An Introduction to Evolutionary Biology

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Generating variation

So, we have already seen that variation is an extremely important component of evolution. And without variation, evolution cannot really proceed. However, there is one more thing about variation that we have to appreciate: it is not sufficient to have a lot of variation. The organism or the species must have ways of generating new variations. Why is that so? That is because we should think about the conditions in which an organism or a population finds itself in nature. Is nature constant? No, it is always changing.

You have new pathogens arising, new predators arising, and the temperature changing. You have rainfall changing, you have the availability of food changing, etc. Therefore, over time, an organism, or rather a population, is continuously facing new challenges. In order to meet these challenges, the population has to have ways to generate new variations.

So, now that is what we are going to discuss. What are the ways in which new variations are generated in populations? Now, there are four major ways in which new variations are generated. These are recombination, horizontal gene transfer (also called lateral gene transfer), introgression, and mutations. Now, these are all topics that you have seen or learned before in a basic genetics course. So, what I will do is go through them a little quickly.

I will not talk about the mechanics of any of the topics, but I am generally going to talk about how these things, These modes of generating new variation and how they relate to evolution. So, first, recombination. So, what is recombination? As we know, recombination is the exchange of parts of chromosomes during the process of meiosis I, also known as crossing over. So, this is showing you the process of recombination. Now, what recombination does is reshuffle the alleles from the parents in various combinations.

Now, we already know that various genes at various loci are interacting with each other in terms of their function. Which essentially means that the traits of an organism are dependent on precisely which alleles are present in what combination. And what recombination does is it continuously reshuffles this. It is continuously creating new genetic combinations, which result in new phenotypes. Therefore, the total diversity of traits in the population goes up tremendously.

Now, it is estimated that in any sexually reproducing organism, this kind of variation occurs. That which is arising due to recombination is a huge portion of the total variation available. Now, one thing one has to keep in mind is that recombination is actually a double-edged sword. What do I mean by that? Remember, recombination can bring alleles in combination with each other. But at the same time, it can also take away from or break favorable combinations.

So, in that sense, it's a double-edged sword. It continuously creates and breaks these favorable allele combinations. The second very important thing is what is known as horizontal gene transfer, or lateral gene transfer. So, as we know, in most organisms, most of the time, the genes that we have come from our parents, right? But there are certain situations under which you can have the transfer of genes without reproduction across organisms. And in many cases, this transfer of genes can even happen across species' boundaries.

So, these are what is known as horizontal gene transfers. Now, one thing we have realized is that horizontal gene transfer is very common in prokaryotes. However, there are an

enormous number of examples also known from the eukaryotic world. For example, much of the drug resistance that we find in microbes today is a huge, huge problem. You go to any hospital, and most of the microbes that you know come up there.

Many of them are actually multi-drug resistant, and it is thought that We are going to run out of antibiotics to give to our patients very soon unless something major can be done. Now, this process, a lot of it is actually happening due to horizontal gene transfer. So, basically, you treat one pathogen with your latest antibiotic, but that pathogen ends up, you know, getting resistance to it. developing resistance to it, and then that gene from that pathogen goes into other pathogens. for which you have never used that antibiotic, but they end up getting that gene anyway.

And therefore, on their own, they end up becoming multidrug-resistant. The same thing happens with pesticide resistance as well, particularly in bacteria. So, what is the implication of horizontal gene transfer for evolution? Sorry, before we go there, there is another form of horizontal gene transfer known as introgression. So, in horizontal gene transfer, what happens? A few genes are transferred, you know, across individuals or across species boundaries. Now, in introgression, you have two different species mating, and there is the formation of a hybrid.

And this hybrid, we typically know that the biological species concept says that if you have two species, they should not be able to mate with each other. Now, obviously, that is not the species concept we are using here. We are primarily talking about the morphological species concept. Now, most of the time, the hybrids are infertile, or they have very low survival rates, or they are able to produce very few offspring. But sometimes you end up having hybrids that are fertile, and when that happens, And if that fertile hybrid is now able to breed with one of the existing, you know, partners.

And then it is able to pass the genes from the donor species into the recipient species. And once this happens, you know this can keep happening generation after generation, and at some point. The genes from the donor species will end up getting incorporated into

the gene pool of the recipient species. In this process, obviously you can have more than one gene, you know, spreading into the recipient species. So, for example, a very famous example, if you look at modern humans, about 1-3% of the genes that make up our genome, Depending on which population you are talking about, about 1 to 3 percent of that genome actually comes from Neanderthals.

