Cell and Molecular Biology
Prof. Vishal Trivedi
Department of Biosciences and Bioengineering
Indian Institute of Technology, Guwahati
Week 07
Concepts of Genetics (Part 1)
Lecture - 25
Law of Inheritance (Part 1)

Hello, everyone. This is Dr. Vishal Trivedi from the Department of Biosciences and Bioengineering, IIT Guwahati. And in this particular course, so far what we have discussed is the origin of life. We have discussed the evolutions. Then subsequent to that, we have also discussed the cellular structures.

Then we talk about how the transport of the material within the cell happens. And then we also discuss how the cell is communicating with another cell. And then we also discuss the genetic material. Right.

So how is the genetic material varying from the prokaryotic system to the eukaryotic system and so on? Now, this genetic material is very important because it carries the information from one generation to another. Right. I'm sure you might have noticed many particular types of phenotypes within the human species or within organisms. For example, you might have seen the attached earlobes. You might have seen that thumb extends.

You might have seen the headless. You might have seen the dimples, right? So you might have seen the dimples in some of the girls, right, or in boys as well. And then you might have also seen the color blindness. So all these and the many more such types of phenotypes are being controlled by the genetic material that is present inside the cell. And these materials are actually governing what kind of color you are going to exhibit, what kind of hair color you are going to exhibit, whether you are going to exhibit curly hair or straight hair, and so on.

So in today's lecture, we are in this particular module; we are going to discuss how you can study this phenomenon and what the guiding principles are that actually control the phenotypic changes in the phenotypic appearances of a particular organism. Before getting into this, we have to discuss the basic information so that you will be able to understand and follow the content of this very complicated subject. The basic material required to carry the information from one generation to the next is the genetic material. And the genetic material is made up of DNA. And you know that DNA has four different types of nucleotides.

It has adenine, guanine, cytosine, and thymine. And the information is carried from one generation to the next in the form of these four nucleotides, whether it is a tiny bacterium or whether it is a big mammal, like the different types of elephants and so on. So all this

information is encoded in the genetic material, and that's how the genetic material is passed from one generation to the next and how it conveys that particular message to the subsequent generation. I'm sure you might recall that when we were discussing cell division, we said that cell division is also precisely controlled and regulated so that there will be a complete synthesis of this particularly important component of the cell, which is the genetic material. So until the genetic material has been completely synthesized, the cell will not enter the next phase of cell division.

So before we get into the details, we have to understand how this particular field of science is being developed and how people have discovered the different types of rules and laws, and some other related concepts. So the information is in the DNA, right? It determines many types of phenotypes. It determines the eye color, right? It determines the height. It determines even the behavioral tendencies. You might have seen that some people are very short-tempered.

Some people are very kind, and so on. Then it also divides cells; then organ functions can actually work in evolution as well. So genetics is also called the blueprint of life, which means genetics is the field of science that actually studies how information from one generation goes into the next generation. So genetics is the branch of biology that is concerned with the study of heredity and variations. The term genetics was first introduced by Batson in 1906, and it is derived from the Greek word for gene, which means to become or to grow into.

It explores how the traits and characteristics are passed from one generation to another through genes and the fundamental unit of heredity. So genes are the fundamental units of heredity. I am sure you will understand more about these things when we discuss the central dogma and other kinds of things. The field of genetics has a profound impact on newer disciplines like agriculture, medicine, evolutionary biology, and biotechnology. Because once you know how the information goes from one generation to the next, you can change that information.

You can make it better. And that's how the people have developed different types of crop plants, different types of medicines, different types of evolutionary advancements, and so on. The history of genetics dates back to ancient times when early humans unknowingly applied genetic principles to the selective breeding of plants and animals, which began between 10,000 and 12,000 years ago, right? So basically, even when we don't know about the laws of genetics, we don't know anything about the scientific principles. People were doing selective breeding so that they could obtain better breeds and better quality plants and animals. Civilizations like those in Mesopotamia, Egypt, China, and India selected the organisms with desirable traits to improve crop yields and livestock over generations.

In India, the early concept of heredity was first mentioned in ancient texts like the Ayurveda and the Manusmriti, which noted the influence of parental traits on the offspring. Then, not using scientific methods, these observations mark the pre-scientific era of genetics. This is all people were doing: simply conducting observational studies.

They were saying that, okay, some of the bulls are very healthy, they are not getting diseases, and they are very strong in terms of using them for breeding and other kinds of phenotypes. And some of the cows are very good at giving milk, and that's how they started selective breeding, and so on, and that's how they have actually changed the varieties.

So even in ancient Hindu texts from about 2000 years ago, they said that traits actually come from the parents and advised avoiding partners with undesirable traits, showing an early awareness of these traits. So there were marriages that actually avoided one particular type of phenotypic parent and so on. Similarly, the Jews demonstrated an accurate understanding of Haemophilia inheritance, and the Greek philosophers also contributed theories like Pangenesis, influencing genetic thought for centuries. In modern times, India began contributing to genetic research in the early 20th century with pioneers like J.

B.S. Haldane later moved to India and significantly influenced genetic studies. Indian scientists also contributed to agricultural genetics primarily during the Green Revolution, which led to the development of high-yield crops using genetic techniques. So before getting into the complicated subject of how, you know, genetics is being studied and how genetic laws and all that, let's first discuss how this particular field is evolving. You might have seen that I have talked about many different types of examples from the Hindu system or the old Indian system where people were doing selective breeding and so on. But let's take account of how the historical aspects are being developed in the field of genetics.

