Wildlife Conservation
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Lecture - 27
Chromosomal & genetics disorders, inbreeding

[FL] In today's class we will have a look at Chromosomal and genetic disorders and inbreeding.

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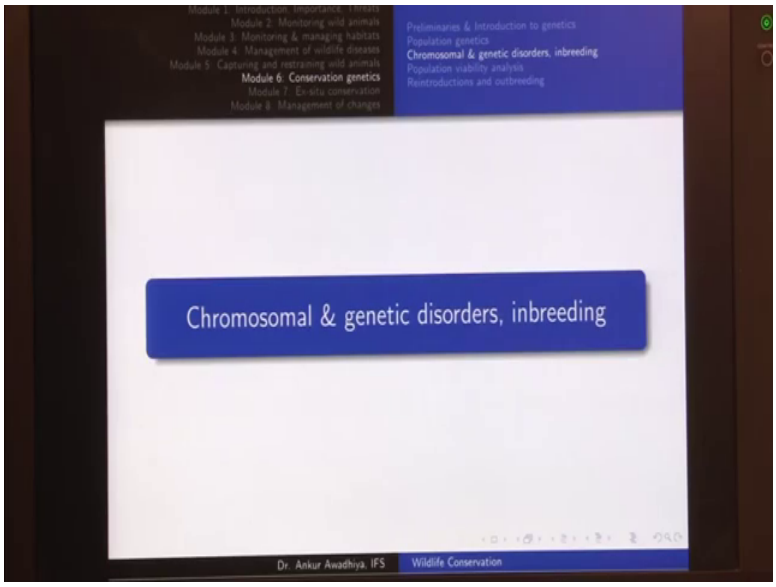

Now, chromosome as.

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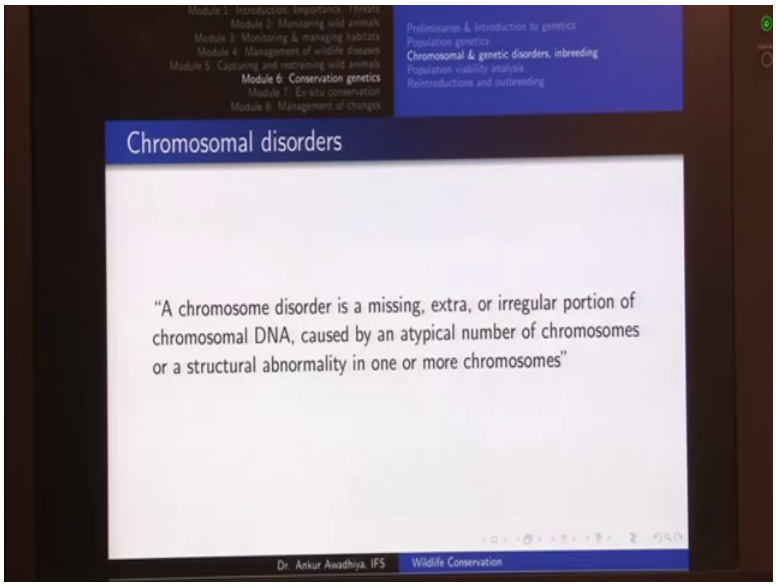

We have seen in one of the lectures before.

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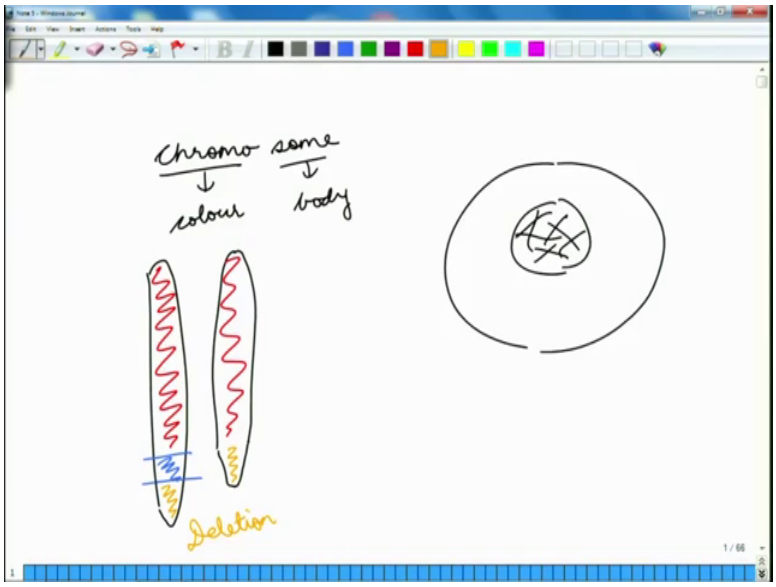

Chromosome. So, chroma is colour and some is body. So, in our in any of our cells we have a nucleus and inside this nucleus we have these fragments that contain all are genetic information and these are known as chromosomes. Now chromosomal disorders are defined like this. A chromosome disorder is a missing extra or irregular portion of chromosomal DNA caused by an a typical number of chromosomes or a structural abnormality in one or more chromosomes.

So, what do we see here? Either there is some part that is missing some part that is extra or some part that is irregular in the chromosomal DNA and this can be caused because of a typical number of chromosomes that is in incorrect number of chromosomes or a structural abnormality in one or more chromosomes.

So, what are all things can we see here?

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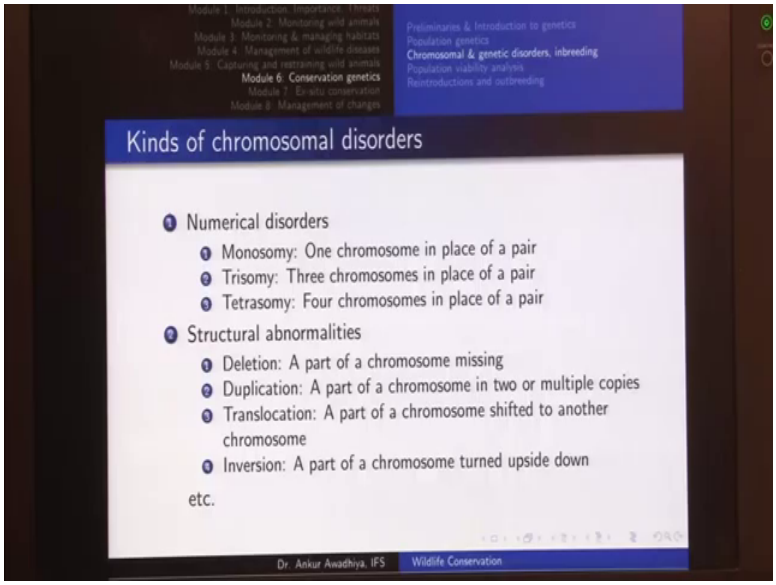

We can observe things like numerical disorders. So, all of are chromosomes are there in pairs. So, for instance if you talk about chromosome number 18 18, there would be one chromosome 18 18 that comes from the father and one chromosome 18 that comes from the mother. And this phenomenon is known as Bisomy. So, bi is two and soma is body. So, there are two bodies of every chromosome that are present in the cell now numerical disorders include Monosomy. So, mono is one somy is body. So, there is only one chromosome in place of a pair of chromosomes. Then we can have Trisomy. So, in place of two chromosomes we can have three chromosomes; Tetrasomy in which in place of two chromosomes you can have four chromosomes and so on. So, these are known as numerical disorders.

