Course Name: I Think Biology

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W5L24_Genetics - I

Hi, in this lecture, we begin a new unit to discuss genetics. Specifically, we will start with an introduction to the vast field of genetics. And after that, we will talk about what our knowledge of genetics has led to in terms of technological benefits, but also in terms the kinds of ethical questions that arise out of new technologies.

So let's start with basically in the last unit, we had discussed that you know, there are cells and these cells go through a cycle, they keep dividing. And by and large, we said that cells are, they contain DNA, right? And since cells are the building blocks of life, all of life is encoded by DNA. So what does this mean? It means that every living thing is united in a way, right? We all use the same language. And by we, I don't just mean humans, but I mean, plants, other animals, insects, including insects, bacteria, etc. Right? So we're united in this way. And so much of the biology that we learn from other living organisms also applies to humans, right? So that's one of the exciting things when you learn about nature is you're also learning about yourself and vice versa, right? We learn about humans, we're also learning about other organisms in nature. So the fact that life is encoded by DNA means that DNA is a way of storing information, and we can store information in the same language, right?

So DNA, these four bases that DNA is made up of ATGC is the alphabet to store information. And because we have the same language across all living things, we can even exchange information across organisms. And that's the basis of all of biotechnology. We will learn more about that specifically when we discuss Bt cotton. Right, so we can exchange information across organisms, right, but also across species and species that seem sort of different, between humans and bacteria, for example. Okay, so going back to genetics, what we should be able to do by the end of this unit is answer some of the questions that are listed on this slide.

So we know that children resemble their parents, right? And we know that this is, we've heard a lot that this is because of genetics, right? We often use the phrase, this is in my genes, right? This is what I've made up. So what does that mean? And you know, another question that always comes to mind is whether children resemble their parents, but why don't the same parents give birth to identical progeny? So I'm not referring to identical twins, but if it's the same combination of parents, why don't they give rise to children that are also identical? So hopefully by the end of this unit, you should also be able to answer that question, right?

We know that genes encode certain traits. So this is again, something that we know, you know, in our everyday language, we say, okay, this is in my genes. That means, you know, I don't know, you might think, okay, somebody can do is a really good high jumper. And we say, okay, this is genetically encoded because people in their families are athletic. So what does it mean for genes to encode certain kinds of traits characteristics or abilities that people have? So there may be physical traits, you know, the way you look, but there may also be things that you're able to do. Okay. And finally, you know, to answer these questions, we've had to study a lot about genes and genetics. But in the process, we've also created a lot of technology, right? By learning about these things. So finally, we will start to think about what are the social, sociological, technological, and ethical consequences of genetic technology.

So all of how we can manipulate genes now, study genes and manipulate them. So we've all heard of Mendel, right, the father of genetics. Mendel did his work in the mid to late 1800s. But it's not that the ideas of heredity, rights, inheritance, and how traits were passed on from one generation to the next. It's not that these were unfamiliar concepts to people before Mendel, right, he wasn't the one who discovered these ideas of heredity.

And this is a kind of funny cartoon that highlights this idea, right, that people used breeding all the time. So this cartoon sort of makes a joke of it that you can, you could breed dogs of, you know, wildly different shapes and sizes. And this was really by mating and continuously and continuing the sort of breeding specific animals that had the traits that you were interested in, and selecting for those traits, right? So, this is called breeding. And this was very, very, very common in farm animals, right?

So for instance, if you found that there was a cow that had a lot of meat, right, that was very bulky, and you wanted more cows that would produce more meat than or more milk for that matter, then you would choose that cow and try to produce, get that specific cow to give birth to more progeny, and so on. That was what breeding was. So, the reason you would do this was because they were notions of heredity, right? So this was done in farming, not just for animals, but also for plants, right?

And cotton in India, for example, has been known to be cultivated for thousands of years, right? This was done because farmers carefully chose the seeds from the plants that were the hardiest or that produced the best kind of material that we wanted, and so on, right. So this was also an example of selective breeding. The idea of breeding comes from the idea of inheritance. Okay.

That was before Mendel. Before really we knew about genes since we knew about inheritance, Mendel was a friar who led a monastic life. He chose a monastic life because he was very poor, and he thought that this would allow him to study because he was, you know, sort of a very academic individual, he wanted to study without having a financial burden. So he studied physics and mathematics. But when he was in the Austrian kingdom, when he was posted to a specific monastery, he decided to work on pea plants to try to understand the principles of inheritance. So what about pea plants? So the specific species that he used was *Pisum sativum*.

And he ended up using, choosing specific traits in the pea plant, right? So everything from the shape of the seed, the form, so whether it was round or wrinkled, the color, whether the cotyledon was yellow or green, the color of the flower, the form of the pod itself, the color of the pod, the type of flowers, and the size of the plant. So he chose these traits and started to look at how these traits propagated. And by propagated, I mean, how are they inherited across different generations, right? And he set up very careful experiments.