So, from Mendel to modern genetics, okay, so in 1856, Gregor Mendel conducted his famous P experiment concerning gene segregation, right? So from 1856 to 1863, this is the time when Mendel was conducting experiments with the pea plants, and these are the experiments that are pioneering in terms of providing different types of information and principles on which the famous theories are based. The Mendelian law of genetics was being formulated, and these are what we are going to discuss in detail. So in the year 1856, the Frederick mixture actually isolated the nuclei; the nucleic acid from the nuclei, and nuclein is also known to be DNA, right? So this was the time when the people actually isolated the DNA, right, DNA from the particular nucleus. Then in 1900, Hugo de Vries, Carl Correns, Eric Vaughan, and all these people independently produced results confirming Mendel's principle of heredity. In 1902, Garrard identified the first human genetic disease, and Walter and Boveri actually proposed the chromosomal theory of heredity.

In 1928, the figurative actually performed the famous experiment on transformations. So he did the experiment on the bacteria and collected the agent that is responsible for the transformation of the bacteria from one type to another. And that's how you might have heard about this famous experiment where he infected the mice with different types of bacteria, both heat-killed and live, and so on. And that's how he actually came up with the idea of transforming principles. Then, in the year 1941, Beadle and Tatum proposed the one gene, one enzyme hypothesis.

According to this hypothesis, it states that one gene actually gives rise to one protein, and so basically, that's how people have started talking about the Central Dogma and so on. And then in the year 1952, Hershey and Chase showed that the genetic material of the bacteriophage is DNA. And in the year 1953, Watson and Crick actually saw the structure of the double helix of DNA. And that's how they have actually taken a lot of information from their DNA. And that has paved the way for the idea that DNA can be used for, or be responsible for, the different types of genetic traits.

In the year 1961, Francis Jacob and Matthew Malthus actually discovered messenger RNA. Then, in the year 1966, Nirenberg and Khurana actually worked out the complete genetic codes. In the year of 1972, Paul Berg constructed the first recombinant DNA molecule in vitro. In the year 1977, Walter Gilbert and D. Frederick Sanger actually developed the method for sequencing DNA.

Then in the year 1986, the Carey-Mullers discovered PCR, and that's how they were able to make multiple copies of a particular DNA. It actually helps to generate multiple copies so that you can use this for subsequent recombinant DNA technology. In the year 1997, the Roslin Institute actually cloned the first cloned mammal, called Dolly, from an adult organism using the technique of transgenic cloning. And in the same year, the E. coli genomic sequences were completed.

From the year 1990 to 2003, the Human Genome Project was completed, mapping all the genes and the genetic content of humans, and that's how we know a lot of information about how this particular gene controls skin color, this particular gene controls hair color, and so on, right? And then from 2002 to date, there will be more advancements in terms of genome editing techniques. So whether it is a ring finger technique or whether it is a CRISPR-Cas. And then we also have a lot of integration of computational tools such as AI into genomic research. Now the question is why we would like to study genetics, right? We would like to study genetics because we want to understand how the different types of factors are controlled and how these factors are passed from one generation to the next. So, genetics is the science of heredity, right? The study of biological properties passed from the parents to the offspring, isn't it? It is important that you understand life, and if you want to understand life, it is important that you understand how life is governed, how life processes are governed, and that's why it is important to study genetics.

So understanding the genetics is essential for explaining the inheritance and the variation among individuals. Remember that many of these things were not known when Lamarck and Darwin actually proposed the theory of evolution. And that's how they could not explain many of their hypotheses. Right. Many of the phenomena they have observed.

They could not explain because of the lack of information on how to explain the phenomena of zebras or giraffes having a particular type of phenotype. Then it actually makes the basis of genetic diseases, because if you understand the information, if you can understand who is causing this particular type of phenotype, you can actually understand

all the genetic diseases. And that's how you can actually control and correct these diseases as well. Then it also helps to study evolution, ecology, and population biology. And it will also be applied in agriculture, biotechnology, and medicine.

Some of the examples we are going to take up by the end of this particular course will be covered by the end of this particular module. Then the genetic research also led to rapid diagnostic tools. You can actually have the DNA-based sensors. You can actually have the DNA-based diagnostic and so on. And then it also helps in the development of different types of pharmaceutics.

For example, we discovered human insulin, right? So earlier, until recombinant DNA technology was known, the genes for insulin were not known. People were actually collecting insulin from other sources. And that's how they were actually purifying insulin from pigs and other kinds of animals. And that is the insulin people were using to control diabetes. Then you can also have identically improved crops and livestock, and you can also use this genetic material for studying forensic DNA.

What are the different types of sub-disciplines of genetics? So you can actually have transmission genetics, molecular genetics, population genetics, and then you can also have quantitative genetics. So within transmission genetics, it deals with how the genes and genetic traits are transmitted from generation to generation and how the genes recombine or exchange between the chromosomes. So how are the variations coming from one generation to the next generation? Because there is the exchange of genes between the homologous chromosomes. For example, analyzing the pattern of trade transmission in human pedigree, in the crosses of experimental organisms, and in examples of transmission genetics will allow you to study how genetic information passes from one generation to the next, and in cases of variation, how recombinations and other kinds of phenomena are actually responsible. Who is making the recombination? Then we also have another field which is called molecular genetics.

So molecular genetics deals with the molecular structure and functions of the genes. It is actually also dealing with genomic analysis. Then analyze the molecular events involved in the gene control of cell division or the regulation of expression of all the genes in a genome. Then we also have population genetics. So studying the heredity in a group of individual portraits that are determined by one or only a few genes is correct.

So population genetics is not for studying the genetics of a single individual, but population genetics is about studying a particular phenomenon within a particular population or area, actually. So, for example, if you consider the frequency of disease-causing genes. Right. So that can be very high in a particular area of the city or within the country and so on. And that's the kind of information you will get when you study population genetics.

