CONGENITAL BICUSPID AORTIC VALVE: 2026 CURRENT STATUS & PARADIGM SHIFTS

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I THE GENETIC AND PATHOPHYSIOLOGICAL SHIFT

- **Oligogenic and Polygenic Architecture:** Bicuspid Aortic Valve (BAV, prevalence 0.5 to 2.0 percent with a 3:1 male predominance) is no longer viewed as a simple single-gene disorder. While autosomal dominant variants with incomplete penetrance occur in approximately 10 to 30 percent of families—involving NOTCH1 (associated with calcific valve disease), SMAD6, ROBO4, GATA4/GATA5/GATA6, ACTA2, and TGFBR1/2 (in overlapping syndromic aortopathies)—most sporadic BAV cases reflect a complex polygenic and oligogenic architecture governing second heart field, neural crest cell migration, and extracellular matrix homeostasis.
- **International Consensus Nomenclature (Michelena et al., 2021):** Standardized international nomenclature classifies BAV into three major types: Fused BAV (90 to 95 percent of cases, featuring two functional cusps, three distinguishable sinuses, and a fibrous/calcified raphe—most commonly Right-Left [R-L] coronary cusp fusion [70 to 80 percent], followed by Right-Noncoronary [R-N] fusion [20 to 30 percent], and rare Left-Noncoronary [L-N] fusion), 2-Sinus BAV (5 to 7 percent, true two-sinus/two-cusp valve without a raphe), and Partial-Fusion (Forme Fruste) BAV.
- **The "Hemodynamic vs. Genetic" Consensus:** BAV aortopathy (present in 40 to 50 percent of patients) is definitively established as a dual-hit phenomenon combining an intrinsic genetic/developmental medial wall defect (vascular smooth muscle cell apoptosis, elastic fiber fragmentation, and matrix metalloproteinase upregulation) with abnormal helical systolic flow and localized Wall Shear Stress (WSS) generated by the restricted bicuspid orifice.

II ADVANCED MULTIMODALITY IMAGING: 4D-FLOW CMR

- **Current Clinical Role:** Time-resolved 3-dimensional phase-contrast MRI (4D-Flow CMR) has evolved from a research tool into an advanced clinical imaging adjunct in specialized aortic centers for non-invasively mapping 3D helical flow eccentricity, flow displacement, and regional Wall Shear Stress (WSS).
- **Cusp Fusion-Specific Flow Jets:**
  - **Right-Left (R-L) Cusp Fusion:** Directs an eccentric anterior/right-handed helical systolic jet against the right-anterior convexity of the mid-ascending tubular aorta, predominantly driving the Ascending (Tubular) Aortopathy Phenotype.
  - **Right-Noncoronary (R-N) Cusp Fusion:** Directs a high-velocity jet posteriorly and superiorly toward the posterior ascending aorta and proximal aortic arch, more frequently associated with distal ascending/arch dilation and earlier valvular stenosis/regurgitation in children.
- **Prognostic Utility:** Regions of elevated systolic WSS on 4D-Flow CMR correlate directly with localized elastic fiber thinning and extracellular matrix dysregulation on surgical histopathology, helping identify patients with borderline aortic diameters (45 to 49 mm) at higher risk of rapid aortic growth.