We even have a situation that is known as Nullsomy. So, Nullsomy would mean that in place of a pair of chromosomes, you do not have any chromosome that belongs to that pair. So, for instance chromosome number 18 could be missing from any cell or n organism.

So, these are numerical disorders next we have structural abnormalities. Structural abnormalities include deletion duplication translocation and inversion. So, division means that a part of a chromosome is missing.

So, for instance if this is your chromosome, then it is possible that this portion is missing and so, your chromosome will become shorten.

So, this position is missing and then it just has the other end. So, it is shortened up. So, this is a deletion a part of a chromosome that is missing. Next you have duplication a part of a chromosome in two or multiple copies. So, let us represent this with different colour.

So, let us call this as the red region, this is the blue region and say this is the yellow region. Now in the first case what we had was that we have that red region and the yellow region and the blue region was missing so, this a deletion. Now in the case of duplication what we can have is that, we have this chromosome.

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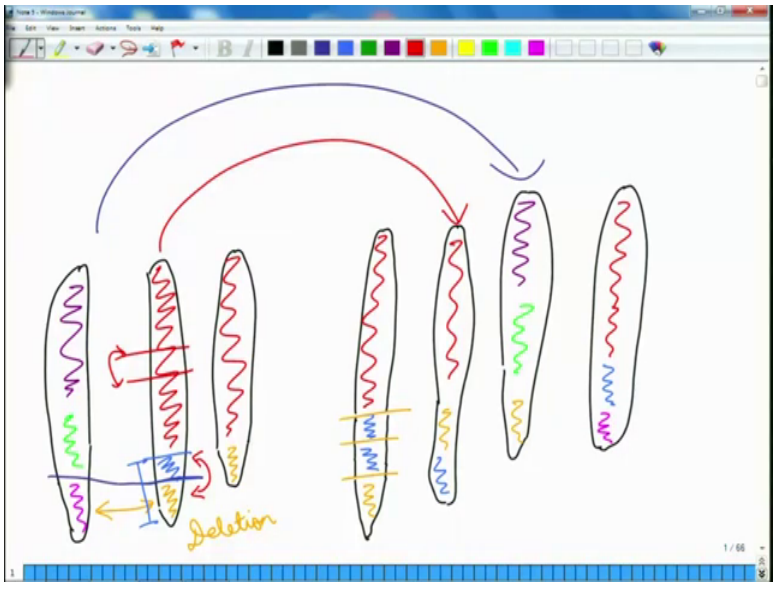

The red region is there then we have a blue region that comes that as duplicated. So, it has increase units length and then we have the yellow region.

So, in this case we will see that, this is the red region then we have one blue region and then we have another blue region and then we have the yellow region. So, this would be known as duplication because this region of the chromosome is present in two copies

when place of two copies we can have even multiple copies. Next is translocation. So, translocation is that a part of a chromosome gets shifted to another chromosome.

So, in that case, it is take another chromosome. And in this chromosome we have see the purple colour followed by light green colour followed by say pink colour. So, now, in the case of a translocation, we could have a situation in which say this portion gets trans located this portion is getting trans located.

So, that would result in a situation in which the first chromosome and we have the second chromosome. So, the first one now has the purple region on the top followed by the light green region followed by the yellow region because these two portions got translocated from one to the other and so, the second one would have the red region followed by the blue region and then followed by the pink region. So, this thing is known as a translocation.

So, a part of a chromosome has shifted to another chromosome. And the fourth situation is in inversion. So, in the case of an inversion, suppose we are talking about this chromosome only. So, in the case of inversion it is possible, that these two portions get interchanged.

So, in this situation what we will have is that in this chromosome we have the top is the red religion followed by the yellow region followed by the blue region. So, what has happened in this case is that this portion of this chromosome it is turned upside down. Now it is not necessary that this should only occur in the end of the chromosome, but it can occur somewhere in between as well. So, for instance in the red portion we can have a situation that this portion gets upside down.

So, such a situation will be known as an inversion. Now why are all of these important? Now chromosomes contain DNA that has all the genes. Now if there is a situation in which there is a deletion of a part of a chromosome, then it is possible that some genes or may be a single gene or a set of genes they are deleted along with this part of the chromosome.

So, in that case any of the functions that were being done by that gene will now no longer be in the organism. So, it is possible that there would be some proteins that are

now completely missing from the animal. So, that mean lead to a disease or that might lead to death duplication a part of a chromosome is in two or multiple copies.

So, if there was a gene that was performing a function. So, there was a gene that was producing some protein. So, we had a gene that was producing protein.

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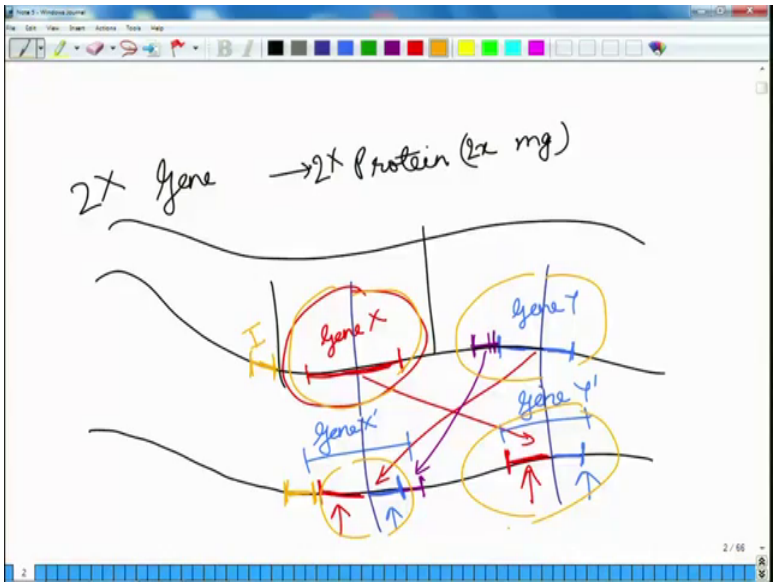

And let us say that it was producing x amount of protein. Let us call it x mille grams of protien. Now if you have two times of this gene then it may produce two times of the protein. So, we get a 2 x mille gram of protein it is also possible that if we have more and more copies of the gene in the chromosomes then it is possible that are proteins that had to be present at say this level are now presented this level.

So, now, that would also lead to some amount of abnormality in the body that would also lead to some amount of disease in the body. Next is translocation. So, a part of a chromosome has shifted to another chromosome. So, translocation and inversion now in both of these situations your genes are there in the chromosome. Whether in that particular chromosome or in some other chromosome, but then why are these important? Because these may break some of the gene.

So, for instance if you had this DNA and this DNA if you had this portion as say a gene that is call it gene x. Now when this portion is getting inverted then it is possible that your inversion occurs at this region.

