**Comprehensive Molecular Diagnostics and Advanced Gene Expression Analysis**

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**Lecture 58 : Ethical concerns in molecular diagnostics**

Namaskar. Hello students. Welcome back to our lecture series on comprehensive molecular diagnostics and advanced gene expression analysis. We are heading towards the end of this course, we are in module 12 and our today's topic is ethical concerns in molecular diagnostics and we will be covering that topic under these following headings. We will be discussing ethical issues around various types of genetic testing. So presymptomatic test, susceptibility test, carrier testing, prenatal testing, newborn screening right.

You already know what these types of genetic tests are right and after that we will also be discussing the various polarizing view of the society regarding this ethical concerns and also we will be discussing some regulatory programs which gives us the guidelines around what we need to do and what we should not write. So let us briefly review the various types of disease in very brief right. So you already know predictive genetic tests are that one which are performed on healthy or apparently healthy individuals. It is predictive.

I do not have the disease right now but I may develop the disease or I may have the risk of developing the disease in the future right. So those type of predictive genetic testing or predictive molecular testing are broadly divided into two subtypes. Number one is presymptomatic test and number one is number two is susceptibility test right. So what are presymptomatic test? The name itself is quite easy to understand. So presymptomatic test involves that we are looking for genetic mutations that have got a high penetrance.

So usually autosomal dominant means if I have got that mutation it is almost certain that I have a high chance of developing the disease right. So I am searching for a test even I am searching for symptoms I mean I am searching for the disease occurrence whether this is present or not even before I start to develop the symptoms so presymptomatic test. So these tests as you already know needs to be highly sensitive and specific. So very few false negatives or false positives right. So one example being I mean examples include

test genetic testing for Huntington's disease or genetic variants of early onset Alzheimer's disease I mean for example for Alzheimer's disease preseniline 1, PS1, PS2 mutations if they are present then is a very strong component that the individual will ultimately lead to Alzheimer's disease in later life right.

So neurodevelopmental disorders are one of the very common example of presymptomatic test. But we should know that we can identify healthy individuals right. So now when we are trying to perform the test the individual we know we can see as of now is healthy, but if the mutation is present I mean that we know in our back of the mind while you are going for the test that if it is found it is virtually 100 percent likely that the individual will develop a devastating or debilitating disorder in future at some point. And mainly for example the diseases that I have discussed right now at this time there is no cure right. So for those diseases so what we need to do we need to the patients the relatives can be mentally prepared, palliation etcetera can be arranged in time right.

So these are the various presymptomatic tests. So what are the ethical issues that might creep up? You will find these ethical issues these headings, these headers or the verticals they are quite similar in almost every type of ethical concern in any type of test. So if you at least know or if you pay attention to one type it will be much easier for you to construct your answer on the various other types of the test. So number one very very very important informed consent. So as you can read up presymptomatic genetic testing raises concern about obtaining truly informed consent.

Why? Why it is due to the fact I am asking the patient for permission but there might be an ethical consent to the fact there is potential psychological impact about learning the future of health risks or wise man said it is never wise in knowing the future. Why because if you know that the future is very good you might stop acting right now that actually made your future good right. And if you already know that the future is bad will become so sad in the present that will stop working all together right from this moment that later is the scenario in this type of test. So if someone has got fear basically so what if I do have the mutation, what if I develop the disease is a huge psychological pressure that may creep up right and that is one major concern about getting a consent from the case or the patient to whom it needs to be tested. So it is very important that ethical concern needs to be taken care of we are just discussing the concerns about what can happen.

There are remedies how to take care of that definitely. Next privacy and confidentiality. Almost with every test huge concern. So genetic information as you know is highly sensitive right. So it raises an ethical question about the privacy and confidentiality due to the fact that the test result right if someone has a positive result right he or she might

think there might be a social stigma right there might be some discrimination even if that person or that individual is healthy right now the in future if the results are disseminated right if the results are in public the society may develop very differently to that person.

So it is very very very important that the result should always be absolutely private and confidential. So this is also another major ethical concern if there is any breach in privacy and confidentiality right. As I was discussing that is related to every step psychological impact very important. So learning about one's genetic predisposal right without the symptoms I don't have the symptoms but I know deep inside me that I do have the disease lingering inside me. So that invariably if that is a devastating or debilitating disorder I know that in future I might be helpless I might not be able to move out from the bed I might need a helper to change my bed sheet or bed pan right.

