Q&A with a Pediatric Geneticist

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Q&A with a Pediatric Geneticist:
Interview with David Kronn, M.D.
By Kyle Flattery

David Kronn, M.D. is an Associate Professor of Pediatrics at New York Medical College, and serves the Greater New York region as Director of the Medical Genetics Center at Children’s and Women’s Physicians of Westchester in Valhalla, NY. He established the Inherited Metabolic Disease Center at Westchester Medical Center (WMC), and has been named not only to Westchester’s Best Doctors, but also to New York’s Best Doctors.

In my conversation with Dr. Kronn, we discussed how he combines his training in pediatrics and genetics, how genetic diagnosis impacts medical practice in many fields, and how Dr. Kronn adapts to rapid advances in genetics research and knowledge.

Q: Can you describe your typical week as a Medical Geneticist?

A: We sometimes round in the NICU [neonatal intensive care unit] – one of our common sites for referral. We treat all the hospital patients who may have some developmental delay, or some feature or signs of any number of disorders, so it is all over the map. You cannot really describe the average patient since we see a variety of patients. And we do not only see pediatrics, but we see adults as well. I am trained primarily in pediatrics, and genetics, which as a specialty not only covers infants and toddlers, but all child age groups. We would see consultations for cancer, or for, perhaps, Marfan Syndrome, or patients who have severe signs in the ICU who we do not know what is going on. We would sometimes consult those patients. So, it is a variety.

We also do outreach sites. We do outreach genetics where we see patients in Vassar Brothers Hospital, in Danbury, Connecticut, and Good Samaritan Hospital, so we can improve the accessibility of patients to our service. Sometimes crossing the bridge is a geographical barrier to care, and so we cover the whole Hudson Valley, but we also cover parts of Connecticut and New Jersey, so we try to make it easier for patients to reach us.

That is the morning. In the afternoon, we tend to see scheduled patients. We tend to see a lot of patients. Maybe around 10 patients in an afternoon will be referred. We have a counselor and a nurse practitioner, who can assist in the process and work with the patients directly. Because there is a huge demand for patients, we have about a 6-month waiting list to see patients. This is a lot more intense than even when I was in training, because in training, we would see maybe 10 patients a week. And now we are seeing about 50 patients a week or more, depending. Our
program now sees around 100 patients a week. The demand for genetic services has increased dramatically.

**Q: Can you explain why that demand has gone up recently?**

**A:** There are several things. There have been huge advances in genetics recently. The Human Genome Project has been one thing, but now we know so much more about the genetic etiology of disease. A lot of diseases have a gene associated with them, so we can do diagnostic testing – that is the first thing. And we are interested in metabolism, so we are interested in the *treatment* of diseases as well. There is a lot more treatment for diseases that we can now do for patients. If there is a treatment for a disease, it becomes more and more important to screen for the disease, so we don’t miss it.

The other thing that we are involved with is newborn screenings. When I started here with this practice in 1996, we were only screening for seven diseases in newborn screening. And since then, the number of diseases have increased to about 55. We can screen for more diseases now, and we are making that many more diagnoses in the newborn period for patients. We are getting a lot of referrals for newborn screening. So that is an important part of how we are getting more diagnoses; we are getting patients earlier.

Also, we have access to advanced genetic testing. One of the things that is really developed in terms of advances is exome sequencing. We can sequence every gene in the body now, and that has become a very important clinical tool. We can now make diagnoses that were never possible before. And we use this quite a lot. There are some diseases where there are only one or two people diagnosed in the entire world. We have the *technology* now to make more advanced diagnoses.

And then we must follow up with these patients. We continue to follow the patients we have diagnosed – we have hundreds who we are following who we have diagnosed over the years. So that adds to the number of patients that we are seeing. And genetics does not really have a time frame, so we will see patients forever. Once we make a diagnosis, we do not really discharge a patient. We continue to follow the patient on a periodic basis – maybe a yearly basis – so those are the patients who we see all the time.

**Q: So it sounds like the demand is going to continue to rise, yet you still have the same amount of resources when you started out?**
A: We have a little bit more. But because the demand has increased throughout the country… to hire somebody is very difficult, because everybody needs another geneticist. And we have been trying to hire another geneticist for two years now, for somebody who can help me see patients. We have recently started hiring more nurse practitioners and training them to help manage patients. There is a growing demand for what we are doing.

Q: Are you aware of any attempts to bring Medical Genetics into other specialties, and incorporated into those practices rather than being kept separate?

A: We interact with all practices because we see every kind of patient from say neurology, or spinal muscular atrophy, hematology, cardiology – because now we know some of the genetic risk factors for cardiomyopathy – so we interact with all specialties.

