

Using Genetic Testing for Diagnosis of Congenital Bilateral Absence of the Vas Deferens (CBAVD)

Indications:

- Bilateral absence of the vas deferens in males
- No or only one heterozygous *CFTR* mutation detected during targeted mutation analysis in patients with CBAVD

Benefits:

CFTR sequencing

- Can confirm a clinical diagnosis of CBAVD and identify the familial mutation(s)
- Can assist in genetic counseling with regard to CF carrier status of at-risk blood relatives of CBAVD patients, depending on the known familial mutation(s)

Background:

- Congenital bilateral absence of vas deferens (CBAVD) is a recessively inherited disease caused by mutations in the *CFTR* gene.^{1,2}
- A definitive diagnosis of CBAVD can only be achieved by genetic testing.^{1,2}
- Mutations in *CFTR* are also associated with CF, an often life-limiting multisystem disease affecting the respiratory, digestive, and male reproductive systems.^{1,2}
- CBAVD is associated with the presence of a “mild” *CFTR* mutation on at least one chromosome copy. The second chromosome copy may carry another mild mutation, but can also harbor a “severe” mutation. Knowledge of the familial CBAVD mutation(s) can therefore help predict the risk of CF in blood relatives of patients with CBAVD.^{1,2}

References

1. Moskowitz SM, Chmiel JF, Stern DL, et al. Clinical practice and genetic counseling for cystic fibrosis and *CFTR*-related disorders. *Genet Med*. 2008;10:851-868.
2. Moskowitz SM, Chmiel JF, Stern DL, Cheng E, Cutting GR. (Updated March 26, 2001.) *CFTR*-related disorders. In: *GeneReviews* at GeneTests: Medical Genetics Information Resource (database online). Copyright, University of Washington, Seattle. 1997-2010. Available at <http://www.genetests.org>. Accessed February 21, 2010.

Ordering Information: Please see other side.

Ordering Information for Congenital Bilateral Absence of the Vas Deferens (CBAVD) Testing

Indications for Testing

- Bilateral absence of vas deferens in males
- No or only one heterozygous *CFTR* mutation detected during targeted mutation analysis in patients with CBAVD

Ordering Information

Gene(s)	Test Code
<i>CFTR</i>	150101

Family Testing (single amplicon)

Note: Family testing for CBAVD can only be used for males. For family testing in female relatives of CBAVD patients, 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.