So that invariably will lead to anxiety and depression right it may lead to altered perception of identity or posing or the multiple ethical dilemmas for the individual as well as their family will be there right even if the case is pre-symptomatic I do not have any symptom I just have the mutation that will lead to symptom at some point later in life may be it is Alzheimer's disease the disease will develop after 50s I am considering about early onset Alzheimer's usually develops around 60 to 70s still if I know that I am inevitable I am destined to have this disease there will be psychological burden that will lead to all of these symptoms. Again healthcare disparity this is another ethical concern because the pre-symptomatic genetic testing may exacerbate alright testing existing healthcare disparity what do we mean by healthcare disparity in a country like India specially other third world countries where there is a huge difference in multiple layers of socio economic strata the wealthy or the ones with high higher socio economic status have got easy access to multiple healthcare facilities they have got their financial means to procure their own treatment right in multiple good private setups whereas this mutation can actually be found in any individual and if the individual the concerned person belongs to lower socio economic strata it might be difficult for him to regularly follow up to regularly avail good palliative care so basically it is an issue about equality, equity and justice that raises another concern so health disparity again healthcare disparity very very very important it is much more minimized in western countries where they have got systems like either private setup like US where also not everyone has access to good healthcare but specially in European countries and Schengen countries also in Canada our healthcare is free and taken care by the government for every citizen this concern is a bit less compared to other ethical concerns right. Again very important utilization of test results what do you mean by that so ethical considerations arise how individual healthcare providers utilize this pre symptomatic genetic test result alright means suppose I have got five cases who have got the mutation right we know that they will develop the disorder so in order to probe or in order to check whether they are actually having the disorder or not the things might be at the two extreme spectrums

number one they may be subjected to over medication right unnecessary intervention early intervention right continuously or it might be so that the person actually upon getting the test result does not turn up to follow up right so there might be two spectrum number one inadequate follow up care that is a bit of negligence in terms of the healthcare policy provider or the healthcare provider because we know yet we fail to track the person that is the major ethical concern right malpractice and negligence issue although it is voluntary still again if we know we may become over aggressive so both of this there should be a balance there should be completely defined policy when to follow up care when to intervene what are the symptoms need to counsel the patient so I hope you have already gone through the class of genetic counseling and patient education so we need to make the patient aware so that they actually voluntarily turn up for proper monitoring for proper time intervention for proper timing timely screening and routine check ups right so these are the ethical issues regarding pre symptomatic very similar susceptibility testing see susceptibility testing what is susceptibility testing it involves looking for genetic mutation so susceptibility whether I am at risk right that confer a higher risk of developing the disease right so disorders are mainly multifactorial so the disease if it develops is not only due to this mutation but actually multifactorial but pre symptomatic means if I have got this mutation I will develop the disease 100% there is no other contributory factor right so these tests since they are multiple I mean there are multifactorial sporadic Alzheimer's disease I am giving Alzheimer's disease example for both genetic variation of Alzheimer's disease for example as I told you pre seniline 1 and pre seniline 2 these type of mutation leads to genetic variation of Alzheimer's disease and it is almost certain that it will develop right apo e4 polymorphism is one of the contributor of sporadic Alzheimer's disease there is no known cause of sporadic Alzheimer's disease right again mutation brca 1 and brca 2 involves a high risk of developing breast cancer not 100% of the patient will develop breast cancer right so this is known as susceptibility testing so I become very susceptible so for these type of test they have got some relaxation when it comes to sensitivity and specificity right because we can always repeat the test someone may be negative at some point they become again positive at some point right because these are mutation that may happen over time whereas in case of autosomal dominatrix transferred from generation to generation person will always have the mutation to start with that is we are comparing both pre symptomatic and susceptibility so what are the ethical issues believe me almost everything is similar we have just altered the content but if you take care of the heading you will find we can construct the heading in almost similar so regarding informed consent definitely if we already know that we might become susceptible this is absolutely similar to pre symptomatic just a bit milder right so informed consent if I know that I am susceptible I might not be agreeing to give you the consent or the healthcare provider might not be explaining me properly so that I am ready to give the consent whatever be the situation informed consent is one of the major ethical concern in case of any genetic test specially susceptibility test again same heading privacy and

