Comprehensive Molecular Diagnostics and Advanced Gene Expression Analysis

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Lecture 50 : Genetic counselling and patient education

Namaskar. Hello students, welcome back to our lecture series on Comprehensive Molecular Diagnostics and Advanced Gene Expression Analysis. We are in module 10 lecture 50 and today's topic is Genetic Counseling and Patient Education. Rather it is a part of the ongoing topic that we have started in last class where we discussed about various aspects of infertility, there is reproductive genetics, what are the genetic factors, but dividing I mean driving infertility we discussed about both the male and female aspects of genetic reasons of infertility. We will continue that is and you know with that we need to order genetic tests, genetic testing which already have been taught in detail in this module right. So, we are now moving into the aspect of genetic counseling right.

We were exposed a bit to this topic in the second lecture of this module where we discussed about genetic testing in primary care. So, there was a bit of information regarding counseling which needed to be done in the primary center, but here are the detailed topics. We will be dividing our lecture based on these sub headings we will discuss what is the need purpose of genetic counseling indications, why do we do it, who are the beneficiaries right, what are the phases I mean there are definite steps in counseling where we can apply the counseling. We will be again briefly recalling what are the role of the primary care provided the nurse especially and also we will be touching on the patient education aspect.

So, the term genetic counseling actually was coined by Reed Sheldon Clark, Reed coined this term back in 1947 right and he published his data in the book medical genetics right his concept so, then the term was coined right. Since then the term has been there, but it has been actually adopted much later when the human genome project the genetic sequencing all those things the answer to all disease lies in the genes that after that thing was established after genetic testing was done then the genetic counseling came much more into importance. So, what is genetic counseling? Basically it is the process by which the patients or relatives right mind these are the few keywords you can always I mean describe thing a concept in your own language once you know what it is right. Patients or relatives at risk of an inherited disorder right it can be inherited as well

as genetic. Now at this point you know that all genetic disorders might not be inherited right are advised of what of the consequences and the nature of the disorder they are told about the disorder and what outcome there can be right.

The probability of transmitting it whether I can I mean affect someone else right someone else be maybe my sibling, maybe my son, my daughter, the next generation right. So, people might not be aware right. So, it is the job of genetic counseling to educate them right and the options that they have in managing the disorder right in family planning right in order to what do what in order to prevent. So, they can prevent the disorder from happening avoiding the disorder right or ameliorate even if it is diagnosed they will take definite steps to treat it or take care of it right make lifestyle modification. So, these are the broad way how we can define genetic counseling it is not definition it is a factual statement all right.

So, if we know the definition well we can actually jot down the purpose. So, the purpose if we have to jot it down point by point number one will be to state the medical facts regarding the diagnosis, how it will be diagnosed, what happens, how the test will be positive, what is needs to be done right probable course of the disorder, what if what will happen if you get diagnosed right, what are the changes in life, what are the disease manifestations right all these are to be told in genetic counseling and the available management what are the treatment options. Again explain the pattern of inheritance. So, not only a disease if it is genetic it is fine if it is a inherited disorder all genetic disorders are not inherited right. So, the pattern of inheritance based on a disease should also be explained a mitochondrial disorder is inherited maternally, a x linked disorder is inherited paternally, an autosomal dominant disorder can be transmitted from generation to generation and autosomal recessive disorder will only be transmitted when someone of the same recessive gene if he is a carrier gets married to someone who is also a carrier even then there is a chance of only 25 percent.

X linked recessive disorder only expressed in females only expressed in males, but the females are carrier all these options and knowledge about the pattern of inheritance needs to be clearly explained in a disease specific way to the patient who is being addressed to regarding the disorder. Again understand the options of dealing with the risk of occurrence right. So, the treatment option, the management option, lifestyle option everything should be clear. Again course of action what if the disease diagnosed a thalassemia carrier right when he or she is being counseled about what will happen if he or she marries a thalassemia carrier positive partner the baby if he has she has got major thalassemia what can be the problem what are its course of action right. So, everything needs to be given clear cut picture also if someone is diagnosed of a genetic disorder to make the best possible intervention the make possible make possible adjustment to the disorder right of the affected members right or the risk of recurrence of that disorder

right.

