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Original Article

Frequency of Bicuspid Aortic Valve and Coarctation of Aorta in Young Patients Presenting with Aortic Stenosis

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ABSTRACT

Background: Aortic stenosis in young patients is frequently associated with congenital structural abnormalities rather than degenerative valvular disease. Among these, bicuspid aortic valve and coarctation of the aorta are clinically important because their coexistence may increase aortic complications, ventricular loading, and long-term cardiovascular morbidity. Objective: To determine the frequency of bicuspid aortic valve and coarctation of the aorta in young patients presenting with aortic stenosis. Methods: This cross-sectional study was conducted at the emergency and outpatient departments of Shaikh Zahid Bin Al Nayyan Institute of Cardiology, Quetta, from January 2025 to December 2025. A total of 136 young patients with aortic stenosis were enrolled after informed consent. Demographic and clinical data were recorded, and echocardiography was performed by a consultant cardiologist to identify bicuspid aortic valve and coarctation of the aorta. Data were analyzed using SPSS version 26.0. Quantitative variables were summarized as mean ± standard deviation, qualitative variables as frequencies and percentages, and post-stratification chi-square testing was applied with $p \leq 0.05$ considered significant. Results: The mean age was 31.86 ± 8.21 years, and 58.8% of participants were male. Bicuspid aortic valve was identified in 34 patients (25.0%), while coarctation of the aorta was present in 52 patients (38.2%). Diabetes mellitus showed a significant association with bicuspid aortic valve ($p = 0.025$), whereas age, sex, hypertension, and smoking were not significantly associated with bicuspid aortic valve or coarctation of the aorta. Conclusion: Bicuspid aortic valve and coarctation of the aorta were common among young patients presenting with aortic stenosis. Early recognition of these associated congenital abnormalities may improve diagnostic assessment, surveillance, and management in this population. Keywords: aortic stenosis, bicuspid aortic valve, coarctation of aorta, congenital heart disease, echocardiography, young patients.

INTRODUCTION

Aortic stenosis is one of the most clinically significant valvular heart diseases and is characterized by progressive narrowing of the aortic valve orifice, resulting in obstruction to left ventricular outflow, increased transvalvular pressure gradient, compensatory ventricular remodeling, and, in advanced cases, substantial cardiovascular morbidity. Although degenerative calcific disease is the predominant cause in older adults, aortic stenosis in younger patients is more commonly linked to congenital structural abnormalities, particularly bicuspid aortic valve, which predisposes affected individuals to earlier valvular dysfunction, accelerated calcific change, and progressive stenotic lesions (1-4). Bicuspid aortic valve is the most frequent congenital cardiac valvular anomaly, affecting nearly 1% of the general population with a recognized male predominance, and it is clinically important not only because of its

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association with premature aortic stenosis and regurgitation but also because of its close relationship with aortopathy, aortic dilatation, and other congenital cardiovascular defects (1,3,6-8).

Among the congenital abnormalities associated with bicuspid aortic valve, coarctation of the aorta is particularly relevant because both conditions may coexist within a broader arteriopathic phenotype involving abnormal aortic wall structure and altered aortic hemodynamics. Coarctation of the aorta contributes to increased afterload, abnormal vascular shear stress, systemic hypertension, and long-term risk of aneurysmal change or other aortic complications, while the simultaneous presence of bicuspid aortic valve may further intensify these risks through combined valvular and vascular pathology (5,9-17). Previous literature has shown that bicuspid aortic valve is highly prevalent among patients with coarctation of the aorta, and the coexistence of these abnormalities has been associated with a greater burden of aortic complications, including ascending aortic dilatation, aneurysm formation, left ventricular strain, and adverse long-term outcomes even after repair of coarctation (5,10-18). These observations support the need for early recognition of associated congenital lesions in patients who initially present with aortic stenosis, especially in younger age groups where congenital etiologies are more likely to predominate than age-related degenerative causes.

