Introduction to Genetics

How is it possible that you look like your mother and have your father's characteristics? What makes siblings look and perhaps behave similarly? Genetics and heredity!

Genetics is defined in many ways. It is defined as the study of genes, genetic code heredity, and variations. It is that field of biology that describes how characteristics and features pass on from the parents to their offsprings in each successive generation. The unit of heredity is known as genes. Before we understand more about genetics, we need to understand what the genetic materials are.

Introduction to Genetics

There are two classes of genetic materials that are responsible for the transfer of information from one generation to another in animals:

- DNA or deoxyribonucleic acid
- RNA or ribonucleic acid

It is in the DNA or RNA sequences that biological information is stored and passed on.

DNA

Most organisms contain DNA except some viruses which contain RNA as their genetic material. DNA was discovered by two scientists- Watson and Crick and their model of the structure of DNA are called the Watson and Crick model. The structure of DNA is said to be a double- helical structure with two strands of DNA that are wound around each other. Each strand of DNA is made up of a sequence of nucleotide monomers. Each nucleotide is made up of:

- Deoxyribose sugar
- One of the four nitrogen bases
- Phosphate group

The nitrogen bases found in a DNA molecule are Adenine, Thymine, Cytosine, and Guanine. The nucleotides create a chain via covalent bonds that are formed between the phosphate of one nucleotide and the sugar of the adjacent one. The two strands of DNA are held

together by hydrogen bonds between complementary nitrogen bases i.e. Adenine with Thymine and Cytosine with Guanine.

A DNA molecule is said to be stable enough to be able to replicate itself. DNA replication involves RNA intermediates. The part of the DNA which codes for specific proteins during the replication is called as the gene.

RNA

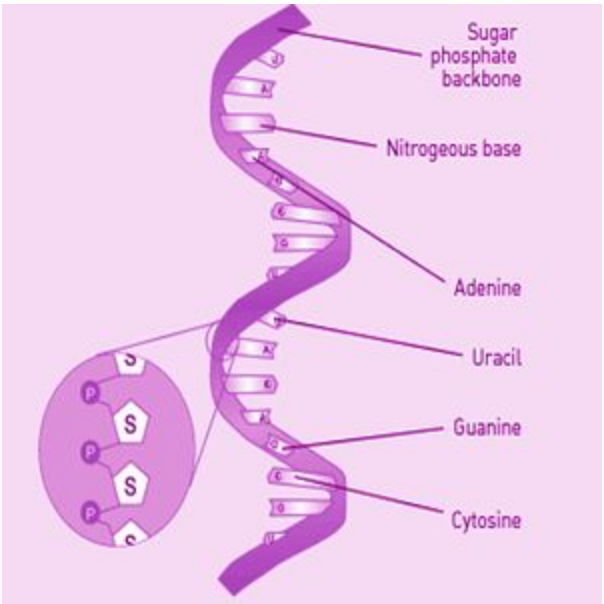

Image Source: javatpoint

Unlike the DNA, RNA is a single-stranded genetic material. The nucleotide bases present in RNA are similar to those in DNA except that thymine is replaced by uracil and pairs with adenine. While DNA

is the genetic material in most organisms, RNA is found in a few viruses. RNA is of three types depending on their function:

- tRNA or transfer RNA- helps transfer the amino acids from the mRNA to the ribosomes.
- mRNA or messenger RNA- helps to carry the codes for amino acids from the DNA to the ribosomes
- rRNA or ribosomal RNA- are found on the ribosomes and help in protein synthesis.

Video on Genetics

Introduction to Chromosome

Image Source: pinterest

Now that we know what the units of heredity are, let us understand what chromosomes are. The DNA that is found in the nucleus of each cell occurs as a tightly coiled package around proteins called as histones. These thread-like packaged structures of DNA are called as chromosomes.

Humans have 23 pairs of chromosomes(or 46 chromosomes). 22 pairs are called autosomes and one pair is called the sex chromosomes. Females in humans have 2 X(XX) chromosomes whereas males have

one X and one Y (XY) chromosomes. The number of chromosomes varies in different animal species.

Each chromosome is said to be divided into two unequal halves by a centromere into two arms. The short arm is called as ‘p arm’ and the longer arm is known as the ‘q arm’.