So, see intervention in preventing the risk right. So, if I know something and I do not do something that is an intervention that is an adjustment that I do in order to avoid the disorder. So, it is very important patient education patient knowledge is extremely important and that is why genetic counseling is so much important because once the disease happens we have got very less to do the main thing is about prevention the main focus about prevention. So, what are the indication I mean why we should consider in which cases we do consider genetic counseling. Number one confirmation of screening test we have now known many screening tests that are done in order to filter out.

So, those diseases I mean those tests those marker gives us some rough idea about the disease. So, for example, neural tube defect or down syndrome these are all inherited disorder these are all genetic disorders. However, if the triple test or quadruple marker are positive they give a suspicion as to this disease might be happening in the fetus in the baby right. So, before confirming the screening test by invasive test right the couple needs to be concerned genetically that is what is the risk right what if I mean they should be allowed to explain I mean they should be explained the thing in order to have options open they should understand what is the gravity of the disorder. So, what are the risks of the confirmatory test the risks of miscarriages increase as we go for invasive test all these have been discussed all these just as we discussed also should be made known to the patient via genetic counseling this is one criteria right.

So, confirmation of screening test if the marker is an abnormal result right again if so, from screening test we are now we have now counseled the patient that we should go for an invasive test because often the screening test might be positive there are false positive, but an invasive test always gives a positive I mean confirmatory much confirmatory outcome. Then amniocentesis is one such example of an invasive test gives an unexpected result. So, now, we know that there is a chromosomal defect in the unborn baby what is the now this case again should go to genetic counselor why because now the genetic counselor should explain the risk. So, this is a chromosomal abnormality the baby will have so and so problems. So, this decision should be taken to terminate the pregnancy right now because if the the baby is born with the symptom there will be so much problem.

So, all the health problems all the lifestyle disorders are to be born this pain has to be born by the child and the parents right. So, the best thing will be to treat the condition before the baby is born right. So, all these needs to be counseled needs to be informed to the patient. Again candidate for I mean indication of genetic counseling. So, either parent or close relative has got an inherited disease or birth defect all right or parent already has a children with birth defect right.

So, either any of the partner has got defect disease themselves or they already have an issue who has got birth defect all right who has got genetic disorder the child. So, one child with genetic disorder definitely the couple is a candidate for counseling genetic counseling that again is an indication why they should be considered about I mean they should be considered for genetic counseling. Very important the next point the mother had two or more miscarriage and the or a baby who dies in infancy. So, a very high suspicion of genetic disorder is there again genetic counseling is must be for anything any right high maternal age right specially if the patient is having pregnancy for the first time I introduce this term to you primigravida. A primigravida who is more than 35 years of age should always be genetically counseled along with all the provision of the prenatal diagnostic markers right.

And consanguineous marriage the partner is blood relative all right. So, in case of consanguineous couple or the partner who is a blood relative there is a high chance of genetic disorder. So, again that is also an indication a case where the couple should be subjected or should be sent for genetic counseling. Now, if we need to jot down the beneficiaries the point that I just told you the indications basically the couple who are having those indications are actually the beneficiary. So, you can easily deduce it on your own even if I do not I mean enumerate the slide.

So, see condition might be different noted the indications are basically the cases of genetic counseling. Beneficiaries are the people to whom genetic counseling is done. So, they are basically two same sides of I mean both sides of the same con right. So, people who have got genetic condition or birth defect definitely they are beneficiary of genetic counseling. People who have a child with birth defects either having own or having a child right.

Parent who have child with developmental dealer mental retardation any other problem with growth and development right again. So, documented genetic defect in a child again whose parents for the next pregnancy should be counseled not only for the next pregnancy. In case of any developmental delay a direct genetic testing for the baby also needs the genetic counseling of the parents what will be the outcome, what will be the lifestyle modification, what can be done everything is a part of genetic counseling and again they become a beneficiary. So, two right I consider it two the guidelines often change text books depending on which country you are at generally accepted terminologies two or more miscarriages foreign or infertility of unknown cause. So, definitely we need to do genetic counseling, we need to explore the genetic factors of infertility and whenever we are exploring the genetic factors genetic counseling is must right.