So, in the resulting chromosome you will have a situation in which your half of the gene is here then this portion was inverted. So, now, when that inversion happens. So, this portion gets to this side and this portion get to this side. So, in that situation you will have that this portion has not turned into this side and is now here. Now what you are saying here is that we have a fragment of gene X here and we have a fragment of gene X here. Now in the earlier situation when you had the complete gene X this was producing some protein, but now we have got two different fragments of gene x.

So, probably your protein is no longer been produced. Or it is also possible that in these situations you may have (Refer Time: 09:29) for instance this portion on the right it was having say some other gene. So, now, this was say gene y. When this inversion occurs, now you have a situation in which you have these two fragments here and these two fragments here.

So, now in place of your gene X you have another portion of information. Let us call it gene X prime and let us call this one is gene Y prime. Now there could be situations in which your gene X prime or gene Y prime are just non functional. So, they are not producing anything or they are producing something that has minor amount of aberration, but then because this is coding for an entirely new sort of a protein it is also possible that is produces some protein that is completely harmful to the body.

So, probably it produces a protein that goes and attaches itself to an enzyme and it stops the functioning of that enzyme. So, in those situations the life of the animal would become much more critical. So, what are the impacts of these chromosomal abnormalities they depend on which genes are being impacted the level to which they are being impacted and also any new genes that get created in this manner. So, the impacts may vary, but then these are the basic chromosomal disorders that would lead to such an impact.

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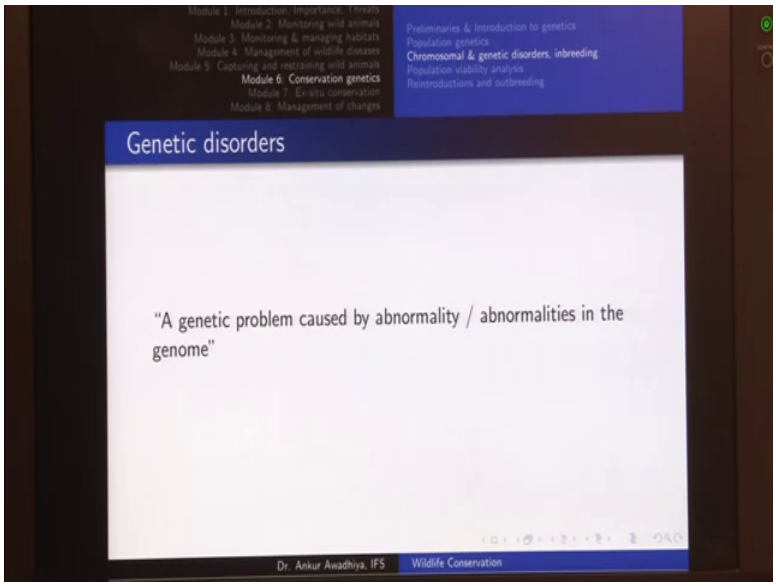

Next we have genetic disorders. Now a genetic problem that is called by an abnormality or abnormalities in the gene home. So, it is very similar to the chromosomal disorders but here we are looking at the genetic level.

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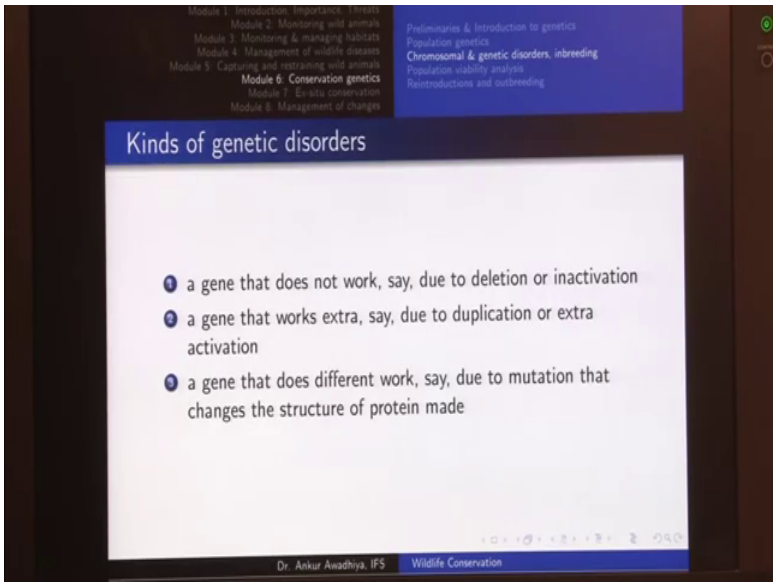

So, now, the kinds of genetic in disorders could include a gene that does not work say due to deletion or inactivation. Now deletion is something that we have already seen. So, essentially you had a gene X here. Now if this portion see got deleted so, now you would

have a chromosome that was not have any gene x. So, that is deletion, but then what is inactivation. So, if you consider any gene.

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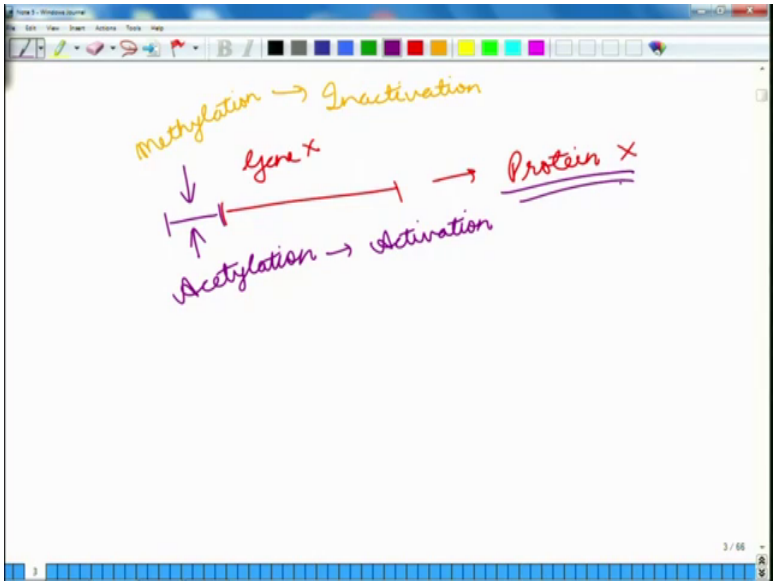

So, this is your gene X that is producing a protein X. Now the amount of protein that needs to be produced in the body has to be very carefully regulated. So, for instance if there is any enzyme that is being produced if there is no enzyme or very little amount of enzyme then the body will not be able to function properly, but at the same time if this enzyme is present in a very large amount then to it will not be able to function properly. Now to control that there are a number of activating and deactivating regions in the whole of the geno.

So, for instance if you have this gene and if you have sequences before it that say have Acetylation. So, Acetylation would mean that there are acetyls groups that get attached here. So, that would lead to an activation of this gene. On the other hand if there are some groups that get Methylated. So, Methylation is when you have your CH3 a group that it is attached here. So, in that case your gene will become inactivated. So, Methylation leads to inactivation and Acetylation leads to activation of the gene.

