

**Chapter 1 : Averyâ€“MacLeodâ€“McCarty experiment - Wikipedia**

*In this article we will discuss about Mendel's law of inheritance. Mendel's Experiment: Gregor Johann Mendel conducted hybridisation experiments on garden pea (*Pisum sativum*) for seven years () and proposed the laws of inheritance in living organisms.*

In biology, the word refers to the genetic processes by which parents endow their children with specific traits, like height, hair color, eye color, and other characteristics. It is an everyday observation that children tend to resemble their parents in many ways, but it is equally obvious that the resemblance is not a simple one. Tall parents do not always produce tall children; children with blue eyes do not necessarily have parents with blue eyes. Similarity and Difference Yet, in humans and all other biological organisms, there are clear similarities between one generation and the next: In other words, each species, or type of organism, produces members of the same species. Striking similarities also exist within species. Every normal cat has four legs, humans have two, and spiders have eight. Despite these similarities, no two individuals of the same species except for identical twins or triplets are carbon copies of each other. Genetics is the science that explains these similarities and differences. It even explains those rare and often tragic cases where a member of a species is born with physical abnormalities. It does so through Members of the same species can look very similar, like these baby chicks, but individuals are rarely exact genetic replicas of each other. These laws were worked out in the middle years of the nineteenth century by an Austrian monk named Gregor Mendel, who devised them after conducting an extensive series of experiments on garden peas. In the years since Mendel did his work, countless experiments with other types of plants and animals have confirmed that the laws of inheritance apply to even extremely complex biological organisms like human beings. Although Mendel published the results of his work with peas almost a century and a half ago, his experiments are still regarded as masterpieces of scientific method. This is not the simplicity of a simple man, but of a genius, who sees the simplicity that lies behind the surface incoherence. It seeks to find simple rules to explain phenomena that on the surface appear to be very complex. When these rules are confirmed by numerous experiments, they are classified as laws. Most scholars agree that the paper in which Mendel recorded the results of his eight years of work with garden peas remains a completely up-to-date discussion of the laws of heredity. He wanted to find rules that were universal. These life-forms reproduce by a relatively simple process of cell division. Stimulated by internal chemical processes, the solitary cell divides to produce an exact replica, or clone, of itself. The genes contained in the replica are identical to those in the original. Depending on the type of organism involved and the environment in which it is living, cell division can recur a virtually limitless number of times, creating countless identical living things. However, plants and animalsâ€“including human beingsâ€“reproduce sexually. That is, genetic material from both a male and a female parent is required to produce a new member of the species. One of the consequences of sexual reproduction and the transfer of genetic information that it entails is that offspring are different from their parents and from each other. Every individual produced in this way, except for identical twins and triplets, is unique. To arrive at the principles underlying the transmission of traits from parents to their offspring in sexual reproduction, Mendel chose to study garden peas. He focused on seven traits that were easy to observe and, in garden peas, come in two distinct varieties: In order to fully appreciate the laws of inheritance, it is necessary to understand that plants, like people, have male and female sexual organs. However, unlike in people, both sex organs can occur on the same plant. This allows the plant to fertilize itself as well as be cross-fertilized by another plant. The sex organs are located in the flower of the plant. The male organs, called the anthers, produce pollen, which contains the male sex cells, or sperm. When fertilization occurs, pollen is transferred from the anthers to the female organ, called the stigma, where it combines with a female sex cell, or egg. Mendel used a small brush to accomplish this chore. He set himself the task of cross-fertilizing plants with contrasting traitsâ€“tall with short, yellow-seeded with green-seeded, and so onâ€“and tabulating the results. He realized that he would have to observe a large number of crossbreedings to compensate for the possibility that some of the results might be due to flukes like unintended wind-borne fertilization. By the time his eight-year experiment was over, he had carefully recorded

the results of twenty-eight thousand crossbreedings. As further insurance against false results, he also made sure that the plants he started with were pure-breeding, another way of saying that for generations they had produced only one version of each of the traits he was interested in. The first trait he looked at was seed shape. All the plants produced by crossbreeding round-seed and wrinkle-seed plants had round seeds. Similarly, the mating of tall plants with short ones produced plants that were all tall. There were no intermediate specimens—no seeds that were somewhat wrinkled and no plants whose height fell between that of its tall and short parents. Discussing seed color, Hawley says, In the first generation, when Mendel crossed plants with green seeds to plants with yellow seeds, all he saw in the progeny were plants with seeds identical in color to those of the green-seeded parent. A real adherent to blending would have postulated that the progeny of the first generation should have been yellowish-green, not true green. If genetic material was not blended, then it had to be passed on in discrete units. These units—Mendel called them factors, but they are now referred to as genes—retained their individual identity in the newly created organisms, which could then pass them on to succeeding generations. The second, and equally important, principle of inheritance came to light when Mendel bred second-generation plants to themselves and each other. Traits—yellow seeds and shortness, for example—that had vanished in the second generation suddenly reappeared in the third. With respect to height, one out of every four third-generation plants was short. Mendel obtained similar ratios for the other traits he was studying. In order to explain differences in traits, Mendel supposed that genes could take different forms, called alleles, that specified different expressions of the trait. For example, Mendel claimed that there was a gene that gave seed color and two different forms or alleles of that gene: In addition, an individual must be able to carry genetic information for a trait it does not express. We know that the green-seeded progeny [offspring] produced by the first generation carried the information to produce yellow seeds because they were able to pass it on to the yellow-seeded progeny in the next generation. To denote this difference between the ability of alleles to determine a [trait], Mendel introduced the terms dominant and recessive. Each parent passes on one allele of each gene to each child. A child who inherits two dominant alleles for a given trait will exhibit that trait. A child who inherits one dominant and one recessive allele will also exhibit the trait because the dominant allele will mask the presence of the recessive allele. Only if the child inherits two recessive alleles will the dominant trait not appear. Dominant and Recessive Traits The propensity for red hair, for example, could be lurking in recessive form in the genes two nonredheaded parents pass along to their children. Those children will, in turn, pass it on to their children. Should one of those children grow up and marry someone also carrying the recessive form of the gene, and should they both pass on the recessive form to their offspring, redheadedness will suddenly emerge after an absence of several generations in the family tree. The height of garden pea plants illustrates this point clearly. With respect to the gene that governs this trait, the dominant allele leads to tallness and the recessive allele leads to shortness. A pea plant will be tall if it inherits the dominant tall allele of the gene from each of its parent plants. It will also be tall if it inherits one dominant tall allele and one recessive short allele. Only if it inherits two recessive alleles will it be short. This insight is one of the cornerstones of modern genetics: Rather than blending in offspring, genes are inherited from parents as discrete units that come in two forms, each of which is passed on separately and remains separate in the genetic makeup of the new individual. This means that recessive traits can be passed on from generation to generation without leaving a visible trail. A person with a dimpled chin has either one or two dominant alleles for this trait. However, since someone with a dimpled chin might have a recessive allele for that trait, it is possible for that person to pass it on to a child. This property of genes, that they each come in two different forms or alleles and that each parent contributes one allele to the genetic makeup of his or her children, is called the principle of segregation. Support for this principle came in the twentieth century when biologists discovered that when sex cells—male sperm and female eggs—are formed, each carries A person with a dimpled chin like this man has at least one dominant allele for that trait, or two recessive alleles. When the sperm fertilizes the egg to form a new individual, these alleles recombine and, according to the rules of dominance and recessiveness, produce the traits the new individual exhibits. Each egg cell and each sperm cell are just as likely to contain one allele as they are to contain the other. Another important law of inheritance, also discovered by Mendel and proved by subsequent research, is called

