

Medical Home Telephone Conference November 11, 2005

Lisa Samson Fang, MD and Pilar Lenglet Genetic Counselor U of U. What can genetic counselors offer the medical home, when to refer to a counselor and what resources are there for genetic counseling.

Who are genetic counselors? Genetic counselors are health professionals with specialized graduate degrees in science with specialized experience in the areas of medical genetics and counseling. Usually come from different backgrounds such as psychology, biology, social work and nursing. Genetic counselors provide information and support to families who have members with birth defects or genetic disorders and to families who may be at risk for a variety of inherited conditions. Genetic counselors:

- **Identify families at risk** – try to ask appropriate questions when looking into family history. Families may not know if they have members of their extended family with mental retardation but know that someone is different or slower. Another example is looking for a history of osteogenesis imperfecta they may not know that terminology by itself but when you are trying to get to that kind of history ask about frequent fractures, hearing loss, heights of individuals in the family.
- **Investigate the problem present in the family** – who else needs testing, who else may be at risk of having a child with a certain condition or birth defect and try and get that information out to those families by writing a generalized letter to give to all family members.
- **Interpret information-** about the disorder in a way that it understandable to them. A genetic counselor is in a way a liaison between the physician and the families. Take the medical aspect and interpret in a way that the families are able to use that information. Provide management and anticipatory guidance, what should we be expecting in the future?
- **Analyze inheritance patterns and risks of recurrence** – this is a big deal for families. Especially if the family is coming in for a child that has an unknown mental retardation, multiple congenital anomaly condition they want to know what the chances are for having this happen again. We try to look at the family history and pattern and give answers to that.
- **Review available options** with the family. Whether it is treatment or management
- **Provide psycho social supportive and counseling to families.** Many of these conditions are very rare and for the family this is new to them and no one in their family has ever heard of the condition so we try to help the parents know the feelings they are going through are normal and try and validate everything they are going through.
- **Serve as patient advocates.**
- **Refer individuals and families** to community or state support services that are in their area or even national support organizations.

Types of genetic counselors: When you are thinking about referring to genetic counseling it really depends on the patient population that you are thinking about referring to but most genetic counselors are trained in all areas so should be able to handle any of these areas. There are many different subspecialties within genetic counseling, therefore, the specific genetic counselor that you refer to will depend upon the type patients that they see in their practice. Currently in Utah:

- Pediatric – 4
- Prenatal – 3 (2 at U of U, 1 at LDS)
- Cancer – 4 (Huntsman)
- Cardiovascular (Myriad genetics Lab)
- Adult
- Neuromuscular
- Research
- Laboratory

When referring, think about your population Is it an adult, child, with a known syndrome or you are questioning the diagnosis, is it a family history of cancer, those kinds of things

When should you refer to a genetic counselor? When you have a family or individual who is concerned about their risk of occurrence or recurrence of a specific condition.

Indications for referral:

- Individuals who are suspected of having a genetic condition:
 - Developmental delay/Mental retardation
 - Growth delay
 - Seizures
 - Dysmorphic facial features
 - Multiple congenital anomalies birth defects.

Genetic counselors as a whole are not able to bill for their own services, so we need a physician as a back up for us. Until this changes they are unable to see patients on our own. Genetic counselors always see patients with a geneticist or oncologist so there is someone in the background to sign off on the cases. In prenatal they always see pts with a maternal/fetal medicine specialist.

Abnormal prenatal history

- A woman over 35 who is concerned about her risk of down syndrome
- Ultrasound anomalies, heart defect or cleft lip, refer for counseling to talk about what conditions are associated with the ultrasound finding and what testing is available and what would they do if there baby were to have a condition such as Trisomy 18
- Abnormal maternal serum screening to find out what their options are
- Teratogenic exposures - Medications, alcohol, illicit drugs. Pregnancy risk line works a lot with these exposures.
- Individuals with a family history of birth defects

Lisa

I think the more common places that a pediatrician is going to be thinking about referring to a genetic counselor would be the child that already has a diagnosis but it has been a number of years and the parent has never really processed the information about recurrence. Are those the situations that we could refer directly to you?

Pilar:

If a child was diagnosed when they were 2 and now the child is 10 they can be referred to a genetic counselor for a refresher wondering if this behavior is related to my child's diagnosis or 10 year-old behavior. Discuss what the condition is again and go over the medical management because with genetics things can change over time. Healthy siblings may also want to know what risks they have.

