

Using Genetic Testing to Diagnose Atrial Septal Defect with Atrioventricular Block

Indications:

- Clinical diagnosis of ASD, in combination with at least one of the two following symptoms:
 - conduction delay (such as prolonged PR interval or AV node conduction delay or block)
 - family history of sudden cardiac death
- Unexplained AV block, especially in presence of family history of congenital heart disease
- Family history of ASD
- Family history *NKX2-5*-related congenital heart disease

Benefits:

Genetic testing for mutations in the gene *NKX2-5* can:

- confirm a diagnosis of ASD.
- alert patients and physicians to the risk of AV block later in life.
- determine which family members of an individual with a known *NKX2-5* mutation also harbor the mutation, facilitating early diagnosis and preventative treatment.

Background:

- Atrial Septal Defects (ASDs), which occur in approximately 1 in 1000 individuals, are characterized by an opening in the atrial septum that creates a connection between the two atria.¹
- If diagnosed early, ASDs can be effectively treated by surgical closure.^{1,2}
- Clinically significant ASDs are usually well tolerated in children and adolescents and may not be recognized until adulthood, after irreversible damage to the heart and lungs or a stroke caused by blood clots that arise from the venous circulation has already occurred.^{1,3}
- 8-19% of familial ASDs are associated with autosomal dominant mutations in the gene *NKX2-5*. Mutations in *NKX2-5* may also lead to AV block, often developing in adulthood.⁴
- The initial presenting symptoms of ASD with AV block may be related to either the ASD or the AV block.⁵

References: 1. Webb G, et al. (2006) *Circ.* 114:1645-1653. 2. Gruber PJ, et al. (2004) *Circ. Res.* 94:273-283. 3. Warnes CA, et al. (2001) *J. Am. Coll. Cardiol.* 41:2072-2076. 4. Hirayama-Yamada K, et al. (2005) *Am. J. Med. Genet.* 135A:47-52. 5. Smits JP, et al. (2005) *Europace* 7:122-137.

Ordering Information: please see other side

Ordering Information for Testing for Atrial Septal Defect with Atrioventricular Block

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Ordering Information

Gene(s)	Test Code
<i>NKX2-5</i>	190401

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.