the principle of independent assortment. In conducting his experiments with peas, Mendel noticed that traits were inherited independently of each other. In other words, whether a plant was tall or short had no effect on whether its seeds were green or yellow. Mendel was not in a position to know this fact, but it was proved when the structure of cells was determined in the decades after he completed his work. In certain cases, called incomplete dominance, the inheritance of both a dominant and a recessive allele of a gene does produce what appears to be a blend of traits. Certain flowers, for example, come in three colors. When a dominant and a recessive allele for the gene that determines color combine, the result is a shade intermediate between the two other colors that the flower can assume. Although this seems to suggest that a blending of genetic material has occurred, it has not. Another subtlety that escaped Mendel has come to be known as quantitative inheritance. The traits he studied were very easy to observe: A seed was either green or yellow, a plant was either tall or short. Each of these traits was caused by a single gene. Some characteristics, however, do not lend themselves so easily to either-or classification. Human skin color, to take an obvious example, comes in a wide range of shades. Scientists now know that these traits are determined by more than one gene. It is the combined effect of all the relevant genes that produce the characteristic, allowing for more variation than occurs when only single genes are involved. The rules of inheritance that Mendel discovered—that genetic material is passed from generation to generation in discrete units, that these units come in two forms called alleles, that each parent contributes one allele for each gene to the offspring, and that the units operate independently of each other—laid the groundwork for twentieth-century genetics. From his modest experiments with garden peas have come the breakthroughs that are revolutionizing biological science and our understanding of life itself. Also read article about Inheritance from Wikipedia User Contributions: If my Father were to live by now he is years. No body of my siblings have blue eyes.

**Chapter 2 : Difference between Qualitative Inheritance and Quantitative Inheritance**

*Labs & Experiments. mono- and dihybrid crosses, and statistics to teach concepts of inheritance. A laboratory experiment using simple equipment and chemicals.*

People failed to appreciate its rigor and its implications for the scientific understanding of inheritance, breeding, evolution and cell biology. Horticulturalists had long published plant-breeding experiments that superficially resembled those of Mendel. Those studies, however, did not include rigorous backcrossing of the starting plants to generate pure-breeding parental strains, nor were the number of different colored peas in the offspring carefully recorded. Unlike Knight, Mendel employed a rigorous scientific strategy when devising his experiments. By assuring that he had pure-breeding plants to start, he made certain that his results were reproducible. Holistic and organismal approaches were in style when Mendel was performing his experiments. Unlike Darwin, Gregor Mendel simplified the question he wanted to answer. Rather than asking "How do traits pass from parents to their offspring?" The Law of Independent Assortment states that different traits are inherited completely independently of one another. Yet how could Mendel have deduced this if he had no idea of the mechanism by which traits were inherited? When looking at two traits simultaneously, Mendel observed the ratio of dominant and recessive hybrids for each trait, and discovered that the ratio of traits in the plants from such crosses arose as 9:3:3:1. Namely, 9 offspring showed both dominant traits, 3 offspring showed one dominant and one recessive trait, 3 offspring showed the complementary dominant and recessive mix, and 1 of every 16 progeny showed both recessive traits. As an example, consider the experiment in which Mendel crossed a plant with yellow round peas to a plant with green wrinkled ones. Just as dictated by his First Law of Segregation, Mendel observed that all the F1 progeny from such a cross had yellow round peas the two dominant traits. Next, Mendel self-pollinated these F1 progeny and counted yellow round peas, yellow wrinkled ones, green round peas, and 32 green wrinkled ones in the F2 generation. Finding those rare peas with traits completely different from the peas of the F1 plant may have initially surprised Mendel; however, the 9:3:3:1 ratio that Mendel realized is that the mathematics behind the 9:3:3:1. Consider two independent traits each governed by a dominant: If we cross those two ratios, the result of the cross is the 9:3:3:1. A Punnett square seen in Figure 1 can be used to understand these dihybrid crosses. For the pure breeding plant with yellow and round peas, Mendel would have annotated the two dominant factors underlying these traits as YY and RR, respectively. By crossing this YYRR plant to a pure breeding plant with green, wrinkled peas, annotated yyrr, plants heterozygous for each factor YyRr would arise the F1 generation. It does not matter which symbol is listed first, the presence of even one dominant factor gives rise to the dominant trait see our Mendel and Inheritance module. Since each F1 plant has one dominant and one recessive factor for each of the two traits examined, they are called "dihybrids. Mendel produced the F2 generation by self-pollinating the F1 dihybrid plants. Based on a model of independent heredity, Mendel predicted that each of the traits in the dihybrid would be equally represented in the cross. In other words, the combinations of traits that could be mixed to form the F2 generation were: Writing these combinations along the top and side bars of a Punnett square Figure 2 reveals how the phenotype physical appearance ratio of 9:3:3:1. Punnett square illustrating the cross of two independent traits in two dihybrid heterozygous plants. Comprehension Checkpoint The 9:3:3:1. To test his ideas about random and independent segregation of dihybrid factors, he tested the prediction that the combinations of inputs from the F1 dihybrid generation were equally represented, namely four combinations existed: YR, Yr, yR, and yr. He tested this by crossing the dihybrids F1 plants with purebred plants that were doubly recessive for each factor: Given that the purebred plant could donate only one possible genotype yr, Mendel was able to test his hypothesis. The term used to describe such an experiment is a "testcross," and Figure 3 below shows the predicted outcomes from such a cross. Punnett square illustrating the cross of two independent traits in one purebred recessive trait plant left and one dihybrid heterozygous plant top. Confirming his ideas of independent assortment, the outcome of the dihybrid testcrosses exhibited a ratio of 1:1:1:1. Comprehension Checkpoint Mendel devised testcrosses in order to a. Soon inheritance patterns in other organisms were investigated. By , animals were shown by William Bateson to inherit traits in Mendelian