So, if this gene is activated it will produce protein X if this gene is inactivated it will not produce protein X. Now when your chromosomal abnormalities are leading to a situation in which your gene was not deleted, but it gets inactivated.

So, what is happening in this case is that suppose in our previous example. We had this is gene X that was inactivated and your gene Y that was having an activation area here. Now once you have this translocation.

So, what will observe is that this region the purple region now comes to the side because this side shifted to this side. Now in that case and here your inactivation region remain (Refer Time: 13:58) because this is outside the translocation (Refer Time: 13:59). Now in this case what is happening is that this gene X that was earlier inactivated. Now this gene X prime also happens to be an inactivated gene, but then your gene Y that was activated now does not have any of these activation sequences. So, your your gene Y prime even if it is able to (Refer Time: 14:21) for a correct protein it will be inactivated in this case because it does not have the activation sequences before it.

So, it is a gene that does not work. Similarly you can have a gene that works extra because that is present in multiple copies or there is an extra activation sequence that is present because of the genetic disorder. Or you can have a situation in which there is a gene that does different work say due to mutation that changes the structure of the protein made. So, in this example we had seen that are a gene Y had shifted had converted into gene Y prime.

So, now, this would be having a very different function and a very different sequence, but then it is also possible, but if you have a gene then there are some region that get changed and in that case this would also lead to a mutation.

So, your kinds of genetic disorders include a gene that is not working a gene that is doing an extra work or gene that is doing a different work making a very different protein.

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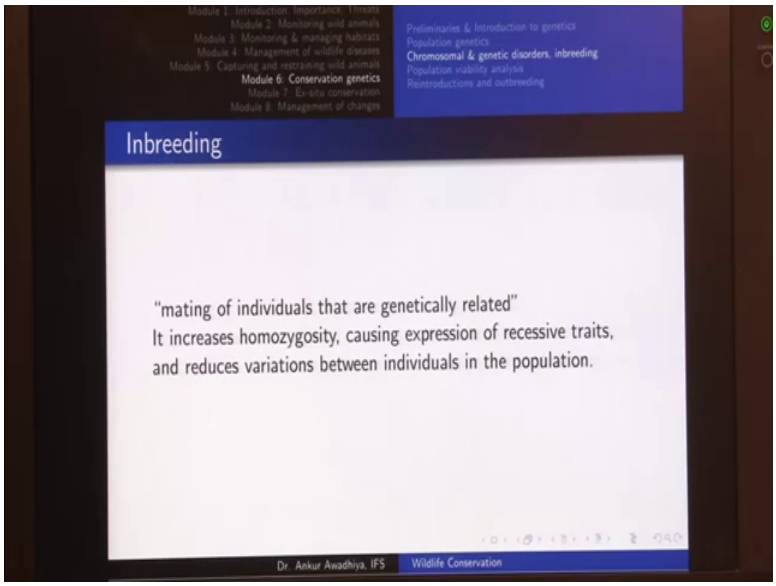

Now, let us have a look at Inbreeding. Inbreeding refers to the meeting of individuals that are genetically related. So, essentially it means meeting of individual say that are brothers and sisters or say parents and children. So, that would called as an as a very extreme level of inbreeding. Now it increases homozygosity causing expression of recessive traits and reduces variation between individuals in the population why? Because both of these individuals were genetically related; so, if there is a high possibility that both of them are have a this same genes in a number of locations. Now when both of these individuals made together when there is a high possibility that any of the recessive traits start showing themselves. So, as we had seen in our previous class if you have a situation in which you have say.

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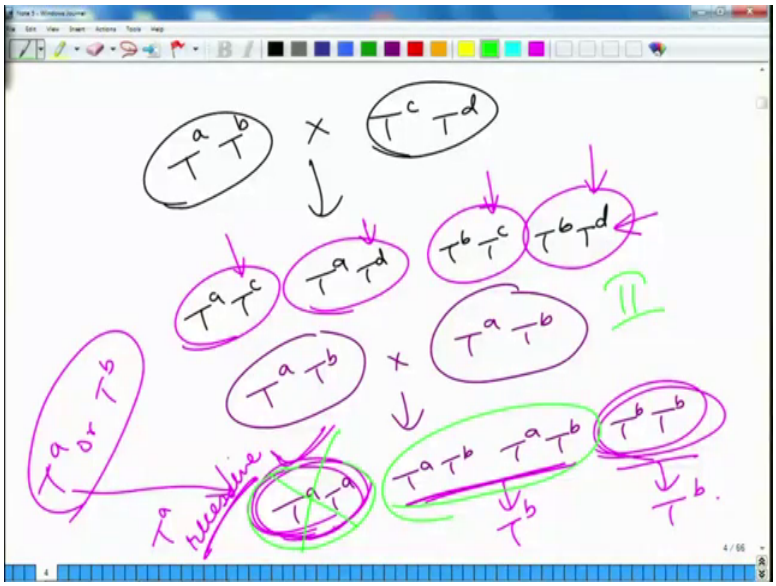

T a T b and c T c and T d. If these are four different alleles and you have these two individuals then the progeny would be T a T c T a T d T b T c and T b T d.

So, there would be these four different kinds of individuals because both of these individuals the parents are not related. So, they are having very different alleles among themselves, but then if both of these are related then you can have a situation in which you have T a T b cross with T a T b. Now what would that result in? That result in T a T a T a T b T a T b and T b T b. So, this are the four individuals. Let a form when both of your parents are genetically related. So, they are having the same alleles on this particular gene. Now if we look at the results we have the situation T a T a and T b T b.

So, these Progeny. So, now, in the previous situation we did not have any gene that was homozygous. So, we did not have a situation with T a T a or T b T b or T c T c or T d T d, but in this situation in a when our parents are genetically related we have a situation in which the offspring is T a T a or the offspring is T b T b. Now why is it important this is important because say your T a or T b was a recessive allele.

So, in this case the phenotype will be that of T c in this case the phenotype will that be of T d. In this case your phenotypes is T c and here your phenotype is T d. And if this these alleles are coding for say some recessive disorders. So, let us say that only your T a was a recessive gene here. So, in this situation these two individuals will be expression the phenotype of T b. So, here we are saying that T a is recessive.

So, these two individuals are showing the phenotype of T b because they are heterozygous. This one is homozygous, but it is still showing the phenotype of T b, but what happens in this case is that this individual has not started showing up the phenotype that was coded by the T a gene other T a allele. So, what is happening in this case is that we are seeing an expression of a recessive trait that was not seen before. Now these recessive traits might be coding for some diseases.

Now this also reduces variation between individuals in the population because what we are saying here is that in our second scenario in this second scenario we saw that both of these individuals were the same and if you look at their progeny. So, this one is say coding for a recessive disorders. So, this dies off. So, now, in the second generation also we are seeing the same alleles in the same order that are seen in the next generation. And if this thing continues then the amount of variation between the individuals we will go on reducing with every generation.

