



Will Genetic Testing Make Some Blood Tests Unnecessary?

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Esther Schorr:

Hi there. This is Esther Schorr, co-founder of Patient Power. We have with us today Dr. Susan Leclair, who is joining us from ASH in San Diego. Thank you for being with us, Dr. Leclair.

Dr. Leclair:

It's very nice to see you again.

Esther Schorr:

With the advent of precision therapies, immunotherapy, combination therapies, it seems as though genetic testing is going to become more and more important for patients, and a CBC might not be sufficient. Can you comment on this?

Dr. Leclair:

Well, in many ways, I think you could say that it was never enough. It is, however, and will remain the most important first step. You have to know a general sense of what the cells are doing, feeling, and it's only after that that you can determine which now of many different ways that you can go to evaluate them.

If I'm looking at granulocytes that are looking a bit, well, odd, would be a good one at the moment, then you might want to do testing on what kind of cells they are or what kind of activity they're doing. And if it's suspicious of a malignancy, well, then you're going to go to certain genetic tests right away.

If you're talking about lymphocytes that are looking, well, odd is going to be my key word for the day, then you're not going to talk about genetic testing right away. You're going to talk about flow cytometry and go down that road before you get to genetics.

If you're talking about the myeloproliferative disorders, you won't be doing flow most likely, and you'll go immediately to a very small number of very specific genetic tests.

I think you have to see the CBC as that important step. Everything starts from there, and the direction you go is based on what that says, even to the point of whether or not you're going to have a bone marrow.

Esther Schorr:

Let's talk a little bit about a newly diagnosed patient, one who has been diagnosed with a blood cancer. What are the questions he or she should be asking her medical team in terms of testing that should be done to help them evaluate whether they should enter a clinical trial or what their treatment plan should be?

Dr. Leclair:

Well, the first thing I would say is when you hear the words, the first thing that's going to happen, and pretty much all hematologists will tell you and others will tell you this is true, you will go deaf for a few moments. The shock of hearing it, give yourself some time, and unfortunately there's not enough time in a physician's office but try and give yourself some time to breathe, to kind of collect yourself because the next several minutes are going to have a lot of intonation in them, and you need to be able to hear it.

If you suspect this is going to be a bad thing, bring somebody with you. It can be a tape recorder, I suppose, it can be your spouse, it can be anybody who's taking notes you for you because while you will be interacting with your physician you will be at such a high level of tension, that long-term memory is probably not going to work so well. So, first and foremost, take a couple of breaths.

If your physician is the one you're going to keep, he or she will be the one who will let you take those couple of breaths and kind of get yourself kind of organized. Nothing is going to happen immediately. There will be no, okay, we're going to finish this conversation, and you're going to get started treating immediately, so there's time for you to ask questions. Tell me more about this disease.

Science, thanks to the—thanks to the Renaissance period where scientists used to get burned at the stake, developed a whole different set of languages to hide themselves and to make their communication with each other more precise. Someone says to you, you have chronic lymphocytic leukemia. Well, what does that mean? You have acute myeloblastic leukemia. First of all, the only word I heard was leukemia, and I missed the other two, and so I need to know.

And so the first thing you want to say to your physician is spend the next five to 10 minutes, show me pictures, draw it out on a piece of scrap paper if you have to, tell me about this disease. It will give you a chance to breathe. It will give you a chance to understand what's going on.

The next thing you want to say is, what are the next steps you think are possible for me to do, and why? Because once I understand the why, then I as a patient can decide issues. So ask them routinely, what and why. And if the answer comes out the way you expect it to, what do you think the next steps are going to be, if the answer comes back not what you expected, what do we do then? Just get yourself a set of ground rules.

By that time, you should also have a sense that your physician can, because some can't, or will, because sometimes there's a choice in it, be the kind of conversation partner that you want. So those would be the first and most important things. Get control of your—I know, I know that the emotions are real, so I don't want to belittle them. But sometimes they don't work for you so take your time, even come back for a second conversation with them, just take your time.