Genetic Inheritance

Gregor Mendel, a monk, is known as the father of modern genetics. He postulated few laws, known as the Laws of Inheritance. Mendel’s Laws are:

- Law of Segregation
- The Law of Independent Assortment
- Law of Dominance

With the help of experiments performed on a pea plant(Pisum sativum) and the use of Punnett squares, Mendel explained heredity and inheritance of characters and laid the foundation of genetic inheritance as we know it today.

Solved Example for You

Q1: What keeps the two strands of DNA linked to each other?

a. Hydrogen bond
b. Covalent bond
c. Ionic bond
d. The bond between the sugar molecule and the phosphate molecule

Sol. The correct answer is the option ”a”. The hydrogen bonds between complementary bases in the two DNA strands keep it together.

Laws of Inheritance

Gregor Johann Mendel was a scientist who is recognized as the Father and Founder of genetics. Mendel conducted many experiments on the pea plant (Pisum sativum) between 1856 and 1863. He studied the results of the experiments and deducted many observations. Thus, laws of inheritance or Mendel’s laws of inheritance came into existence. Before learning about Mendel’s laws of inheritance, it is

important to understand what the experiments performed by Mendel were.

Mendel's Experiments on Pea Plant

Mendel after carefully study selected the pea plant for many reasons:

- The pea plants were easy to grow and maintain
- It has many clearly distinct and contrasting characters.
- The pea plant is an annual plant and so many generations of the plant can be studied in a short period of time.
- Peas are naturally self-pollinating but can also be cross-pollinated.

Mendel made a list of contrasting characters which he studied:

Image Source: bioninja

Mendel structured his experiments in a way that he would observe one pair of contrasting characters at one time. He began his experiments using purebred lines for contrasting characters.

He cross-pollinated two pure lines for contrasting characters and the resultant offsprings were called F1 generation(also called the first filial generation). The F1 generations were then self-pollinated which gave rise to the F2 generation of second filial generation.

Browse more Topics under Principles Of Inheritance And Variations

- Introduction to Genetics
- Linkage and Recombination
- Mutation and Chromosomal Disorder
- Sex Determination

Understand the concept of Genetics here in detail.

Results of Mendel's Experiments

Let us look at the results of Mendel's experiments on crossing a pure tall pea plant with a pure short pea plant.

- In the F1 generation, Mendel observed that all plants were tall. there were no dwarf plants.
- In the F2 generation, Mendel observed that 3 of the offsprings were tall whereas 1 was dwarf.
- Similar results were found when Mendel studied other characters.
- Mendel observed that in the F1 generation, the characters of only one parent appeared whereas, in the F2 generation, the characters of the other parent also appeared.

- The characters that appear in the F1 generation are called dominant traits and those that appear for the first time in the F2 generation are called recessive traits.

Learn more about Linkage and Recombination here in detail

Conclusions

- The genes that are passed from the parents to the offsprings exist in pairs. These pairs are called alleles.
- When the two alleles are the same, they are called homozygous. When both the alleles are different, they are called as heterozygous.
- Dominant characters are described using capital letters and recessive using small letters. For example, the dominant genes for tallness in a pea plant are written as TT and recessive genes as tt. The heterozygous genes are written as Tt where the plant appears tall has the recessive gene which might express itself in the future generations.
- The appearance of the plant is known as the phenotype whereas the genetic makeup of the plant is called the genotype. So, a

plant with Tt genes appears tall phenotypically but has a recessive gene.

- During gametogenesis, when the chromosomes become half in the gametes, there is a 50% chance of either of the alleles to fuse with that of the other parent to form a zygote.

Understand the concept of Sex Determination here in detail.

Based on these observations, Mendel proposed three laws.

Laws of Inheritance

Mendel proposed three laws:

- Law of Dominance
- The Law of Segregation
- Law of independent assortment

Law of Dominance

Image Source: gladewaterbiology

This law states that in a heterozygous condition, the allele whose characters are expressed over the other allele is called the dominant allele and the characters of this dominant allele are called dominant characters. The characters that appear in the F1 generation are called as dominant characters. The recessive characters appear in the F2 generation.

Law of Segregation

Image source: wikipedia

This law states that when two traits come together in one hybrid pair, the two characters do not mix with each other and are independent of each other. Each gamete receives one of the two alleles during meiosis of the chromosome.

Mendel's law of segregations supports the phenotypic ratio of 3:1 i.e. the homozygous dominant and heterozygous offsprings show dominant traits while the homozygous recessive shows the recessive trait.

