice that have been genetically engineered to have one or more genes "knocked out" or deleted. These mice are used to study the function of specific genes and to model human diseases.

Linkage analysis
Linkage analysis is a statistical method used to map the location of disease-causing genes in families with inherited genetic disorders. It involves identifying genetic markers that are linked to the disease gene and analyzing their inheritance patterns.

Genome-wide association studies (GWAS)
GWAS is a method used to identify genetic variations associated with a particular trait or disease. It involves comparing the genomes of large groups of individuals with and without the trait or disease to identify common genetic variants.

Pharmacogenetics
It is a field of genetics that studies the genetic variability in individual responses to drugs and drug metabolism.

Metagenomics
Metagenomics is the study of genetic material obtained directly from environmental samples, such as soil or water. It allows for the identification and characterization of microbial communities in their natural habitats.

Reference:

Morris-Rosendahl, Deborah. (2010). A glossary of relevant genetic terms. Dialogues in clinical neuroscience. 12. 116-20. 10.31887/DCNS.2010.12.1/dmrosendahl.