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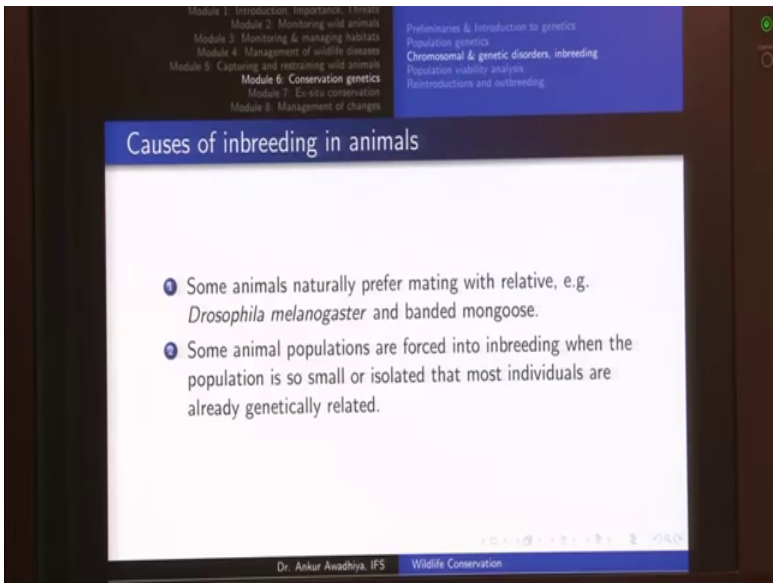

Now, in some organisms inbreeding is seen naturally. So, in the case of drosophila melanogaster or banded mongoose the animals have a tendency to prepare meeting with one of their relatives, but then in the case of some other animal populations, they have forced into inbreeding when the population is. So, smaller isolated that most of the individuals are already genetically related, there is not much of a meeting choice available for those animals.

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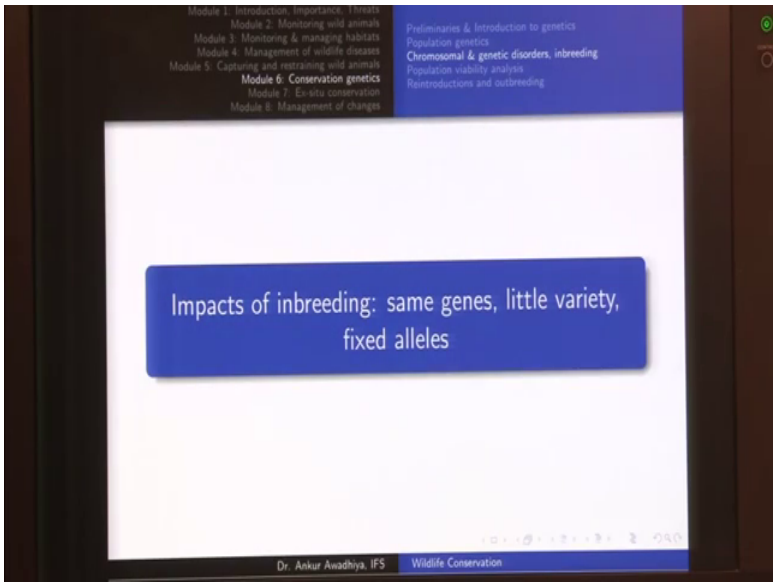

So, what are the impacts of inbreeding? Suppose you have a very small population most of the individuals are already genetically related. So, there is inbreeding. So, why should we be concerned about inbreeding? So, there are three kinds of changes that this can bring about. You can have same genes, you can have little variety and you can have fixed alleles.

So, we will have a look at the impacts here. So, what we are observing is that there are a group of animals in which most of the animals have the same genes. So, essentially all the animals are very similar to being clones of each other.

So, what happens is that when animal gets a disease that the other animal can also get that disease very fast. Because the pathogen that was able to infect one organism will be very easy able to infect another organism because the immune response that is being set up by both of these individuals are one and the same. Also if there is a little variety and also there is there are fixed alleles to fixed alleles is a situation and which you know coming back to example. Here we were observing situations in which all these you have four different alleles in the population, but then what happens.

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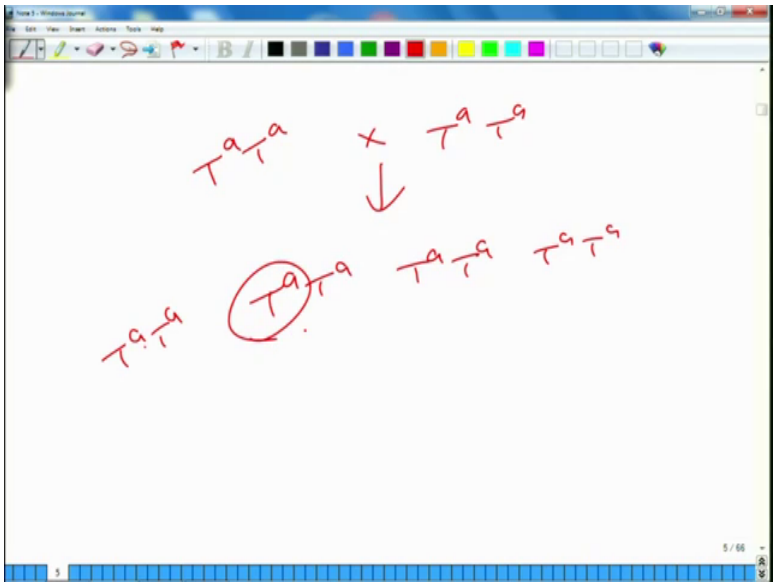

If all your organisms are see a T a T a crossed with T a T a. So, in this situation all the progeny that are formed will be T a T a only.

So, in this situation we will say that are allele T a has become fixed in the population there is no way that we can have another allele in this population till we get a mutation or till we get some other population from outside. So, these are known as fixed alleles.

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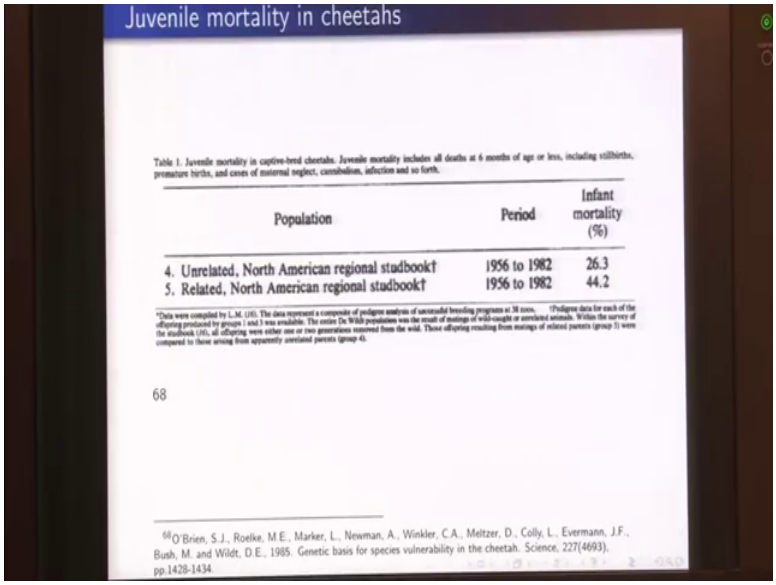

So, the impacts are seen in a number of ways such as Juvenile mortality. So, this is one paper in which they studied cheetahs with their studbooks. A stadbook is a collection of the information of their parents and the children.