Again people concerned that they may have an inherited tendency to develop cancer very important we are not only confined to reproductive genetics, but also any disorder that might have a genetic component I told you BRCA mutation for example, breast and ovarian cancer there was a case in my family do I have a cancer, do I have a tendency to develop cancer, do I have the mutation again I now become a beneficiary of genetic counseling. So, these are the various cases you can think about they are the beneficiaries of genetic counseling, it does not end here people concerned again I have a suspicion that in my family there was a development disorder neurological disorder for example, Huntington disease, Alzheimer's disease there are definite genetic components. So, am I predisposed so that I may develop the disease again I need to do genetic testing for myself I need genetic counseling. A person or doctor whose care giver has recommended genetic evaluation or genetic testing this is a basically of course, a no brainer suppose I may not have any suspicion, but my physician family physician has recommended me for genetic counseling now definitely now I become the beneficiary. And pregnant women as the cases that I have already told you 35 years old consanguineous marriage testing with pregnancy confirm that their baby may have a genetic disease that is markers, family history of genetic diseases, mental deterioration, birth defects.

So, basically I may be repeating things, but this is for you so that all the most common causes that are the candidate of genetic counseling should be I mean you should be aware of all of them. So, we move on to the phases of genetic counseling. So, they are done in step by step manner. So, genetic counseling also has got definite phases. So, the phases are assessment phase, diagnostic phase, analysis phase, communication phase and referral and support phase right.

So, we will and it comes one after another right. So, we will start with this one. So, assessment phase what is being done in assessment phase happens mostly in primary care. So, this is done. So, initial interview so we need to make sure after at the end of assessment phase all these tasks are done.

So, initial interview with counseling and family for preparation of counseling right they need to made aware right. So, they need to made aware whether I mean they have they might have been referred or they may be have come on their own right. So, out of curiosity so we need to collect all detailed history collection of family history, relevant history, consideration of any potential diagnosis right based on the history because from many historical patterns very important, recalling of who had the disorder in family. So, searching of collection of family history and relevant history is very important I mean if we can roughly construct a pedigree chart. Suppose my mom has this disease, my maternal uncle has this disease etcetera etcetera then it will give a high suspicion that the lineages from the maternal side right genetically.

So, the all these things are done in assessment phase mind it very important, seeking question might prop up pedigree chart construction is done in the assessment phase of genetic counseling. Next when do we do the tests in the diagnostic phase. So, the goal of this phase is to make a diagnosis of a particular genetic condition or syndrome right based on various test right. So, when it is done mainly when a child is born with multiple defects or problems it can be done in many phases mind it is a part of counseling only right. So, the whole process of counseling involves the complete package right.

So, during counseling only after the assessment phase when things are I mean he or she is made aware of what can happen. So, with the help of I mean when we are seeing the results and we are making a diagnosis that is basically the diagnostic phase. So, what are the tests basically any chromosomal analysis biochemical test x ray or any tissue biopsy molecular DNA testing there might be many samples that can be collected for confirmatory test and all these come under diagnostic phase just the testing right. Now the test results have come now comes the analysis phase. So, this result is positive this marker is positive not all marker might be positive someone might have only alpha phytoprotein ray someone might have only two marker histone inimine someone might have all the markers normal, numerical translucency increase someone might have PAPPA increased all the markers are normal right.

Again no changes in USG only changes in triple marker right many combinations can happen because science I mean medical science there is no one plus one that becomes two patient to patient case to case reports vary a lot there is a lot of diversity in normal physiology. So, you can expect that things might not always go as per the guidelines. So, how we need to explore the thing how we need to analyze we do our literature search and review of the information right we do extensive I mean what are the cases with these type of findings that have been documented we need to gather a team of experts and we need to consult with them right. So, basically we need to compile all the information and ultimately determine the risk. So, that is done in analysis phase all right communication phase right.