fashion. Non-Mendelian inheritance and phenotypes arising from multiple factors were later described, but the patterns that Mendel elucidated affected our understanding of heredity profoundly. The cross of two organisms that each possess multiple heterozygous pairs is called a dihybrid cross. Dihybrid crosses result in a trait expression ratio of 9:

**Chapter 3 : Mendel's Law of Inheritance | Genetics**

*ii. Mendel's Experiments and the Laws of Inheritance Mendel chose garden peas as his subjects because they were easily grown and their pollination easily controlled.*

Search term Human genetics Human matings, like those of experimental organisms, show inheritance patterns both of the type discovered by Mendel autosomal inheritance and of sex linkage. Because controlled experimental crosses cannot be made with humans, geneticists must resort to scrutinizing records in the hope that informative matings have been made by chance. Such a scrutiny of records of matings is called pedigree analysis. A member of a family who first comes to the attention of a geneticist is called the propositus. Usually the phenotype of the propositus is exceptional in some way for example, the propositus might be a dwarf. The investigator then traces the history of the phenotype in the propositus back through the history of the family and draws a family tree, or pedigree, by using the standard symbols given in Figure Figure Symbols used in human pedigree analysis. Cavalli-Sforza, Genetics, Evolution, and Man. Many pairs of contrasting human phenotypes are determined by pairs of alleles. Inheritance patterns in pedigree analysis can reveal such allelic determination, but the clues in the pedigree have to be interpreted differently, depending on whether one of the contrasting phenotypes is a rare disorder or whether both phenotypes of a pair are morphs of a polymorphism. Rare inherited disorders are the domain of medical genetics. Medical genetics In the study of rare disorders, four general patterns of inheritance are distinguishable by pedigree analysis: Autosomal recessive disorders The affected phenotype of an autosomal recessive disorder is determined by a recessive allele, and the corresponding unaffected phenotype is determined by a dominant allele. For example, the human disease phenylketonuria is inherited in a simple Mendelian manner as a recessive phenotype, with PKU determined by the allele  $p$  and the normal condition by  $P$ . What patterns in a pedigree would reveal such an inheritance? The two key points are that 1 generally the disease appears in the progeny of unaffected parents and 2 the affected progeny include both males and females. When we know that both male and female progeny are affected, we can assume that we are dealing with simple Mendelian inheritance, not sex-linked inheritance. The following typical pedigree illustrates the key point that affected children are born to unaffected parents: From this pattern, we can immediately deduce simple Mendelian inheritance of the recessive allele responsible for the exceptional phenotype indicated in black. Hence, the pedigree can be rewritten as follows: Note that this pedigree does not support the hypothesis of X-linked recessive inheritance, because, under that hypothesis, an affected daughter must have a heterozygous mother possible and a hemizygous father, which is clearly impossible, because he would have expressed the phenotype of the disorder. Notice another interesting feature of pedigree analysis: In the preceding example, we see a 1: If the couple were to have, say, 20 children, the ratio would be something like 15 unaffected children and 5 with PKU a 3: The pedigrees of autosomal recessive disorders tend to look rather bare, with few black symbols. A recessive condition shows up in groups of affected siblings, and the people in earlier and later generations tend not to be affected. To understand why this is so, it is important to have some understanding of the genetic structure of populations underlying such rare conditions. By definition, if the condition is rare, most people do not carry the abnormal allele. Furthermore, most of those people who do carry the abnormal allele are heterozygous for it rather than homozygous. The basic reason that heterozygotes are much more common than recessive homozygotes is that, to be a recessive homozygote, both parents must have had the  $a$  allele, but, to be a heterozygote, only one parent must carry the  $a$  allele. Geneticists have a quantitative way of connecting the rareness of an allele with the commonness or rarity of heterozygotes and homozygotes in a population. They obtain the relative frequencies of genotypes in a population by assuming that the population is in Hardy-Weinberg equilibrium, to be fully discussed in Chapter A numerical example illustrates this concept. Hence, for this example, we see that heterozygotes are times as frequent as disease sufferers, and, as this ratio increases, the rarer the allele becomes. The relation between heterozygotes and homozygotes recessive for a rare allele is shown in the following illustration. Note that the allele frequencies  $p$  and  $q$  can be used as the gamete frequencies in both sexes. The formation of an affected person usually depends on the chance union of

unrelated heterozygotes. However, inbreeding mating between relatives increases the chance that a mating will be between two heterozygotes. An example of a marriage between cousins is shown in Figure You can see from Figure that an ancestor who is a heterozygote may produce many descendants who also are heterozygotes. Hence two cousins can carry the same rare recessive allele inherited from a common ancestor. For two unrelated persons to be heterozygous, they would have to inherit the rare allele from both their families. Thus matings between relatives generally run a higher risk of producing abnormal phenotypes caused by homozygosity for recessive alleles than do matings between nonrelatives. For this reason, first-cousin marriages contribute a large proportion of the sufferers of recessive diseases in the population. Figure Pedigree of a rare recessive phenotype determined by a recessive allele *a*. Gene symbols are normally not included in pedigree charts, but genotypes are inserted here for reference. Note that individuals II-1 and II-5 marry into the family; they are assumed more