Lisa:

Is that something that we could do if we had an older sibling who is approaching adulthood and we wanted that sibling to know that information we could refer that sibling directly for a genetics consult

Pilar:

Yes and sometimes that sibling will call. Every week we have a counselor on-call.

Example: an individual called me and he has a cousin with cystic fibrosis and he's wondering about carrier testing for that for him and his wife because they are planning to have children. A lady also called me about her husband with Crohnes disease so it doesn't just have to be a genetic condition that we think about recessive inheritance versus dominant inheritance but something like Crohnes disease where we know there is some genetic component, she was wondering about the recurrence for their children. She also had a history of depression so it doesn't have to be strictly genetic but things that may seem to run in the family. A lot of times people have questions about those such as schizophrenia or bipolar, where the facts aren't as clear as a 25% recurrence risk but we are often able to give them some number as a risk estimate.

Lisa:

So you often get calls from families when one child is diagnosed with a disorder wondering about a sibling?

Pilar:

Either worried about their other children or a distant family member. We saw a gentleman with beta thalasemia trait who is completely healthy with no sign of the disease, he is wondering about he and his wife's risk of having a child with beta thalasemia major. I was able to talk to him about carrier testing for he and his wife to talk about the risks if both are carriers and their options available to them.

So they can give us a call or we may want to schedule them in clinic if they want more of an extensive visit to get more information. If they need testing they will need to come in but most things can be discussed on the phone.

Chuck:

You are happy to do preliminary or counseling evaluations on the phone?

Pilar:

Definitely. This family with cystic fibrosis, he knew his cousin had CF and when I talked to him on the phone he was wondering about carrier testing and if it is the case where someone wants testing then we usually want them to come in and meet with us but if its just there talking to us about their chances of having a disorder. E.g. "I have an aunt with schizophrenia what are the chances of my child having that", then that is something we don't necessarily need to see them in person, it is hard to bill for those kinds of things but we do it all the time, phone counseling is not a problem. If they wanted to come in to have blood drawn then we would want them to come in to talk to us and we could bill for that.

Chuck: Can you tell us a little about the billing? Do you know how often your services are or aren't covered under insurance?

Pilar:

Usually we have certain codes that we bill for and I think from when I have talked to my billing person that they are pretty well covered. One of the other counselors see patients that come in for asymptomatic or pre-symptomatic genetic testing for Huntingtons' disease and these individuals are not affected at this time but want to know if they have this gene change that they know they will develop symptoms in the next 10-15 years and so insurance companies are generally comfortable covering these kinds of counseling sessions but some families don't want their insurance to know they are having genetic testing because of the way that sounds so they opt to pay for it out of pocket for either the testing or the visit. Usually the visit is not a problem for insurance because we can bill for an outpatient clinic visit but sometimes families will pay for the testing rather than have it on their medical records or go on their insurance for fear of discrimination. It is a definite fear but an unrealistic fear. I don't think this is one that has actually happened as there are laws to protect against it but I don't think anyone is willing to test those laws'.

Chuck:

You are not aware of any patients getting in trouble because they have had testing?

Pilar:

I am not familiar with cases of patients getting in trouble with their insurance for having genetic counseling. The HIPPA laws are really for health insurance so genetic discrimination is protected for health insurance but it may not prevent discrimination for life insurance for those trying to obtain that.

Lisa:

You must sometimes see young teenagers who have a condition themselves whether it is fragile bone disease or some other genetic condition so they are going to have fertility and so they need to make their own decisions about recurrence risk. What is an appropriate age to refer a teenager who may be concerned about having children with genetic disorders?

Pilar:

I think it depends how much the teenager has been told by their parents. Sometimes I think parents of kids who have genetic conditions don't know how to talk to their child about it so we can help in talking to the child about what this condition is, what does it mean to them, do they know why they have to see a doctor regularly so I would think around 12 to 13 years of age to help with transition into adulthood which brings a lot of other issues itself or even around age 10 or 11. I think it is important to be completely honest with them about their risks are and answer their questions honestly but not too much at a time.

It is important to support children and families and help them find resources even if the child does not have a known diagnosis and connect them with other families who are going through the same thing. We may know of a family with a child with similar findings to use as a resource for someone to talk to. So if you have a family that could use additional support you could give us a call and we could find families that we have that may be willing to talk to them. From the families standpoint that is the best resource, other families. We could tell you the risks for things, the management and what has been reported, but other parents are the best for dealing with other issues.

Lisa:

The kids we are working with are not just the kids with disabilities, maybe kids with a congenital heart lesion, typically developing kids with medical issues what do you see the role of genetics with kids like that?