So to set up his experiments, he used true breeding strains. What does that mean? This is a really important concept. True breeding strains are defined as strains that when they are pollinated by themselves or crossed with other true breeding strains, the same true breeding strain, always produce the same trait. So for example, if you chose a plant that had, say, white flowers, right, and another plant that had white flowers, and these were pollinated, right, let's say mated, this always gives rise to progeny that has white flowers, right? So let me write down the definition of a true breeding strain.

That is progeny always carry the same trait as parents. Yeah. Okay, so this is the example. So what he was able to do, because he was using these pea plants, right, he was able to breed the plants either by selfing the plants, right, or crossing the plants. And what do I mean by this? He used the process of pollination, which is how plants reproduce, right? And how do plants reproduce? They have pollen, which is the male reproductive gametes, right? That's what is here. That's number 10. And these are produced by the male structures, and they're found on the stamen, right? When this pollen is released, it can be then transferred to another plant.

And when it's transferred to another plant, it's called a crossing because it is crossing, it's not selling, right? And when it's found on another plant, it goes to the stigma. That's what's here. And the stigma is part of the female structure. Yeah. This pollen is the male reproductive gamete. Once it reaches the stigma, it travels down and reaches the ovaries. And that's where fertilization occurs, right? So when you self a pea plant, you take pollen from the flower, and then make sure that it reaches the stigma of the same plant, right? So that's why it's called selfing.

But when you cross, you make sure that the pollen reaches another plant. Okay. So what Mendel did was that he did a crossing between true-breeding pea plants with violet flowers, right? And you can see the violet flowers here, and with white flowers, right? So the definition of true breeding, remember, is if you took white flowers and self them, this is the illustration here of a cross. But if you selfed a white flower, you would always get white flowers in the true breeding strain. If you selfed a violet flower plant in a true breeding strain, you would always get violet flowers. But what he did, he didn't sell them, he crossed these two, right? And in this cross, that means whether, by the way, I should say that whether this is, he used the male of the violet flower, the male gametes from the violet flowers to fertilize the white flowers or vice versa, his results were identical.

Okay. So this was irrespective of the sex of the gametes that were used from each plant. And what he saw was in the first generation, the F1 generation, where now the F stands for filial, right? In the F1 generation, all of the flowers were violet, right? What this means is, right, from the P generation with the parental generation to the F1 generation, all of the flowers were violet. So this was sort of surprising in the sense that the white flowers just sort of disappeared, right?

And then what he did was he took the F1 generation and he selfed them, right? So I just, I'm symbolizing self by this sort of circular thing, right? So he selfed them. So again, you take the male gametes from the plant and then fertilize it on the same plant. And in the F2 generation, so in the second filial generation, what's fascinating is that this white trait reappeared, right?

So the white flowers are back again, whereas in the first generation, they had completely disappeared and they've resurfaced. So this is kind of shocking, right, because it's as if this trait of being white was somewhere hidden in the F1 generation. And one thing that I should point out is that Mendel did this not just with one set of plants, right, but many, many, many plants. In fact, his, all of his experiments, he used over 10,000 pea plants to conduct his experiments, right?

So this is a very, very patient man. And so what he found was that there was this ratio of violet flowers to white flowers, right? So from 700 violet flowers to around 225 white flowers. So the statistics you will see turn out to be important. What I mean by statistics is that he used a large sample size, and that's very important to be able to make the connection that he eventually did.

And we'll talk about that. And so what he did, what Mendel did was that he used the same characteristics that we talked about, right, and repeated the same kind of cross. So whether it was flower color, flower position, plant height, seed texture, color, the pea pod texture, or the pea pod color, right, these were the characteristics that I talked about earlier. In the F1 offspring, it was one trait that always disappeared, right? So when we had the violet versus white flowers, the F1, the first filial generation, they were all violet. But in the F2, you saw that the white reappeared.

So if we take the other another example of the pea pod color, the green versus yellow, if you cross them, in the F1 generation, you had 100% green, but in the F2 generation, you saw that the yellow reappeared. What is most fascinating is that all of these crosses that he did always resulted in a similar ratio of one trait versus the other. So whatever trait was found in the F1 offspring was present in approximately a three. So three out of the four, four were found in the F1 as the F1 trait. And one out of the four had the trait that reappeared in the F2 generation, right?

And so these statistics, these ratios that he was able to come up with, by looking at thousands of plants was really important. So there's one really important idea that comes out of this, which is that you know, the fact that the white disappeared, right, means that the purple and white didn't just blend. It wasn't that when the two were mated, you didn't get a sort of lighter purple color in the F1 generation, right, you got the same purple color. So this is proof that characteristics did not blend, right? So for example, if a black dog and a white dog mate, are right, the idea of blending would be that you always get a gray dog.