Esther Schorr:

In the context of the move towards precision medicine, what are the questions that any patient should be asking with regard to testing that could help them be eligible for the latest treatments for their disease?

Dr. Leclair:

Well, I think you could start with the basic are you, the physician, connected to any of these trials? I might be interested in one. Certainly if you're going to a major medical center, they're probably going to be attached to and participating in any number of trials. But if you're a—if you are a patient who by choice or by location are dealing with a—and I don't mean this again in a belittling sense, it's just availability of choices. If you're—I come from Massachusetts—if you're on Nantucket, you're not going to have easy access to Boston. You have to take a boat.

So you would want to know, if I'm interested in participating in a clinical trial, do you have connections to those, and how might I find those connections out? You want to find out if the person is involved in a clinical trial what are the

requirements of that trial. I don't want to get myself excited about becoming a member of a clinical trial if it turns out they're only taking people between the ages of 40 and 45, and I'm 55.

So you want to find out right off the top if you're interested in a clinical trial, does your physician participate? Will your physician participate with a referral physician to get you into a trial, and are there—what are the typical restrictions that people might have? And again, that's a first step. That gets you into a conversation with your physician.

Esther Schorr:

Let's talk about a patient who's already had treatment, and they're either in remission, or another disease crops up, or they come out of remission. Is there some reason for them to be retested?

Dr. Leclair:

Sure. If you were diagnosed with polycythemia vera in, say, 2000, we didn't know about the JAK2. That was discovered and published 2005-6, somewhere around there. So if you were doing fine with—I always hate that word—if you were doing fine with hydroxyurea (Hydrea) and maybe some phlebotomies every now and then, and now all of a sudden something has been changed. Well, wouldn't you want to know if you do have the JAK2 mutation, not every p. vera patient does, because if you do have the JAK2 mutation, we have brand-new drugs that are out there to help you.

The same thing is true with chronic lymphocytic leukemia. In the old days, you would diagnose CLL just by the morphology of the cells, or if you got a little high-techie on it you also looked at the structure of the bone marrow. Now, well, in the 1980s, this will show you the kind of a movement on this, in the 1980s people said let's look at flow cytometry and see what the major sets of lymphocytes, are they all T, are they all B, or are they in K cells, what are they, because those have only been discovered in the 1960s.

And then in the early 2000s it was, well, now that we know that there are T cells or there are B cells involved in this, what are the genetic abnormalities that are involved? So now you have B cells with a 13q deletion or have just T cell or you have some other presentation, each of those presentations means that you may be eligible for or ineligible for certain medications. And these medications keep coming online every single year or every other year or so, so you need to have information, knowledge in order to be able to choose those drugs appropriately.

Esther Schorr:

Is it also possible that your genetic profile can change over time?

Dr. Leclair:

It is possible. We know that in chronic myelogenous leukemia, particularly when it was being treated—hmm, how does one put this—less well than it is today, we watched the evolution of genetic damage. We watched it go from one abnormal chromosome to maybe getting two abnormal chromosomes or three, that chromosomes would form rings so that we knew that there was a progressive structural disorder or disarray that was going on. So, yes, there could be, not probably in all of the diseases but certainly in enough, that you might every now and then want to be able to choose.

You also want to understand that going—going along with your disease is all this research. Before, really the only thing that you had in polycythemia was phlebotomy. And then the big deal was you could have phlebotomy and hydroxyurea, wow. And for some people that was perfectly wonderful but for some not so much.

And now you have all of these JAK2 inhibitors that—and this one inhibitor number one might be really good if you're biggest problem in life is just the red cells, but inhibitor number two not so good on the red cells but really good on overactive platelets. And so there will be these subtle changes in the way the medications work that has to be taken into account when a physician's looking at a patient. So, yes, unfortunately, expensive, time-consuming and stressful as laboratory testing can be, every now and then it's not a bad idea to have. Particularly when there's a change that seems to be coming, it's a good idea to have it done.

Esther Schorr:

Thank you so much, Dr. Susan Leclair, for joining us from ASH this year. We always appreciate your wonderful insights.

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