Despite the established pathophysiological association between bicuspid aortic valve, coarctation of the aorta, and aortic stenosis, the frequency of these abnormalities among young patients presenting with aortic stenosis remains insufficiently documented in many local and regional settings. Most available evidence originates from international cohorts, while data from Pakistan, particularly from cardiac referral centers managing younger symptomatic patients, remain limited. This gap restricts context-specific risk stratification and may delay timely evaluation for associated congenital aortic pathology in routine cardiology practice. Determining the burden of bicuspid aortic valve and coarctation of the aorta in young patients with aortic stenosis may therefore improve diagnostic vigilance, guide echocardiographic assessment, and support earlier referral for appropriate surveillance or intervention. The present study was conducted to determine the frequency of bicuspid aortic valve and coarctation of the aorta in young patients presenting with aortic stenosis at a tertiary cardiac center in Quetta (19,20).

MATERIALS AND METHODS

This cross-sectional study was conducted at the emergency department and outpatient department of Shaikh Zahid Bin Al Nayyan Institute of Cardiology, Quetta, from January 2025 to December 2025. The study focused on young patients presenting with aortic stenosis who fulfilled the predefined study eligibility criteria and were enrolled during the study period after provision of informed consent. Patients were interviewed using a structured clinical proforma, and baseline demographic and clinical characteristics were recorded by the investigating team at the time of enrollment. Relevant variables included age, sex, history of diabetes mellitus, hypertension, and smoking status, while the principal outcome variables were the presence of bicuspid aortic valve and coarctation of the aorta.

All enrolled patients underwent echocardiographic evaluation performed by a consultant cardiologist with assistance from the investigating doctor. Echocardiography was used as the principal diagnostic tool to assess aortic valve morphology and to identify the presence or absence of coarctation of the aorta in patients already presenting with aortic stenosis. For analytical purposes, age was summarized as a continuous variable and was also categorized into clinically relevant groups. Diabetes mellitus, hypertension, smoking status, bicuspid aortic valve, and coarctation of the aorta were treated as categorical variables and analyzed as frequencies and proportions. Data collection was carried out in a uniform clinical setting to maintain consistency in patient assessment and diagnostic evaluation.

To improve internal consistency of measurement, echocardiographic findings were documented in a standardized manner for all participants, and clinical information was recorded directly by the study team at the time of patient contact. The use of a single-center protocol, direct patient assessment, and cardiologist-performed echocardiography helped reduce information variability across participants.

Data was entered and analyzed using SPSS version 26.0. Quantitative data were presented as mean and standard deviation, whereas qualitative variables were presented as frequencies and percentages. Stratified analyses were performed for age, sex, diabetes mellitus, hypertension, and smoking status to examine their association with bicuspid aortic valve and coarctation of the aorta. Post-stratification chi-square testing was applied, and a p-value of 0.05 or less was considered statistically significant.

The study was conducted in accordance with institutional ethical standards. Informed consent was obtained from all participants before inclusion in the study, confidentiality of patient information was maintained throughout data collection and analysis, and all recorded data were handled solely for research purposes.

RESULTS

A total of 136 young patients presenting with aortic stenosis were included in the analysis. The mean age of the study population was 31.86 ± 8.21 years, with an age range of 18 to 45 years. Males constituted 58.8% of the cohort, while females accounted for 41.2%. Regarding clinical comorbidity profile, 16.9% of participants had diabetes mellitus, 36.8% had hypertension, and 36.0% were smokers. Overall, bicuspid aortic valve was identified in 34 patients, corresponding to a prevalence of 25.0% (95% CI: 18.5%–32.9%), whereas coarctation of the aorta was found in 52 patients, corresponding to a prevalence of 38.2% (95% CI: 30.5%–46.6%). These findings indicate that approximately one in four young patients with aortic stenosis had bicuspid aortic valve, and more than one in three had coarctation of the aorta.