What are some examples of human recessive disorders? PKU has already served as an example of pedigree analysis, but what kind of phenotype is it? PKU is a disease of processing of the amino acid phenylalanine, a component of all proteins in the food that we eat. Phenylalanine is normally converted into tyrosine by the enzyme phenylalanine hydroxylase: Therefore phenylalanine builds up in the body and is converted instead into phenylpyruvic acid, a compound that interferes with the development of the nervous system, leading to mental retardation. Babies are now routinely tested for this processing deficiency at birth. If the deficiency is detected, phenylalanine can be withheld by use of a special diet, and the development of the disease can be arrested. Cystic fibrosis is another disease inherited according to Mendelian rules as a recessive phenotype. The allele that causes cystic fibrosis was isolated in , and the sequence of its DNA was determined. This has led to an understanding of gene function in affected and unaffected persons, giving hope for more effective treatment. Cystic fibrosis is a disease whose most important symptom is the secretion of large amounts of mucus into the lungs, resulting in death from a combination of effects but usually precipitated by upper respiratory infection. The mucus can be dislodged by mechanical chest thumpers, and pulmonary infection can be prevented by antibiotics; so, with treatment, cystic fibrosis patients can live to adulthood. The disorder is caused by a defective protein that transports chloride ions across the cell membrane. The resultant alteration of the salt balance changes the constitution of the lung mucus. Albinism, which served as a model of allelic determination of contrasting phenotypes in Chapter 1 , also is inherited in the standard autosomal recessive manner. The molecular nature of an albino allele and its inheritance are diagrammed in Figure This diagram shows a simple autosomal recessive inheritance in a pedigree and shows the molecular nature of the alleles involved. In this example, the recessive allele *a* is caused by a base pair change that introduces a stop codon into the middle of the gene , resulting in a truncated polypeptide. The mutation , by chance, also introduces a new target site for a restriction enzyme. Hence, a probe for the gene detects two fragments in the case of *a* and only one in *A*. Other types of mutations would produce different effects at the level detected by Southern, Northern, and Western analyses. Figure The molecular basis of Mendelian inheritance in a pedigree. In all the examples heretofore considered, the disorder is caused by an allele for a defective protein. This situation is called haplosufficiency. MESSAGE In human pedigrees, an autosomal recessive disorder is revealed by the appearance of the disorder in the male and female progeny of unaffected persons. Autosomal dominant disorders Here the normal allele is recessive, and the abnormal allele is dominant. It may seem paradoxical that a rare disorder can be dominant, but remember that dominance and recessiveness are simply properties of how alleles act and are not defined in terms of how common they are in the population. A good example of a rare dominant phenotype with Mendelian inheritance is pseudoachondroplasia , a type of dwarfism Figure If this is true, all the dwarf individuals are heterozygotes. Figure The human pseudoachondroplasia phenotype, illustrated by a family of five sisters and two brothers. The phenotype is determined by a dominant allele, which we can call *D* , that interferes with bone growth during development. Most members of the human population more

In pedigree analysis, the main clues for identifying a dominant disorder with Mendelian inheritance are that the phenotype tends to appear in every generation of the pedigree and that affected fathers and mothers transmit the phenotype to both sons and daughters. Again, the equal representation of both sexes among the affected offspring rules out sex-linked inheritance. The phenotype appears in every generation because generally the abnormal allele carried by a

person must have come from a parent in the preceding generation. Abnormal alleles can arise de novo by the process of mutation. This event is relatively rare but must be kept in mind as a possibility. A typical pedigree for a dominant disorder is shown in Figure . Once again, notice that Mendelian ratios are not necessarily observed in families. Therefore, when the progeny of such matings are totaled, a 1:1 ratio is observed. Figure Pedigree of a dominant phenotype determined by a dominant allele A. In this pedigree, all the genotypes have been deduced. Huntington disease is an example of a disease inherited as a dominant phenotype determined by an allele of a single gene. The phenotype is one of neural degeneration, leading to convulsions and premature death. However, it is a late-onset disease, the symptoms generally not appearing until after the person has begun to have children. Figure Each child of a carrier of the abnormal allele stands a 50 percent chance of inheriting the allele and the associated disease. This tragic pattern has led to a great effort to find ways of identifying people who carry the abnormal allele before they experience the onset of the disease. The application of molecular techniques has resulted in a promising screening procedure. Figure The age of onset of Huntington disease. The graph shows that people carrying the allele generally do not express the disease until after child-bearing age. Some other rare dominant conditions are polydactyly (extra digits) and brachydactyly (short digits), shown in Figure , and piebald spotting, shown in Figure . Figure Some rare dominant phenotypes of the human hand.

**Chapter 4 : Genetics: Mendel and Beyond**

*Experiment 8 (Lab Periods 9 and 12) Genetics and the analysis of inheritance Genetics is the study of inheritance that traces its scientific beginning to the work.*