So, basically, the assumption is that at some point, Homo sapiens mated with Neanderthals. And then their genes have now propagated into the human gene pool. And even after so many thousands of years, it is still, you know, identifiable. Now, what are the evolutionary implications of horizontal gene transfer or introgression? So, the evolutionary implications are very interesting. So, normally you remember when we were drawing phylogenetic trees; we got trees like this.

These are what is known as branching trees, where each node has only one ancestor. So, the whole idea is that one species is giving rise to another species, and another species, and so on and so forth. So, in this kind of scenario, you get a tree that looks like this, Which looks, you know, resembles the branches of a tree, you know, as you can see over here. However, when you have horizontal gene transfer, what this implies is that In the context of knowing where your genes are coming from, you can have more than one ancestor. So, some of your genes are, let us say, coming from Homo sapiens, but, Some of your other genes are coming from you know Homo neanderthalensis.

So, if you have a situation like this, then you end up with a tree that is known as a reticulate tree. So, here, for example, you can see not only do you have this kind of bifurcating branches, But you also have connections between the branches. For example, the ones shown in orange and green and these other ones, which are shown in thin blue branches going horizontally. So, all this shows is that the ancestry of one species can be from multiple species. Now, at some point, some people were very, very intellectually bothered by it.

And almost to the point of claiming that you know, oh my God, Darwin's theory is

wrong. However, I do not think that is entirely correct because even though you might end up having a few genes, Although it is coming from a different ancestor, the majority of your genes are still coming from one ancestor. Therefore, you know that is one thing that you can see in this particular branching tree. That the thickness of the branches is very different, right? So, the horizontal branches are much thinner, showing that the contribution along that axis is very, very low. Whereas the vertical branches are much thicker, saying that.

Much of your genome is actually coming from that root through what is known as vertical transmission. So, because of this, in terms of the process of descent with common modification. Horizontal gene transfer does not play any role there. All it ends up showing is that the way you interpret your trees probably needs to be slightly different. And you have to acknowledge that some of the genes that an organism may have, might come from a completely different species compared to the one for which the vertical tree is drawn. So, that is all that is happening there, all that is to it, and as I said. In a nutshell, it implies that phylogenetic trees can sometimes be reticulate instead of branching. And in no way does it pose a challenge to Darwinian descent with modification. So, these are the three you know are relatively less important ones done, only relatively, you know.

But the one that has been most studied and the one that contributes maximally to the genetic variation are mutations. So, what we will do is a quick revision of various kinds of mutations again without going into the mechanics of them. And once we are done in the next discussion, we are going to look at the certain properties of these mutations. that make them very interesting from an evolutionary biology point of view. So, prior to that quick revision of mutations.

So, mutations can happen at various levels. At the level of individual bases, you can have what are known as point mutations. You can have mutations at the level of the entire chromosome, or you can have mutations at the level of the entire genome itself. So we will study these one by one. At the level of individual bases, you can have a change, an alteration at the level of a single base or you can have an insertion, or you can have a

deletion. A new base can be inserted, or an existing base can be moved out. So, functionally speaking in terms of their effects, the mutations can be divided into three kinds, you know, three types. One is what is known as silence. So, when the change in the ATGC sequence does not change the amino acid, that is when you get a silent mutation.

Why does that happen? Because we have redundancy in the genetic code, in many cases the same amino acid is coded for by multiple codons. And therefore, if your mutation is such that you have gotten into a synonymous codon, Obviously, the amino acid is not going to change, and that is why it is going to be a silent mutation. Similarly, you can have a missense mutation. where, due to a mutation from an insertion or, you know, a change, a different amino acid is coded for. And finally, you can have what is known as a nonsense mutation, you know, that there are three codons that are stop codons, and if a mutation is such that a stop codon is generated, then That is where your translation is going to be truncated, and therefore, that is why you will get a smaller length of a polypeptide chain. I am simplifying this; actually, there are many other kinds of nonsense mutations that are possible. But this is, you know, for a basic revision; this should be enough. Then, at the level of individual bases, you can also look at what exactly is changed. So when the number of bases remains the same, it basically means that a change has simply happened.