confidentiality safeguarding of sensitive genetic information is the duty of the healthcare provider if it is not it will lead to ethical concern very important since it is susceptibility test there are multiple markers may be involved now a days there are multiple predictive tests right means there is a scoring I have got these these these factor these these these mutations and there is an algorithm or AI that has been fed multiple patients data of a I mean thousands and thousands and thousands and millions of data by which it has predicted it has got a scoring based on that if I have those disease what is the probability that I may develop the disease in future right so that again the predictive nature of this susceptibility test may give rise to genetic I may give rise to an ethical concern how the data is used right healthcare disparity is always there at the same disparity basically similar to pre symptomatic testing like susceptibility testing we should ensure that there is equitable distribution means everyone do get the proper provision of the genetic test or proper follow up if they are found positive or proper counseling proper treatment everything so there should not be any inequality psychological impact always there just like pre symptomatic testing susceptibility testing if I know I may be in anxiety I may be in tension I may be in depression what might happen right so this anxiety social stigma everything has got tied up in monitoring or in governing the psych of the patient right very important interpretation and actionability you see the susceptibility test are a bit complex interpreting susceptibility test result are bit complex compared to pre symptomatic test why no mutation is there is a probability that you might get the disease I may make the patient aware right but in case of all susceptibility test since these are not highly sensitive compared to pre symptomatic test there is no guarantee even if the test is positive you might not develop the disease even if the test is negative you might develop the disease so all these windows and loop holes the predictive value since they are lower compared to pre symptomatic testing again will give rise to concerns that whether in spite of being positive I might be reluctant or in spite of being negative I might turn up again for retesting even if I am positive in the back of the mind I may turn up repeatedly at the hospital the hospital will might design multiple programs for early diagnostics multiple interventions in timely interventions so all those things that were there with pre symptomatic test do apply for susceptibility testing alright. Next carrier identification what is carrier identification basically the mutation is there dormant recessive right so it is not present in this generation but I am a carrier of the disease you already know what carrier information what carrier status is right so if someone a two carriers marries right and they develop a child there is a probability that the child may have the disease right so carrier testing involves individual known to be at high risk because of positive family history right for example testing a woman whose twin whose sister has a son with cystic fibrosis it means the sister had the disease that actually penetrated in his son now that other sister the other sibling is being tested whether she has got the mutation right so since there is a family history there is a high probability that she might be a carrier although she is not suffering from cystic fibrosis right so carrier testing involves individual also involves individual with no family history for example testing all

Caucasian women of child bearing age for cystic fibrosis so there might be two scenario right number one so who are those that will be tested for example carrier mainly we are concerned about the next generation who are at risk of developing a child with a particular disease so we have included two cases where number one there is positive family history and they are being tested right and there is a policy for example I told you in western countries since CFTR cystic fibrosis trans feminine regulated gene is the most common mutation almost all pregnant women right Caucasian population are being tested for cystic fibrosis tested for cystic fibrosis so consider this case that the mother is in India where cystic fibrosis is not very common right so we almost take for sure that we do not include cystic fibrosis in carrier testing in India thalassemia carrier testing is of course often done in any prior to any marriage it is recommended right by the government however when we change the geographical demography situation then it might become mandatory so in both the cases carrier screening might involve a blanket scenario where all women are tested or someone but the situation is I am being tested I am a parent I am going to be a parent I do not have the disease but what if I have got the mutation that might lead to a disease condition in my child so multiple example of carrier screening that you have already been told you get one example for example asconogy Jewish so Jewish population has got a very high probability of T-shax disease very important mucopolysaccharidosis sickle cell anemia again very common in African American population and as you already know in Caucasian population cystic fibrosis is very common and for India and other south-east Asian countries India Pakistan Bangladesh Sri Lanka thalassemia right so hemoglobin beta chain mutation is very common so what are the ethical issues that will be there in carrier screening you see you can actually broadly divide the headings that I discussed in almost all so what else is there now very important in these type of tests we should always respect the individual or couple believes and values right for concerning test taken for assisting reproductive decisions alright suppose the couple you know asistic reproductive decision are always taken by those couples who have tried or who have got certain obligation due to which they are failing to conceive in regular or they there might be I mean concern regarding adoption so there are multiple situation where assisted reproductive technique might be opted for then again in every case of ART there are few screening tests specially for carriers that might need to be done and that depends on the policy set by the government of the country so whether someone is willing to undergo that test or not is totally up to them mind it is always optional and should never be enforced I am telling this over and over again if the test is done forcefully it is unethical so there will be an ethical concern that is rising there should one the decision of the couple or the decision of the individual what is his or her belief should always be discussed in much detail before going ahead with the test after informing consent by after making the person understand that he or she is willing to do the test on her own right very important mutation for certain diseases may have a higher prevalence in certain ethnic population raising the issue of stigmatization very important so the thing is if I get a CFTR mutation right now right I