Law of Independent Assortment

Image Source: biology-forums

This means that at the time of gamete formation, the two genes segregate independently of each other as well as of other traits. Law of independent assortment emphasizes that there are separate genes for separate traits and characters and they influence and sort themselves independently of the other genes.

This law also says that at the time of gamete and zygote formation, the genes are independently passed on from the parents to the offspring.

Solved Example for You

Q1: What is the genotype of an individual?

a. Physical appearance
b. Genetic makeup
c. Nature of the individual
d. Blended characteristics of the individual

Sol. The correct answer is the option ”b”. The genetic makeup of the individual is known as the genotype whereas the physical appearance of the individual is known as the phenotype.

Linkage and Recombination

Linkage and recombination are phenomena that describe the inheritance of genes. A linkage is a phenomenon where two or more linked genes are always inherited together in the same combination for more than two generations. The recombination frequency of the test cross progeny is always lower than 50%. Therefore, if any two genes are completely linked, their recombination frequency is almost 0%.

The phenomenon of linkage was studied by the scientist T.H. Morgan using the common fruit fly or *Drosophila melanogaster*.

Morgan's Experiment

Image Source: biotech.gsu

Morgan picked *Drosophila melanogaster* as his subject for the following reasons:

- He noticed a white-eyed male drosophila instead of the regular red eyes.
- It was small in size
- They have a short lifespan and so many generations can be studied in a short time frame.
- They have a high rate of reproduction

He crossed a purebred white eyed male with purebred red-eyed female. As expected following Mendel's laws, the F1 progeny were born with red eyes. When F1 generation was crossed among each other, the ratio of red-eyed to white eyed progeny were 3:1. However, he noticed that there was no white- eyed female in the F2 generation.

To understand further, he performed a cross between a heterozygous red-eyed female with a white-eyed male. This gave a ratio of 1:1:1:1 in the progeny(1 white eyed female, 1 red eyed female, 1 white eyed male and 1 red eyed male). This made Morgan think about the linkage between the traits and sex chromosomes. He performed many more crosses and determined that the gene responsible for the eye color was situated on the X chromosome.

Types of Linkage

Linkages are primarily of two types: Complete and incomplete

- Complete Linkage: When the combination of characters appears together in more than two generations in a regular manner, it is called as a complete linkage. Due to this complete linkage, only two types of gametes are formed. Example: Drosophila melanogaster
- Incomplete Linkage: When there is an incomplete linkage, new gene combinations are formed in the progeny or offsprings. This occurs due to the formation of a chiasma or crossing over between the linked genes.

Linkage Significance

- Due to the linkage between genes, desired characters cannot be brought together by breeders. This would be possible only if the genes would sort independently.
- The characters that are linked remain so as there is no chance of recombination of the linked genes.

Sex- chromosome Linked Diseases in Humans

Diseases like haemophilia, color blindness, male pattern of baldness are sex-linked diseases. Where color blindness and haemophilia are X-linked diseases, male pattern of baldness is a Y-linked one. This indicates that the X-linked diseases will express themselves in a male whereas the female is always a carrier until both the genes are recessive in the female. Male pattern of baldness being a Y-linked trait expresses itself only in the males while females are never affected by it.

Crossing Over

This is a phenomenon where genetic material is exchanged between non-sister chromatids of homologous chromosomes which results in a new gene combination. The process of crossing over occurs in a sequence of following steps:

Image Source: socratic

- Synapsis
- Duplication of chromosomes
- Crossing over
- Chiasmata formation
- Terminalization

Solved Example for You

Q1: If the genes are completely or fully linked, what are the chances of recombination?

a. 65%

b. 25%

c. 0%

d. 100%

Sol. The correct answer is the option ”c”. If the genes are completely or fully linked, then the chances of recombination are 0%.

Mutation and Chromosomal Disorder

Mutations and chromosomal disorders are two major drawbacks when it comes to genetic inheritance. Well, not everyone is born ideally, are they? Mutation and chromosomal disorder are different phenomena in genetics. Let us understand them in a bit more detail.

Mutation

A mutation can be defined as a slight change or alteration in the genetic material of an individual that brings about genetic diversity in the species. They can affect a small portion of the gene sequence or a large one. Mutations can be broadly classified into two types:

- Somatic or Acquired

These gene mutations can occur at any time during an individual's lifetime. They are called acquired because they are acquired during one's lifetime due to environmental factors like UV rays. They are called as somatic mutations because they occur in general body cells and not in those that produce the egg or sperm. Due to this, they cannot be inherited or passed on from one generation to another.

