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Evaluating Port-Wine Capillary Malformations in Children

Announcer:
You're listening to *On the Frontlines of Pediatric Skin Health* on ReachMD. Here's your host, Dr. Alexandria May.

Dr. May:
This is *On the Frontlines of Pediatric Skin Health* on ReachMD, and I'm Dr. Alexandria May. Today, I'm joined by Dr. Kristen Kelly to discuss early recognition and evaluation of port-wine capillary malformations. Dr. Kelly is a Professor and the Chair of the Department of Dermatology at the UC Irvine School of Medicine.

Dr. Kelly, we're so glad to have you here today.

Dr. Kelly:
Thank you very much. I'm really pleased to be here.

Dr. May:
To begin, Dr. Kelly, can you give us an overview of port-wine capillary malformations and what clinical features are most important to recognize?

Dr. Kelly:
So, first of all, a variety of different terms have been used for port-wine capillary malformations in the past. We started with port-wine stain. There was some research—including a survey that I had done with patients a number of years back—that indicated that the word "stain" was a derogatory term, and in some cases, made patients feel badly. It really was eye-opening to me, because I certainly never wanted a patient to feel badly, and so I started using the term port-wine birthmark. But most recently, the International Society for the Study of Vascular Anomalies has classified it as port-wine capillary malformation, so utilizing that term is probably most precise.

Port-wine capillary malformations are present from birth. There are a variety of genetic changes that can be associated with them. GNAQ mutations are the most common, but certainly there are other genetic changes that can be associated with them. They tend to be slowly progressive, so not rapidly like an infantile hemangioma, but slowly over decades. They may thicken. You can get nodules, which sometimes will bleed, et cetera. So generally, they're slowly progressive.

And then—unlike an infantile hemangioma, again—they stay in the same area. They don't cover a larger area than they did before, percentage-wise. But as the patient grows, the area may grow with them.

Dr. May:
As we look at these features more closely, how can clinicians distinguish isolated port-wine capillary malformations from lesions that may be associated with syndromic or neurologic involvement?

Dr. Kelly:
So that's a really important question, especially anytime we're seeing a patient, and especially a child, because often if the patient is older, there would've been manifestations that would've become evident. You always want to be thinking about this, because it's important to get the patient set up with the right other specialists if there are other potential complications from involvement of other areas.

For Sturge-Weber syndrome, we have to have two of three different things: a port-wine capillary malformation, which presumably, if you've identified that, you know that. And then there could be brain involvement, which in children, most commonly presents as seizures, but in adults can present in a variety of different ways. For example, they may have migraines, et cetera.

And then also, there can be eye involvement, which most commonly is glaucoma. And so the area that we're going to be most concerned about for the port-wine capillary malformation is if it's in the central forehead; those are going to have extensive involvement.

So in those patients, we really want be thinking about Sturge-Weber syndrome, and then you would want to get them evaluated. I have a pretty low threshold if I have any concern to get them set up with a neurologist and an ophthalmologist just to make sure that everything's okay. I think that's important because, for example, we don't want any loss of vision if we don't identify the glaucoma early.

So I think it's better to get someone set up so that if they have any issues, that they can be helped. I've also had patients who, for the first five or six months had, no seizure activity, but then had very significant seizure activity, so it was good that they were already set up with a neurologist.

So we've talked a little bit about Sturge-Weber syndrome, but there are a variety of other syndromes that one wants to consider—for example, Klippel-Trenaunay. So if there's limb enlargement, we want to think about that. And then there are some rarer ones; for example, if the port-wine capillary malformation is over the midline of the back, then we want to also think about whether there could be involvement, for example, with the spine or something like that.

We want to think about those things as well. So Sturge-Weber syndrome is probably the one people think about most, but there can be other associated syndromes. And then there are also the overgrowth syndromes, which may have a port-wine capillary malformation associated with them. Those are usually a little bit more obvious because the overgrowth is generally pretty apparent.