So, if you look at unrelated populations we had in an infant mortality of 26.3 percent, but if you look at related organisms we had an infant mortality of 44.2 percent. So, the amount of infant mortality goes up.

Now, why does this go up because there could be a number of recessive diseases that are now showing up even in the embryonic stage and. So, this is one defence mechanism that nature has put in that if there is a foetus that is having a number of diseases then it automatically awards itself. Or even if this, this foetus is able to come up to the stage where it is born then because most of these disorders will start showing up at the early age. So, we will see a huge amount of infant mortality. So, they will be more number of stillbirths and also more number of infant mortality.

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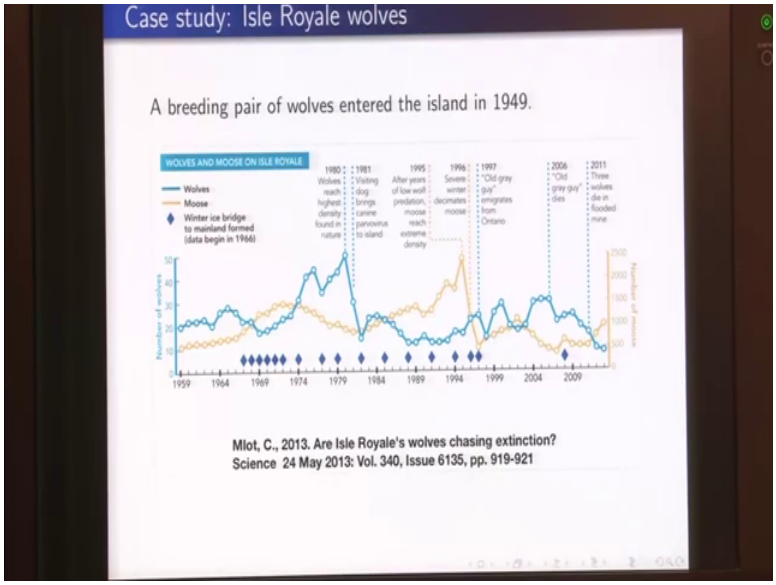

Now, a case study here is the case study of isle royal wolves. Now isle royal is an island in the United States and this island is surrounded by water on all sides and so, it is completely cut off from any other land mass. The only way in which it gets connected to the land masses is in the case of winter seasons where an ice bridge forms that connected to the mainland. Now in the 19th century we had a situation in which some moose came

into this island. And moose are very similar to our deers they are large sized animals and they are herbivorous.

So, because there were no predators on this island so, the moose population started increasing. Then in the early 1900's we had some wolf that came into this island with another ice bridge found.

So, if you look at this blue coloured chart this blue colours tells us the number of wolves and it goes from 0 to 50. The yellow colour chart tells us the number of moose and it goes from 0 to 2500. So, we have a very line number of herbivorous and a very less number of predators and on those islands, but if you look at these predators these wolves; because a very small population came into this island and this is small population was breeding among itself.

So, we started seeing quite a lot of in breeding in this system. So, what happened was that we had this moose population and then we had this wolves population. Now wolves population would decline in certain times because there is a severe winter and that could lead to death of wolves and also that of the moose. Now if you look at wolves chart. So, this wolves density is this the kept on increasing and by 1980 we had wolves that that had these the highest density that is found in nature , but then in 1981 there was a fisherman that visited this island together with his dog and that dog had canine parvovirus.

Now, this parvovirus was spread from that dog into the wolves. And we can see that their population that reach to this height now suddenly decimated. Now why was this decimation possible because most of these wolves work closely related to each other and. So, if one wolves got infected, if there was a very high chance that the other wolves also get infected. Now after this decimation they again started to increase their population, but then this population was kept at a very low phase because of huge amount of inbreeding depression.

So, essentially here we had very less number of wolves that had come inside and had this point we had another bottle neck in which most of individuals died of and. So, a very less number of individuals for lifts. So, any wolves from this point onwards would be having a very high level of inbreeding depression.

Now, when the wolf population is less the moose populations they starts to increase. And then there was a severe winter in 1996 in which we had a severe decimation in the moose population. Now in 1997 we had one individual that came in and that was known as the old gray guy. So, this individual was able to provide some amount of genetic rescue into the inbreed bulls and so, their population increased again. So, from this level its reach to this level. Then this individual also died of and then we had at these wolves that were extremely closely related to each other.

So, this is a natural case study that has come up. Now if we observe these Isle Royale wolves, what sorts of genetic abnormalities do you observe in these.

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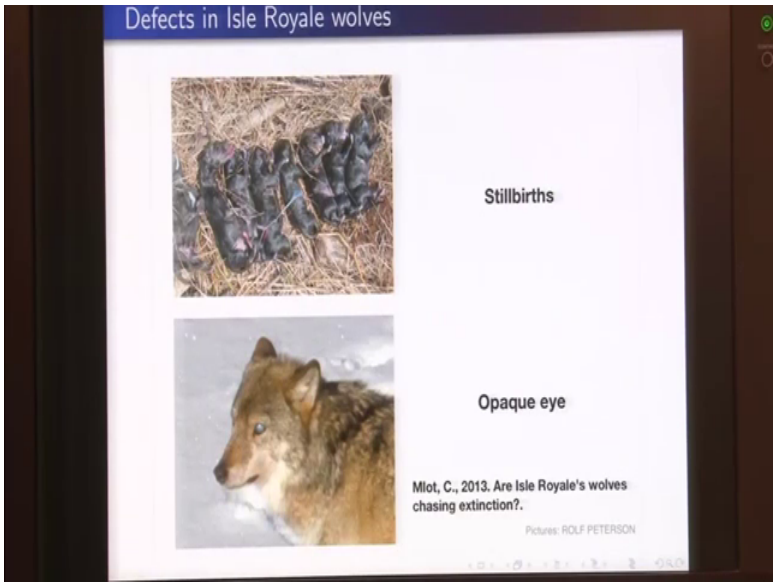

So, one is a very high level of Stillbirths. This stillbirth occurs because most of the foetuses have some amount of recessive disorders some diseases and so, nature express them out nature awards them. So, that these are not born with these diseases.

Now even in the case of the adult wolves. So, these are the wolves that were even to be born and then these (Refer Time: 27:21) to their maturity we can observes genetic disorders such as is opaque eye. Now if there is a wolf whose eye is opaque then it would not be able to hunt efficiently. And in this particular wolf population we are observing a number of wolves that are having opaque eyes which is another genetic disorder.