mean in India cystic fibrosis is not much common in Indian population right however if I get a carrier test positive for thalassemia alright the situation will be dealt differently why because not everyone in and around me knows about cystic fibrosis however the disease thalassemia is known right to non medical person to layman to people of lower cephalon class they know there is a disease where the baby becomes anemic where we need to transfuse blood very frequently right and the baby will might develop a disease fracture or death in an early age similarly they might not be knowing about cystic fibrosis in India however in western population they are much more aware about their health conditions and whenever a baby is seen inhaling any mist or any respiratory disorder it is very common to understand that the person or the baby is suffering from cystic fibrosis. So, all of these ethical concern can be I mean need I mean the patient should be made aware about all of this and the concerns regarding privacy confidentiality of data everything should be considered before going for carrier testing right again very important if the patient or if I am diagnosed as a carrier I have got very few choices I mean I should be made and I should be explained properly that I should refrain there should there are some activities which I cannot do or which should I think twice before doing for example child bearing or donors donation of sperm or egg right pre implantation genetic diagnosis.

So, if I am a carrier I cannot I should refrain from child bearing provided the other person is also a carrier and because my even if the my son or my daughter does not become diseased he or she might be a carrier right. So, that decision has to be taken I should always refrain from donating my sperm or egg if in case of female person because that gamete might have the carrier gene right and that will inadvertently in I mean that might cause some problem in artificial or acid reproductive technique maybe in future. So, it should always be avoidant that should be made aware to the person. So, that might also alter the cycle alter the perception about life right the free will whenever there is a intervention or whenever there is a break in free will there will be problem in any class of citizens right very important obligation to pre implant genetic diagnosis obligation to offer education and counseling very important that person should always be offer genetic education and counseling whenever here either he is being tested for carrier or if he is tested for positive for carrier it becomes a duty of the health care provider to either refer him or to counsel him for genetic counseling and to educate him properly. If it is not done then again the practice becomes unethical and that again is an ethical concern right.

Pre-natal testing I am not going to discuss in detail we have discussed in much detail what is in pre-natal testing again very important number one respect to individual beliefs right and values is crucial potential for increase pressure right on couples not to have children right very important again this should be taken care of the couple should be made aware or this thing might deter him or might prevent him for actually appearing in

pre-natal testing at all right. They if in a pre-natal test the baby is found to have some mutation or there is some problem in the test result the couple might be under pressure not to conceive in future right whereas we know that one genetic disorder diagnosed in a baby for example all the trisomies does not guarantee that the future pregnancy might be having the same event but still that is a concern that always happens with the couple who are being tested that has to be taken care of very important possible decrease tolerance and fewer resources for those with disabilities. A disabled person already has got multiple conscious and subconscious might have all multiple conscious and subconscious burden to start with and if a person with disability with some disability is found to a positive pre-natal test that person will have a immense psychological aspect or psychological trauma that needs to be handled very carefully. Mind these are points in addition to those common points that we already discussed in pre-symptomatic and susceptibility testing I am training you so that you can design a big answer if any blanket question comes in any type of descriptive theoretical exam what are the ethical concerns in molecular diagnostics all right. Again very important ethical concern possible termination of fetus based on ambiguous information I told you while though any genetic test pre-natal specially are not 100 percent guaranteed specially those are based on serum markers right and the decision to terminate the pregnancy should always be taken after reviewing and reviewing of multiple parameters after at least performing I mean confirmatory invasive test and that should include a team of specialist doctors obstetricians including genetic specialist if possible right.