- Hereditary

As the name goes, these mutations can be passed on from one generation to the next. These mutations occur in the egg or sperm cells of the parent. When the two parental cells, one with mutation, unite, the resulting offspring is born with this mutation. This offspring will now have this mutation in all of its body cells.

Another kind of mutation that can occur is called as mosaicism. In this type of mutation, the parental germ cells have no inherent mutation in them. But, once the zygote begins to undergo multiple cell divisions, a mutation occurs. In this case, those cells that divide from the mutated cell or cells will have the mutation while the rest are clear of the mutation.

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- Laws of Inheritance
- Linkage and Recombination
- Sex Determination

Chromosomal Disorders

Unlike mutations that affect the genetic code, chromosomal disorders are caused by the change in number or structure of chromosomes. Both these give rise to different kinds of diseases.

Diseases Caused by Changes in Chromosomal Number

Humans normally have 46 chromosomes or 23 pairs of them. However, some babies are born with either less or more chromosomes than the required number giving rise to different conditions. Sometimes, a chromosome can get duplicated leading to a condition known as trisomy.

In other cases, a chromosome does not get duplicated leading to a condition called as monosomy. At times, the entire set of chromosomes get duplicated called as triploidy and if more than

duplication would lead to polyploidy. Examples of diseases that are caused by changes in chromosomal number:

- Down's syndrome: It is caused by trisomy of chromosome number 21. It is characterized by intellectual disability including mental retardation. They have lots of heart conditions and learning and growth disabilities as well.
- Turner's syndrome: This syndrome occurs due to missing second sex chromosome and is written as XO. Characterised by both male and female genitalia, individuals born with this syndrome have mental disabilities and retarded sexual development.
- Klinefelter's syndrome: This syndrome is characterized by an extra sex chromosome. These individuals are called 'supermales'. The syndrome is represented as XXY where the individuals have an extra Y chromosome. The individuals with this syndrome have similar features like those with Turner's syndrome but they are more aggressive and tend to have a criminal behaviour.

- Other syndromes are Edward's syndrome(trisomy of chromosome 18) and Patau's syndrome(trisomy of chromosome 13).

Diseases Caused by Alteration in Structure of Chromosomes

These diseases arise due to some changes like deletion or rearrangement of parts of the chromosome. They are of various types:

- Deletion: As the name suggests, this disorder arises due to loss of a certain portion of the chromosome during cell division. The amount of deletion decides the severity of consequences. If there is a loss of certain important genes, it can be lethal to the offspring. Example of such a disorder is Cry-du chat syndrome.

Image Source: ontrack-media

- Duplication: When there is a duplication of some part of a chromosome or a replicated part of a homologous chromosome attaches itself to an arm of the chromosome, there can be repeat gene sequences. This is known as duplication. Example: Fragile X syndrome.

Image Source: texasgateway

- Translocation: When a portion of the chromosome is translocated or moved to another chromosome, this phenomenon is called as translocation. They can be of two types: Reciprocal translocation is when segments from two different chromosomes are exchanged and Robertsonian when one entire chromosome attaches to another chromosome. The effect translocations depend upon which segment has been translocated and which area it has been translocated to. They often result in children born with disabilities or worse, miscarriages.

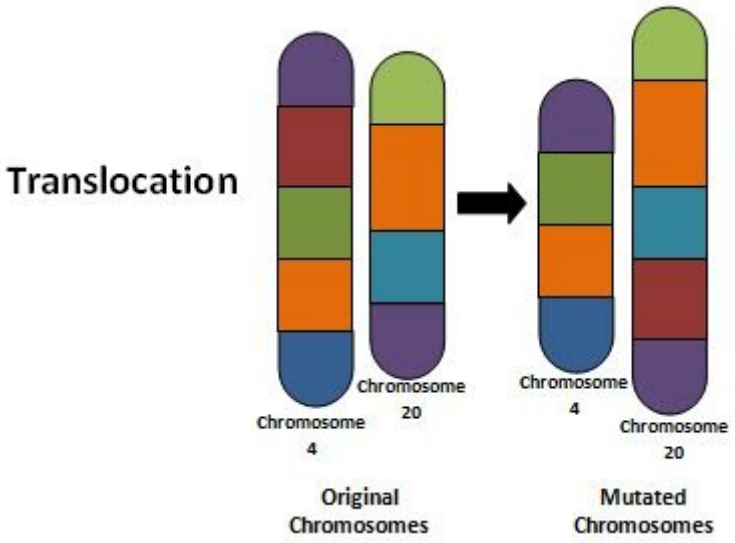

Image Source: ontrack-media

- Inversions: In this type of chromosomal structure defect, a part of the chromosome get inverted such that the gene sequence appears inverted. Its effects are not as severe as seen in other forms of structural defects.

