

Evolutionary Dynamics
Supreet Saini
Chemical Engineering
Indian Institute of Technology Bombay
Week 03
Lecture 13

Hi, welcome back to this video. So, in the last lecture, we saw that in prokaryotic populations, diversity is generated via novel mutations occurring in the population. Mutation rates are small, but the diversity generated in microbial populations is huge because of their large population sizes. We saw that in something as small as just 100 milliliters of a flask in a lab, the population sizes can reach 10^{11} , and the number of mutations that can occur is of the order of 10^8 , which is 100 million different mutations happening in just that flask.

As a result, the variation that natural selection acts on is generated via these mutations. What we want to understand today is that if we are talking about eukaryotic and sexually reproducing organisms such as us, the diversity generated in the populations arises from a different mechanism altogether. I mean, mutations are the ultimate source, but if you're looking at a few generations, the source of diversity is something else, and that is what we want to investigate. As a result, because the mechanisms via which diversity is generated in prokaryotes and eukaryotes are different, the approach to studying evolution for these two organisms is also different. In prokaryotes, which is what we'll be studying in this course,

we will actually be able to follow what mutation arose, at what time it arose, what happened to it, what it did, and understand the molecular underpinnings of how adaptation was facilitated. In eukaryotes, the study of evolution of sexually reproducing complex organisms takes the name of population genetics, which is something we will not be talking about in this course. But it's important to understand the distinguishing feature between the two. This short video is an attempt to understand what happens in sexually reproducing complex organisms, such as us. Why are we saying that, like in the case of prokaryotes, where new mutations are happening and leading to the generation of variation?

Why is that not the case here? So, the first thing that we must recognize is that inheritance is biparental. So, you have parent 1 and parent 2. These two mate to produce an offspring. This is qualitatively different from there being one parent that divides into two offspring.

Now, the DNA of this organism is coming half from one parent—50% of the DNA—and 50% from the other parent. This is qualitatively different from what we've been looking at so far. And before

we go any further, we first need to look at how our DNA is organized inside the nucleus. By 'our DNA,' I mean any sexually reproducing complex organism, but human beings are a good system to look at this problem. So, inside each of my cells—let's say this is a cell—

Our cells are much larger than bacteria. There is the nucleus. Most of our DNA is present in the nucleus. There is some in mitochondria, but that is minuscule, and we will not discuss that in this context. Unlike bacteria, whose DNA is circular in nature, human DNA consists of linear threads of DNA, and there are These linear threads are called chromosomes.

So there is chromosome 1, chromosome 2, 3, going all the way to 22. They get increasingly smaller. The numbering is because of the size. And the 23rd—I am a male, so I have an X and a Y. Okay. Well, let us discuss the 22 first, and we will come to the last one later on.

The interesting part is, because inheritance is biparental, I do not have one copy of chromosome 1. I actually have two copies of chromosome 1. So, the other copy that I am carrying—I inherit one copy from each of my parents. So, I have two copies of chromosome 1. One from my mother, one from my father.

I have two copies of chromosome 2. I have two copies of chromosome 3, and I have two copies of chromosome 22. In addition, the 23rd chromosome is what determines the sex of the individual, and in my case, since I am a male, I inherited the X chromosome from my mother and the Y chromosome from my father. Y is much smaller than X, and this forms the 23rd pair. So, human DNA is organized in these 23 pairs of different threads.

Each one is a double-helical DNA structure. And this inheritance is biparental because the blue-colored DNA is something that I've inherited from my father, and the other color is from my mother. So then the question arises: what do I, as an individual whose DNA is this, pass to my offspring? What is the 50% that I pass on? So obviously, I pass it through the sperm, and I'm going to pass one copy of each of the chromosomes.

When I say that one copy of each of the chromosomes—for chromosome number 1, going all the way to chromosome number 22— I will pass that. In addition, I will pass either an X or a Y for chromosome number 23. Okay. So the question then becomes: since I have two copies of chromosome one—this and this—which of the two copies do I pass to my progeny?

Is the choice completely random? Does one have a greater chance of being passed on than the other? And that's the question that we'll be looking at. And that's the question that makes sexual reproduction quite amazing. And this 23—there is nothing sacrosanct about 23.

It's just that we happen to have 23 pairs of chromosomes. Different species have different numbers. Something like baker's yeast, which is a single-cell eukaryote but capable of sexual reproduction, has 16. You'll find examples of different numbers throughout nature. So let's only look at chromosome 1 and see which of the two copies gets passed to my progeny.

And The chromosome one that I have—the two copies of chromosome one that I have—are arranged like this. Oops, sorry. One copy of chromosome one and the second copy of chromosome one, and we look at chromosome one. So this is one copy

From one of my parents; this is the second copy from my other parent. And the question is: which of the two do I pass to my offspring? And what nature does here is something very clever and quite complex. So the copy that's passed to the progeny is neither this one nor this. What nature does is that it shuffles the two chromosomes back and forth and creates a hybrid, one copy of hybrid chromosomes.