It may so happen I already told you that the positive disease or the positive test does not 100 percent mean a positive diagnosis so there can be case where even if the in the first test or the first screening test the result is found to a positive and the mother or the couple decides to terminate the pregnancy although result might have been ambiguous right it could have been in follow up care for example if the marker of trisomy is high right the decision to continue the pregnancy will always depend upon other findings and the expertise of the obstetrician in follow up it might be found that in anomalies can I am giving a specific example the baby is completely healthy but what if the baby is terminated before that advanced stage. So all of these are ethical concerns specially related to pre-natal testing which needs to be board in mind while designing or offering these tests alright. Newborn screening again all the common points will come one after another so let us see number one voluntary versus mandatory screening right in some cases in some hospital might be if they have included a panel in of newborn screening right taking into account the fact the susceptibility or the prevalence of the disease if it is justified and it is approved by the government fine whereas in major cases the test should be voluntary right. Yes if the family history is high if there is a consent we can go for mandatory screening however in any type of screening test in any type of test for that matter it should always be voluntary and the consent should always be obtained prior to the test. So if we can compare and contrast mandatory test testing versus voluntary

testing you can write your own answer I believe however you should know that there are pros and cons to both for example mandatory screening you will not miss any case right.

So if the policy is decided for phenylketonuria is a metabolic disorder related to phenylalanine test in metabolism. So I will always test every mother for phenylketonuria in my centre where baby is being delivered it is very good I will not miss a case however not all mothers might opt for the test so I give a choice right you can opt for this test you cannot opt for this test suppose a voluntary mother who has got the disease does not opt for the test then what happens that case goes the case is missed and later that baby will develop symptoms of phenylketonuria and it will become difficult for the baby to thrive alright. So lack of informed prenatal consent this is very important this is a major ethical concern that the after the test the mother or the patient relative will often might say that we did not know it was not properly informed we can we cannot simply inform the parents or the relatives that your baby has got this this this test without actually asking seeking permission from them prior to doing the test right everyone expects a healthy baby and a news that their new born baby in a family has got this disorder at such an early age might be devastating right. So the proper everything should be done so prop the patient or the relative should be properly educated this every point is tied to one another right so proper awareness proper education one should be made aware of the risk what might happen if you do not do the test what if the test is positive it is always possible to lead a healthy life with proper intervention so if these things are not done properly then there will be ethical concerns related to various newborn screening. Now very important see technology creep test what do you mean by this term technology creep right technology creep means a new technology is being added and it supersedes what was already existing right.

Now it might be so that in I am a hospital owner and I have installed a extremely new state of the art facility right and it can diagnose very very very rare diseases right. So I make it mandatory that along with all the newborn testing that was being done I will also use this machine to test other new genetic very very very extremely rare genetic disorder for all the babies that are being delivered in my hospital right. So what is the problem over here number one it will increase the cost healthcare cost of all concerned patients and their relatives right and since the disease is so rare in spite of having high technology it is often certain that most of the cases if not all will be negative right. So one needs to assess the benefit of the baby specially in case of newborn screening before applying any newborn technology even if it is available right. So I can give you examples so there are cases which were initially just done to in certain cases for example, I will tell you the gene library or gene bank all right not human genome project you know human genome project actually paved the way for all of this whatever we are discussing let us not go into that.

In America we know whenever DNA testing is done it was initially designed only to be done by the defense service military the only people who are going into war actually were entitled to deposit their DNA in the bank so that in if in case of war casualty dead remains those could be identified by tallying the DNA from the DNA bank I am talking about United States of America specially right. But nowadays you can see it is done in almost all routine investigation all routine convicts right need to I mean are actually counseled or the judge tries to get a DNA sample from every possible convict right. So you see this is basically a technological because it is so easily available because it is done so easily the original lead right has been superseded surpassed by the new system right definitely has its benefits but without assessing not every convict needs DNA right I am deviating from medical scenario over here but you understand my point what is technology creep right very important necessity for treatment and follow up prevent the damage. So for newborn screening if the test is positive one should be always aware to follow up with that mother or follow up with the baby and the information provided by the parents during discharge to proper treat to treat the baby properly to help with proper health care delivery if it is not done again it is a health care concern very important increasing pressure to use residual sample for population based research extremely important for all type of study right what to do with excess sample right. Now we already know that there is always a need of knowing new diseases right if we do not know what are the new diseases then how can you diagnose how can we make our life better for future generations and how do we know disease through research and development right through R and D research and diagnosis and for research and diagnosis we need samples.

