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Genetic Testing in Pediatrics: Identifying Neurodevelopmental Disorders Early

Announcer:
You're listening to *Clinician's Roundtable* on ReachMD. On this episode, Dr. Jennifer Kalish will discuss genetic evaluation of neurodevelopmental disorders, which she spoke about at the 2025 American Academy of Pediatrics National Conference and Exhibition. Dr. Kalish is an Assistant Professor of Pediatrics at the University of Pennsylvania Perelman School of Medicine and the Director of the Beckwith-Wiedemann Syndrome Program at the Children's Hospital of Philadelphia. Let's hear from her now.

Dr. Kalish:
Early genetic evaluation helps us figure out, basically, what could be causing the neurodevelopmental disorder, and there's clues both in the family history potentially, the physical appearance of the child, and their medical history that can help clue us in. And when we use those clues, we can try and put together what could be causing the diagnosis. Thinking about that testing, it can give families a direction to understand the trajectory of what's next and what to expect for their child, and also an answer. In addition, it can also give us information about the recurrence risk in a family if we do make a molecular diagnosis to explain the neurodevelopmental delays.

There are many challenges that pediatricians may face in trying to initiate testing. First is knowing what testing to initiate, how to go through that process, where to order a test, which test to order, and specifically how to get that test done. And there's barriers such as insurance authorization, access to the specific test types that need to be done, and how to interpret and counsel the family for doing that testing.

In terms of educating and having pediatricians understand what they need to do to even get to that step is trying to do all the things I mentioned before: thinking about the family history, the patient's medical history, or if there are physical features that stand out that look different. And a pediatrician doesn't have to know all of those things together, but the idea is that there is something different here that we need to do testing and try and think about whether or not we need to call a specialist to help us with this, or if that's something that they can actually order themselves.

The larger takeaways from all of this is that, as we've learned so much more about genetics and how genetics relates to neurodevelopmental disorders, there's a lot of testing that we can do at our fingertips, but some of that testing is largely guided by our physical exam, taking a careful family history and a careful medical history for the child, and looking at ancillary testing that may help guide us in a direction, because for some of these large tests, like exome sequencing or genome sequencing, we still need clues from the medical records, the medical history, and the family history in order to interpret those results. So it's really that whole entire picture that we need to think about and consider as we're doing this genetic testing.

Announcer:
That was Dr. Jennifer Kalish discussing genetic testing for neurodevelopmental disorders, which she spoke on at the 2025 American Academy of Pediatrics National Conference and Exhibition. To access this and other episodes in our series, visit *Clinician's Roundtable* on ReachMD.com, where you can Be Part of the Knowledge. Thanks for listening!

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