Dr. May:
For those just joining us, this is *On the Frontlines of Pediatric Skin Health* on ReachMD. I'm Dr. Alexandria May, and I'm speaking with Dr. Kristen Kelly about recognizing and evaluating port-wine capillary malformations in pediatric patients.

Turning to long-term considerations now, Dr. Kelly, what changes or clinical progression might we see over time, and why is ongoing follow-up so important in these cases?

Dr. Kelly:
This is a very important point again, so all excellent questions. Keep in mind that we like to start as early as possible when we're treating a port-wine capillary malformation with laser, because we generally get better results when we start early.

However, it is very rare that we're able to get 100 percent clearance, and even when we get close to that, there is generally some recurrence. Now, this doesn't mean that the port-wine capillary malformation will go back to the way it was before. That generally does not happen. But you can get some recurrence, and so it's important to continue follow-up.

And most of my patients, even when we've gotten a very good result in the first year of life, continue to see me for a couple treatments every year to try to maintain that for them. My hope is that eventually we'll have a medication so that maybe won't be the case, but probably, they will continue to need some kind of follow-up.

I think it's also key—we just talked about syndromes a moment ago—that they continue to follow up with those specialists, because while the majority of time, by one year of age you can identify things—unless the other specialist feels like they really don't need to see the patient back—touching base periodically is important just to make sure that nothing else develops.

Dr. May:
With all of this being said, what are some of the most common misconceptions or diagnostic pitfalls that clinicians encounter with these vascular lesions?

Dr. Kelly:
So I think one of the ones that comes up most often is sometimes people will say, "Well, how can I identify a port-wine capillary malformation versus an infantile hemangioma?" Many pediatric dermatologists, of course, are very familiar with this, but depending on the background, etc., some people might have questions.

So, in general, as I say, port-wine capillary malformations are going to be static. They're, at least at the beginning, slowly progressive, but early on, they're not going to change very much. So they're always present from birth, again, so that's often one difference, because infantile hemangiomas may appear shortly after birth, etc.

And then the infantile hemangiomas generally have that rapid growth phase early on whereas a port-wine capillary malformation will not. So I think that's one thing to keep in mind: the differential between those two.

And then we already talked about making sure that you get other specialists involved if it's going to be helpful to the patient. And that can be a pitfall, where maybe someone isn't connected, and they should be. So I think that's another important consideration.

Dr. May:
Finally, Dr. Kelly, as awareness and diagnostic tools continue to evolve, where do you think are the greatest opportunities to improve early identification and coordinated care?

Dr. Kelly:
I love this question, and I'm very excited about the possibilities for the future. we've done a lot of work using non-invasive imaging, like optical coherence tomography, to try to get more definition of the blood vessels for each patient. It turns out, and we've known this for a long time, these lesions are very diverse.

So if you look at one spot and then the spot right next to it, the depth that the vessels start and the diameter of them can be very different. I hope that eventually, we will be able to have an instrument that will be able to give us that information rapidly enough so that each laser pulse could be adjusted based on what would be best for that particular position. We're a long way from that, so that is definitely a hope for the future, but not something we're going to be able to do yet.

Of course, now we're doing genetic testing, because there is the opportunity for medications, and there are some medications that can be helpful. But I think there's definitely more opportunity for that for the future. We may be able to ultimately non-invasively be able to diagnose different genetic changes. We can't do that yet, but there are some clues from the imaging that perhaps in the future, we'll be able to do that. So if we knew early on the characteristics of each spot of a port-wine capillary malformation—we knew the genetic change—that might help us to give better laser treatments, but also perhaps, I hope, to have a medication that'll be more effective.

Right now, for port-wine capillary malformations, we don't have any effective medications. But I really do think that in the next decade, and I hope much sooner than that, that we're going to have a medication that will be helpful for patients.

Dr. May:
Well, that's a great way to round out our discussion. A big thanks to my guest, Dr. Kristen Kelly, for joining me to explore how we can best identify and diagnose port-wine capillary malformations. Dr. Kelly, it was great having you on the program.

Dr. Kelly:
Thank you very much. I really appreciate the opportunity to talk with everyone.

Announcer:
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