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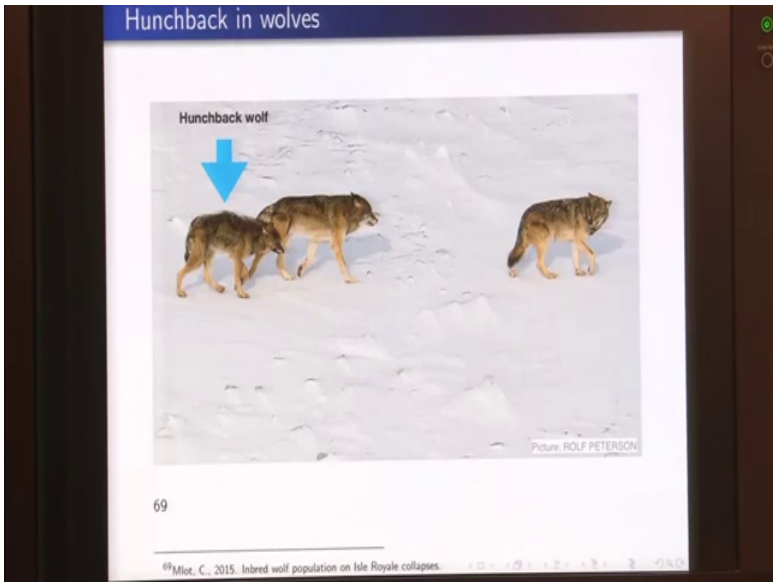

Next we are observing disorders even in their skeletal systems. So, these are the last three wolves that remain in the system. And both of these are extremely closely related and this is the offspring and if you see the offspring then there is a hunchback in this wolf and this wolf is not able to hunt properly it is not even able to walk properly. So, this is one case study.

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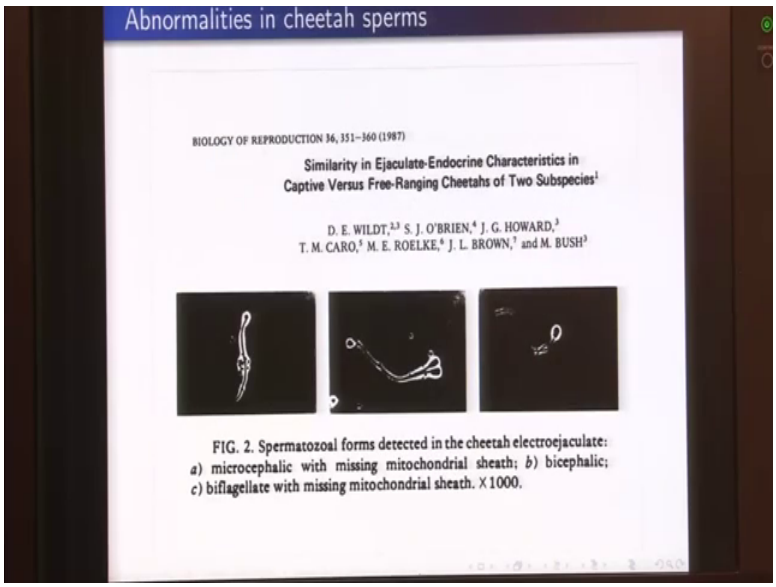

Now, the other kinds of abnormalities that we have observed in (Refer Time: 28:11) inbreed populations are things such as changes in the sperms. So, if we consider a sperm they would have a head region and a long flagellum.

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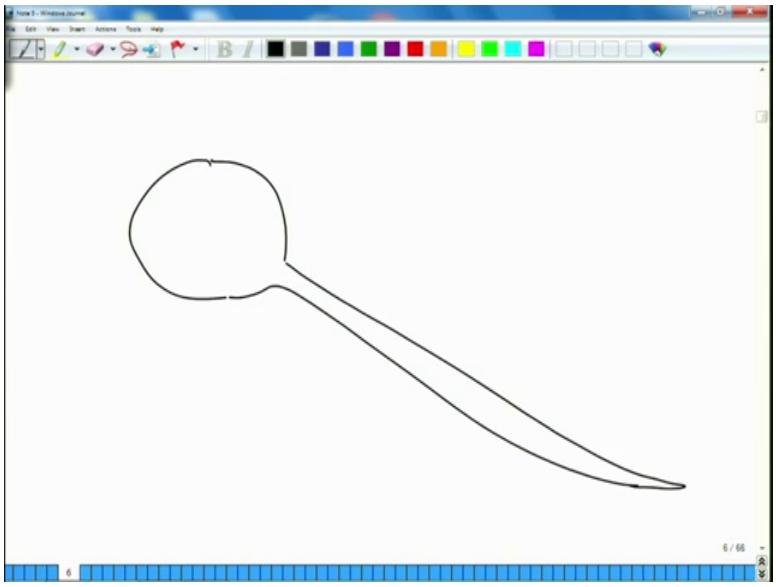

So, this is the structure of a sperm, but if you look at these cheetahs sperms which are very highly inbreed we see abnormalities such as this so, this a micro cephalic sperm in which the head region is very small. Here we have a sperm that has got two heads. Here we have a sperm that has got two tales. So, we are observing genetic or structural abnormalities in a number of cheetahs that are closely inbreed.

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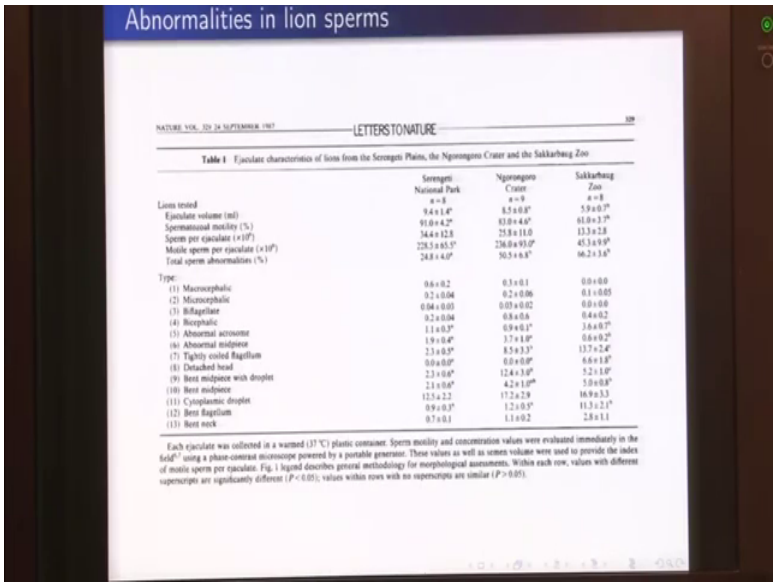

Other kinds of abnormalities that we observe. So, this is in the case of lions sperms because these animals. So, cheetahs and lions are heavily hunted in the past. So, most of the animals that I left out now are closely related. So, in the case of lions sperm as well, we are seeing things like macro cephalic that is lost his head micro cephalic that is small size head in this sperm Biflagellate Bicephalic and then we have abnormal Acrosomes abnormal midpiece tightly called Flagellum, which is not allowing these sperms to move detached head. So, your head is completely separated from the Flagellum bent midpiece. So, in all the portions of the sperm we are now observing abnormalities. So, now, this is also a result of the genetic disorder that have been brought about by inbreeding.