What it might do is that it will start with this one. And then at some point in the chromosome, it jumps to the other copy and then it continues with this one for a while. And then it switches back to the original and then it may copy this for a while. And then it may switch back to the second chromosome again and go all the way. If this is how the chromosome 1 was packaged, then the hybrid that you would get would be something like the blues would be separated and in between them would be this orange colored strand and this would be the hybrid chromosome.

This process of switching back and forth is called recombination. For every chromosome, this happens and you have a hybrid one which gets passed on to the progeny. These switches happen at least three or four times on every chromosome. On an average, they happen every three or four times and create this. Their exact locations are again, these are statistical properties.

Their exact locations are, they're not exact locations, but statistically we know that these regions have higher chance of crossover from one to the other than other regions in the chromosome. Because this process is not certain, the chromosome two might look completely different. Chromosome two might look something like this. Chromosome two is smaller than chromosome one, but this is what it might look like. So you can see, so this is chromosome one, this is chromosome two.

And as you can see in chromosome 1, it's relatively 50-50. In chromosome 2, maybe the blue is a little more. And you can have all sorts of variations. Now, this... The process of shuffling these pairs of chromosomes and generating these hybrid chromosomes—one copy of each chromosome for each one of the 22, and then packing one of X or Y—would be done by me.

The same would also be done by my partner. And then, that's how the fusion of these two would lead to the birth of an offspring. Now, this process of shuffling and recombination matters because imagine the scenario where the two copies that I am carrying... Again, I got one from my father and one from my mother. Let us say we are talking about chromosome 1.

Imagine the two copies that I am carrying are identical to each other. They are completely identical to each other. Which means the DNA sequence of this—let's say this came from my father, and this came from my mother. If these two sequences are completely identical at every single place, which means the Hamming distance between these two sequences is zero, then recombination doesn't really matter because you can go up to this point and then switch and copy from here, but since copying from here is exactly the same as copying from here, it doesn't matter which of the two chromosomes you copy from to create this hybrid because both are identical at every single place. However, chromosomes are not identical.

Even though between any two human beings there is large amount of similarity in the DNA, there are enough differences for something like this to happen. Let's imagine that they are not identical. In fact, they are carrying these mutations. If I identify the locations of the differences in the DNA sequence between the two copies of chromosome 1 that I am carrying, let's imagine that one is represented by a plain line and the other one, every time there is a difference between the two chromosomes, I identify that with the red line on the second chromosome. And now you can see that the number of hybrids that I can create is really really high.

Now as the process of generation of this hybrid starts I start off from here but it matters now whether I am copying from this or this template because there are these mutations here. So if I am copying from here up until this point what that means is that I have missed out on these six mutations. then I switch and now I am copying this but then I am missing on the variation that is here because these two are non-identical. So the number of variations that you can generate via this process is astronomical and remember that this is happening on one chromosome but this is also happening at every other chromosome except the 23rd because X and Y there is no recombination. So the number of genetic variations that you can create just by process of recombination is astronomical.

As a result of that, the variation that is generated in complex sexually reproducing organisms such as us is largely through recombination. Of course, mutations are also happening. Newer mutations are coming up all the time. But their contribution to the variation that exists in a population is relatively small as compared to the variation that already existed and then recombination just creates different permutations and combinations of that entire process. This variation that exists in a population.

So let me just redraw these chromosomes again. Let's say this. The second one is the same color. But let's say the mutations are. These are the places where mutations occur.

That is not to say that only this copy of the chromosome has all the mutations. These red marks are to identify locations where the two strands are not identical to each other. So the mutation could have happened anywhere. The result is that the two are non-identical, and that's what these marks identify. So.

So, the variation that is generated—the key takeaway from here—is that the variation is largely through recombinations. And not via mutations. Of course, what recombination is doing is creating newer genotypes. It's creating newer genotypes out of existing mutations in the population pool that exists. Remember that if the two copies of the DNA were exactly identical to each other, then recombination does not generate diversity.

Then mutation is the only source of diversity. Recombination is able to generate newer and newer combinations of mutations because there exists considerable variation in the population. This existing variation, existing mutation, and that leading to recombination and generating newer and newer variation—this existing variation in the population is called standing genetic variation. Complex sexually reproducing organisms rely on standing genetic variation to create further diversity, genotypic and phenotypic diversity, and to respond to the environmental challenges they face. This is also important because the generation time of complex sexually reproducing organisms, such as us, is really large.

So, transitioning from one generation—suppose an offspring is born today in a human population—you have to wait 25-30 years before it leads to the next generation. The waiting time between generations is really large, and hence you need newer variations, newer mechanisms to create variation. Simply waiting for new mutations to occur and then testing those out by natural selection to see which ones increase fitness may not be the best strategy. Hence, recombination is a great strategy that speeds up the ability of a population in terms of its adaptive speed and how it responds to environmental challenges. The point is that there are two different ways of looking at the evolution of a population.

It depends on whether you are looking at asexual populations, where evolution happens via novel variation—newer mutations—or at sexually reproducing organisms, where we look at standing genetic variation, the variation that already exists in the population. For the purpose of this course, we will stick with prokaryotes, but it's important to understand and distinguish these two scenarios, and we will not return to sexually reproducing complex organisms. So, we'll continue our discussion of prokaryotes and how they evolve in the next video. Thank you.