We are interested in partnering with generalists to help with the management of some of these patients, because we are stretched on our resources to help the patients in an ongoing basis. So that is something we are thinking about now, because we know it is going to be hard to fill the positions with additional medical geneticists. That is something that is in the process: improving genetics with primary care. It is improving, but it is still limited. And genetics is changing so rapidly that it is difficult to even keep up with what is happening. Even for us, it is hard for us to keep up with the changes that can occur. So it has hard to be on top of everything.

Q: With so much research going on right now into genetics, how quickly does the field change every few months, or every year?

A: Oh, all the time. The translational research occurs very rapidly, and we get involved early on. Some of our patients have helped contribute to the cloning of genes. Some of our patients were the first patients cloned with a certain disease. So once the gene is identified, we are able to do genetic testing to confirm the diagnosis in other patients almost immediately. So that is the research process.

So once that technology has been adapted, it has been transferred into a laboratory which has been certified, and that testing can be offered to everybody. And that translational research happens rapidly. And since we are doing exomic sequencing, we can sequence every gene. Although we can sequence all 25,000 genes, we only know the function of about 30% of the human genome. So there is still a lot of things we do not know. That changes all the time as more and more genes are identified, because patients develop symptoms and are proven to have these genetic defects. There is a research aspect that happens all the time, so that helps with patients coming down the pike. So it is a to-and-fro. We are very involved clinically, but we collaborate a
lot of the time with research laboratories, in helping them find particular diseases. We collaborate with laboratories all over the world in this sense. So even though we are clinical, we are very research-oriented. We see so much pathology here that we should collaborate and help with patients with this type of diagnosis. It is exciting since so many things are happening all the time.

A patient might come in with a very rare diagnosis, and I will make the diagnosis on an exome level. Then, this data gets shipped and shared on international databases, and we sometimes get approached by the laboratory to see whether they can share the clinical status of the patient so they can understand more. Then they will collaborate – they will find more patients around the world – where they can find what the actual condition is based on the genetic mutation.

Q: And in sharing that information about some of the patients, is your field having any trouble in figuring out if public access to genetic information will be an issue further down the line?

A: Yes, we are always concerned about that. That is a major concern. These are all anonymous databases. The researcher would contact us, and we would contact the patient to see if they want to join. The databases do not have any identifiers of the patient. There is no way an individual can identify the patient. The researchers would first contact us and ask if the patient would be willing to participate, and then we would contact the patient to see whether they would. There is no direct access to the individual patient.

Genetics non-discrimination is a huge thing in this current environment. People are reasonably concerned that their genetic information will be used against them. It is very important that we want to protect that information. We are concerned about that since we are about to go onto electronic medical record (EMR). We delayed going onto EMR because we wanted to have a firewall preventing others from easily accessing people’s genetic information. We are very concerned about that.

And there are security protections, but we are all concerned in this environment about what will happen in the future. So we will be cognizant of that. And people ask us all the time. No genetic testing is done without patient consent. People will come in and they will understand the implications of the testing because genetic testing is not like doing a blood count; it is always there. If you have a BRCA1 mutation, that is never going to change. And that has a lot of implications for you, and your family. Genetics is interesting because it is not only the individual that is affected, but other individuals in the family can be identified. Also, in the process of this testing you might uncover other things that people did not even understand. Or other risk factors that we are not aware of.
Q: Of the people you see, there are some who have a symptom, and they want to find out the genetic, underlying cause. But for other people, they may come in because they know something that’s within their family?

A: Exactly. Or we will identify an individual within the family with a particular disease, and then we start testing other members of the family who did not realize they may be at risk, and discover other members who have the disease as well.

Q: And for people coming in who are planning to have kids?

A: Exactly. And they might have some members of the family who have the disease, so it’s a new baby, and they are concerned about what the risk factors are for future pregnancies. What can we do? What other members of the family are at risk? All of those questions come up. The counseling becomes very important in that. Something very important that we do is transfer information to make sure our patients and families understand the implications and all the information we are providing to them. Because it is not so easy, we have to spend quite a bit of time with them. And that is why we have genetic counselors, who really drill down with the families and the patients, to make sure they understand the implications and what they want to do. We never direct patients on what they want to do, but simply help them make an informed decision about the information.

Q: Lastly, what drew you to working with medicine and genetics, together?

A: I think the basis for genetics was to understand the molecular pathology of disease. And that is what I was interested in, because I started in genetics training in 1990. And at that point, we had not even cloned the cystic fibrosis gene. There has been a tremendous expansion on even just the time I have been in practice. And we have a greater understanding of the human genome, and how it functions. That leads to a lot of potential opportunities for treatment that we have not had up until now. And that is exciting because if we understand on a molecular basis what causes a disease, then I think you will have the best chance at understanding how we can treat it.