In that case, there are two possibilities. One is what is known as a transition, where you have a purine to purine or a pyrimidine to pyrimidine change. So adenine becoming guanine or guanine becoming adenine, that is one. Similarly, C to T or T to C; all these are transitions. When you have a purine to pyrimidine or a pyrimidine to purine change, that is what you call a transversion. So in this particular diagram, A, C, T, and G are the four nucleotides.

So if you go along the diagonals, which are A to G, G to A, C to T, or T to C, then that is what is called a transition. Anything else along the four sides of the square is going to be a transversion. So just so you know, if you remember this diagram, alpha (α) is along the diagonals. And beta (β) is along the sides; then alpha is equal to transitions, and beta is

equal to transversions. This diagram is a simple way of keeping this in mind.

What happens when the number of bases is altered? How does this happen? This will happen if you have an insertion or a deletion in del, as we call them. And when an insertion or deletion happens, sometimes, but not always, you have what is known as a frameshift mutation. What is a frameshift mutation? So, you know that our codons are in triplets, right? So three nucleotide bases together form a codon. If you have deletions or insertions that are not in multiples of three, Then the reading frame for your polypeptide chain is going to be altered. And when you have that, then there can be a huge change in terms of what is translated in the end.

So this is what is known as a frameshift mutation. And you know a frameshift mutation is rather critical many times. For example, certain kinds of cancers are caused by these kinds of mutations. So from this, we move to mutation at the level of chromosomes. There are seven major types we are showing you here.

So you can have what is known as a deletion. So in deletion, you can see that a particular fragment of the chromosome simply gets deleted, or you can have a duplication. where a particular fragment of the chromosome is duplicated. And then it gets added right in the same place as you can see in the second figure over here. Or you can have an inversion where a particular fragment of the chromosome is cut out and then inverted. And then it gets inserted in the same place in the chromosome, as you can see here in the middle figure or you can have a fission where a chromosome divides into two parts and becomes two separate chromosomes. Or, opposite of that, you can have fusion, where you have two chromosomes that come together and form a new chromosome. And then you have what are known as insertions and translocations. So in insertion, a full segment of one chromosome cuts out and gets inserted into a different chromosome at a different position. And in translocation what happens is the same thing, but now you have reciprocal insertion.

In other words, you have two chromosomes: a portion cuts from here, a portion cuts from

here. Then that portion is exchanged, which is what I am showing you here in this figure. So these are the seven major types of changes that can occur at the chromosomal level. We have already seen what happens, you know, when you have a mutation at the level of the entire genome. Polyploidy, the condition in which a normally diploid cell, An organism acquires one or more additional sets of chromosomes; "set" means the entire set.

So, for example, let us say you have a haploid, which is N; here we are talking about 3 chromosomes. And let us say then it has, you know, doubles; that is, a diploid condition: you have 3 + 3, 6 chromosomes. where each chromosome is in 2 copies, and now let us say the ploidy level goes up by 1 more. Then everything, every chromosome, is in 3 copies, which is triploid, or 4 copies, which is tetraploid. So, these two below are the ones that are the polyploids, the 3n and the 4n ones.

We have already seen examples of this; remember we were talking about, you know, speciation in Primula kewensis. This is exactly the process you know: generating a full, you know, 2n became 4n through a process of polyploidy. That is what led to the generation of that new species. We know that there is a lot of variation in terms of how many chromosomes different species possess. So, an automatic question that comes to mind is that If some species have more chromosomes, then does that indicate that it is a more complex species? To understand, or rather to investigate, this question, let us look at four different species and examine their chromosome numbers.

So here we have four species: the jack jumper ant, the human fern, and some kind of ciliate; it is a one-celled organism. As you can see, the ciliate is a one-celled organism; therefore, it is obviously very simple. This fern is a plant; the jack jumper ant obviously has more kinds of tissue types, etc. than the fern. And in terms of complexity, one can argue that humans are the most complex yet if you look at the number of chromosomes that they have, here we are showing you the haploid numbers, the n. The jack jumper ant has only one chromosome, the human has 23 chromosomes, and the fern has 630 chromosomes. Whereas the ciliate, which is a one-celled organism, is probably the

simplest of the lot that has 16,000 chromosomes. Therefore, this gives us a very important message: the chromosome number does not really predict how complex an organism is. So after this, we are going to look at how mutations impact evolution.

So we are going to study certain properties of mutations that are interesting in this context. And that is what will happen in the next discussion. See you.