Pilar:

I did my thesis on adults with congenital heart disease and I specifically chose those that were non-syndromic so under isolated congenital heart defects in adults to try and find out their understanding of their heart lesion and their risk for recurrence. Most of them knew about their heart condition but didn't know about the implications for their children. So if you have a child with an isolated defect like cleft lip and palate or heart defect and they are wondering about their risk they can give us a call and that is something we could do over the phone. If it was more extensive where we needed a bigger family history to find out about other relatives than we might have them come in for a formal genetic counseling evaluation.

Lisa:

How do we access you?

Pilar:

I have on the second page of the handout has resources for a referral. I broke it down into little goals prenatal, preconception, and cancer. On the handout is also our main division phone number 801-581-8943 which is a phone tree so if you hit 0 you will get our main office and ask to speak to the genetic counselor on-call. My direct line is 801-585-5945 and my email address is on the handout as well. If we don't know the answer to your question we will look it up and get back to you.

Also the U of U, LDS genetic counselors numbers are there. So if someone is taking a family history with their pediatric patients and the Mom states she has breast cancer and her aunt died of ovarian cancer that should send off some red flags and you should refer for some cancer genetic counseling. Cancer genetic counselors can look at whether it is unilateral or bilateral and the ethnic background to see if testing is indicated for one of the hereditary forms of cancer. And even if it is not hereditary they can give screening and management guidelines. The practical things when taking a history are there may be a lot of people with Type 2 diabetes or coronary artery disease that looks familial but not clear cut dominant inheritance where there is 50/50 chance of having it so referring to cardiovascular genetics at the U of U. They don't have counselors but they may have a research study that is collecting these families. If you have a family that is willing to participate in a study to try to find out the underlying cause of their family history of coronary artery disease we can identify those as well.

Question – Bob Teresashima A difficult situation is when a provider is worried about a child and the family isn't at all. For example if you are looking at a child with dysmorphic features but the families thinks the child is cute and looks like uncle Harold. Do you have ways of approaching this situation and encourage the family into genetic counseling? If there were more than dysmorphic features then they will look into if there is anything underlying them all.

Pilar: That is always difficult for me because we see a child in clinic that we see looks different than the family background but parents may not recognize that. If it was just dysmorphic facial features that is one thing but if there are other things going on I try to put it as we all have different minor variations but putting all these together that we start thinking there is something underlying all of these. Such as distinct ears, coupled with feeding delays, and speech delays so I try to create this puzzle for them. One of these things is not a concern until you start putting them together. Using the example of down syndrome where there is a characteristic facial feature but if you point out all of these individual features you may not think that slanting eyes may not mean anything until you put it all together you lean towards an underlying condition so I would try to put it in that kind of context.

Question – For adolescents who are transitioning into the adult health care system and are thinking about their future in reproductive lives, is there a recommendation that they all seek a counselor, when should you refer siblings of someone with a disorder?

Pilar:

Definitely refer siblings of someone with a disorder whether or not this condition is a known syndrome or not because things may have changed since their brother or sister was originally seen and we may be able to offer additional testing and be able to pinpoint a diagnosis and even if we don't have one we may know if this is an X linked type of inheritance so you know sisters you may be a carrier and have a 50% chance of having a son with this condition versus a disorder that is more recessive. Referring the individuals whether the siblings or

the individual themselves that really gears us in terms of recurrence risk because that is the main question of families.

Question: What about the individual themselves who does not have a cognitive impairment

Pilar:

One that I think of is turner syndrome, the girls usually have fertility problems and mild learning problems, if you can follow them in clinic and refer them around adolescence to talk to them about why they are going through all these hormone treatments. Around 10-14 years old start talking to them about all the issues and let them know that turners syndrome is not a good way for contraception. We have a metabolic clinic where we follow PKU from birth to adult and educating them very early on to help them monitor their own diet. Women with PKU tend to have babies with severe mental retardation and microcephaly if their phenylalanine levels are too high so instilling in them early on as an early teenager the importance of staying and remaining on diet if you are thinking about having children talking to us and we will have you talk to a high risk OB. Those kids do not have any learning problems it is more their diet management.

Next conference call January 2006.

Attendees: Lisa Samson-Fang, Gina Pola-Money, Barbara Ward, Pilar Lenglet, Lisa Lee, Chuck Norlin, Diane Behl, Al Romeo, Robert Terashima, Val Jones, Donna.