And that's not always the case, you might get progeny that are white that are black, that have patches. So it is in that characteristics always blend. And that's exemplified in this example. Further, what is also shown is that since in the F1 generation, the white trait has disappeared, right, this trait, Mendel identified as being recessive, meaning that the dominant trait is a violet color, right?

And the white is somehow hidden, right? And so he called this a recessive trait. And in the F2 generation, the recessive trait reappears, where the F2 generation is a result of an F1 selfing. So what can explain this ratio, this three-to-one ratio of the dominant versus the recessive trait? And this is the key connection that Mendel made, right? He said that each parent, so in this case, the violet flower plant and the white flower plant has two copies of a trait, right?

And each offspring gets one copy of that trait, such that the offspring in turn has one has two copies, one from each parent. Okay. So, for example, let's say that the violet flowers represent this trait, as capital P and capital P. And we capitalize on them because we've already just said that it's a dominant trait. And the white, the white flowers, as we said, are small p, small p, right.

So let's represent it with these, with these letters. So let's try to then understand what this means, right? The fact that each parent has two copies of the trait and transmits one to its offspring. So let's think about this. We already said that the parents are P, P, capital P, capital P, and we've crossed it with a small p, small p.

Okay. And now let's work this out with what is called a Punnett square. Punnett is named after a scientist whose last name was Punnett. So let's say that one of the colored plants has given a capital P, capital P, capital P. And the white colored ones, I'm going to write these as a small p so that you recognize them, right? Now if each offspring gets one copy from each parent, that means that the first offspring would get capital P, small p, the second one likewise, and so on, right?

And so no matter what, right, every offspring from this cross, right, will have, now these two copies of the trait, right, capital P, one capital P, and one small p because they're getting them from the parents, right? And now you start to see why the white was hidden, right, because the capital P is the dominant trait, which is the violet flower. And it sort of overshadows, let's say, it dominates the small p, right? And that's why you see the purple, the violet color showing up without any of the white. Now if you take this parent, right, the capital P, small p, and you self it, which means again, you have a capital P, small p, right.

So what's the result of this? So one of the children will have capital P, capital P, the other will have capital P, small p, capital P, small p, small p, small p. Okay, so this doesn't mean, right, in the F2 generation, if Mendel had just looked at four plants, it doesn't mean that he would have seen this exact ratio, right, where this plant is violet, this plant is violet, this plant is violet, but this plant is white because it does not have any of the dominant copies of the trait, right. It's not that you will just see four progeny, where three are violet and one is white. The reason that Mendel had to do these large sample sizes, right, is to look at the probability of F2 offspring having the purple flowers or the violet flowers, right?

So that's what the statement is saying, right? So Mendel demonstrated through the use of statistics, right, through the use of thousands of plants to show that the probability of the F2 offspring getting the purple flowers, right, was 75%. That's why he had to use those thousands of flowers. He was able to show or demonstrate why the 75% or three is to one ratio that we observed, and where it comes from. And he was able to do this really with this idea of, you know, each parent providing two copies of the trait, having two copies of the trait and transmitting one to the offspring.

Yeah. Okay, so there are a couple of connections here that are important. One is Mendel didn't know about genes, right? He didn't know that cells have DNA. He just said that we have two copies of our traits, right? He didn't know in what physical form these traits were. He just knew that they were passed on from parent to offspring. But how did we figure out that these traits were carried on genes? We didn't figure out too much later, but how did we figure out the traits that were carried on chromosomes in the first place? And that was done by somebody named Thomas Morgan Hunt. He was an American scientist and he used fruit flies, right? So what he did, he discovered that you know, in his fly collection, he used to grow flies in his lab.

He had fly cultures, right, in his fly collection. He found that some flies, male flies, had white eyes. You see that indicated in this image. And so if he crossed these male flies with female red-eyed flies, right, he found that the first generation, all of them had the red eyes, right? There were no white-eyed flies. But if you repeat it across now with the same two flies from the F1 generation, so a red-eyed female fly and a red-eyed male fly from this particular cross, he ended up seeing that you could see males that had both white eyes and red eyes, but the females always had red eyes.

Okay. So the key connection that Thomas Morgan Hunt made was that this pattern of inheritance matched how sex chromosomes were inherited. Okay. So in other words, he said, they knew already that females had an XX. So they had, females who had two X chromosomes, right?