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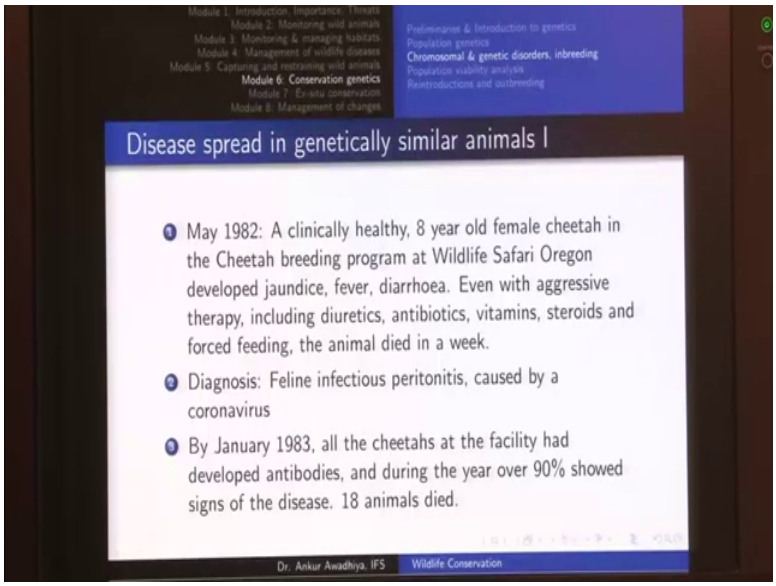

Another case study is that of the spread of diseases. So, in May 1982, a clinically healthy 8 year old female cheetah in the Cheetah breeding program at Wildlife Safari Oregon in the United States developed Jaundice fever and diarrhoea and because it was there in a Cheetah breeding program facility.

So, it was treated aggressively. So, even with aggressive therapy including diuretics antibiotics vitamins steroids and post feeding the animal died in a week.

So, we had a healthy looking cheetah that suddenly developed jaundice fever and diarrhoea and then died in a week. The diagnosis was feline infectious peritonitis caused by a corona virus. Now into this was in May 1982 and this is a viral disease. So, now, in by January 1983 all the cheetahs at the facility had developed antibodies and during the year over 90 percent should sign of the disease and 18 animals died. So, when we say that there is an animal that is showing that has developed antibodies it means that the virus has infected that animal because of which the immune system is not putting up a response to the virus. So, this response is in the form of antibodies.

So, in say around seven months we saw that all the cheetahs in the facility had been infected by the virus and in the year over 90 percent of these started showing signs of the disease. And 18 animals died out. That is a very high level of mortality. Now if you have a high level of mortality there could be a number of reasons the most common reason is that we have a virus, that is extremely virulent and that has a very high level of lethality.

So, this is something that we had discussed before whenever we are talking about a pathogen we are looking at it is virulence we are looking at its lethality. Now in this situation, we could have a virus that had a very high level of virulence a very high level of infectivity. So, infectivity which mean that it infects all the virule animals. Virulence means that it shows signs of a disease that has a level of severity and then it also have a very high level of lethality because of which a number of animals are died out.

But if you look at some other evidences from this area then because this was already a facility.

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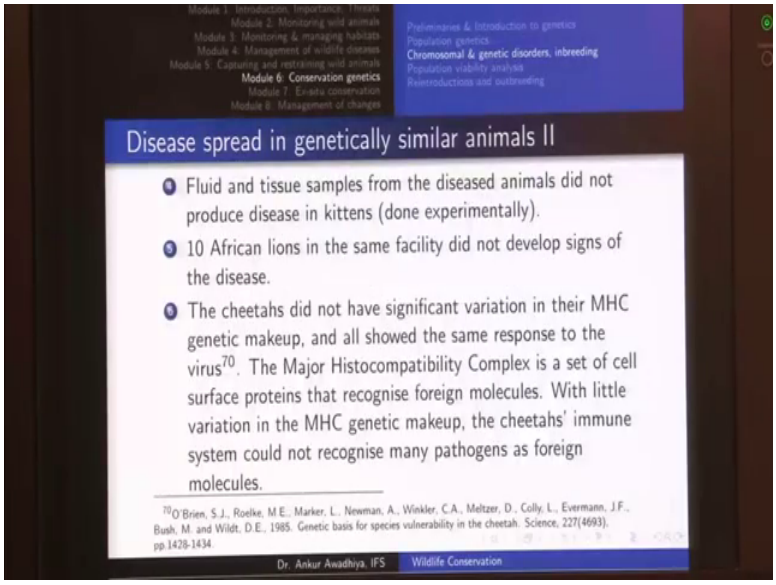

So, the scientist took out fluid and tissue samples from the diseased animal and then injected them into kittens experimentally.

So, if this virus was a virus that had a very high level of infectivity virulence and lethality, then the kitten should also have died because they are also closely related to the cheetahs they belong to the same feline family, but in this case these kittens did not produce did not get the disease.

So, this virus was not able to infect these kittens and when we are talking about kittens they already have a very small age. So, they are extremely young and we scientist is used kittens because very young animals and very old animals have higher susceptibility to be

infected by the disease, but still it did not produce disease in the kittens. Also 10 African lions in the same facility did not develop science of the disease.

So, lions are also members of the cat family and they were also in the same facility, but they also did not develop any science of the disease. So, then it was figured out that the cheetahs that were there in the facility did not have a significant variation in the Major Histocompatibility Complex genetic makeup and all showed the same response to the virus. The major histocompatibility complex is a set of cells surface proteins that recognise foreign molecules. And with little variation in the MHC genetic makeup the cheetahs immune system could not recognise many pathogens as foreign molecules.

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So, what was happening in this case is that when you have a virus inside the body you have an immune response that all it is call it field virus and so, the disease gets treated by itself. Now when we talk about the immune response then this response could be against immune organism that has come into the body. So, whenever we are talking about any immune response, this has to differentiate between self versus foreign. Because if you have this immune response against our own cells, then it would lead to a condition that is known as autoimmune disorders. So, essentially are immunes response will start killing our own cells. Now to prevent that this immune response has a mechanism of differentiating between self molecules and the foreign molecules so, the foreign antigen.

So, a virus needs to be identified either foreign antigen. Now this identification occurs with the help of certain cell surface proteins. Now these proteins are able to recognise a virus as foreign antigen, but if in case of a very low level of variation in the major histocompatibility complex.

The number of cell surface proteins that are available their varieties very less. So, they are not able to recognise this virus either as a self or as a foreign. So, basically in those situations the immune response is not shown up by the animal. So, in this lecture we will looked at chromosomal disorders genetic disorders and inbreeding. And inbreeding is especially important in today's scenario because we for a number of species we have a very few number of individuals left.

Now, if it go for a very (Refer Time: 35:35) a very aggressive amount of breeding program then it is possible that we might increase the number of animals, but then in that situation all of these animals will be very closely related and we will also be showing a number of recessive disorders. And when these animals have recessive disorders that that is that (Refer Time: 35:55) functioning such as in the case of the wolves with opaque eyes or with Stillbirth, but even when this animals are closely related and if we are able to remove all those animals that were having any of the recessive disorders.

Even then because all these animals will be nearly clones of each other we will observe a very high level of mortality if there is any infection in this population. So, these factors become extremely important in the conservation of wildlife. So, that is all for today.

Thank you for your attention [FL].