So, all the results have been analyzed we now know the team who is doing it right they know that whether I mean what is the clinical picture. Next part is communication phase where the result needs to be communicated to the beneficiary. So, communication of the result and the risk to the counselee and the family all right. So, the one who is being counselled the one who is being tested upon we need to communicate the result to him and his relatives and the family right also discuss the natural history of the disorder if diagnosed I mean now you have been diagnosed or now that you have seen that the baby is having chromosomal disorder what can happen what is the natural history of the disorder right most of these babies for example, dies at 3 to 4 years of age.

So, this is a natural history. So, what are the treatment option the in case of thalassemia the every month or every 2 or 3 month the patient has to be given blood sample I mean blood transfusion again that will lead to splenectomy, splenomegaly. So, all these things in the language in which they can understand that needs to be explained right. A patient will have a lot of question those questions I mean and needs to be answered right. So, review of questions again assess the counselee's understanding about the fact very important a one way communication just by informing the person or the count beneficiary this this will happen does not serve the purpose we again have to reevaluate ask him or her have you understood it you we should always ask him or her to explain. So, I mean can you tell me what did you understand.

So, that I at all is he aware of the thing it may. So, happen if an after test is positive patient might be in shock or he might be in denial. So, all these things needs to be considered. So, assess the counselee's understanding about the facts relevant hereditary pattern and diagnostic management. So, everything what we know as a physician as a primary care physician or genetic counsellor a whether the patient has understood or not right and all the explanation should be culturally appropriate right.

So, there should not be any miscommunication. So, in his or her own language I mean the mode of communication channel of communication in which he or she is familiar everything should be properly communicated right. So, that he understands everything that is a part of the communication phase. Next part is referral I mean this counselling can be done at a physician level can be done at primary care level right very few cases there might be a need for referring to a super specialist genetic centre or further intervention right or maybe any special test that might not be there at the centre then this referral and support phase will come right. If a centre is already equipped with a state of the art facility then the intervention can then done at this centre only then there might not be a need for referral, but for lower centres when a patient is being sent to higher centre especially in United States in multiple western countries and in few higher centres of India they have got specialised genetic unit.

So, suppose in a gynecological obstetric unit the mother or in an oncology unit they are being counselled about these factors, but final decision is always they are being sent to a genetic clinic where these specialised tests are done where these operations are being treated. So, all these come under the referral and support phase. So, refer to an individual to a specialised genetic for further intervention that is for prenatal diagnosis or treatment modalities again. Next what happens support decision made by the counselling very important there might not be referral, but there should always be support right a patient might emotionally break down after hearing the result might be a very valuable precious pregnancy and a role of I mean a news of chromosomal disorder might be heartbreaking right. So, emotional support provide psychological support not only that once the patient

is gone it is the responsibility of the counselling centre to regularly follow up and evaluate what is the outcome right.

It might so happen that they may conceive another pregnancy even if this pregnancy is terminated they may not go to a genetic counsellor because there is an notion that the genetic counsellor always does the tests and give you a bad news right. So, let us get done with it again that that is a common mentality that may happen in specially lower socioeconomic classes, but then again a baby born with genetic abnormalities much more painful to bear compared to terminate the pregnancy when there is still time. So, all these factors emotional support psychological support support of the decision psychiatry counselling all these things are a part of the extended genetic counselling which is a part of the whole package right. Now, regarding applications of genetic counselling these are the four major areas in which genetic counselling can be applied. So, prenatal genetic counselling when a baby is not born, pediatric genetic counselling in children, adult genetic counselling in case of genetic counselling when a disease is being diagnosed in adults right and again cancer genetic counselling itself whole new branch because the when the diseases are malignant the cancerous then again genetic counselling will be needed.

So, let us touch them one by one I will not be going into much details in case of prenatal genetic counselling you already know higher I mean a old mother old I mean more than 35 years or old, old enough to have a high probability of genetic disorder right. So, previous history I mean change in chromosome may lead to mental retardation, birth defect this you already know. So, basically any such suspicious case again for prenatal genetic counselling they should be offered other choices for example, ultrasound serum, AFP, chronicle sampling, amniosteum. So, basically all these things that you know you can now write your own answer if you are given a short note about genetic prenatal genetic counselling. So, any suspicion whenever there is a probability that there might be a genetic disorder even if the baby is not born prenatal again consanguineous marriage very important falls under prenatal genetic counselling.