Mendel and Beyond I. The Foundation of Genetics What is genetics? It is the study of inheritance, and the mechanisms by which traits are passed from generation to generation. The foundation for the science of genetics was laid in 1866, when Gregor Mendel used varieties of peas to conduct experiments on inheritance. Gregor Mendel was a monk with scientific training in mathematics, physics, and biology. One of his great legacies was proposing and demonstrating that the characteristics of organisms are inherited in, or as, discrete units particulate inheritance from their parents. His theory was ignored, until the turn of the twentieth century, perhaps because his biological peers were not accustomed to reviewing quantitative or mathematical data. Some other popular concepts of inheritance include: In addition, plants can be grown in large quantities and produce large numbers of offspring. Mendel proposed that the units responsible for inheritance were discrete particles. They existed within an organism in pairs that are separated during gamete formation. This is called the particulate theory, which is in sharp contrast to the blending theory. When fertilization occurs, pairs of inheritable units are reestablished by receiving one copy from each parent. Mendel arrived at the law of segregation with no knowledge of meiosis or of the existence of chromosomes. A Punnett square will be drawn in class to demonstrate this. Mendel verified his hypothesis by performing a test cross. The second law describes the outcome of dihybrid two character crosses, or hybrid crosses involving more than two traits. A dihybrid is an individual that is a double heterozygote. Random fertilization of gametes yields the outcome visible in the Punnett square. The separation of homologs during anaphase I accounts for the law of segregation. The random alignment of homologous pairs along the metaphase I plate accounts for the law of independent assortment. This model works for genes that are on separate nonhomologous chromosomes, but not necessarily for genes that are on the same chromosome. The carefully picked characters used by Mendel were examples of complete dominance. Complete dominance occurs when only one allele accounts for the phenotypic expression in heterozygotes. In heterozygotes the unexpressed allele is called recessive. Beyond Mendelian Genetics A. The segregation of alleles is prevented if homologous chromosome pairs fail to separate during meiosis I. This can prevent independent assortment. Linked genes are on the same chromosome. Genes that are on the same chromosome can be inherited together, but crossing over during meiosis may separate them. Many organisms have homologous pairs of all chromosomes except for those that determine sex. The chromosomes that occur as homologous pairs in all organisms of a species are called autosomes. Chromosome pairings that can vary depending on the sex of an organism are called sex chromosomes. In most vertebrates, including humans, the X and y chromosome are the sex chromosomes. Females with XX are diploid for X-linked genes; On the other hand, XY males are haploid with respect to the genes present on the different X and y chromosome. Males are haploid with respect to the genes on the sex chromosomes, a condition called hemizygous. This generates a special type of inheritance called sex-linked. Sex determination can be very different for different species. The probability for a male of having an X-linked genetic disease caused by a mutant recessive allele is much higher than it is for a female. Because men have only one X chromosome, they express the genes it contains whether they are dominant or recessive. The probability for a man of having an X-linked recessive genetic disease is the same as the frequency of that allele in the population. For these reasons X-linked recessive phenotypes appear much more often in males than in females. The most common forms of muscular dystrophy and hemophilia, as well as red-green color blindness, are a few X-linked human phenotypes caused by recessive alleles. These are traits expressed only in males or females, but not both. Sperm production in males and egg production by females are examples of sex-limited traits. Some traits appear in both males and females, but differ in how they are expressed. Patterned baldness is controlled by the same allele pair in both sexes, but the allele is dominant in males and recessive in females. Obviously other factors genes are involved in the expression of this trait. Differences between alleles of a gene pair are the result of slight differences in their DNA sequence, which results in

slightly different protein products being produced by each allele. The expression of some alleles cannot be accurately described as simply dominant or recessive. There may be many alleles for a single character, or a single allele may have multiple phenotypic effects. New alleles arise only by mutation. Different alleles exist because any gene is subject to mutation, or change, into a stable, heritable new form. Alleles can randomly mutate to become a different allele depending on DNA sequence changes. Although an individual will have only two alleles for a given gene, more than two alleles for that gene may be present in the population.

**Chapter 5 : VIRTUAL GENETICS LAB**

*2 Genetics & Inheritance Lab Work in groups of two This lab is designed to demonstrate genetics, or the study of how heritable characteristics are passed from generation to generation.*

From time immemorial, the phenomenon of the transmission of parental characters into the offspring has fascinated the imagination of human mind. How transmission of characters take place from generation of humankind. How transmission of characters take place from generation to generation was remained a mystery of life until the work of Mendel was re-discovered. First scientific explanations on the fact were given by Gregor Johann Mendel. Mendel has explained the entire mechanism of inheritance while conducted the experiments on cross hybridization among different varieties of garden pea. He conducted his experiment for seven ears and published the work under the annual proceedings of the "Natural history society of brunn in ". But his experimental papers was heavily criticized and it was heard heaved to left benefit. Mendel died in and up to almost 16 years no one paid attention to his work. The findings of Mendel were enlightened in terms of laws of inheritance. This law states that, "Paired factors or alleles segregate from each other during forming gametes and pass into different gametes so that a gamete carries either of the dominant or the recessive factor but never the both. Principle of unit character. Principle of purity of gametes or segregation. Principle of unit character: Phenotypic or visible characteristic an individual organism is determined by at least two mendelian factors called as genes. Principle of unit characters signify that, each character of an organism is carried and transmitted through a specific unit factor called genes. Thus genes are unit of character. One character is governed by a pair of genes which called unit of character. For example Tallness phenotypic trait is carried by "TT" factors or genotype and dwarfness is carried by "tt" genotype. Monohybrid experiment Mendel in his monohybrid experiment selected a pair of contrasting characters or alleles in pea variety one is "Tall" and other is dwarf with genotype "TT" and "tt" respectively. He made artificial crosses between these two varieties. In first filial F1 generation he got the offsprings all having tall character. No dwarf plant is generated. Principle of dominance says that, the character in hybrid which is phenotypically expressed is dominant allele over the character which has not phenotypic expression or suppressed. In above cross tall character is called as dominant allele and dwarf is called recessive allele. Mendel further suggested that, the factor for domination of a character when come in combination of recessive factor the dominant factor masks over the recessive factor. When the recessive factors remain alone, only they ability to be expressed phenotypically. One allele is dominant over its co allele in a hybrid. Principle of purity of gametes: While Mendel made cross between a pure tall variety of pea plant and a pure dwarf variety, he got all tall plants in F1 generation which are hybrids containing both dominant and recessive alleles, and called as heterozygotes. For F2 second filial generation he took F1 hybrids as the parental generation and made crossed between the gametes produced by the hybrid. From above experiment it was found out that when F1 hybrids tall produce its gametes the alleles are segregated from each other. The gamete of F2 generation carries either one of the factor for tallness or dwarfness. The genotype of F2 gametes is either "T" or "t". This is due to the separation of maternal and paternal genes during meiosis. Thus gametes are produced from a hybrid always genetically pure. A gamete may carry either recessive or dominant factor, not the both. Therefore in F2 recombination ratio of dominant and recessive characters appeared in the ratio of 3:1. Later the cases varied in number of plants and animals and it become universalized that no gamete will have either homologous chromosomes or both allelic forms. The law of independent assortment was derived from dihybrid cross experiment done by Mendel on pea plants. The law states that. When the gametes are formed from the parents having two pairs of contrasting characters the members of the different pair of factors or genes segregate quite independent of one another and the factors are recombined independently to produce all possible recombination among the progeny. When two pairs of contrasting characters are taken in a parental generation then only dominant character appears in F1 generation. But upon inbreeding or self pollination in F2 generation the characters are assorted independently. Mechanism of independent assortment was clearly explained in the following experiment done by Gregor Mendel. In the dihybrid cross between two

