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FIRST-DEGREE RELATIVES OF PATIENTS WITH THE MOST COMMON CARDIAC BIRTH DEFECT SHOULD BE SCREENED FOR LARGER-THAN-NORMAL AORTAS, NEW STUDY SHOWS

WHAT: Bicuspid Aortic Valve (BAV), a condition in which patients' aortic valves have just two leaflets instead of the normal three, is the most common cardiac anomaly, affecting up to two percent of the general population. The defect can result in calcification deposits on the heart valve, leakage of the valve and may results in a feeling of tightness in the chest as well as shortness of breath. The condition is easily diagnosed; often physicians can hear a "click" or a murmur when they listen to a BAV patient's heart with a stethoscope.

Studies have shown that BAV is likely genetic, although the gene has not been identified, and in some families, incidence of this defect could run as high as 20 percent.

A new study, published in the Journal of the American College of Cardiology, suggests that nearly a third of first-degree relatives (siblings, children or parents) of BAV patients are likely to have enlarged aortas, a potentially serious condition that can only be detected by undergoing transthoracic echocardiograms. This was found even in the absence of any abnormalities of the heart valve itself.

According to the study, 32 percent of first-degree relatives with no heart valve abnormality had significantly larger aortas that expected for age, gender and body size as compared to no enlargement seen in control patients. Also, the study found that the aortas of the first-degree relatives had abnormal stiffness similar to the patients with congenital bicuspid valve. Generally, when aortas are 50 millimeters in diameter, surgery is recommended in order to prevent a rupture of the aorta.

"If you know that a relative does have bicuspid aortic valve, then you know that you should be screened," said study author Kirsten Tolstrup, MD, assistant director of the Cardiac Noninvasive Laboratory at the Cedars-Sinai Heart Institute. "BAV appears to be a genetic condition that has many different manifestations, so we will be studying the genes."

WHO: [Kirsten Tolstrup, MD](#), assistant director of the Cardiac Noninvasive Laboratory at the

(more)

[Cedars-Sinai Heart Institute](#), is available to discuss the study's findings and provide additional details.

- DETAILS:** This study, conducted among 54 patients with bicuspid aortic valve and 48 first-degree relatives of those patients as well as 45 matched controls found:
- 32 percent of apparently healthy first-degree relatives have enlarged aortas
 - 53 percent of BAV patients had enlarged aortas
 - 9.4 percent of first-degree relatives had BAV

RAMIFICATIONS: The findings suggest that patients with bicuspid aortic valve and their first-degree relatives should have a screening echocardiogram to be evaluated for dilated aorta and bicuspid aortic valve.

The study abstract can be accessed at <http://content.onlinejacc.org>

For additional information, call Sandy Van at 800-880-2397 or Sally Stewart at 310-248-6566.

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