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Making a Definitive Diagnosis

Announcer:
Welcome to CME on ReachMD. This episode is part of our MinuteCE curriculum.

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Dr. Samson:
This is CME on ReachMD, and I'm Dr. Susan Samson. Here with me today are Dr. Eliza Geer and Dr. Kevin Yuen. Today we're discussing what it takes to make a definitive diagnosis of acromegaly.

Dr. Geer, what biochemical, imaging, and visual field tests should we be using?

Dr. Geer:
So we know that while it's hard to identify the patients who need screening, once we have identified a patient, diagnosis is usually pretty straightforward, and it includes a high IGF-1 level that's stratified for the patient's age in the setting of classic signs and symptoms, and these things, the clinical presentation plus the high IGF-1, is usually sufficient for making the diagnosis. In borderline cases, so people who have kind of very mild clinical presentation or just mildly elevated IGF-1 and we need to confirm the diagnosis, these are patients that we would do glucose suppression on. So this is a 75-g glucose load. We measure growth hormone at baseline and 30, 60, 90, and 120 minutes after giving the glucose load. And we have BMI-based cutoffs because we know that people with higher BMIs tend to have lower growth hormone and IGF-1 levels. So we know that in someone without acromegaly, glucose will suppress growth hormone. Someone with acromegaly, either there's inadequate suppression or there's a paradoxical rise, and we use the cutoff of 0.4 µg/L for people with a BMI of less than 25 and a cutoff of 0.2 µg/L for people with a BMI of greater than 25. So if we have a high IGF-1 plus inadequate growth hormone suppression after a glucose load, that will confirm the diagnosis.

In terms of IGF-1 assays, they vary among reference labs, so it's preferable to try and use the same assay in each patient. We know that there is these values. The normal ranges vary by age, so we have to take that into consideration. In terms of the assays, I don't think there's any data to suggest that mass spec is superior to immunoassay, so either of those can be used.

And once we have a high IGF-1 and a clinical suspicion, we would then get an MRI. So this is a gadolinium-enhanced pituitary MRI. Ideally, the report – and ideally, we're looking at the images as well – will include information about tumor invasion, and NOS score. And for patients who have a tumor that is encroaching on or involving the optic chiasm, they should see a neuro ophthalmologist to have formal visual field testing to look for deficits.

So along with testing, clinical exam is also important. Dr. Yuen, maybe you can discuss a little bit what the clinical exam should entail.

Dr. Yuen:
I think being a doctor is very important. Listening to the patient's problems, to the patient's symptoms is extremely important because that will then dictate which part of the clinical exam that you will like to focus on, given the fact that acromegaly is a multisystem and multiorgan disorder. So I think listening to the patient very clearly, getting a handle of how long the symptoms have been developing, and

then, depending on how you elicit that history and then you perform the clinical exam, particularly looking at clinical features. Perhaps also asking if the patient has any old photographs that you can compare. That's certainly one of the least expensive tests that one can do. Examining the patient's face for any acro disorders, prognathism, a large nose, large tongue. Examining the patient's neck for any possibility of thyroid enlargement, heart for any possible heart failure signs and symptoms. Examining the patient's abdomen for hepatomegaly and also not forgetting the hands and feet. Again, don't forget this is the first time we are also seeing the patient, so perhaps it is important to note that, you know, the patient's ring size and the patient's shoe size and sometimes even the patient's hat size. So I think these are important cues, and certainly the history will certainly help you focus on which part of the exam that you need to be focusing on.

Dr. Samson:
So I think we have learned a lot from Dr. Geer and Dr. Yeun about the diagnosis of acromegaly, both with biochemistry as well as with a really careful clinical exam. You know, importantly, the longer the delay in the diagnosis of our patients, the more comorbidities they have. For example, 10-20 years ago, a patient might have a 10- to 20-year delay in their diagnosis from their first symptom. We know that that delay in diagnosis has decreased over the years, now probably in the range of 4-7 years. And the longer the delay in the diagnosis, the more the comorbidities. For example, a delay in diagnosis over 10 years might mean that the patient has 6 different aspects of their health that have been impacted by acromegaly. This could be their heart, cardiac function, arrhythmia, hypertension, diabetes. So it is imperative that we diagnose patients early so that we can normalize their growth hormone and IGF-1 and reduce the comorbidities from acromegaly.

Well, this has been a great micro-discussion, and our time is up. Thanks for listening.

Announcer:
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