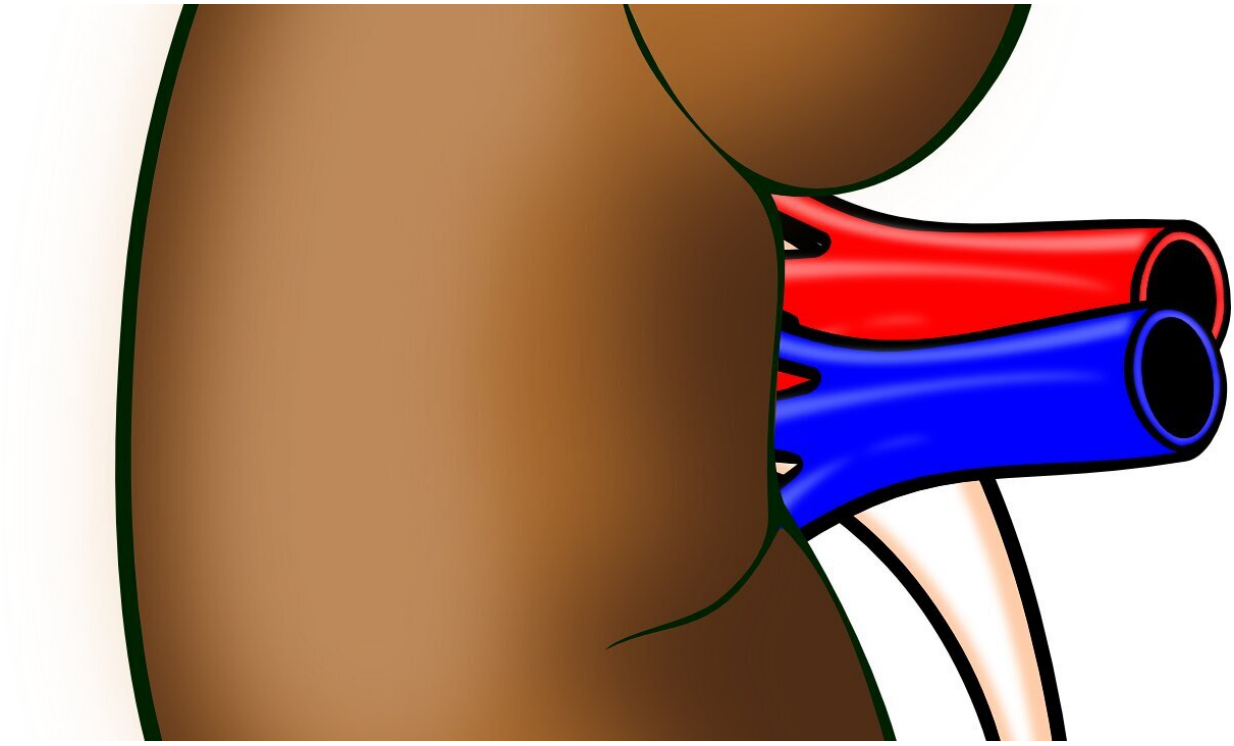
Genetic testing for kidney diseases in embryos from in vitro fertilization

August 27 2020



Credit: Pixabay/CC0 Public Domain

Preimplantation genetic testing (PGT)—a technique used to identify genetic defects in embryos created through in vitro fertilization before pregnancy—helps prospective parents to prevent passing on disease-causing mutations to their children. An analysis in an upcoming issue of *CJASN* provides a clinical overview of the past 25 years of PGT for

kidney diseases in the Netherlands.

The analysis, which was conducted by a team led by Albertien M. van Eerde, MD, Ph.D. and Rozemarijn Snoek, MD (University Medical Center Utrecht), included information on couples counselled on PGT for monogenic kidney diseases (which are caused by mutations in a single gene). The procedures were performed through the Dutch PGT consortium—with as lead center Maastricht UMC+ - from January 1995 until June 2019. Most procedures for kidney disease were performed in the last decade. The PGT procedure in the Netherlands is part of reimbursed healthcare.

"PGT is a relatively recent addition to the options couples at risk for having a child with monogenic kidney disease have," said Dr. van Eerde. "There was no literature on the range of indications PGT has been performed for, success rates, and reasons for opting out after having been informed."

The team found that the number of couples counselled on PGT for monogenic kidney disease steadily increased over time. Ninety-eight couples were counselled regarding PGT, of whom 53% opted to proceed with testing, 38% chose not to proceed, and 9% had not yet decided whether to proceed as of June 2019.

The most frequent indications for testing were autosomal dominant polycystic kidney disease (38%), Alport syndrome (26%), and autosomal recessive polycystic kidney disease (9%). Also, 45% of procedures were performed for adult onset kidney disease, with an increase to over 50% in recent years. Of couples with at least one PGT cycle with egg retrieval, 65% experienced one or more live births of a child without kidney disease.

The authors noted that as the number of kidney diseases for which a

monogenic cause can be identified continues to grow, it becomes increasingly important to counsel patients on their reproductive options to enable them to make informed choices.

"Choosing how to start a family can be a complex decision if one has a genetic disease. Through PGT, a genetically unaffected pregnancy can be established, through an in vitro fertilization procedure," said Dr. van Eerde. "Insights from our study may help doctors in informing patients with monogenic kidney disease who wish to start a family, and allow for adequate counselling."

An accompanying editorial noted that in the absence of insurance coverage, costly genetic services are primarily available to those who can pay out of pocket, and limits in the coverage of assisted reproductive technology disproportionately burden racially marginalized groups. The authors stressed that there is a need to address moral questions about the acceptable use of PGT, and to ensure access based on need rather than ability to pay. "Payers, including government programs, may be more likely to include coverage of preimplantation genetic testing if professional organizations offer stringent guidelines for its use that have the dual goal of preventing the birth of children with serious disease while supporting equitable access," they wrote.

More information: "Preimplantation Genetic Testing for Monogenic Kidney Disease," DOI: 10.2215/CJN.03550320

"Preimplantation Genetic Testing for Genetic Kidney Disease: Addressing Moral Uncertainties and Access Inequity," DOI: 10.2215/CJN.11790720

Provided by American Society of Nephrology