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GENE SEQUENCING APPROACH MAY HELP TAILOR TREATMENTS FOR YOUNG KIDNEY TRANSPLANT RECIPIENTS

Method revealed kidney disease–causing gene mutations in one-third of pediatric patients.

Highlights

- Whole-exome sequencing of blood or saliva revealed a genetic diagnosis of kidney disease in 32.7% of pediatric kidney transplant recipients.
- The findings indicate that such a sequencing strategy may help individualize pre- and post-transplant care for many young kidney transplant recipients.

Washington, DC (January 17, 2019) — Nearly one third of pediatric kidney transplant recipients in a recent study had a genetic cause of their kidney disease that investigators identified through a technique called whole-exome sequencing (WES). The findings, which appear in an upcoming issue of the *Journal of the American Society of Nephrology* (*JASN*), suggest that such genetic analyses may be useful for guiding the care of many young kidney transplant patients.

Causes of chronic kidney disease (CKD) in children and young adults differ greatly from those in older individuals. Preliminary research suggest that WES may help identify mutations that contribute to kidney dysfunction in young patients with CKD or kidney failure, which could help clinicians offer more personalized treatment. To assess the potential of WES for use in such patients, a team led by Friedhelm Hildebrandt, MD and Nina Mann, MD (Boston Children's Hospital) studied 104 children who had received a kidney transplant from 2007 through 2017. Patients' WES data from blood or saliva were examined for deleterious variants in approximately 400 genes known to cause CKD.

Through the use of WES, the researchers identified a genetic cause of CKD in 34 of the 104 (32.7%) patients. The likelihood of detecting a genetic diagnosis was highest for patients with urinary stone disease, followed by renal cystic ciliopathies, steroid-resistant nephrotic syndrome, congenital anomalies of the kidney and urinary tract, and chronic glomerulonephritis.

“In this study, we found that almost one third of children who underwent kidney transplantation had a genetic cause for their kidney disease. In several cases, identifying the genetic etiology could help to guide the care of these patients,” said Dr. Hildebrandt.

Study co-authors include Daniela Braun, MD, Kassandra Amann, Weizhen Tan, MD Shirlee Shril, MS, Dervla Connaughton, MD, Makiko Nakayama, MD, Ronen Schneider, MD, Thomas Kitzler, MD, Amelie van der Ven, MD, Jing Chen, MD, Hadas Ityel, MD, Asaf Vivante, MD, PhD, Amar Majmundar, MD, PhD, Ankana Daga, MD, Jillian Warejko, MD, Svjetlana Lovric, MD, Shazia Ashraf, PhD, Tilman Jobst-Schwan, MD, Eugen Widmeier, MD, Hannah Hugo, Shrikant Mane, PhD, Leslie Spaneas, RN, Michael Somers, MD, Michael Ferguson, MD, Avram Traum, MD, Deborah Stein, MD, Michelle Baum, MD, Ghaleb Daouk, MD, SM, Richard Lifton, MD, PhD, Shannon Manzi, PharmD, Khashayar Vakili, MD, Heung Bae Kim, MD, and Nancy Rodig, MD.

Disclosures: Dr. Hildebrandt is a co-founder of Goldfinch Bio. The authors reported no other financial disclosures.

The article, entitled “Whole Exome Sequencing Enables a Precision Medicine Approach for Renal Transplant Recipients,” will appear online at <http://jasn.asnjournals.org/> on January 17, 2019, doi: 10.1681/ASN.201806057.

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