

Using Genetic Testing to Identify Patients with Renal Cysts Who are at an Increased Risk of Diabetes

Indications:

- Renal cysts of unknown etiology
- Family history of Renal Cysts And Diabetes syndrome (RCAD)

Benefits:

- Genetic testing for RCAD can:
 - identify RCAD as the cause of renal cysts.
 - alert patients and physicians to the risk of other RCAD-associated manifestations, such as MODY5.
 - allow family screening for RCAD, facilitating the early diagnosis of RCAD in family members of patients.

Background:

- RCAD was identified in 36% of children and 18% of adults with renal cysts not associated with other known hereditary causes.^{1,2}
- RCAD is also associated with maturity onset diabetes of the young (MODY5) in about half of patients.^{2,3}
- Other manifestations of RCAD include pancreatic atrophy and urogenital malformations.^{2,4}
- RCAD is inherited in an autosomal dominant manner.^{3,5,6}
- RCAD is due to loss-of-function mutations in the gene *TCF2*.^{3,5,6}

References: 1. Edghill EL, et al (2006) *J Med Genet* 43:84-90. 2. Ulinski T, et al (2006) *J Am Soc Nephrol* 17:497-503. 3. Horikawa Y, et al (1997) *Nat Genet* 17:384-5. 4. Bellanne-Chantelot C, et al (2004) *Ann Intern Med* 140:510-7. 5. Lindner TH, et al (1999) *Hum Mol Genet* 8:2001-8. 6. Nishigori H, et al (1999) *Diabetes* 47:1354-5.

Ordering Information: please see other side



Ordering Information for the *TCF2* DNA Sequencing Test (Renal Cysts and Diabetes Syndrome)

Indications for Testing

- Renal cysts of unknown etiology
- Family history of Renal Cysts And Diabetes syndrome

Ordering Information

Gene(s)	Test Code
<i>TCF2</i>	170201

Family Testing (single amplicon)

Family Testing is available. Please contact Client Services at 1-866-647-0735 for requirements.

Test Methodology

- Amplification by polymerase chain reaction (PCR); sequencing of entire protein-coding region

NOTE: Specimens must be accompanied by a completed consent form. In the case of family tests (ie, known mutations), a copy of the result of the first patient tested in the family (the index case) must be submitted unless that test was performed at Correlagen. Other family members are subsequently tested for the specific mutation found in the first patient tested.

**For test information, sample requirements, or to request a sample shipping kit,
please contact Client Services at 1-866-647-0735 or visit us on the web at
www.correlagen.com.**