

JSPCCS-AHA Joint Session

Sat. Jul 11, 2026 9:10 AM - 10:50 AM JST | Sat. Jul 11, 2026 12:10 AM - 1:50 AM UTC | Room1 (Conference Hall A)

[AHAJS] JSPCCS-AHA Joint Session (III-AHAJS)

Basic Science in Congenital Heart Disease: Morphology, Genetics & Molecular Mechanisms

Chair: Antonio Cabrera (The Ohio State University, Nationwide Children's Hospital)

Chair: Yoshihide Mitani (Perinatal Care center, Mie University Hospital)

[III-AHAJS-2] Notch1 and Bicuspid Aortic Valve Disease: From Human Genetics to Mouse and Cell-based Models

○ Vidu Garg (Nationwide Children's Hospital and The Ohio State University)

Bicuspid aortic valve (BAV) is the most common type of cardiac malformation with a prevalence of ~1% in the population. BAV is associated with congenital aortic valve stenosis found in infancy along with later onset aortopathy and calcific aortic valve disease. Pathogenic variants in *NOTCH1* were discovered to be a cause of BAV in humans. Here, we will review evidence supporting the independent roles of Notch1 signaling in aortic valve cells and in smooth muscle cells of the proximal aorta that contribute to the associated diseases of congenital valve stenosis, ascending aortic aneurysms and valve calcification using a combination of mouse and induced pluripotent stem cell models. These findings suggest that genetic contributions to a congenital heart valve malformation are also associated with adult onset cardiovascular diseases.