So mainly major genetic centers are usually contacted for samples those samples are mainly samples for those who already have been tested therefore it is very common in any genetic in almost all if not all in major genetic centers a column is written or a point is written I voluntarily provide my I agree to voluntarily provide my sample for future research knowing without knowing I mean after my job is done basically right. So I know that after my sample is tested I will provide my sample for further research and diagnosis. So someone may feel ok with that someone might not right and someone might be too inhibited just by seeing it they might feel that oh your main concern is research and diagnosis you are not concerned with my well being this is a very common scenario. So all of the aspects of sample or the consent status should be clearly discussed with the patient what type of research might be performed right it will be purely confidential the data will not be traced back to the patient the patient is not answerable to anything that might happen in future right. So all of these are very important with and specially in relation to newborn screening it is not all number one parents I mean to continue parental anxiety about false positive results right even if just because if the patient is educated that a false positive result does not necessarily mean the baby will be absolutely healthy then even if after having false positive result the patient might be

anxious right.

So how to go ahead with that definitely very important one should always explain the patient that there are multiple ways to determine the health status of the baby so subsequent follow up scans for example ultra sonography anomaly scan etcetera will give you a convincing information you do not need to be anxious even if something is false positive right. For example very important the test the what is false positive right the situation I have explained is false negative that the test is negative but the patient is still anxious right. See whenever I have the disease but I do not have positive test that is known as false negative the test is negative still I developed the disease or the disease still there it was missed right what is false positive I do not have the disease however the test is positive that is a much more aspect of being anxious. So what if my I mean the couple have got high markers in triple tested quadruple test they will become very anxious to start with that my baby of course will have a trisomy 21 or down syndrome etcetera to start with again the measure is same you should reassure the patient you should always follow up before taking drastic decision invasive confirmative test can be done right. So all of these are very much should be taken care of in case of specially newborn screening right harm to parent child relationship by parental misperception about meaning of child's carrier status.

Again this thing is very difficult and situational to deal with a baby right when he becomes adult right. This is very special and sensitive user case scenario that the baby knows that he or she is a carrier right and where from he got the disease he got the disease from one of their parents right and the baby might blame or accuse the parent or might think that I got the disease from him or her right. So that might lead to some friction or relationship gap between or misconception in parental parent child relationship right and there is nothing to be done and this thing should be discussed with anyone who has appeared for any type of testing newborn testing carrier testing what not right. Because in newborn screening also something that might have been overlooked may be evident right again very important possibility that the children will be subjected to needless and potential risky medical intervention or monitoring. This is very common whether it is an adult who has got positive susceptibility or pre symptomatic test or be it a newborn screening where the baby appears to have a positive disease everything might lead to an over intervention status right and that may actually jeopardize the normal health of the baby right extremely important.

So that actually brings us to the end of all the ethical concern that is possible in relation to multiple genetic testing. So now let us in few slides discuss about what are the views of the society as I told you in the very first slide in the concept the view is actually very polarizing in certain cases right. So the society the many literatures actually have divided the community into two types based on how they feel. Number one is genetic

libertarians. So who are they? They feel that the patient have got a have a right to a full and complete accounting of all possible risks right conveyed by both established and noble variants found through genetic testing right or even variants of unknown significance in disease genes.

You understand my point? Those who believe that I have got the right to know right I have got the right to know everything. So I every testing should be done and should be informed right because what happens to me is my own property right. I have got full right to know and genetic testing should be done even if it is very rare or novel variant everything should be done. Empiricists who are they? Who believe that this is of no use or not much has happened therefore why do this right? So those who believe there is insufficient evidence about the penetrance of most pathogenic variants in general population to warrant the sharing of incidental finding right and it is irresponsible to create a psychological burdens of being a patient in waiting or to expose the patient to any unnecessary surveillance or diagnostic testing. So see a situation where I have got an incidental finding right very rare disease right.