Table 1. Baseline Characteristics and Overall Frequency of Study Outcomes (n = 136)

Variable	n	%	Additional Statistic
Age (years), mean ± SD	136	—	31.86 ± 8.21
Age range	136	—	18–45
Male	80	58.8	—
Female	56	41.2	—
Diabetes mellitus	23	16.9	—
Hypertension	50	36.8	—
Smoking	49	36.0	—
Bicuspid aortic valve	34	25.0	95% CI: 18.5–32.9
Coarctation of aorta	52	38.2	95% CI: 30.5–46.6

The age distribution showed that 44.9% of the cohort fell within the 18–30-year category, 34.6% were aged 31–40 years, and 20.6% were aged 41–45 years. The proportion of bicuspid aortic valve was numerically highest in the 31–40-year group, where 14 of 47 patients (29.8%) were affected, compared with 13 of 61 (21.3%) in the 18–30-year group and 7 of 28 (25.0%) in the 41–45-year group. However, this variation was not statistically significant (p = 0.601). By sex, bicuspid aortic valve was present in 25.0% of males and 25.0% of females, showing no measurable sex-based difference.

Table 2. Stratified Association of Baseline Factors with Bicuspid Aortic Valve

Variable	Category	BAV Present n/N	% with BAV	Odds Ratio (95% CI)	p-value
Age	18–30 years	13/61	21.3	Reference pattern only*	0.601
	31–40 years	14/47	29.8	Reference pattern only*	
	41–45 years	7/28	25.0	Reference pattern only*	
Sex	Male	20/80	25.0	1.00 (0.45–2.20)	—
	Female	14/56	25.0	Reference	
Diabetes mellitus	Yes	10/23	43.5	2.85 (1.11–7.30)	0.025
	No	24/113	21.2	Reference	
Hypertension	Yes	13/50	26.0	1.09 (0.49–2.42)	0.837
	No	21/86	24.4	Reference	
Smoking	Yes	13/49	26.5	1.13 (0.51–2.53)	0.757
	No	21/87	24.1	Reference	

Diabetes mellitus was the only factor demonstrating a statistically significant association with bicuspid aortic valve: 10 of 23 diabetic patients (43.5%) had bicuspid aortic valve compared with 24 of 113 non-

diabetic patients (21.2%), yielding an odds ratio of 2.85 (95% CI: 1.11–7.30; p = 0.025). Hypertension and smoking were not significantly associated with bicuspid aortic valve.

Coarctation of the aorta was observed in 52 patients overall. Age-specific proportions were 39.3% in the 18–30-year group, 36.2% in the 31–40-year group, and 39.3% in the 41–45-year group, showing minimal variation across age strata (p = 0.937). The prevalence of coarctation of the aorta was 40.0% in males and 35.7% in females, corresponding to an odds ratio of 1.20 (95% CI: 0.59–2.43; p = 0.613). Among diabetic participants, 34.8% had coarctation of the aorta compared with 38.9% of non-diabetic participants, which was not statistically significant (OR 0.84, 95% CI: 0.33–2.14; p = 0.709). Similarly, hypertension and smoking showed no statistically significant associations with coarctation of the aorta, with effect estimates close to the null. Collectively, these results suggest that while coarctation of the aorta was relatively frequent in this population, none of the examined baseline factors demonstrated a significant association with its occurrence.

Table 3. Stratified Association of Baseline Factors with Coarctation of the Aorta

Variable	Category	CoA Present n/N	% with CoA	Odds Ratio (95% CI)	p-value
Age	18–30 years	24/61	39.3	Reference pattern only*	0.937
	31–40 years	17/47	36.2	Reference pattern only*	
	41–45 years	11/28	39.3	Reference pattern only*	
Sex	Male	32/80	40.0	1.20 (0.59–2.43)	0.613
	Female	20/56	35.7	Reference	
Diabetes mellitus	Yes	8/23	34.8	0.84 (0.33–2.14)	0.709
	No	44/113	38.9	Reference	
Hypertension	Yes	19/50	38.0	0.98 (0.48–2.02)	0.966
	No	33/86	38.4	Reference	
Smoking	Yes	20/49	40.8	1.19 (0.58–2.43)	0.642
	No	32/87	36.8	Reference	

*Age-stratified counts were reported in three categories in the source manuscript; a single overall chi-square p-value was available, but no directly reported pairwise odds ratios were provided for those three groups.