Pediatric genetic counselling for example, the baby is born families of pediatric patients seek genetic counselling when the child has features of an inherited condition might be mental retardation, might be dysmorphic features a body abnormality, might be many mentally retarded growth abnormality. So, when all the known reasons are being explored a genetic testing might be done again genetic counselling plays a big role mainly in case of counselling the parents of the pediatric population. Adult genetic counselling definitely I told you not all genetic diseases are inherited, but a genetic diseases on exposure to any chemicals any radiation may easily happen right. So, basically an adult it may or may not be inherited right. So, maybe the mutation is such that the disease only exhibits the symptoms they become symptomatic only in adult

condition not in pediatric condition right.

So, that patient will always be oblivious will never feel the need of visiting a genetic counselling centre unless he is very much aware that he had the history of disorder, what is the query I mean there is a chance all these things of course, they are the beneficiaries they also fall in the category of adult I mean genetic counselling cases. However, the one of the typical cases are we did not have any disease, but suddenly I am becoming symptomatic and it is not explained by any other factors then the probing into genetic reasons again role of adult genetic counselling right. Again see birth of a child with obvious features of genetic disease now I am a parent it is not a pediatric genetic counselling right. So, see this has got overlapping domain right. Adult genetic counselling mainly consists of adult parent.

So, it may be so that I had a baby with a genetic disorder now or my brother had a baby with genetic disorder now being a married man I want a genetic counselling whether to I mean explore whether if I marry whether my son or daughter will also have the genetic abnormality or not right. So, basically may lead to consideration of pre symptomatic genetic testing. So, basically whenever someone is curious about something or a disease that has expressed in adult phase adult genetic counselling right. Again cancer genetic counselling whenever there is a propensity of a cancer inherited cancer for example, early onset breast ovarian or colon cancer in multiple generation right or even in case of non malignant condition in adult condition suppose the patients have got multiple kidney disease many patients have got kidney disorder polycystic kidney genetic reasons might happen right. And coming back to cancer centre I mean cancer genetic counselling all those cases I mean the genetic when they come to a genetic counsellor what the genetic counsellor needs to discuss about the chances of cancer in family whether it is related to dominantly inherited gene that gene counsellor discuss the options what is the probability of developing a cancer right what are the treatment options what are the drug that are available course I mean lifestyle modifications everything again comes under the purview of cancer genetic counselling if there is a cancerous condition involved in the disease process.

So, what is the role of primary care providers next we move on to primary care provider that is nurses mainly in the peripheral centre very important see these are very easy. So, you can just place yourself in a situation where you are the one who is sitting in the other side of the table and a patient has come to you for genetic counselling what we need to do being a genetic counsellor. So, if you have attended the previous classes till now it will be very easy for you receive the client and make them comfortable in assessment room for genetic counselling this is nurse what the nurse should do in a genetic counselling centre right obtain various history right for from family conduct I mean some physical examination right the any anything the patient might not be aware that if

the disease process already started to express right again. Pedigree chart preparation to establish information of heredity pattern right if the patient has come in a state of I mean psychological depression again I mean providing some psychological support to individual family throughout the counselling process right. So, hand holding in every step it is very important and again as I told in the primary care this is all repetition of information roughly right.

So, what is to be done in primary care centre again this informed consent everything patient should be made aware they should be given an open option that this test is not mandatory this is optional very important right. So, encourage the family to ask questions related to genetic testing treatment model they might not be asking anything they might be mum and listening to you should always being a counsellor one should always encourage the beneficiary to ask a question to solve any query she or she has anything. So, it is very important to answer all queries before hand rather than I mean addressing on any unanswered question later right. Establish a plan of care with the family and coordinate with other health care professional very important. Respect the decision making of individual or couple even if the couple says of even after it has been well explained that the continue pregnancy will only lead to trouble they are they are not ready to give up being a counsellor one should respect that right.