The disease the mutation is there, but it may be so that the it is proved in literature that not everyone who has got the disease will develop or who has got the mutation will develop the disease. So now there are two classes of people, two classes of scientists or two classes of what I can say I mean social beliefs. Number one yes this thing should be informed to the patient alright because they have got the right to know whatever there is if it is supported by literature he or she is entitled to know the same and those who believe in that are not genetic libertarians. Whereas empiricists they are more empathetic right they feel that just by informing this to the patient it will unnecessarily create a psychological impact, it will create a burden, it might create anxiety all those negative points over medication unnecessary risk right depression social stigma can be avoided just by not telling right. So it is very difficult to comment what is right or what is wrong right you might belong or you might feel that one is right or one is wrong or you might actually feel that both are right in their own aspect right.

So it is important to just know these two terms and future the policies will tell I mean for certain diseases definitely the approach of the libertarians are much better and for certain diseases even I feel the person who has got social difficulty I mean economic status that might burden the site might be altered right. So I support both the views of libertarians in some cases and empiricists in some cases now it is up to you who you find suitable enough right. Now above all these are few regulatory programs number one is ELSI so ethical social ethical legal social and research implication implication research program ELSI that is actually by NIH national institute of health so what is this program this is a western program I will be discussing two one is western and one is Indian. So ELSI program that basically this fosters the basic and applied research on all the ethical

social legal implication related to any genetic and genomic research right for individuals families and communities. Why this program was founded basically the goal is to develop new program to develop new disease to know about new disease right so concept is that the new technology of gene identification may give rise to problems that can be minimized if anticipated and dealt with properly very important on one side as much as we know that knowing about a disease more and more research will give rise to more and more answers right.

However there might be a situation where something might go wrong and anticipating that knowing that problem dealing at the proper time may prevent any problem to any ethical concern. So basically all of these guidelines are clearly described in this program so basically if we just look into the multiple aspects of the program you can follow the link or you can simply search ELSI program in any browser and it will direct you to the website. So number one is ethical scrutiny so what does what is the is based on what number on assessing the moral implication of technological advancement right morally whether it is right or wrong very important right. Number one whether proper legal I mean adherence is followed right whether everything is done lawfully so the existing law and proposing new rules and regulations when necessary again ELSI program takes care of that social equity I told you health discrepancy should always be avoided and ELSI program has a clear guidelines about that. Again cultural sensitivity recognize and diverse perspective and value in technology adoption are being done by ELSI program so depending on what specific geographic location or what population I need this type of test right or I need this type of technology or whether I do need at all do need this type of technology again they are governed by this number one and they are continuously formulating policy guidelines those who are responsible for I mean to improve the innovation and societal well-being right.

So these are the basic major headings based on I mean based on which the ELSI program is standing alright and comparing similar such regulatory program in Indian context we can quote PCPNDT act that is preconception and prenatal diagnostic techniques act right. So basically this is already present in the this is the screen shot of the ministry of women and child development government of India right NIC. So basically again if we just review the PCPNDT act mainly it is to prevent sex determination and prevent selective abortion there is a huge stigma and taboo that if a female child is diagnosed in utero there have been there are multiple evidences female feticide is a big concern based on which government was suppose I mean was actually decided government decided to enact this rule and now sex determination prenatal sex determination is banned in India alright because it is a major unethical thing that was being done. So prohibition strictly prohibition sex selection before and after conception limits prenatal diagnostics to medical necessity. So in our cases only and only if there are there is a need for some prenatal diagnostics only then the medical diagnostic or

prenatal testing will be done.

There are multiple details in the PCPNDT act again you can go through all of them beyond the scope of this lecture discuss the entire PCPNDT act but this name you should be familiar that regarding genetic testing in India our regulatory act is PCPNDT act very important diagnostic centers must register under the act before ensuring before doing a genetic test so that they are adhering to their rules and regulation. There are harsh penalties if some testing center even does any or any violates anything that is beyond the scope of PCPNDT act there are multiple harsh penalties that are imposed upon both the technician, the consulting doctor, the radiologist as well as the owner of the center and there have been multiple instances of the entire license being revoked and the center permission being suspended. Basically and a huge part of PCPNDT act is basically performing multiple awareness campaigns and enforcement to empower women and combat gender discrimination. This is one aspect of prenatal testing in for which regulatory program do exist in India. So to summarize in our today's class we have discussed multiple ethical issues related to all type of genetic and molecular testing right we have we have known now what do we mean by empiricists and libertarians in case of genetic test and we have also briefly discussed various regulatory programs in NIH as well as in India.

So these are my references for today's class and I thank you for your kind attention. Thank you.