Columbia Technology Ventures

Genetic test for heritable pediatric congenital malformations of the kidneys and urinary tract

Technology #cu13044

Nearly half of all cases of pediatric renal failure can be traced to heritable abnormalities. These abnormalities are known as congenital malformations of the kidney and urinary tract (CAKUT), and include kidney hypoplasia and dysplasia, multicystic kidney dysplasia, posterior urethral valves, and ureter abnormalities. Additionally, they are frequently associated with co-morbidities such as hypertension and cardiovascular disease. CAKUT is thought to be caused by genetic mutations. However, attempts to identify the causative mutation have been unsuccessful or focus on a very small subset of patient populations. This technology has identified a loss-of-function mutation in patients with CAKUT that affects urinary tract development. It allows for diagnostic testing as well as genetic counseling for couples trying to conceive.

DSTYK mutations provide the first identifiable genetic cause of congenital urinary tract malformations

This technology identifies the first gene mutation shown to cause a form of CAKUT birth defects. Research into the genetic causes of various forms of CAKUT is ongoing but few specific genes have been found due to of the complex nature of urinary development. Mutation analysis of this gene can be used to diagnose patients with this abnormality prenatally and provide a basis for screening families and providing genetic counseling for at-risk parents.

Linkage analysis has revealed that nearly 2.3% of CAKUT patients display a mutation in Dual Serine/Threonine Kinase (DSTYK), suggesting that it is a major contributor to urinary tract development.

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Applications:
- Antenatal testing for CAKUT
- Prenatal testing for CAKUT
• Diagnostic testing for CAKUT
• Screening tool for high-risk families and populations
• Research tool for studying congenital abnormalities of the urinary tract
• Investigation of the role in DSTYK in other tissues, as both growth factor mediator and driver
• Research tool for developing animal models of congenital malformations of the urinary tract

Advantages:
• Identifies first gene mutation involved in the development of congenital malformations of the urinary tract
• Provides genetic basis for specific type of CAKUT abnormalities
• Enables research into the molecular mechanisms and development of urinary and renal development

Patent Information:
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Related Publications:

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