There should not be any enforcement or one should not I mean state so much term that actually emotionally traumatizes the couple right. We should always be sensitive enough to assess the decision making of the couple and we should always respect that. Again maintaining privacy and confidentiality of all information related to individual very important for ethical and legal ground. Confidential latent privacy is very important because it is not may be not be the first time right. Once a patient finds respect about confidential latent privacy he or she will definitely come back again or refer his or her other friends who needs counselling to that centre only otherwise legal suits will follow.

Again provide referral guidance for if any referral is needed for specialized genetic centre that should always be role of the primary care provider to have the information so that they can take an informed decision. Lastly aspects about patient education so patient how should patient be educated so that they should understand number on the inheritance of genetics right. So, basically what we are trying to teach the patient at the end of the day we should make sure by asking the patient whether they should know this or not right. So, it is our job to educate the patient and the patient should be educated about these things right it is same it is almost similar.

So, things about the disease outcomes everything right. So, number on understanding genetic inheritance how the disease is happening across the family what is the inheritance pattern they should know at least about that disease. So, the awareness about

their family history how the generic testing is done what are the risks false positive false negative benefit what if the test is positive what if the test is negative right which all should be I mean whether the patient is aware or not they should be educated about the possible outcomes with the genetic testing what if the test is positive what can happen right. Reinformed consent they patient should be made aware that only because you are giving us consent that then we are doing the test right. So, this is a very important is a part of patient education we cannot just perform the test with just by giving him a form and letting him fill up it might be.

So, that is not explained clearly what is written in the form. So, very important each and every part of the form should be checked and cross verified whether the patient has understood or not is a part of informed consent also interpreting test results are a part of patient education right. So, you see again it continues. So, risk assessments psychological psychosocial impact of all the is a again a part of patient education one should be aware you see if your baby has got down syndrome this will be the problem at school he or she may have a learning disability if he or she is born with this disorder right. So, very important so, special baby with autism anything that is genetically possible to diagnose or maybe cystic fibrosis what are the psychosocial impact again a part of patient education discussing of the coping of strategies support system treatment management right very important. In case of infertility what are the reproductive option artificial insemination the in vitro fertilization we are not going into all the details, but again a part of genetic counselling when it is for reproductive diagnosis again reproductive genetics includes patient education about reproductive options again.

And all these terms confidential term privacy the patient should also be made aware that these things are confidential and private right it is his or her choice to discuss all these about to others right. However, again that is an issue that I mean should work in both ways again a part of patient education patient education the primary caregiver should be aware that these things need to be dealt with the patient. Long term follow up not only it is the job of the responsibility of the centre to I mean follow up with the patient, but patient should be also be educated that whenever you are also having another pregnancy you can always contact us. So, this is a also known as reverse follow up that also happens that is very important in case of genetic counselling you should someone has a got a genetic disease. So, our centre is offering a drug free of course, that comes in the support resources.

So, this drug trial which is available you contact us this month later we will also be able to tell you what are the treatment options whether you will get this drug for free or whether you will be able to get this drug in a nominal cost. So, provision of information about support groups in case of specific genetic diseases there are support groups there are communities which support their treatment right. So, advocacy of organisation

resources for individual family dealing with genetic condition right there might be multitude of resource a resource about knowledge, resource about the society, resource about financial support, resource about resource about treatment option, resource about specialised schools, resources about development centre, resources about palliative care there are so many things that are a part of patient education and that is a part of genetic counselling which should be taken care of. So, to summarise in this class we discussed about the purpose, indication, what the beneficiaries of genetic counselling in what phases genetic counselling is done, what are the major four application of genetic counselling, what is the role of primary care provider in genetic counselling as well as many patient education aspects regarding genetic counselling. So, these are the references for today's class and this module ends here and we will be continuing with another important aspect of genetic diagnosing module 11 and I will be meeting you again in module 12 with many newer advances in molecular diagnostics as well as future directions. So, I thank you all for your patient hearing.