Then we also have quantitative genetics. So quantitative genetics is considered the heredity of traits in a group of individuals, but the traits of concern are determined by many genes simultaneously. For example, the fruit weight and the crop yield in

agricultural plants are examples of quantitative genetics. Now, before getting into the complicated details of the law of genetics, which has been proposed and formulated by the extensive experiments done by the Mendels, it is important to introduce you to the different types of terms that will be used in explaining those laws. So let's first understand the different types of terms you are going to encounter when we discuss the law.

So the first is traits. Traits are characteristics, right? A characteristic of an individual is called traits, right? It could be like skin color; it could be hair color. It could be eye color, so all of these are actually a part of traits. A heritable trait is transmitted from generation to generation. This is synonymous with the character. Character in terms of visible features, not in terms of moral values or something.

Its character means in terms of the features, like what is the skin color, what is the color of the hair, whether the hair is straight or curly, or whether the hair is oily, and those kinds of things. Right. Examples of collectors include hair color, height, eye color, flower color in plants, and so on. Then we also have heredity or inheritance. The process by which traits are passed down from parents to their offspring is called heredity or inheritance.

It's one reason why your offspring tend to resemble their parents. The transmission of genetic information occurs during reproduction, either sexually or asexually. Then we have the dominant traits. So traits could be of two types.

It could be dominant traits or recessive traits. What are dominant traits? A dominant trait is one that shows up in the organism's appearance; these are the traits that are going to be visible and will appear even if only one copy of the allele is present. Even if some organisms have only one gene, it will actually mask the effects of the recessive alleles. And it is representative for the sake of argument. It has always been represented by a capital word. For example, when you are actually going to have the two different types of trades, one is a dominant trade, and the other is a recessive trade.

The dominant trade has always been written with a capital letter. So it is going to be a capital T for the tall and a lowercase t for the dwarf. So, for example, in a TT heterozygous plant, it is tall because T tall is dominant over T short, or the dwarf. Then we also have recessive traits. So, recessive traits only appear when both copies of the genes are recessive, right? It is marked by the dominant alleles and only shows up when the dominant allele is absent.

Represented by a lowercase letter, for example, the small "t" for the short or the dot. A plant is short only if its phenotype is tt, right? So, recessive traits are only going to show up when they are actually present. If the dominant trait is also present, then it is actually not going to show up. Then the third, we have the haploids or the diploids, right? A cell is an individual with one set of chromosomes; therefore, it is called a haploid. The haploid cell contains one complete set of chromosomes, correct? This means there is only one copy of each chromosome, right? So, for example, if humans actually have 46

chromosomes, right? But these 46 chromosomes are divided into two groups of 23, right? This 23 is a part of the haploid.

This 23 is another part of the haploid. When they come together, it is going to be called diploid. So in sexually reproducing organisms, the gametes, sperm and eggs are typically haploid. This occurs during fertilization, when two haploid gametes are used to form a diploid zygote. So haploid is always represented by a small n, right? And with a single n, whereas diploid is always shown with a 2n, 2n, 3n like that, okay? So the haploid number, or N, is half the diploid number.

In humans, for example, the haploid number is 23. That is what I have shown you, right? Two haploids are coming, and that's how you actually have the 426 number of chromosomes. So a human sperm contains 23 chromosomes, and a human egg actually contains 23 different types of chromosomes. And when they come together, it is actually going to give you a diploid zygote that contains 46 chromosomes. Similarly, we have diploids. Diploid means a diploid cell contains two complete sets of chromosomes, with one set inherited from each parent.

This means there are two copies of each chromosome forming a homologous pair. Now, homologous chromosomes have the same genes in the same order, but may have different versions of those genes. The diploid number is the total number of chromosomes in a somatic cell. In humans, the diploid number is 46, which is two sets of 23 chromosomes.

Then we have the DNA. So DNA, which we have discussed in detail, is the genetic material, and it carries the information in the form of the four nucleotides, which are ATGC. And then we have the chromosomes. A chromosome is a long thread-like structure made up of DNA and proteins, mainly histones, and it carries genetic information in the form of genes. Chromosomes are found in the nucleus of most cells, or chromosomes are present in bacteria.

Each chromosome contains many genes that determine its traits. Humans have 46 chromosomes, 23 pairs, right? There are 22 pairs of autosome chromosomes and one pair of sex chromosomes. So it is XX for females and XY for males. Then we have the genes. So what is meant by the gene? Right. So a gene is a specific segment of DNA located in a particular position on a chromosome, which is called a locus.

Humans have two copies of the gene, one inherited from each parent, located on the homologous chromosomes. Genes are the basic unit of heredity and are located on the chromosome. So, every person has about 20,000 to 25,000 genes, right? Gene is responsible for eye color, isn't he? And the gene is responsible for a person's height. So a gene is actually the main portion of the DNA that is going to carry the information, right? So, for example, in a cell, you are going to have the nucleus; within the nucleus, you are going to have the DNA, and DNA is going to be present in the form of chromosomes, and within the chromosomes, you are going to have the linear DNA. And that linear DNA, a small portion of that linear DNA which may be responsible for providing a particular type of phenotype, is being called a gene.

So a gene is actually the executory molecule or the smallest unit that is actually responsible for a particular phenotype. Then we have the alleles. An allele is a variant or a different form of a gene. While everyone has the same set of genes, the alleles can be slightly different between individuals, leading to different traits. For example, the eye color gene may have different alleles: one for blue eyes and another for brown eyes, right? So if you have the alleles for blue eyes, you are going to have blue eyes.

If you have the alleles for brown eyes, you are going to have brown eyes, which means a single gene, but it is actually going to be modified; there will be variations. Then it is actually going to have different types of alleles, so it can have alleles for blue, alleles for brown, and alleles for, you know, the other kinds of phenotypes, right? So while you have one gene for eye color, you may have inherited one allele from the mother and one allele from the father. So that's how you can actually have the different variations within the populations. Then we have the loci.