III TAVR IN BICUSPID ANATOMY: THE 2026 LANDSCAPE

- **Anatomical and Procedural Challenges:** Compared with tricuspid aortic stenosis, BAV anatomy presents an elliptical or non-circular annulus, asymmetric cusp dimensions, bulky eccentric calcification (especially a calcified raphe), concomitant horizontal/dilated ascending aorta (45 mm or greater), and anomalous coronary ostial heights—increasing the risk of asymmetric stent frame expansion, paravalvular leak (PVL), aortic root/annular rupture, and permanent pacemaker implantation.
- **Current Evidence (Registry Data & NOTION-2 Trial, 2024):**
  - **Observational Registries (STS/ACC TVT Registry):** With contemporary third/fourth-generation devices (SAPIEN 3 Ultra / Resilia and Evolut PRO+ / FX), 1-year all-cause mortality in anatomically selected low- and intermediate-risk BAV patients is comparable to tricuspid TAVR, though BAV patients with both a calcified raphe and excess leaflet calcification have significantly higher rates of aortic root injury, moderate-to-severe PVL, and 1-year mortality (Yoon et al. BAV TAVR Registry).
  - **NOTION-2 Trial (European Heart Journal 2024):** The first randomized trial comparing TAVR versus SAVR in young (age 75 years or younger, mean age 71 years), low-risk patients including a prespecified BAV cohort showed a higher rate of the composite of death, stroke, or rehospitalization (14.3 percent vs. 3.9 percent) and higher non-disabling stroke and paravalvular regurgitation with TAVR than SAVR in the bicuspid subgroup, reinforcing caution against routine TAVR in young low-risk BAV patients.
- **CT Sizing Strategies (BAVARD / LIRA / Casper Methods):** Pre-procedural MDCT planning incorporates supra-annular sizing at the intercommissural distance (ICD) 4 mm above the annulus: in tubular configurations (annulus equal to ICD), standard annular sizing is used; in flared configurations (ICD greater than annulus), annular sizing is used; and in tapered configurations (ICD smaller than annulus) or heavy calcified raphes, supra-annular sizing with slight downsizing prevents fatal root/raphe rupture.
- **Guideline Stance (2020 ACC/AHA & 2021 ESC/EACTS):** SAVR remains the Class I recommendation for young patients (under 65 to 75 years), patients with concomitant ascending aortopathy (45 mm or greater), or unfavorable BAV anatomy (calcified raphe plus excess leaflet calcium), whereas TAVR may be considered as an alternative to SAVR in older or intermediate/high-surgical-risk BAV patients with favorable anatomy after Heart Team evaluation (neither guideline assigns a specific class to bicuspid TAVR, as BAV patients were excluded from the pivotal TAVR trials).

IV THE SURGICAL RENAISSANCE: THE ROSS PROCEDURE AND AORTIC VALVE REPAIR

- **Current Status of the Ross Procedure:** The Ross Procedure (replacing the diseased aortic valve with the patient's own pulmonary autograft and reconstructing the right ventricular outflow tract with a cryopreserved pulmonary homograft) has experienced a major evidence-based resurgence for young and middle-aged adults (under 50 years of age) when performed in high-volume expert centers.
- **Rationale and Survival Advantage:** Unlike mechanical valves (which require lifelong vitamin K antagonists and carry a 1 to 2 percent annual risk of thromboembolism or major bleeding) or bioprosthetic valves (which undergo accelerated structural deterioration in patients under 50), the living pulmonary autograft provides physiological hemodynamics, zero anticoagulation requirement, very low endocarditis risk, and restores 15-to-20-year survival to match the age- and sex-matched general population.
- **Guideline Stance and Valve-Sparing Repair:** In the 2020 ACC/AHA Valvular Guidelines, the Ross procedure holds a Class IIb recommendation for patients under 50 years of age with appropriate anatomy when performed by experienced surgeons at a Comprehensive Valve Center (Aortic Valve Repair / Valve-Sparing Root Replacement [David reimplantation] is a reasonable alternative in experienced centers for young patients with pure BAV aortic regurgitation and pliable cusps).

V PHENOTYPE-DRIVEN AORTIC MANAGEMENT (2022 ACC/AHA & 2024 ESC AORTIC GUIDELINES)

- **Two Distinct Aortopathy Phenotypes:**
  - **Ascending (Tubular) Phenotype (70 to 80 percent of BAV aortopathy):** Dilation predominantly above the sinotubular junction in the tubular ascending aorta; typically associated with older age, R-L cusp fusion, and aortic stenosis, with a slower progression rate.
  - **Root Phenotype (15 to 20 percent of BAV aortopathy):** Dilation predominantly at the Sinuses of Valsalva (annuloaortic ectasia); occurs predominantly in younger men with pure aortic regurgitation, behaves more like a genetically triggered Marfan-like aortopathy, and carries a significantly higher risk of acute Type A aortic dissection, mandating a lower surgical threshold.

Clinical Scenario in BAV Aortopathy	Ascending/Root Diameter Threshold	2022 ACC/AHA & 2024 ESC Class of Recommendation
Any BAV Patient (Without Risk Factors)	55 mm or greater	**Class I:** Elective surgical replacement of ascending aorta/root
BAV with "Root Phenotype" or High-Risk Features (Family history of dissection, growth rate 3 mm/year or greater, coarctation, uncontrolled HTN, or low surgical risk at a Comprehensive Valve Center)	50 mm or greater	**Class IIa:** Elective surgery recommended at 50 mm or greater
Concomitant Surgery (Patient already undergoing SAVR for severe AS or severe AR)	45 mm or greater	**Class IIa:** Concomitant replacement of the ascending aorta/root during SAVR

KEY REFERENCES

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