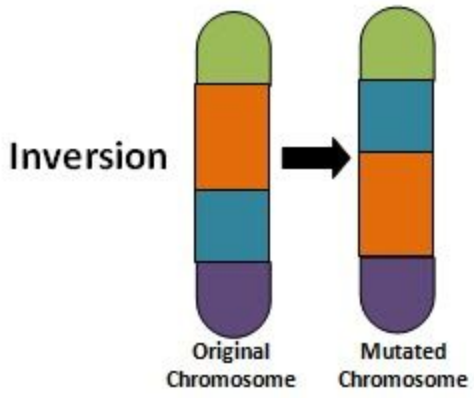

Image Source: ontrack-media

Solved Example for You

Q1: What causes Edward's syndrome?

a. Trisomy of chromosome 13
b. The Trisomy of chromosome 15
c. Trisomy of chromosome 18
d. Monosomy of sex chromosome

Sol. The answer is option "c". Trisomy of chromosome 18 leads to Edward's syndrome. Patau's syndrome is caused by trisomy of chromosome 13.

Sex Determination

Sex determination, in biology, is a system which decides the sexual characteristics of an organism or offspring. It helps to determine whether the organism will be male or a female, which are the two most common sexes. The widely used technique is chromosomal sex determination, in which sex chromosome of male i.e. X or Y chromosome, decides the sex or gender of the offspring. Female carries XX chromosome and male carries XY chromosome, in most cases. It is also possible to perform genetic tests to eliminate any chromosomal or genetic disorders. Sex determination is the genetic process of determining the sex of the organism. Let us study in more detail about this.

Chromosomal Sex Determination

Image source: study.com

Before we understand how sex determination is done, we need to understand the genetic makeup of a human being. As we all know, humans have 23 pairs or 46 chromosomes. Of these 23 pairs, 22 pairs are known as autosomes whereas 1 pair is known as the sex chromosome. It is this one pair that helps in determining the sex of an individual.

This was first studied by the German scientist Hermann Henkingin 1891. He first noticed a different nuclear material in some of the male gametes in the insect he was studying. It was named the X chromosome. He also noticed that a large number of insects had only one chromosome and were denoted as XO. From here began the studies on sex determination of both sexes in all animals.

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Sex Determination in Humans

Image source: studypage

Females in humans have 2X chromosomes- 1 each is inherited from either parent and denoted as XX. Males in humans have 1X and 1 Y chromosome, where the X is inherited from the mother and the Y from the father. In a way, we can say that it is the father who determines the sex of the unborn child. This can put to shame a lot of history in which people believed it is the woman who was responsible for not giving birth to a male heir. We can also say that the absence of the Y chromosome makes the individual a female.

At the time of spermatogenesis in males, both types of gametes are produced- one carrying the X chromosome and one carrying the Y. At the time of fertilization, the sex of the resulting zygote will depend on which gamete of the father will fuse with the X of the mother. We can,

therefore, say that there is a 50% chance that the child will be a male and 50% that it can be a female.

Types of sex determination

- The XX-XY system as seen in human beings, where, XX is the female and XY is the male. This is also seen in a few insects.
- The ZW-ZZ system as seen in birds, where, ZW is the heteromorphic female and ZZ is the homomorphic male. This is also seen in some fishes and few insects.

It is important to note that pre-natal sex determination is an offense in many countries in the world, including India. A process known as Amniocentesis is done that examines the amniotic fluid which determines an abnormality in the fetus. This process also reveals the sex of the unborn child. Due to abortions of the female fetus in countries like India, the government has imposed a strict ban on this process of sex determination. In many western countries, sex determination is legal.

Solved Example for You

Q1: What is the genetic makeup of female birds?

(a) XO (b) ZW

(c) ZZ (d) XX

Sol. The correct answer is the option ”b”. In humans and few insects like Drosophila, the females are denoted as XX. We can say that the females are homomorphic. In birds, the females are denoted as ZW and males as ZZ. So, in birds, the females are heteromorphic and the males are homomorphic. In humans, the mere absence of the Y chromosome makes the individual a female.