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Expert Insights on Congenital Adrenal Hyperplasia Care From an Adult Endocrinologist

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You're listening to ReachMD. This medical industry feature, titled "Expert Insights on Congenital Adrenal Hyperplasia Care from an Adult Endocrinologist," is sponsored by Neurocrine Biosciences, Inc.

Here's Dr. Sara Lubitz.

Dr. Lubitz:

I am Sara Lubitz. I'm an adult endocrinologist at Rutgers Robert Wood Johnson Medical School.

5, 10% of my patient population has classic congenital adrenal hyperplasia (CAH). It's a really rare condition, but if you go looking for it it's actually pretty common.

There's not that many adult endocrinologists who specialize in congenital adrenal hyperplasia. I think it's something that's focused on more in the pediatric endocrinology fellowship, and there's a lot of experts in that area, but people grow up. Everyone turns over 18, and you need adult endocrinology experts to take care of these patients.

The biggest misconception is that when you find a good treatment regimen, that that's going to be the treatment regimen for the rest of your life, because unfortunately as times change, as somebody's needs change, as they're interested in fertility or they're starting to develop metabolic syndrome, the goals of therapy keep changing.

One of the difficulties is that the patient might feel really well, but they walk in there and you look at their labs, their sugar, their cholesterol, their hormone levels, and it's not what you've been taught is the ideal levels for these things. It's not what's going to prevent long term heart attack, strokes, osteoporosis, mood disorders. And then on the other hand, there's times where the patient feels terrible and you're shocked because the labs look fantastic, all the metabolic parameters look great.

You think you're walking into that visit saying, "we're great, here's a prescription for renewal," and the patient feels terrible. It's really difficult to reconcile those two things, because the way the patient is feeling doesn't always match up with what you're seeing on the lab results.

We know what we're doing because we've been doing the same thing for the last 70 years, but it's not the best option. There has to be a better way.

I really try to follow guidelines to guide our clinical practice, but in this condition, congenital adrenal hyperplasia, the most recent guidelines are from over five years ago.

Those guidelines are great about giving us really good rules for, "don't normalize the 17-hydroxyprogesterone," or "try to focus on your androstenedione testosterone levels," but I know the newer stuff is actually even looking at all these adrenal precursors that, like, I never even learned about in medical school as ways to treat the disease better.

As an adult endocrinologist, I'm going through guidelines which are not focused on adult endocrinology that much. The guidelines really talk about growth and puberty, but that's not part of my clinical practice. I do have to still worry about adrenal rest tumors. But again, the guidelines, they're not differentiated between what's important for a newly diagnosed child with this condition and an aging, elderly adult with this condition.

So, I have to pick and choose when I'm going through the guidelines what actually relates to me and my clinical practice.

When we talk about treatment of congenital adrenal hyperplasia, we're trying to replace the glucocorticoids that are missing. So, if somebody is not able to make cortisol or they're not able to make cortisol and aldosterone, we need to replace those hormones.

Ideally, you would want to replace those hormones in the most physiologic way, the way that the body naturally makes those hormones, and you would want to completely mimic what the body naturally does, because that's how the hormones are supposed to be made. However, in congenital adrenal hyperplasia, we have another whole goal of treatment, which is to reduce the adrenal androgens. And in order to do that, we have to take everything that we know about physiologic glucocorticoid replacement and throw it out the window and use this really supra-physiologic amounts of glucocorticoid in a really timed pattern, which is, like, not normal diurnal variation, and give a huge dose of glucocorticoid in order to trick the brain into not making as much corticotropin releasing factor, which then hits the pituitary and tells the pituitary to make ACTH, which then hits the adrenal glands. And you're trying to reduce that ACTH, you're trying to reduce that corticotropin releasing factor, and that's a whole other part of the treatment.

So, if you're not doing a good job with that, the adrenal androgens are too high and you're going to have side effects of hirsutism, infertility, sexual dysfunction, acne. If you're doing a really good job of that or you're over treating it, you're going to have glucocorticoid side effects, which is weight gain, obesity, cardiometabolic risks, insulin resistance, high triglycerides, high cholesterol, musculoskeletal complaints. Just giving high doses of glucocorticoid at night can affect sleep patterns, can affect metabolic response, so you're just in this balancing act always of "which is worse, the treatment or the disease?" And trying to balance that out.

The most critical unmet need in the treatment of classic congenital adrenal hyperplasia is finding some sort of treatment option that gets to the root of the problem. We are using glucocorticoids to both treat the glucocorticoid deficiency, as well as to kind of trick the negative feedback system into allowing the body not to make so much adrenal androgens. And there just has to be a better way to balance that.

My goal for classic CAH patients is to have an improved quality of life. If you could decrease the pill burden and improve the glucocorticoid side effects without still having them afraid every day of going into adrenal crisis, that would be a wonderful goal. It would just be a wonderful goal for patients with classic congenital adrenal hyperplasia to be able to replace the hormones in a more physiologic way.

My approach is being willing to try new things. I think that's the key to success. There's never going to be, "this is your glucocorticoid treatment regimen, and this is going to be the exact same doses and the exact same timing that somebody is going to need for the rest of their life."

You have to be willing to try different timings, try different steroid doses, try new things, and everything is trial and error. So, a patient is not going to do that unless you've earned their trust in the idea that, hey, we're going to try something new.

It might be worse. If it's worse, we can always go back to what we were doing before. But it might also be better.

And every time you're willing to try something new, it's going to take weeks to months to see if it's really working. But you're never going to achieve the best quality of life and the best long-term outcomes without being willing to try something new.

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CP-CAH-US-0294 09/2024