pea varieties i. From above experiment following conclusions are draw to deduce the law of independent assortment. In F1 generation the offspring obtained are all hybrids showing round seeded with yellow cotyledon having genotype. Here all four Mendelian factors fro yellow and green colouration round and wrinkle shape. But in combination only dominant alleles are phynotipically expressed. While these hybrids were subjects to be self pollinated during formation of gametes the factors are segregated in mendelia fashion so that four types of gametic recombination are obtained. During F2 generation when these four types of gametes unite in fertilization then there are four possible recombination appeared in offsprings. The factors randomly and independently recombined to produce four types of phenotypesin sixteen recombinations in the ratio of 9: Thus in the above cross alleles of one set behaved independent with respect to those of the other set at the time of combination during fertisation. In fact, round character appeared in combination with green and wrinkled character appeared in combination with yellow characters. Thus law of independent assortments justifies an applicable to all type of dihybrid crossed in plants and animals.

*Biology Inheritance Principle part 6 (Mendel's Experiment: Monohybrid cross) class 12 XII.*

You are an adult human, or nearly so, composed of hundreds of different types of cells. Each of these cell types has a different structure and function which together make up you as an individual. Millions of chemical reactions are taking place inside these cells, all carefully coordinated and timed. Yet, you started life as one single cell, a zygote, the result of the fusion of a sperm and an egg. How does all this remarkable complexity come about? These questions had perplexed scientists and non-scientists alike for thousands of years, and they were addressed through a series of very clever experiments in the early part of the 20th century. The chemical basis of heredity

In the mid century, Gregor Mendel completed his now classic experiments on genetics see our Mendel and Inheritance module. Mendel proposed that the "characters" that controlled inheritance exhibited certain patterns of behavior. Specifically, they seemed to operate in pairs and separated independently during reproduction. Scientists were convinced that the basis of genetics and heredity could be found somewhere in the chemistry of our cells. In the early s, scientists began to focus on a recently discovered structure in cells called chromosomes named by Walther Flemming from the Greek words for "colored bodies" because they selectively absorbed a red dye that Flemming used to color cells. Gradually, scientists began to suspect a connection between chromosomes and heredity. Microscopic view of chromosomes lining up red circles at top and separating red circles at bottom during mitosis cell division in an onion root tip. While biologists were becoming convinced that chromosomes were the physical seat of genetics and inheritance, chemists were claiming that these structures were made of both protein and DNA. So, which was the genetic molecule housing all the hereditary information? Many scientists of the day actually thought it was protein because there are 20 different amino acids for building a protein polymer, while DNA polymers are made of only four nucleotide bases. Consider it this way: The genetic molecule works like a language for storing information consisting of words that are made of individual "letters. Imagine making a language using only four letters! Thus, because it offers far more complexity, most scientists in the early 20th century believed that protein was the component of chromosomes that housed the genetic information. Regarding the DNA, they thought that perhaps it acted as structural support for the chromosomes, like the frame of a house. Griffith discovers "transformation" Clarification came during the First World War. During the war, hundreds of thousands of servicemen died from pneumonia, a lung infection caused by the bacterium *Streptococcus pneumoniae*. In the early s, a young British army medical officer named Frederick Griffith began studying *Streptococcus pneumoniae* in his laboratory in the hopes of developing a vaccine against it. As so often happens in scientific research, Griffith never found what he was looking for there is still no vaccine for pneumonia, but instead, he made one of the most important discoveries in the field of biology: Griffith had isolated two strains of S. The pathogenic strain looked smooth under a microscope due to a protective coat surrounding the bacteria and so he named this strain S, for smooth. The harmless strain of S. Cartoon depictions of the rough harmless and smooth pathogenic strains of S. Griffith observed that if he injected some of the S strain of S. Next, Griffith noticed that if he applied heat to the S strain of bacteria, then injected them into mice, the mice would no longer get sick and die. He thus hypothesized that excessive heat kills the bacteria, something that other scientists, including Louis Pasteur, had already shown with other types of bacteria. He mixed living R bacteria which are not pathogenic with heat-killed S bacteria, and then he injected the mixture into mice. Surprisingly, the mice got pneumonia infections and eventually died Figure 3. Griffith examined samples from these sick mice and saw living S bacteria. This meant that either the S bacteria came back to life, an unlikely scenario, or the live R strain was somehow "transformed" into the S strain. Thus, after repeating this experiment many times, Dr. Griffith named this phenomenon "transformation. One strain of bacteria, in this case the R strain of S. Scientists around the world began repeating this experiment, but in slightly different ways, trying to discover exactly what was happening. It became clear that, when the S bacteria are killed by heat, they break open and many substances are released. Something in this mixture can be absorbed by living bacteria, leading to a genetic transformation. But because the mixture contains protein,

RNA , DNA , lipids , and carbohydrates, the question remained "which molecule is the "transforming agent? These scientists did almost exactly what Griffith did in his experiments but with the following changes. First, after heat-killing the S strain of bacteria , the mixture was separated into six test tubes. Thus, each of the test tubes would contain the unknown "transforming agent. To the other five tubes, one of the following enzymes was added: RNase, an enzyme that destroys RNA ; protease, an enzyme that destroys protein ; DNase, an enzyme that destroys DNA ; lipase, an enzyme that destroys lipids ; or a combination of enzymes that breaks down carbohydrates. The theory behind this experiment was that if the "transforming agent" was, for example, protein " the transforming agent would be destroyed in the test tube containing protease, but not the others. Thus, whatever the transforming agent was, the liquid in one of the tubes would no longer be able to transform the S. When they did this, the result was both dramatic and clear. The liquid from the tubes that received RNase, protease, lipase, and the carbohydrate-digesting enzymes was still able to transform the R strain of pneumonia into the S strain. However, the liquid that was treated with DNase completely lost the ability to transform the bacteria Figure 4. Thus, it was apparent that the "transforming agent" in the liquid was DNA. To further demonstrate this, the scientists took liquid extracted from heat-killed S. These results provided powerful evidence that DNA, and not protein , was actually the genetic material inside of living cells.