So, a locus is the specific physical location of a gene on a chromosome. So loci is kind of an address of a particular gene on the particular chromosome. So we can actually say gene X is on chromosome number 22 like that. So it is actually going to tell you about the position of a particular gene on a particular chromosome. Then we have the genotype.

So, the genetic makeup of an organism is the combination of alleles it carries. For example, you can have the double T, you can have the small t, or you can have a small double T, right? All of these are for the plant height, but the genotype is different, right? In these cases, you have all the chromosomes, right? You have two chromosomes, for example. So both the chromosomes are, if both the chromosomes are actually containing the capital T, then it is going to be written as capital T, capital T. If it is on one chromosome, it is capital T; on the other chromosome, it is small t; then it is going to be called T small t. And then you're also going to have a third type of chromosome where you're going to have small t and small t on both chromosomes, and that's how it is going to be called Tt, right? So this is going to be homozygous, this is going to be homozygous, and this is going to be called heterozygous, right? This is anyway we are going to discuss in the subsequent slides. So, homozygous means both chromosomes contain capital T, capital T or small t, small t.

Whereas heterozygous means one of the chromosomes is going to contain capital T, and the other one is going to be small t. So, you cannot always see the genotype just by looking at which alleles are present, can you? Then we have the phenotype. So, genotype means genetic makeup or genetics, whereas the phenotype means these features, right? For example, you can have brown eyes, you can have blue eyes, and so on. So, the physical expression or the observable traits resulting from the genotype, for example, the tall or short characteristics that you see, are influenced by other genotypes and sometimes by the environment.

Then we have the homozygous and heterozygous genotypes. So homozygous means that you have both genes present on the chromosome of the same type. So if you have alleles

that are of the same type, then it is going to be called homozygous. A homozygous genotype has two identical alleles for a particular gene. For example, the homozygous dominant is represented as capital T, capital T, while the homozygous recessive is represented as small t, small t.

Then we have the heterozygous genotypes. A heterozygous genotype is one where you have two different types of alleles for a particular gene, one dominant and one recessive, which means on one chromosome you will have a capital T and on the other a small t. So this will be recessive. This is going to be dominant. And that's why this particular type of combination or this type of genotype is being called heterozygous. For example, the tt is a small t, right? which carries both dominant and recessive alleles, and the dominant allele is expressed in the phenotype.

Then we have the different types of generations. So, the P generation, which is also going to be called the parental generation. Then we have the F1 generation, which is the first generation, and then the F2 generation, which is called the second generation. So this is the original pair of organisms involved in a genetic cross, and they are usually purebred or homozygous for a particular trait. An example is a monohybrid cross of height in a plant like tt versus TT, which are the P generation.

So these are actually going to be called pure generation or pure organisms. Then we have the F1 generation. So once you breed the P generation. They are actually going to give you the offspring, and that offspring is going to be called the F1 generation. So the offspring of the P generation is going to be called the F1 generation.

These individuals are usually heterozygous and show dominant traits. For example, TT and small t, when they are going to give you the offspring, it is going to be a heterozygous Tt, which is going to be tall. Even though they carry the recessive gene, it is not expressed in the phenotype. Then we have the F2 generation, which is the offspring of a cross between the two F1 individuals. This generation reveals the three to one phenotypic ratio, showing both dominant and recessive traits. For example, if you have t and t, when they crossbreed, it is actually going to give you four different types of offspring.

Capital T, capital T, capital T, small t, capital T, small t, and the small t, small t. This means it is going to give you three talls and one short, right? So this is going to be part of F2. Then we have the cross, right? So we can actually have the different types of cross, which leaves what the parents are or what the plants you are going to take into the cross that is going to decide whether it is a monohybrid cross or a dihybrid cross. A cross refers to the mating or breeding of two individuals to show how traits are inherited. For example, crossing a tall tea plant with a short plant, right? It could be a monohybrid cross.

It could be a dihybrid cross. So a monohybrid cross actually involves one trait with two alleles, right? So it can be only one trade, right? A monohybrid cross means it will deal with one gene, which means it will actually discuss only one phenotype. For example,

tall. So it is only going to talk about this particular type of gene, or it is going to talk about this particular type of genotype. Crossing a tall plant with a short plant.

So, we are only dealing with the one phenotype, which is height. So, when you deal with one phenotype, it is going to be called a monohybrid cross. And when you talk about, for example, having the TT and small t, small t, it is actually going to give you this kind of offspring. So you are going to have a genotypic ratio of one small t, two small t, and one small t. Whereas the phenotypic ratio is going to give you three tall and one short.

Then we have the diabetic cross. So dihybrid cross involves the two traits at the same time. So you are actually going to talk about two individuals, like, for example, the tall one and the color of the flower. Okay, so if you say it like this, then it is actually going to be a dihybrid cross, which means if you are dealing with the two phenomena or the two phenotypes, then it is going to be a dihybrid cross. For example, crossing a pea plant with round yellow seeds with wrinkled green seeds will show all the dominant traits in the first generation, and in the F2 generation, it will show you the different types of phenotypic changes or different types of phenotypic organisms, depending on the different types of yields. Now, since you understand the basic phenomena and have information about the different types of terms, what are you going to use? So, let us now move on and understand the different types of laws that are being formulated to understand how genetic information is passed from one generation to the next, and all these laws are being formulated by the experiments that are being conducted. The pea plants by Gregor John Mendel, right? And so that's why these laws are being called Mendel's law of inheritance.

