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ReachMD
www.reachmd.com
info@reachmd.com
(866) 423-7849

Navigating Complex CAH Care from Newborn to Adult

Announcer:
You're listening to *Clinician's Roundtable* on ReachMD, and this episode is sponsored by Neurocrine Biosciences Incorporated. Here's your host, Dr. Brian McDonough.

Dr. McDonough:
This is *Clinicians Roundtable* on ReachMD. I'm Dr. Brian McDonough, and joining me to discuss strategies for optimizing care for patients with congenital adrenal hyperplasia, or CAH, is Dr. Phyllis Speiser. She's the Emeritus Chief of Pediatric Endocrinology at Cohen Children's Medical Center for New York and the Emeritus Professor of Pediatrics at the Donald and Barbara Zucker School of Medicine at Hofstra Northwell Health. Dr. Speiser, thanks for being here today.

Dr. Speiser:
Thank you.

Dr. McDonough:
Dr. Speiser, when starting CAH patients on treatment, how do you ensure their needs are at the forefront of care?

Dr. Speiser:
Yes, thank you, Brian. So nowadays, most patients with classic CAH, and when we speak about CAH, we're talking about the most common form, which is a deficiency of the steroidogenic enzyme 21-hydroxylase, they are going to be diagnosed through a newborn screening program. So in the U.S. today, every state has a panel of congenital disorders for which they test in the newborn nursery, usually at about 48 hours of life or just prior to discharge from the newborn nursery. And among those tests in every state in the U.S. today is a heel stick blood test for 21-hydroxylase deficiency CAH.

So once that diagnosis is made by the newborn screening lab, the primary care provider is notified, and the primary care provider will make the initial contact usually with the family and advise them to seek out their local pediatric endocrinologist. And often that's a setting where the state has certified that particular institution and set of practitioners as experts in the area of CAH, so they will make an appointment as soon as possible, often within 24 hours, to see the pediatric endocrinologist, and they will undergo a full evaluation with a history, physical, and additional lab tests in many cases.

And if the practitioner, the specialist, is convinced that the child suffers from CAH, even before the complete set of laboratory tests come back, they will start the child on a medication regimen. And that medication regimen will include glucocorticoids, such as hydrocortisone, which is a replacement for the adrenal cortex product, cortisol, and in addition, in many or most cases, a mineralocorticoid product, which is an oral medication replacing the other major adrenal steroid product called aldosterone. And between these two medications, they will set the balance straight so that the hormonal milieu is not lacking in the elements that support vascular tone and energy levels, as well as sodium balance. So the newborn will be started on a medication dose, but this will require frequent readjustment, depending on the levels of lab tests that are going to be monitored very frequently in infancy. And the family, who generally has no real prior knowledge of how to manage CAH, will be given an intensive set of written instructions, and given instructions additionally for how to deal with emergencies, when to call the physician's office, and when, if necessary, to even visit the emergency department for further intensive care.

And I should add here that beyond the pediatric endocrinologist, of course, there's a need for lifelong treatment, and it's not just the endocrinologist, but it's really a team of physicians. And again, that's a best practice guideline that in most instances, in a large children's hospital, or in any academic medical center, there should be a team of physicians who are familiar with this disorder and can

work as a team to provide the patient and the family with the best possible care in all areas of need.

Dr. McDonough:
Dr. Speiser, you laid out something that's obviously very complex. It's a lot for parents to process when, really, they're bringing their baby home and just thinking about purchasing diapers, formula, breast milk, all those things. What strategies do you recommend to the caregivers of these patients to provide them with the optimal care? Because obviously they're going to need support.

Dr. Speiser:
Right. So for the caregivers, if they are primary care healthcare providers, they may not be so familiar with congenital adrenal hyperplasia. The classic form of 21-hydroxylase deficiency is present in about 1 in 15,000 births. So for any given healthcare provider, they may have seen a couple of patients in their practicing lifetime and may not be totally familiar with the current guidelines for care. So that's one rationale for the medical society, The Endocrine Society, putting out these clinical practice guidelines, which are available on The Endocrine Society's website, and there's a lay summary for people who are not healthcare providers. But people are encouraged to make referrals to people who are board-certified endocrinologists in the pediatric or adult realm and to make sure that they work in tandem with that healthcare team to understand the needs of their patients, make sure that they are going back for specialty care when needed, and that's usually recommended at frequent intervals in infancy and childhood, and then perhaps less frequently as the child matures and finishes adolescence and gets into the adult realm.

One particular concern we have is that as individuals age out of pediatric care, there are relatively few adult endocrinologists who are prepared to take care of CAH. And one of the reasons for that is because a generation or two ago these patients were not living much beyond infancy and childhood, and so they didn't have an opportunity to seek adult care, and now they are searching for places where they can get that care. So I would urge anyone who's taking care of these patients to be familiar with the clinical practice guidelines, to communicate regularly with an academic medical center or a CAH Center of Excellence where they can get advice if they're in a remote location where there isn't a local team to provide that care, and in general, to keep the lines of communication open to make sure these patients are not left without medication renewals and are making their visits frequently and know to call in in case of an emergency or they need adjustments in their medication and sometimes require hospitalization.