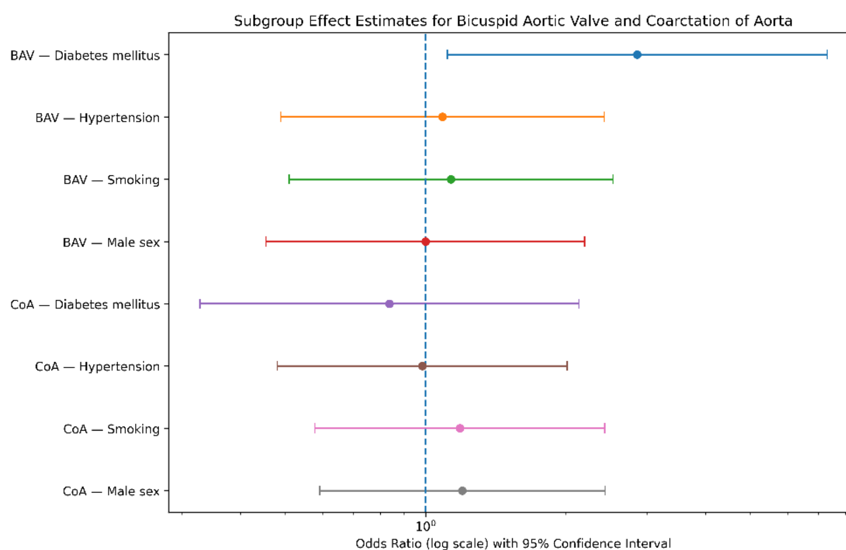

Figure 1 Subgroup Effect Estimates for Bicuspid Aortic Valve and Coarctation of Aorta

The figure demonstrates that diabetes mellitus showed the strongest positive association with bicuspid aortic valve, with an odds ratio of 2.85 and a 95% confidence interval of 1.11–7.30, making it the only subgroup effect whose confidence interval excluded the null. In contrast, the effect estimates for male sex, hypertension, and smoking in relation to bicuspid aortic valve remained clustered near unity, ranging from 1.00 to 1.13, with all confidence intervals crossing 1.00. For coarctation of the aorta, all subgroup estimates were similarly non-significant, with odds ratios ranging from 0.84 for diabetes

mellitus to 1.20 for male sex, again with broad confidence intervals overlapping the null. This pattern indicates that within the available baseline clinical variables, diabetes mellitus was the only factor showing a measurable excess likelihood of bicuspid aortic valve, whereas no comparable predictor signal emerged for coarctation of the aorta in this cohort.

DISCUSSION

The present study evaluated the frequency of bicuspid aortic valve and coarctation of the aorta among young patients presenting with aortic stenosis at a tertiary cardiac center and demonstrated that bicuspid aortic valve was present in 25.0% of patients, while coarctation of the aorta was identified in 38.2%. These findings reinforce the clinical importance of investigating associated congenital aortic abnormalities in younger patients with aortic stenosis, a group in whom congenital structural causes are more likely to underlie valvular obstruction than degenerative senile processes. The observed burden of bicuspid aortic valve in this cohort is clinically meaningful because bicuspid morphology is a recognized substrate for earlier stenotic degeneration, abnormal cusp biomechanics, and progressive valvulo-aortopathy, all of which may influence long-term cardiovascular outcomes and the need for surveillance or intervention (21-24).

The frequency of bicuspid aortic valve observed in this study is broadly consistent with the known role of congenital bicuspid morphology as a major cause of aortic stenosis in younger individuals. Earlier literature has shown that bicuspid aortic valve is the most common congenital valvular abnormality and is strongly associated with premature valvular dysfunction, aortic dilatation, and proximal aortic pathology (21,23,24). The present findings therefore support the concept that young patients with aortic stenosis should not be evaluated solely for the severity of valvular narrowing, but also for underlying congenital valve morphology and associated aortic disease. From a practical standpoint, the finding that one in four patients had bicuspid aortic valve suggests that systematic echocardiographic assessment of cusp morphology in this patient population is justified and may prevent under-recognition of a clinically relevant structural abnormality.