So before getting into the details of Mendel's law of inheritance, let's first understand the experimental design. So this is Gregor John Mendel, the founder of the science of genetics. The work of Gregor Johann Mendel is considered the foundation of modern genetics. In the year 1843, he was admitted to the monastery in Bro, now in the Czech Republic. In the year 1852, he began a series of breeding experiments with the green Pisum sativum to learn something about the mechanism of heredity.

As a result of his creativity, Mendel discovered fundamental principles of genetics. So from the results of crossbreeding pea plants with different traits involving seed shape, seed color, and flower color, Mendel developed a simple theory to explain the transmission of hereditary traits from generation to generation. Mendel had no knowledge of mitosis or meiosis. So remember that at that time there was no information available about the cells, how cell division works, and so on. And we also don't know about the genes, chromosomes, and all that.

So he did not know that the genes segregated according to the chromosomal behavior. So Mendel reported his conclusion in the year 1856, but its significance was not fully realized until the late 1800s and early 1900s. So Mendel did all his significant genetic experiments with the garden pea, which is called Pisum sativum. The garden pea was a good choice because it fits many of the criteria that make an organism suitable for use in genetic experiments. It is easy to grow, bear flowers, and produce fruits in the same year

when a seed is planted, resulting in a large number of seeds. So basically, the reason Mendel used the pea plant is that it is easy to breed, there are many different types of traits present in the pea plant, and it is easy to collect the seeds and regrow them, and so on.

So crossing the pea plant began with a cross section of a flower showing the stamens and the pistils, and the pea normally reproduces by cell fertilization, which is the entry of the same stamens producing the pollen. and which actually lands on the pistil of the same flower and fertilizes the plants. So basically, the pea plant is doing the self-fertilization within the same flower.

You are actually going to have the statement. You are going to have the pistol. So the pollen grains are going to come out from the stamen and then they are actually going to fall onto the pistil. And that's how it is actually going to perform the fertilization. Fortunately, for the success of his experiment, Mendel was able to prevent the self-fertilization of the pea by removing the stamen. So it is actually going to remove the stamens from a few of the plants, from a few of the flowers.

And that's how you can do the crossbreeding. Because if it is self-breeding, then you will actually not have any control over how the breeding is going to work. Right. So basically what he did is he first, because the pea plants are doing self-fertilization, he actually removed the pollen tube. So it is actually going to pluck all the pollen tubes off the plants.

So that he can actually take out the pollen grains from the plant for which he wants to do the breeding. And then he can actually add those pollen grains to the desirable female plants. So next he took the pollen from the stamen of another flower and dusted it onto the pistil of the encapsulated flower to pollinate it. Cross-fertilization, or simply cross, is the fusion of male gametes from one individual to the female individual of another, right? Once cross-fertilization has occurred, the zygote develops into the seeds, right? Certain phenotypes are analyzed by inspecting the seeds. Others are analyzed by examining the plants that grow from the seeds. The primary work ensured that the Mendel worked only for the peer strains, correct? The traits under investigation remained unchanged from parents to offspring for many generations.

Such strains are called true-breeding or pure-breeding strains. So basically, Mendel used the pea plant and found that the plant has 34 different types of strains that are pure in nature, which means they do not show any kind of deviations or variations. From generation to generation, they are actually showing similar types of phenotypes. And that's how he actually used those phenotypes to understand the basic mechanism of inheritance. What he did is what he explained here, right? So, he removed the stamen from this plant, right? Then he actually took the pistil, so this is the pistil of this particular plant.

Then he took out the pollen grains from another plant, and then he did the cross-fertilization. And that is how he actually produced the, you know, the crossbreeds, and

these crossbreeds he studied to see what kind of phenotype they are actually showing. So, Mendel used the different types of traits, right? As I said, you know, he has used the pea plant which is actually showing the true breeds, right? So, he has used different types of phenotypes. So, what are the different types of phenotypes that he has used? So he has used the flower and seed coat color.

So gray versus white and purple versus white flowers. Then he also used the seed color. So the seed color could be yellow or green. Then he also changed the shape of the seed.

So, seed shape could be smooth or wrinkled. So this is what is shown here. Seed colors should be yellow or green. Seed shape could be smooth or wrinkled. Then we also have the pod's color. So, pod color could be green or yellow, right? So this is the pod color: green or yellow.

Then we also have the pod-shaped. So the pod shape could be inflated or pinched. So this is the pod shape, right? And then we also have the stem's height. So it could be tall or short. So you can have the plants which are tall or the short ones.

And then you also talk about a flower's position. So the flower position could be axial or terminal. So this is the flower's position. You can have either the axial or the terminal position. So these are the seven different types of phenotypic combinations that he has used.

And then he started to study the traits. So first, what he did was make a simple system. Right. A simple system is where you actually study the genetic movement of one particular gene. Right? Or one particular elite. Right? Basically, whenever you design scientific experiments, it is important that you first work with the simple system and then work with the complicated system. So the first thing is that he studied how the genetic information of a single gene moves from one generation to the next.

So, the inheritance of a single gene is how he came up with the first law, which is the law of dominance. So crossing the two different tool breeding strains brings together one factor from each strain in the F1. The eggs, which are haploid, meaning they contain only one set of chromosomes with one factor of one strain, and the pollen grains, which are also haploid, meaning they contain one set of chromosomes with the one factor from the other strain. Furthermore, because one of the traits was seen in the F1 generation, the expression of the missing trait must have been masked by the visible trait.

This masking is called dominance, right? So for the smooth versus wrinkled, when he crossed, the F1 seeds were all smooth. Right. So in the log dominance, what he did was crossbreed single traits between the contrasting features. Right. So what he did is smooth rather than wrinkled. And what he found in the F1 generation is that it is all smooth, which has appeared. Thus, the alleles for smoothness are masking or dominant to the alleles for wrinkledness, and the smooth seed trait is considered to be the dominant trait, with the allele associated with it called the dominant allele.

