



Patient Power

The Winding Road to Diagnosis: How Everyone's MPN Journey Is Different

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Andrew Schorr:

So let's talk about diagnosis a little bit, because we may have people watching who it's all new. And we're talking about more rare conditions. So, Nick, you are how old now? Are you still in your 30s?

Nick Napolitano:

Barely hanging on, yeah, 39.

Andrew Schorr:

And you have two young children. I think your youngest is just 8 months old.

Nick Napolitano:

Yes.

Andrew Schorr:

So you had a routine physical. What did it show, and what came out of that routine physical?

Nick Napolitano:

Yeah. So it showed my red blood cells were high around 50, and my platelets were around 675.

Andrew Schorr:

Hold on. Dr. Michaelis, what's normal?

Dr. Michaelis:

So, normal, generally, the hematocrit or hemoglobin—so, hemoglobin is your measurement of a certain kind of protein in your red cells. And the hematocrit is the percentage of that protein in your blood. We like to see people lower than that. Hemoglobin over about 16.5 in men. And in his case, he's telling me his hematocrit, and that hematocrit of 50 is too high in men. It should be closer to under 50 more or less. And then, platelets are also elevated. In this situation, platelets should top out about 430,000 or 450,000. It's important that everybody know that each lab has different normal.

And that's because those normals are derived from the patients in your area. So, if your doctor says this isn't normal, ask what the normal is in your lab.

Andrew Schorr:

Okay. Back to you, Nick. So, Nick, so, the numbers were out of whack. What did the doctor say? And what came out of that?

Nick Napolitano:

Well, the doctor, basically, said let's come back for another round of blood work, which I did, and it showed similar numbers that were elevated. And they gave me a choice to monitor it for a year or follow up and see a hematologist oncologist. So, I thought about it, but not too long, and I said I have kids, and let me go follow up with the hematologist oncologist locally. And so, that's what I did. And so...

Andrew Schorr:

...and then, you hear these words, polycythemia vera. Nobody knows those words until you're told it's probably you or a family member. It must have sounded strange. And also, with a young family, pretty worrisome.

Nick Napolitano:

Yeah. My mindset going in, and I put off the—the doctor called me back because he tested me for leukemia and the JAK2 mutation. And I put off going in for a while, because I didn't think it was anything to worry about.

And so, I put it off for several weeks. And they wouldn't give me the results over the phone. And so, I finally went in, and I still had this mentality that nothing was wrong. So, it was a shock. I was confused. I didn't know exactly what it was, and the doctor didn't over explain it, didn't use the C word, didn't use cancer. So, as I mentioned before, I did the worse thing I could possibly do, which was started Googling stuff, when I walked out of the office. And I was so shocked, I didn't ask a lot of questions about it, and I should have.

Andrew Schorr:

Right. And here you are, and you've come a long way now.

So, Jill, let's hear a little bit about your story. So you had been having kind of clot issues, right? You had kind of blood issues.

Jill O'Brien:

Yes. I had had strange medical issues really ever since my 20s.

In my mid-20s, I had Bell's palsy on the left side of my face, which is still residual. Then, I ended up I was having strange pain issues, and I would get very little painful bumps in different areas of my body. That was in my early 30s. And they did a biopsy of that, sent it to Mayo. It came back with well, we think it's vasculitis, but we don't really know what type. So, through a rheumatologist, I was on methotrexate (Rheumatrex) and steroids. And that controlled it for a while. And but my blood numbers just kept inching up. My red blood cell count, my white count, my platelets. But they would always explain it away.

Like my white count, well, you're on steroids, so you know, that's probably why your white count is a little elevated.

And when my platelets, the vasculitis is inflammation. And, finally, once we moved here to Memphis, my rheumatologist here said I really think maybe you should go to a hematologist and see if there's something else going on.

Andrew Schorr:

And you, ultimately, had a clot, right?

Jill O'Brien:

Yes. And it was right after I went to the local hematologist here, and he just went into well, watch and wait. Well, then, I ended up with this blood clot in my iliac artery and had to have two stents. And so, then, he decided to do a bone marrow biopsy. And that's when the diagnosis of primary myelofibrosis came back.

Andrew Schorr:

Okay, in 2014. Hold the story for one second. I want to go to Dr. Michaelis. So, Dr. Michaelis, people have these winding roads to these MPNs.

And I'm sure you see it all. And then, she mentioned a bone marrow biopsy, which might tell more of the story. Why does it take so long for people to get an accurate diagnosis?

Dr. Michaelis:

Well, one thing is that a lot of medical conditions can cause elevations in blood counts. And it doesn't have to do with some kind of underlying bone marrow disorder. It's because illnesses affect the bone marrow in different ways. Rheumatological disorders can increase your white count. Certain medicines can increase your white count or even sometimes your red blood cell count. People can have elevated platelets after surgeries or sometimes after delivering a baby. So, it's not always a disease when your blood counts are high. So, that's the first thing.

The second thing is that some of the symptoms of myeloproliferative neoplasms, whether or not it's fatigue, whether or not it's itching, those can also be associated with much more common problems like allergies or, for example, some people have fatigue because of heart problems or because of lung problems.

So very common conditions can sometimes mimic these unusual blood conditions. But, in general, if somebody has consistently elevated blood counts, predominantly elevations in their red blood cell count or their platelets, and that elevation occurs even when the other medical issues or other things are under control, then that's a reason to sort of figure that out. One of the other things we now know is that the bone marrow is actually looking at the bone marrow, determining whether or not the bone marrow has fibrosis, what the early blood cells look like that are supposed to make the platelets that are supposed to make the white blood cells.

That's also an important part of our diagnosis, because that can help separate myeloproliferative neoplasms out from either a disease that's happening in the rest of the body and affecting the bone marrow or other bone marrow conditions like myelodysplastic syndrome.

Okay. Now, Nick, have you had a bone marrow biopsy?

Nick Napolitano:

I have. And so, just to pick up on one of the doctor's points on how elevated numbers can be sort of crossed over, I had pulled my numbers from the previous two physicals that I had done, which was about five or six years prior to that initial diagnosis. And they were elevated then, too. And they were in the high 40s, and one was 50. My red blood cell count and my platelets were high as well. So, I've sort of experienced that where they were high. How long did I have it? I don't know if I had it back then, but possibly.

Andrew Schorr:

Right. Let me just mention, I'll just chime in for a little bit of my story. So, I had a clot, a DVT, deep vein thrombosis, Jill.

And then, I was actually in a trial for a blood thinner. And it was while I was being monitored in the trial, and I'm a big proponent of trials both to maybe have tomorrow's medicine today but to be monitored carefully. And that's when they said, and it wasn't a hematologist who was doing that trial, said you need to go back to your hematologist, and we think something is going on. And that led to the mutation being discovered, the JAK2V617F mutation that's active for me, and a diagnosis of primary myelofibrosis.

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