



Expert Perspective: How Genetic Discoveries Are Changing the Future of MPNs

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Andrew Schorr:

Hello and welcome to Patient Power. I'm Andrew Schorr. There is a lot of work going on to understand the genetics and epigenetics of myeloproliferative neoplasms, MPNs. And one of the leaders in the field is Dr. Alison Moliterno from Johns Hopkins in Baltimore. She joins us now to help us understand more about genetics and MPNs.

Dr. Moliterno:

This is something that's near and dear to my heart because when I started my fellowship in hematology many years ago this was a focus of a research project, was to try to understand why blood cells grew well in patients with MPNs. We suspected there was something intrinsic in the cells, the stem cells in patients with MPN that allowed them to grow excessively, and we thought that maybe there was a genetic basis for this, something encoded in the DNA of MPN stem cells.

Over many decades, many researchers have looked at different genes that control growth. And as we all know, in 2005 this really watershed discovery of the JAK2 mutation occurred, and that has really changed the complete landscape of MPN, of how we view it as a disease and view it as—when it changes its pattern and try to understand why some patients have one end of a spectrum, and some patients may have another end.

And so a lot of my research in the last several years has been trying to understand the roles of JAK2 levels, how much JAK2 mutation, and how many other mutations are required and how they changed the natural history of the disease.

Andrew Schorr:

What excites you about your current research?

Dr. Moliterno:

What really excites me about my research is bringing the knowledge of genetic discovery and my own research and others' research into these disorders right to patients and to share it with them. Giving them the knowledge and the understanding of how we—how these breakthroughs have helped us understand these diseases is very gratifying. The more knowledge everyone has and can share that, the more we understand and can move forward in attacking this.

I have long relationships with a lot of my patients. I've been following patients now almost 20 years, and it's been wonderful to share these breakthroughs and learn together as we make these leaps forward.

Andrew Schorr:

One question that's often asked about blood cancer and cancer in general is, is the cancer acute or chronic? What's your opinion when it comes to MPNs?

Dr. Moliterno:

I think that's an interesting question. What I've found in many patients is that they may be diagnosed, meaning their blood counts are identified as abnormal maybe on a routine screening, but then when you look back in their record they may have evidence of blood count abnormalities and evidence of this entity for years to decades.

So if anything, it's chronic, and sometimes these entities then can have an acute phase where things may change, and their disease now is more acute, meaning more active. So I think in every patient there's the potential for both. You mentioned earlier, can these diseases ever remit spontaneously? And we're hoping that someday they will when we have treatments that address the root cause of their entities, but so far I have not had that experience. So I guess I would say these are really chronic disorders and that sometimes they can have an acute phase.

Andrew Schorr:

Other than JAK inhibitors, what's next on the horizon?

Dr. Moliterno:

Well, I think the biggest genetic discoveries that we have to implement now aside from recognition of JAK2 and the CALR mutations, which is a new discovery, is to really apply—to allow physicians to understand how to apply this knowledge in staging disease. It's becoming apparent that the more mutations you have and the higher burden of mutations you have is significant to how the disease—how symptomatic the disease may be and how it may affect prognosis.

So we need to pull that information into our clinical assessments, and that's going to take us sharing knowledge of our research with others and also getting diagnostic testing to reflect that knowledge.

Andrew Schorr:

One last question, Dr. Moliterno, In your opinion, how important is it for a patient to seek out a specialist in MPNs?

Dr. Moliterno:

I think it's critical. These aren't as rare diseases as we once thought they were, and we need to organize our knowledge to share amongst physicians and specialists, and that can be done through events such as this and events that the MPN Research Foundation supports.

But many patients aren't living in large cities or have the capacity to travel, and so I've always been impressed with how getting knowledge through the Internet and helping patients educate their own physicians and hoping that those physicians then reach out to perhaps more specialists. I think really organizing to that fashion is the way we can bring everyone's expertise to every patient.

Andrew Schorr:

Dr. Alison Moliterno from Johns Hopkins, thank you for being with us on Patient Power. I'm Andrew Schorr. Remember, knowledge can be the best medicine of all.

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