Conversely, the wrinkled trait is recessive to smooth because the factor for wrinkled is masked, and the wrinkled seed trait is considered to be a recessive trait, with the allele associated with it referred to as the recessive trait. So when the two alleles are different, one dominates the phenotype as well. So this is what is shown here, correct? So if you take the two pure breeds, one is round, the other is wrinkled, right? So you take the seeds that are round and smooth versus wrinkled, right? Then in the F1 generation, it is only going to show you the seeds that are smooth, right? That says that the smooth is a dominant trait and the wrinkled is a recessive trait. The genotype of the parent grown from the smooth seed is ss, then the small ss small cell is the genotype of the parent grown from the wrinkled seeds, right? An individual that contains two copies of the same allele of a particular gene is considered to be homozygous. When the deployed plant produces the haploid gamete by meiosis, each gamete contains only one copy of the gene, which is either the S-bearing gametes, and the plant from the wrinkled variety is going to contain the small S-bearing gametes.

So when the gametes fuse, they actually produce the diploid zygote, which has one small capital S and one small S allele, and the genotype would be small S capital S. Plants that have two different alleles of a particular gene are said to be heterozygous because of the dominance of the smooth capital S allele. The small capital S and small s produce the smooth seeds. So basically, in a genotype, you are going to have the dominant alleles, or you're going to have the recessive alleles, right? So this is going to be the capital S, capital S, which is going to be a dominant allele, right? And this is going to be a small s, which is going to be a recessive allele, and when both of these are present, then it is actually going to be, you know, the capital S that is going to dominate over the small s, and that is how it is actually going to produce, you know, the smooth teeth.

So dominant and recessive alleles of a gene determine the seed shape in the P generation. So the two breeding smooth seed plants have the genotype SS, and the two breeding single seed plants have the genotype ss. Because each parent is two breeding and deployed, each must contain two copies of the same alleles. And all the F1 plants produce the smooth seed and are called S, small s, heterozygous. Then we have the law of segregation.

So the plant grown from the F1 seeds differs from the smooth parents in that it produces an equal number of 2L types of gametes. S, capital S-bearing gametes and the small s-bearing gametes. All possible fusions of F1 gametes are shown in the matrix called a Punnett square, named after its originator, Reginald Punnett. These fusions give rise to a zygote that produces the F2 generation.

So, in the F2 generation, three types of genotypes are produced. Capital S, capital S, capital S, small s, and small s, small s. So, as a result of the random fusion of gametes, the relative proportion of those zygotes is 1, 2, 1 respectively, right? So, however, because the S factor is dominant, both the capital S and the small s seeds are smooth, and the F2 generation seeds only show a phenotypic ratio of 3 to 1. So this is exactly what it is showing here. So in the P generation, when you are doing the cross-fertilization, you have smooth seeds and wrinkled seeds when they are actually undergoing fertilization in

the first generation. So you have the capital, as in capital, as small as small, and that's how you are actually producing the capital as small as capital as small as capital.

So all are actually capital S and small s, and all are going to be smooth seeds because the capital S is going to be dominant over the small s. Right. So basically in this one, the genotypic ratio as well as the phenotypic ratio is going to be the same, which means all are going to be smooth seeds and all are actually going to produce the heterozygous disc. Now, when you take two heterozygous individuals of this type, like the capital S and the small s, and if you do the crossbreeding for the capital S and the small s, when the SS is going to breed, it is going to give you the capital S capital S, which means it is going to be smooth seed, right? When the capital S crosses with the small s, it is actually going to give you the smooth seeds because of the dominant nature of the smooth beads; therefore, it is going to be heterozygous. Similarly, here it is also going to give you the smooth seeds, but in the smaller sizes, it is going to give you the wrinkled ones, right? So in the F2 generation, what you are going to have is the genotypic ratio, which is going to be like this: if you say genotypic ratio, it is going to be like ss, small s, small s, so it is going to be 1 to 2 to 1, whereas the phenotypic ratio is going to be.

3 is to 1 because these three are actually going to show you the dominant traits, which are the smooth traits. So, this is what is called the law of segregation, which means that even when these two molecules or genes come together from the two individuals, in the second generation they get separated, so they are actually separable, which means the characters do not get mixed with each other. They are pure and they will remain pure and whenever there will be a chance of segregation they will get segregated. So the law of segregation is that the principle of segregation states that the recessive traits, which are masked in the F1 from a cross between the two breeding strains, reappear in a specific proportion in the F2 generation. So in modern terms, this means that the two members of a gene pair segregate from each other during the formation of a gamete.

As a result, half the gametes carry one allele, and the other half carry the other allele. In other words, the gametes carry only one type of gene from each gene. The progeny are produced by a random combination of gametes from the two parents. Mendel also analyzed the behavior of six other pairs of traits. Qualitatively and quantitatively, the same results were obtained from the seven sets of crosses.

He made the following general conclusion about his data. The results of the reciprocal crosses were always the same. All the F1 progenies resembled one parent strain, indicating the dominance of one allele over the other. And in the F2 generation, the parental trait that had disappeared in the F1 generation reappeared. Furthermore, the traits seen in the F1 were always found in the F2 at about three times the frequency of the other traits, right? This means that in the F1 generations, all dominant teams are going to appear, right? Whereas in these F2 generations, the dominant and recessive traits are going to appear in a ratio of 3 to 1, the genotypic ratio is going to be 1 to 2 to 1. So remember that in the F1 generation, the genotypic ratio as well as the phenotypic ratio is all going to be the same, right? Because they are all going to be heterozygous.

