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

Communicating Genetic Testing Results to AMKD Patients: The Impact of Early Diagnosis

Announcer:
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Dr. Hung:
Hi. My name is Adriana Hung, and I am an Associate Professor of Medicine in Nephrology at the Vanderbilt University. The topic that we're going to discuss is communicating genetic testing results to AMKD patients and the impact of early diagnosis. As we discussed before, having two risk variants for the APOL1 gene, or a high-risk genotype, increases the risk of CKD progression, or developing end-stage renal disease in individuals of African ancestry. We have shared previously this study by Parsa et al, the African American Study of Kidney Disease and the chronic renal insufficiency cohort that shows that having these two high-risk alleles increased by 90%, the risk of developing a 50% decline in your GFR or developing ESRD.

So the identification of individuals with these high-risk genotypes is important. But it's also important to understand that not everybody with a high-risk genotype will develop kidney disease. This is important when we are genotyping individuals, or when individuals get genotyped by choice, for example, 23andMe, and then they wonder, ‘what does this mean? And so this is important because then when we talk to our patients, we can tell them about a second hit hypotheses. So environmental or other genes that will increase the risk of developing kidney disease. And also the opportunity to intervene early to prevent the development of kidney disease or all the consequences of kidney disease.

And so for example, in this particular study which is done also in the African American Study of Kidney Disease, we can observe that individuals with a high-risk APOL1 genotype, the curves in yellow and black, if they receive strict control of their blood pressure, they have a 50% decrease in the risk of dying. This is particularly important and compelling as to a reason why we need to genotype patients that we have reasons to suspect that they may have this gene and to inform the patient and the physician so that they can be more strict in their interventions in an early stage.

It's been shown that having two high risk alleles also it's associated with an early-onset hypertension and higher levels of systolic blood pressure. This particular study was done by Girish Nadkarni from BioMe, and it was replicated in BioVU in the Northwestern University Genetic Biobank. And so, again, in this case, these individuals may not have kidney disease yet, but knowing that they have the high-risk genotype may provide a reason as to why to monitor their blood pressure from an earlier age.

And so one of the aspects that we face in many of the studies that we've done is that most of the genetic testing tools today for genes that are actionable, such as APOL1, have been in the context of research studies. And so, whenever we want to return a result, that has to be repeated in a clinical graded laboratory, so that we feel certain that the information that we are providing is accurate.

The other things that we can see in this particular algorithm is that it's important to understand if you're going to be able to answer all your patient questions, or if you should be always upfront, be ready to offer genetic counseling. I personally always offer genetic counseling to my patients. I think they need many answers that I won't be able to provide in the time of the clinical visit. For example,

what will they tell their family members? Will this affect their insurability? Will they be labeled? There are so many questions that come with a return of results. But what we have to tell our patients too is that we're providing to them the opportunity of being monitored from an early stage of the disease, or even prior to developing the disease, or that we're giving their physicians the opportunity to provide a more aggressive monitoring and change outcomes, as we see in the study that I shared with you done in the African American Study of Kidney Disease.

So this is all important information that we have to give to our patients, the benefits that they will get from these, and try to talk to them about the risks that
come with learning this information. It's fair to always inform the patients about their risk. And so that's why I think genetic counseling is very important, because we as clinicians don't have the window of time required to answer all these questions. And genetic counselors are also very well prepared to handle many of the questions that come with returns of results.

And this was just a study that was in CJASN as to how do we do return of results when genes are actionable. But what I really wanted to say is that there's still no standard of care in this regard, and many studies are ongoing to learn the barriers and the best practices when returning results.

But I think we have all learned throughout this process, that this is common, that this affects outcomes, and that even if we genotype people that do not have kidney disease, this can be effective in preventing the onset of kidney disease. And because of that, if we want to tackle this issue, we have to offer genotyping, be ready to do return our results, and be ready to offer genetic tests.

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