The proportion of patients with coarctation of the aorta in this study was also notable. Although coarctation is less commonly discussed than bicuspid aortic valve in routine adult valvular practice, its coexistence with bicuspid valve disease is well established and has major implications for cardiovascular risk. Previous studies have demonstrated that bicuspid aortic valve and coarctation may coexist as part of a broader congenital aortopathy, with increased susceptibility to altered aortic flow dynamics, ascending aortic dilatation, aneurysm formation, persistent hypertension, and late complications even after anatomical repair of the coarctation segment (25-31). The high frequency of coarctation found in this cohort highlights the importance of looking beyond the valve itself when young patients present with aortic stenosis, particularly in settings where delayed diagnosis of congenital lesions may still occur.

With regard to subgroup analysis, diabetes mellitus was the only baseline factor significantly associated with bicuspid aortic valve in the present study, whereas age group, sex, hypertension, and smoking status were not significantly associated with either bicuspid aortic valve or coarctation of the aorta. This pattern should be interpreted cautiously. Because bicuspid aortic valve and coarctation of the aorta are fundamentally congenital abnormalities, the lack of strong association with conventional adult cardiovascular risk factors is biologically plausible. The statistically significant association observed between diabetes mellitus and bicuspid aortic valve may reflect sample-specific clustering rather than a causal relationship, especially given the relatively small number of diabetic participants and the cross-sectional nature of the study. It is therefore more appropriate to interpret diabetes mellitus in this dataset as an observed correlational signal rather than an established etiological determinant. Larger multicenter studies using multivariable modeling would be needed to clarify whether this finding persists after more robust adjustment for potential confounding.

Another important implication of the study relates to clinical screening and risk stratification. Young patients with aortic stenosis may initially present with symptoms or echocardiographic findings that focus clinical attention on the valve lesion alone. However, the coexistence of bicuspid aortic valve and coarctation of the aorta may increase left ventricular afterload, amplify hemodynamic stress across the aortic root and ascending aorta, and contribute to long-term morbidity beyond the stenotic lesion itself (26-31). In this context, early diagnosis is essential not only for defining the anatomical basis of aortic stenosis but also for identifying patients who may require closer imaging follow-up, blood pressure control, surgical planning, or staged management of combined valvular and aortic pathology. The study therefore adds practical value by supporting a more comprehensive echocardiographic and aortic assessment strategy in young patients presenting with aortic stenosis.

The findings should also be interpreted in light of several limitations. This was a single-center cross-sectional study, which limits generalizability beyond the source population and precludes temporal or causal inference. The study sample, although adequate for descriptive estimation, was relatively modest for detecting subgroup differences, and multivariable regression analysis was not performed, limiting the ability to account for confounding. The manuscript also relied primarily on echocardiographic assessment without a broader multimodality imaging framework, and detailed operational definitions for severity gradation or diagnostic standardization were not fully described in the source text. In addition, because the data were collected from a tertiary referral cardiac setting, the frequencies observed may reflect referral enrichment rather than population-level prevalence. Despite these limitations, the study provides clinically relevant local data in an area where regional evidence remains limited and offers a useful basis for future prospective and multicenter research.

Overall, the study supports the view that congenital structural abnormalities are common among young patients with aortic stenosis and that both bicuspid aortic valve and coarctation of the aorta warrant active consideration during diagnostic workup. Future studies should aim to validate these findings in larger and more diverse cohorts, apply standardized imaging criteria, and evaluate how the coexistence of these abnormalities influences intervention timing, long-term aortic complications, and survival outcomes.

CONCLUSION

Among young patients presenting with aortic stenosis, bicuspid aortic valve and coarctation of the aorta were identified with notable frequency, affecting 25.0% and 38.2% of the study population, respectively. These findings underscore the importance of comprehensive structural evaluation in younger patients with aortic stenosis, as associated congenital aortic abnormalities may substantially influence morbidity, surveillance needs, and therapeutic planning. Early echocardiographic recognition of bicuspid aortic valve and coarctation of the aorta may improve risk stratification and support timely intervention in this clinically important population.

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