In this case, it is going to get segregated. To confirm this, Mendel also conducted different types of test crosses. So at the time of Mendel's experiment, the presence of segregating factors that were responsible for the smooth and wrinkled phenotypes was only a hypothesis. So Mendel was not aware of the concept of genes, was he? He could not understand what kind of factors are controlling and how these kinds of things are happening. So to test his hypothesis regarding this factor, Mendel allowed the F2 generation to self-pollinate. As he expected, the plant produced from the wrinkled seed breeds true, supporting his conclusion that they were pure for the small s factor, right? So he actually uses the term that is called the factor governing the wrinkled seeds or the factor that is governing the smooth seeds.

Selfing the plant derived from F2 smooth seeds produces the two different types of progeny. One third of the smooth F2 seeds produces all smooth-seeded progeny, while the other two thirds produce both smooth and wrinkled seeds in each pod in a ratio of three to one. And the same ratio was seen in the F2 progeny. These results support the principle of the gene segregation. So he actually did the test cross. Test crosses mean when you are actually breeding the offspring together, and so when you do the cell fertilization, for example, when he did the cell fertilization for the SS or the heterozygous ones and you get the F3 progeny, it is actually going to be all smooth, right? Because both of these are true breeding, whereas in this case it is actually going to show you the pure population of 3 to 4 for the smooth or 1 to 4 for the wrinkled.

So the capital S and the small s plants have a different phenotype but the same dominant phenotype. The cell fertilization test of the F2 progeny proved to be a useful plan to determine whether a plant with the dominant phenotype was heterozygous or homozygous. So a test cross is a very good method by which you can determine whether you actually have a pure breed or whether you are going to have the cross breeds. Because if it is a purebred, then it is actually going to give you only that phenotype; if it is a crossbreed, then it is actually going to show you a phenotype in a three to one segregation, which means it is actually going to show you the dominant traits of three plants and the recessive traits. So if it is a capital N and a small s, it is actually going to show you the plants of both types of seeds.

But if it is a small "s," then it is actually going to show you only the wrinkled seeds. A common test to perform this is a test cross. A cross of an individual expressing the dominant phenotype with a homozygous recessive plant is conducted to determine its genotype. In sum, the test crosses of the F2 progeny from Mendel's crosses are shown to yield the dominant phenotype, resulting in a 1 to 2 ratio of homozygous dominant to heterozygous genotype in the F2 progeny. That is, when you cross the homozygous SFC, one third of the F2 progeny with the dominant phenotype gives rise to a progeny with the dominant phenotype and is therefore heterozygous for the dominant allele. The other two-thirds of the F2 progeny with the dominant phenotype reduce the progeny to a 1 to 1 ratio of dominant phenotype to recessive phenotype and therefore include the heterozygous.

But there are exceptions. There are exceptions that do not follow such rules of dominant

traits, right, where the dominant traits also get mixed with the recessive traits. So Mendel's principles apply only to the deployed eukaryotic chromosome and form the foundation for predicting the outcome of processes in which segregation and independent assortment occur. Whereas all the allelic pairs Mendel studied showed a complete dominance or complete recessiveness relationship.

Many allelic pairs do not. As more geneticists also conducted experiments, they found that Mendel's principle did not apply exactly. So you have complete dominance, right? It's the phenomenon in which one allele is dominant to another so that the phenomenon of heterozygosity is the same as that of the homozygous dominant. With complete recessiveness, the recessive allele is phenotypically expressed only when it is homozygous. Complete dominance and complete recessiveness are the two extremes of a range of dominant relationships for the two alleles. But in between, you also have incomplete dominance, right? So, when one allele of a gene is not completely dominant over another allele of the same gene, it is said to exhibit incomplete dominance, also referred to as semi-dominance or partial dominance.

In incomplete dominance, the phenotype of the heterozygote lies in the range between the phenotypes of the individuals that are homozygous for either allele. For example, if you have red and white, when you crossbreed them, ideally it should show you red because red is dominant over white, but what happens is different. Because of incomplete dominance, it is actually going to show you a mixing of both phenotypes, and it is actually going to give you the pink color. So the phenotype of the heterozygote C is typically referred to as an intermediate phenotype, though it may not be exactly in the middle between the phenotypes of the two heterozygotes. So incomplete dominance is also exhibited in the fruit color of eggplant. When a homozygous plant that produces purple fruit is crossed with a homozygous plant with white fruit, the heterozygous F1 generation produces violet fruits, right? This is actually a very simple example where these two species are two breeds, right? Two breeds for purple.

And white, and when they fuse right, it is actually going to give you a violet because violet is the mixture of these two. So it is not showing purple, it is not showing white; it is showing the violet. So when the F1s are crossbred with the other one-fourth of the F2, one-half are purple, one-half are violet, and one-fourth are white.

So this one-to-one ratio is different from the three-to-one ratio that we would observe if a plant could exhibit dominance. So there are simple differences. Right. If it is complete dominance, then it is actually going to show you a ratio of three to one; if it is complete or incomplete dominance, then it is actually going to show you a phenomenon of one to one. Whereas one is actually going to be red, this one is going to be white, and this one is going to be pink, right? So it is actually going to show you an incomplete dominance wherever you have the heterozygous alleles. So when a trait displays incomplete dominance, the genotypic ratios and the phenotypic ratio of the offspring are the same because each genotype has its own phenotype. So remember that if there is incomplete dominance, then in the F1 generation or F2 generation, you will actually have a similar ratio of genotypic and phenotypic ratios in both cases.