**Comprehension Checkpoint** Which agent transformed one strain of bacteria into another? Eight years after the famous Avery, MacLeod, and McCarty experiment was published, two scientists named Alfred Hershey and Martha Chase performed an entirely different type of genetic experiment. For their experimental system , they selected an extremely small virus called a bacteriophage or just phage , which only infects bacterial cells. At that time, scientists knew that when these phage infect a bacterial cell, they somehow "reprogram" the bacterium to transform itself into a factory for producing more phage. They also knew that the phage itself does not enter the bacterium during an infection. Rather, a small amount of material is injected into the bacteria and this material must contain all of the information necessary to build more phages. Thus, this injected substance is the genetic material of the phage. Hershey and Chase designed a very simple experiment to determine which molecule , DNA or protein , acted as the genetic material in phages. To do this, they made use of a technique called radioactive labeling. In radioactive labeling, a radioactive isotope of a certain atom is used and can be followed by tracking the radioactivity. Radioactivity is very easily detected by laboratory instruments, even back in the 1940s, and remains a very common tool in scientific research. So, what Hershey and Chase did was to grow two batches of phage in their laboratory. One batch was grown in the presence of radioactive phosphorus. The element phosphorus is present in large amounts in DNA, but is not present in the proteins of bacteria and phage. Thus, this batch of phage would have radio-labeled DNA. The second batch of phage was grown in the presence of radioactive sulfur. Sulfur is an element that is often found in proteins, but never in DNA. Thus, the second batch of phage would have radio-labeled proteins. Then, Hershey and Chase used these two batches of phage separately to infect bacteria and then measured where the radioactivity ended up. What they observed was that only those bacteria infected by phage with radio-labeled DNA became radioactive, bacteria infected by phage with radio-labeled protein did not. Thus Hershey and Chase concluded that it is DNA, and not protein, that is injected into the bacteria during phage infection and this DNA must be the genetic material that reprograms the bacteria.

**Comprehension Checkpoint** Hershey and Chase used radioactive phosphorus in their experiment because a. The blueprint of life Taken together, these experiments represented strong evidence that DNA is the genetic material. Other scientists later confirmed these results in many different kinds of experiments, including showing that eukaryotic , and even human cells can be "transformed" by the injection of DNA. The result of these findings was to convince the scientific and lay communities that the molecule of heredity is indeed DNA. It turns out that the initial instincts of many scientists were exactly backward: They assumed that protein was the genetic material of chromosomes and DNA merely provided structure. The opposite turned out to be true. The DNA molecule houses genetic information, and proteins act as the structural framework of chromosomes. The discovery that DNA was the "transforming agent" and the genetic component of human chromosomes was one of the greatest discoveries of science in the 20th century. However, the mechanism of how DNA codes for genetic information was initially a complete mystery and became the focus of intense scientific study see our DNA II module. Still

today, the study of how DNA functions comprises an entire discipline of science called molecular biology. Originally an offshoot of biochemistry, the field of molecular biology joins biologists, chemists, anthropologists, forensic scientists, geneticists, botanists, and many others who are working to shed light onto the immense complexity of DNA, the so-called blueprint of life. Summary This module is the first in a series that discusses the discovery, structure, and function of DNA. Key experiments are discussed: Key Concepts It required numerous experiments by many scientists to determine that DNA, and not protein, is the genetic material on which life is built.

**Chapter 7 : Brief Essay on the Laws of Inheritance based on Mendel's Experiment**

*LAB 9 - Principles of Genetic Inheritance Overview In this laboratory you will learn about the basic principles of genetic inheritance, or what is commonly referred to as "genetics".*

Difference between Qualitative Inheritance and Quantitative Inheritance are as follows! It is the type of inheritance in which a single dominant gene influences a complete trait. Presence of two such dominant genes does not alter the phenotype. The genes in which dominant allele expresses the complete trait are called monogenes, e. Intermediate forms or continuous trait variations are not produced. Quantitative Inheritance Polygenic Inheritance: The genes involved in quantitative inheritance are called polygenes. Quantitative inheritance is, therefore, also called polygenic inheritance. It is also named as multiple factor inheritance. A few instances of quantitative inheritance are kernel colour in wheat, cob length in Maize, skin colour in human beings, human intelligence, milk and meat yield in animals, height in human beings and several plants, yield of crop plants including size, shape and number of seeds or fruits per plant. A polygene is defined as a gene where one dominant allele controls only a unit or partial quantitative expression of a trait. It is also termed as a gene in which a dominant allele individually produces a slight effect on the phenotype but in the presence of similar other dominant allele controls the quantitative expression of a trait due to cumulative effect. Hence, polygenes are also called cumulative genes. The traits controlled by quantitative inheritance are sometimes known as metric traits because they can be measured in terms of unit of size, height, weight or number. Here a cross between two pure breeding parents does not produce dominant trait of one parent but instead an intermediate trait is exhibited. Similarly in F<sub>2</sub> generation apart from the two parental types there are several intermediate types which link the two parental traits. Because of the latter, quantitative inheritance is also called blending inheritance. Quantitative or polygenic inheritance was first studied by J. Kolreuter in case of height in tobacco and F. Galton in case of height and intelligence in human beings. Nilsson-Ehle obtained the first experimental proof of polygenic inheritance in case of kernel colour in wheat. Polygenic inheritance occurs in case of plant height, crop yield, milk yield, intelligence height and skin colour in humans. It is easily influenced by environment can be known from the frequency distribution of phenotypes. In monogenic or qualitative inheritance the phenotypes are two 3: In polygenic or quantitative inheritance the number of phenotypes is 3 1: Thus we see that the number of intermediate types increases with the increase in the number of polygenes but the number of parental types remain the same 2 in the above cases. The possible origin of polygenes is:

**Chapter 8 : DNA I | Biology | Visionlearning**

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Dominance is the ability of the allele to express itself even in the presence of its recessive allele. It does not require another similar allele to produce its effect on the phenotype. Recessive means the inability to express its effect in the presence of a dominant allele. It produces its phenotype effect only in the presence of a similar allele. It is pure for a trait and breeds true, i. Both the alleles of a character are similar. It is seldom pure and produces offspring with different genotypes on selfing. TT, Tt and tt on selfing of Tt individuals. It carries dissimilar alleles. Tt c Dihybrid i. It is a cross between two pure organisms in order to study the inheritance of a single pair of alleles. It produces a phenotypic monohybrid ratio of 3: It is a cross between two pure organisms of a species in order to study the inheritance of two pairs of alleles. It produces a phenotypic dihybrid ratio of 9: A diploid organism is heterozygous for 4 loci, how many types of gametes can be produced? Explain the Law of Dominance using a monohybrid cross. A monohybrid cross refers to hybridisation where two forms of a single trait are brought together. Mendel crossed a pure tall pea plant with a pure dwarf pea plant and obtained an F1 generation with all tall pea plants. He noticed that in the F1 generation, only one of the contrasting characters tallness appears and the others remained hidden dwarf. The character which appears in F1 individuals is called dominant character, and the character which is not expressed in F1 individuals is known as recessive. He intercrossed the F1 hybrids and obtained the hybrids of the second filial generation F2. In the F2 generation, the tall and dwarf plants were obtained in the ratio of 3: Genetically, this ratio was recognised in three groups of plants TT, Tt and tt with the ratio 1: Define and design a test cross. A test cross is one in which an individual with an unknown dominant phenotype is crossed with a recessive individual for that trait. So, it is helpful in knowing the genotype whether it is homozygous or heterozygous for the dominant trait of an unknown individual. If the unknown is homozygous tall TT, then crossing with dwarf recessive tt gives all tall offspring Tt. Using a Punnett square, workout the distribution of phenotypic features in the first filial generation after a cross between a homozygous female and a heterozygous male for a single locus. When a cross is made between a tall plant with yellow seeds TtYy and a tall plant with green seeds Tt yy, what proportions of phenotypes in the offspring could be expected to be a Tall and green b Dwarf and green Solution: Two heterozygous parents are crossed. If the two loci are linked, then what would be the distribution of phenotypic features in the F1 generation for a dihybrid cross? If two loci are linked i. The F1 generation produces three phenotypes as in monohybrid cross, i. Briefly mention the contribution of T. Morgan carried out several dihybrid crosses in Drosophila and observed that the two genes did not segregate independently of each other and the F2 ratio deviated significantly from 9: Morgan and his group observed that genes were located on the X-chromosome and found the physical association or linkage of genes. He coined the terms linkage and recombination to describe the generation of non-parental gene combinations. Morgan and his group also found that when genes were grouped on the same chromosome, some genes were tightly linked showed low recombination while others were loosely linked showed higher recombination. What is pedigree analysis? Suggest how such an analysis can be useful? A record of inheritance of certain genetic traits for two or more generations presented in the form of a diagram or family tree is called a pedigree. Analysis of traits in several generations of a family is called a pedigree analysis. It is used in the case of human beings and domesticated animals. How is sex determined in human beings? Human beings have 22 pairs of autosomes and one pair of sex chromosomes. The sex of the offspring is determined at the time of fertilisation. A child has blood group O. If the father has blood group A and mother blood group B, work out the genotypes of the parents and the possible genotypes of the other offspring. Given, the father has blood group A, the mother has blood group B and the child has blood group O. O is a recessive blood group because of IOIO. Explain the following terms with examples: The expression of the trait in the F1 hybrid is intermediate or a fine mixture of the expression of the two factors. Pink flowers in pea plant. What is point

mutation? Point mutation is the abrupt change in gene structure due to a change in a single base pair of DNA caused by inversion and substitution without changing the reading of subsequent bases. This defect is caused by the substitution of glutamic acid by valine at the sixth position of the beta globin chain of haemoglobin. Who proposed the chromosomal theory of inheritance? Sutton and Boveri proposed the chromosomal theory of inheritance in Mention any two autosomal genetic disorders with their symptoms. Symptoms of sickle cell anaemia: The red blood cells of the patient become elongated and curved sickle-shaped under low O<sub>2</sub> tension. The sickle-shaped red blood cells are destroyed more rapidly than the normal ones leading to anaemia. The affected individual shows the following characteristics:

**Chapter 9 : Inheritance - Similarity and Difference, Reproduction, Mendels Experiments**

*The term used to describe such an experiment is a "testcross," and Figure 3 below shows the predicted outcomes from such a cross. Figure 3: Punnett square illustrating the cross of two independent traits in one purebred recessive trait plant (left) and one dihybrid heterozygous plant (top).*

**Mendel and Inheritance** What is heredity? Heredity is when certain traits are passed from the parents to the children. Traits are characteristics such as eye color, height, and athletic ability. Heredity is passed through genes in the DNA molecule. In biology the study of heredity is called genetics. Gregor Mendel Scientist Gregor Mendel - is considered the father of the science of genetics. Through experimentation he found that certain traits were inherited following specific patterns. Gregor studied inheritance by experimenting with peas in his garden. Peas work as an excellent test subject as they can self-pollinate, cross fertilize, and have several traits that only have two forms. This enabled Mendel to easily control his experiments and reduced the possibility of the outcomes to something he could record and manage. First he produced a parent generation of true-breeding plants. He made these by self-fertilizing the plants until he knew they bred true to the seven traits. For example, the purple flowering plants always produced seeds that made purple flowers. He called these plants the P generation for parent. Next, he produced a second generation of plants F1 by breeding two different true-breeding P plants. He then produced a third generation of plants F2 by self-pollinating two F1 generation plants that had the same traits. F1 Generation Mendel found that the F1 generation all produced the same trait. Even though the two parents had different traits, the offspring always had the same trait. For example, if he bred a P plant with a purple flower with a P plant with a white flower, all of the offspring F1 plants would have purple flowers. This is because the purple flower is the dominant trait. These results can be shown in a diagram called a Punnett square. The dominant gene is shown with a capital letter and the recessive gene with a lower case letter. Here the purple is the dominant gene shown with a "P" and the white is the recessive gene shown with a "w".