Dr. McDonough:
Dr. Speiser, you alluded to it, but more specifically, how do you suggest ways for the care teams to work together to provide essentially seamless collaborative care?

Dr. Speiser:
Right. So ideally and what the guidelines recommend is that in a Center of Excellence, there be a multidisciplinary team of healthcare providers, including physicians of various specialties, generally led by the endocrinologist, but also including a geneticist to help with genetic counseling a pediatric urologist and/or gynecologist to help with issues related to the genitourinary system, which may be affected, particularly in the females who are affected with classic CAH. We sometimes will consult with reproductive endocrinologists and certainly psychiatrists who can help with the mental health issues, a social worker who can help maintain access to the medical care, and to just have a session a few times a year where all of these specialists get together and talk among themselves to make sure that everybody's on the same page, that the recommendations to the family and to the patient are clear, and to allow the family and the patient to ask questions. And for pediatric patients, that has to be an age-appropriate type of communication because obviously, for the infant, the main people you're communicating with are the parents. And as the child gets older, we expect them to ask questions about why they're coming to see this team of doctors, why they're taking their medication. And it's very important to be upfront and transparent, explaining everything about the care, so that the family and the patient are motivated to continue with their care and not drop out from seeing their specialists.

Dr. McDonough:
For those just joining us, this is *Clinician's Roundtable* on ReachMD. I'm Dr. Brian McDonough, and I'm speaking with Dr. Phyllis Speiser about congenital adrenal hyperplasia care.

So with those best practices in mind for collaborative care that you discussed earlier, Dr. Speiser, could you share a real-world patient case where these strategies were used?

Dr. Speiser:
Yes. So as you may know, Brian, there are relatively few specialists in the adult realm who are familiar with CAH. And so I think a lot of the consultation requests that I get will be through email or telephone from adult providers saying, 'I have this patient who came to me with a diagnosis of CAH, I'm not sure how to manage them.' And often, if it's a straightforward question, I can answer those questions without necessarily seeing the patient, although for specific management issues very often the patient does need to see somebody and I

can make a referral.

For the pediatric population, the same may hold true if the patient lives in a remote area and there is no local pediatric endocrinologist, so sometimes the patient or the family will need to travel to seek out an opinion from somebody who has familiarity. So the questions that come up frequently would be is my child taking the right medication or the right doses of medication? How do we manage stressful situations? When should we give extra doses of glucocorticoid? When do we need to go to the emergency department to seek more advanced care? And should we come in for a consultation in person and review the medical records, and then go back to the primary care provider who can work in tandem with the specialty team and maintain good health for the CAH patient?

Dr. McDonough:
We've talked about the care teams, and I'm interested in what tools or resources you would recommend to the care teams that could help them provide the best care for CAH patients?

Dr. Speiser:
Right. So I think nowadays there's a lot of talk about shared decision-making. And our team in particular has worked with mental health professionals to develop a shared decision-making tool so that the family and the patient can lay out their own values and priorities and say what's important to them in making medical decisions. So we try to establish a good rapport of communication with the family, with the patient. If need be, we'll do that with a patient advocate or sometimes with a translator if they're not primarily English speaking. So I think that all of these tools and a team approach are critical to establishing good rapport and to maintaining continuity of care within the family and sometimes connecting them with a support organization that can also help the communications if they meet and speak with other people who have lived with this condition and attend in-person meetings or virtual meetings that can give them some sense of confidence about helping manage their own health condition.

Dr. McDonough:
Dr. Speiser, before we finish our conversation today, how could fostering seamless collaborative care between the patient, caregivers, and care teams impact the quality of life and outcomes for these CAH patients?

Dr. Speiser:
So I think, Brian, that this is an issue not just for CAH, but it's an issue in any patient disorder where they require care throughout the lifespan. So we know that establishing good communication, fostering communication among the members of the team who are caring for a family, making sure that the specialists maintain contact through various channels, whether it's phone calls, emails, letters with the primary care provider so that they're on the same page. They're aware of what's going on. They know when to refer the patient back. And to be alert to make sure that the family or the patient is not being lost to follow-up. And this is particularly important in the adolescent population as they age out of the family plan, where they're going to have to make their own medication renewals, their medical appointments, and make sure that they are in a place where they're safe and well provided for for medical care, whether it's being away at college or starting to live on their own. This is something we emphasize and prepare the adolescent patient for transition to individual adult care.

Dr. McDonough:
With those impacts in mind, I want to thank my guest, Dr. Phyllis Speiser, for sharing her insights on optimal congenital adrenal hyperplasia care. Dr. Speiser, it was great having you on the program.

Dr. Speiser:
Thank you, Brian. It was a pleasure talking with you.

Announcer:
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