It is impossible to maintain the eggplants that are pure breeds for the violet flutes because all plants with the violet flutes are heterozygous. Some human diseases show incomplete dominance. One example is sickle cell anemia, in which the homozygotes for the sickle cell mutant alleles have sickle cell anemia, while the heterozygotes have a milder sickle cell trait. So heterozygotes should show very strong alleles, right? Strong anemia occurs because sickle cell anemia will manifest in the homozygotes, right? Incomplete dominance is exhibited when the heterozygotes have a phenotypic intermediate between the phenotypes of the two homozygotes. When the trait exhibits incomplete dominance, the cross between the two heterozygotes produces a one to one phenotypic ratio in the progeny.

And this is what is shown in this particular figure. So you have the purple fruits, you have the white fruits, and these are the P generation, right? So, when you do the first generation fertilization, it is actually going to generate violet-colored fruits. So, it is not going to generate the purple fruits or the white fruits. Instead, all these heterozygous individuals are going to generate violet fruits, and then in the F2 generation, when these are crossbred, it is actually going to give you all sorts of genotypes, like for example, capital P capital P or small p small p, and then you also have a capital P and a small p. So capital P is going to give you purple-colored fruits, small p small p is going to give you white-colored fruits, and this combination is actually going to give you violet-colored fruits.

So it is actually going to give you the fruits in the ratio of one to one, right? So this is the one. This is 1, and this is going to be 2, right? So, this is going to be 1 plus 1. So, it's going to be 2, right? And this is all happening because of incomplete dominance. And then we also have co-dominance, right? So, co-dominance.

Another modification of the dominance relationship is co-dominance. So, in co-dominance, the heterozygotes exhibit the phenomenon of both homozygotes. This means the phenotype of the heterozygote is not intermediate between the phenotype of the homozygote. Rather, the heterozygotes simultaneously express the phenotype of both homozygotes. By contrast, in complete dominance, the heterozygotes exhibit a phenotype intermediate between the two homozygotes. The ABO blood group system is a classical example of co-dominance, where both alleles contribute equally to the phenotype in a heterozygous individual.

So, you remember that the blood group, right? The blood group is, you know, having two different types of alleles. You have the A alleles and the B alleles. So if you don't have any of these and you have both, then it is actually going to give you AB. If you don't have any of these, then it is actually going to give you the O blood group. So that's how you are going to have four different types of blood groups.

So if you have A alleles, you're going to have A blood group. If you have B alleles, it's going to be the B blood group. If it is both, then it's going to be AB. And if you don't have any of these, then it's going to be the O blood group. So this is the phenotype, right?

This is the O blood group, A blood group, B blood group, and AB blood group, right? And this is the genotype. Genotype is I by I, IA by IA, or IA IB.

So this is actually in the absence of the A or B genotypes. In the A, you're going to have the A phenotype; B is A or B, and then AB is actually going to have the mixture of A and B. But in this one, both A and B are actually going to express and will give you the hybrid phenotype. So the gene responsible for determining the blood group has three alleles: IA, IB, and small i, right? So if you have IA, it is going to be an A blood group. If you have IB, it is going to be a B blood group. Neither of these, but if you have IA and IB both, then it is going to be AB blood group, and if you have IEI, then it is going to be O blood group.

An individual with the genotype of IAIA or IAI will have a blood group of A. Those with the IB and IB or the IB and I will have blood group B, and an individual with the genotype of II will have blood group O. which lacks both A and B antigen. In contrast, the i allele is recessive to both IA and IB. So, in this kind of co-dominance, the A antigen is produced from the IA, and the B antigen is produced from IB, and that's how both antigens are expressed on the RBC, which is why it does not follow Mendelian genetics. So IA and IB alleles are co-dominant, which means the individual with the genotype IA and IB expresses both A and B antigens on the surface of their red blood cells, resulting in the AB blood group.

This inheritance pattern demonstrates how co-dominance allows both alleles in a heterozygote to be fully and simultaneously expressed. So this is just a checkerboard analysis. It shows how the offspring are going to work and so on. Then we also have another example of pleiotropy. So pleiotropy, as the name suggests, occurs when two or more phenotypes are correlated; the traits do not vary independently.

For example, fair skin, blonde hair, and blue eyes are often found together in the same individual. The association is not perfect. Sometimes we see individuals with dark hair, fair skin, and blue eyes. But the traits are found together with enough regularity for us to say that they are correlated. The phenotypic correlation between the two quantitative traits can be computed by measuring the two phenotypes on a number of individuals and then calculating a correlation coefficient for the traits. One reason for a phenotypic correlation among traits is pleiotropy, where the multiple phenotypic effects result from a single locus determining the traits.

Genes rarely affect only a single trait, and this is particularly true for the polygenes that influence the continuous traits. For example, in humans, many body structures respond to the growth hormone, and there are genes that affect the amount of growth hormone secreted by the pituitary gland. People with certain genes produce a high level of growth hormone, which increases both height and hand size. But the other processes, the lower level of growth hormone, lead to both short stature and small hands. So height and hand size are phenotypically correlated in humans.

And this correlation is due to the genetic correlation and the fact that both characteristics

are affected by the same gene that controls the amount of growth hormones. Genetically speaking, height and hand size are the same characteristic because they are the phenotypic manifestations of a single set of genes. So, when the two characteristics are influenced by the same gene, they are genetically correlated. So far, what we have discussed is how genetics is evolving as a field, right? So we discussed how people have discovered DNA, how Gregor John Mendel performed different types of experiments with the pea plant, why he chose the pea plant, and so on.

And then, based on these experiments, Gregor John Mendel formulated the different types of the law of inheritance. So, we discussed the first law, which is the law of dominance. And then, subsequent to that, we have also discussed the exceptions to the law of dominance. So we discuss incomplete dominance, we discuss core dominance, and then we also discuss pleiotropy. So, with this brief discussion about genetics, we would like to conclude our lecture here. In the subsequent lecture, we will discuss some more aspects related to the law of inheritance. Thank you.