LABupdate

Atrial Septal Defects

Affecting more than 1 in 1000 live births[1,2], atrial septal defects (ASDs) account for about 10% of cases of congenital heart disease.[1] ASDs are characterized by an opening in the atrial septum that creates a connection between the systemic and pulmonary circulation, which allows oxygenated blood to be shunted into the lower pressure pulmonary circuit. Over time, this shunting can cause irreversible damage to the heart and lungs, which may lead to pulmonary hypertension, right-sided heart failure, and life-threatening atrial fibrillations. In addition, patients with unrepaired ASDs often develop atrioventricular (AV) blocks and are at high risk for sudden cardiac death unless implanted with a pacemaker. The average life expectancy for patients with significant shunting is 45 years.[1]

Most patients with ASDs become symptomatic at some point in their lives.[3] The age of onset of symptoms is variable and is not solely related to the size of the opening in the atrial septum.[3] The most common initial presenting symptom is exercise intolerance in the form of fatigue or exertional dyspnea.[3]

Treatment of ASDs with surgical or percutaneous device closure holds an excellent prognosis for young patients[3]; however, ASDs are often not recognized until adulthood, after irreversible damage to the heart and lungs or stroke injury may have already occurred.[1,2,4]

In some individuals with ASDs, a positive family history for that disorder or other congenital heart malformations exists.[4,5] Genetic mutations associated with ASDs could be a major cause of familial cases.[6] Genetic testing for the presence of a germline mutation in the genes known to be associated with ASDs may[6]:

- Confirm a diagnosis of familial ASDs.
- Assist with clinical management of ASDs.
- Facilitate identification of at-risk individuals in affected families.

Relevant Assays*

Test Name	Test No.
GeneSeq®: Cardio Familial Congenital Heart Disease Profile	451402
GeneSeq®: Cardio Gene Specific Sequencing, NGS**	452053
Mutation-specific Sequencing, Whole Blood†	451382

*Visit the online Test Menu at **www.LabCorp.com** for more information, including a current list of included genes, test methodology, and specimen requirements. To request a sample shipping kit, please call 866-647-0735.
**Full Gene Sequencing for any gene(s) on any of the GeneSeq: Cardio panels
†Known mutation testing for any gene(s) on any of the GeneSeq: Cardio panels

References
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