And males had only one X chromosome. Okay. And so he was able to say then, again, given the idea of dominance that Mendel had already passed on, right, that if the male had only one X chromosome, right, and the male was red-eyed, the male would receive its X chromosome from the mother and the female would receive its X chromosome from both, each of its X chromosomes, one from each parent, the mother, and the father, right. And because of this, right, if you look at this diagram carefully, it shows that the red-eyed trait is dominant over the white-eyed trait, right? Wherever the fly, the male fly has the white eye trait that is carried on the X chromosome, the male will have white eyes. So basically this cross demonstrated that the way that the white-eyed trait was passed on from one generation to the next matched exactly the idea that this trait was being carried on an X chromosome and that red eyes were dominant over white eyes.

Okay. And you see that demonstrated here, the female has two X chromosomes and has received its X chromosomes from them, one each from the male and the female parent, right? And there are two possibilities. In both possibilities, since the red-eyed trait is dominant over the white-eyed trait, you see that the female has red eyes. The red-eyed male has received one copy from the mother of the X chromosome, and that happens to be the red trait. The white-eyed male has received one of the copies, which is the white-eyed trait.

And hence the male has white eyes, right? So basically this demonstrated not only the idea that some traits are sex-linked because they are on the X chromosome but also that they must be carried specifically, more importantly perhaps that traits as Mendel had defined them are carried on chromosomes, right? Remember they still hadn't figured out that chromosomes are made up of DNA and what the structure of DNA was. Okay. So now that we know that genes are encoded, echoed traits and that they lie on chromosomes, right, what are the inheritance mechanisms from parent to offspring? And this is seen really through meiosis, right? And meiosis is the process of producing gametes, so either sperm or egg in our case, right?

And what happens during the process of meiosis is in the first step in your nucleus, DNA is copied, right? And so as a result of this copying, you have, so these are homologous chromosomes, right, the red and the blue, and these are the copied pairs. So these are called sister chromatids, right? And these are homologous. The homologous chromosomes, one each are received from the parents, right, and the sister chromatids arrive from having DNA copies or DNA replication.

Then this is a key event here, which is recombination, which means that between homologous chromosomes, there occurs a process called recombination, where there's an exchange between chromosomal segments, right? And this is the reason why eventually when these separate, right, when these homologous chromosomes separate after recombination, you have chromosome pairs that are drawn apart, they separate into two cells, right, this is meiosis one, and then at meiosis two, it results in four gametes, each with one copy of the chromosome. So that means that each of these four cells has half as many as we started within the initial parent cell, right? An important thing to keep in mind is that one of the reasons that the same parents don't produce identical offspring is because the gametes arise from recombination events, right?

So it's not that every gamete that a parent produces is identical, right? So whether it's a male parent or a female parent, the gametes that are produced, every gamete will be different because of the recombination events, right? So now this gamete, or these gametes contain the traits that are then passed on to the next generation, right? So suppose this is the male, this is the female gamete, and the male gamete eventually, right, this is the sperm that I've drawn, these two come together. And the sperm also carries its DNA, these two come together then to fertilize, right, and produce the zygote. So now we know the mechanism of inheritance of chromosomes as well, right?

So now we just to recall, we've gone from knowing the traits are passed on, we've gone from knowing the traits are carried on chromosomes. And now we know how chromosomes are inherited from one generation to the next. Since Mendel and Hunt, there have been many, many, many more discoveries in genetics. So Mendel had said that if you have trait A and trait B, right, how trait A sorts into the gametes is independent of how trait B sorts into the gametes. It was later shown that that's not the case for all traits and that they can also be linked.

Further, we see that there are sort of layers to the ideas of dominance. So there can be things like codominance, and incomplete dominance. Incomplete dominance refers to something similar to what we talked about in blending, right? Some genes can be more penetrant than others. Epistasis is the idea that some genes affect the function of other genes, right?

So the production of say a protein can affect how another gene is expressed. We also have mobile genetic elements on our chromosomes, we have elements that jump from one chromosome to another. And of course, a very important discovery is the double helical structure of DNA, right? So this is just a short list of the important discoveries, but there have been many, many more. So in summary, I just like to finish by just looking back at what we've learned.

So we discussed the idea of breeding, right? And this was our first understanding of heredity, how traits are passed on from one generation to the other. We talked about Mendel and his pea plant experiments, his ideas of dominance, right, and how parents have two copies of each trait, right? And we also talked about the hunt and the idea of traits being carried on chromosomes. Note here I've not used the word gene very much because we've not really, rather hunt and Mendel did not know about genes, right, or what chromosomes were made of, right? Traits are on chromosomes. We talked about X-linked traits or sex-linked traits, let's call them. And we talked about a few other discoveries, right, or in genetics. Not in much detail. But this gives you the basis for understanding some ideas of heredity. And in the next class we will discuss a little bit, we will sort of skip in time, right, skip forward in time, and then discuss more of modern-day genetics, right? So we are going from the 1800s and coming to the 2000s.