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Bicuspid aortic valves (BAV) Registry (RE-BAV): Clinical and echocardiographic characteristics of patients with BAV according to novel classification of bicuspid aortic valves

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WHAT'S NEW?

The publication presents the results of the first Polish Registry of Bicuspid Aortic Valves (RE-BAV) in adults, conducted between 2021 and 2023 across 23 tertiary centers using the latest BAV classification. The registry included 814 patients, providing updated insights into BAV phenotypes in the Polish population. Clinical and echocardiographic data were analyzed, with phenotypes and aortopathy adjudicated using the new classification. The most common phenotype was typical valvulo-aortopathy with valve dysfunction and/or aortic dilatation without major associated diseases. The occurrence of the three BAV phenotypes differs from

previous publications, including a higher prevalence of the partial-fusion BAV type. The fusion type of BAV, particularly right-left cusps fusion, was the most frequent phenotype, while the partial-fusion type also showed a notable presence. Clinical characteristics and comparison of the three BAV phenotypes is provided. The most prevalent aortic malformation was extended aortic dilatation, involving the root and ascending aorta, which differs from previous studies.

ABSTRACT

Background: Bicuspid aortic valve (BAV) is a common congenital heart defect linked to abnormal valve structure and aortic dilatation.

Aims: To present BAV types and valvulo-aortopathy in the Polish population using the latest 2021 classification.

Methods: RE-BAV is a registry of adult ambulatory and hospitalized patients with BAV evaluated in echocardiographic laboratories at 23 tertiary centers in Poland (2021–2023).

Results: The study included 814 patients — 72.7% male, average age — mean (SD) 50 (17.4). Common symptoms included dyspnea (54.1%) and chest pain (17.5%). Hypertension (54%) was the most frequent comorbidity. Left ventricular ejection fraction was normal (median 60%, interquartile range: 55–65), but global longitudinal strain was mildly reduced — mean (SD) –16.8% (3.7). Moderate/severe aortic stenosis was found in 34.2% and regurgitation in 44.1% (P <0.001).

The most common phenotype was typical valvulo-aortopathy (69.9%), followed by uncomplicated BAV (19.3%) and complex valvulo-aortopathy (10.8%). Among 640 patients with specified subtypes, fusion was the most frequent BAV type (79.4%), followed by 2-sinus (15.8%) and partial-fusion (4.8%) (P <0.001 for all comparisons). Patients with the 2-sinus type were the youngest and had the least comorbidities, contrary to the partial-fusion group. Right-left cusp fusion was the most common subtype (80.4% of fusion BAV). Within the 2-sinus type the latero-lateral and antero-posterior phenotypes had similar prevalence. Aortic dilatation occurred in 63.6%, with extended aortic dilatation being most prevalent (26.3%).

Conclusions. The RE-BAV registry provides updated insights into BAV phenotypes and aortopathy in the Polish population, reflecting the latest classification advancements.

Key words: aortopathy, bicuspid aortic valve, valve phenotypes, valvulopathy

INTRODUCTION

The prevalence of bicuspid aortic valve (BAV) in the general population is estimated at 1%–2%, making it the most common adult congenital heart defect [1]. In patients with BAV, in addition to changes in the structure and function of the valve itself (i.e. stenosis or regurgitation), there is often a dilatation of the proximal part of the aorta (root and ascending aorta), known as BAV-related aortopathy. It is a common and heterogenous clinical problem most often recognized during standard echocardiographic examination. This publication presents the results of the first Polish Registry of Bicuspid Aortic Valves (RE-BAV) of the adult population and reports BAV morphology, according to the new latest classification [2].

MATERIAL AND METHODS

The registry was conducted in 23 Polish tertiary cardiological centers, mainly university based, with the participation of echocardiography experts who held the Echocardiography Association Certificates of the Polish Cardiac Society. The registry included consecutive inpatients and outpatients presenting to the echocardiographic laboratories in each of the 23 participating centers between 2021 and 2023. Patients with acute coronary syndrome, hemodynamic instability, or recent valvular heart surgery were excluded. Data on comorbidities, including coronary artery disease, history of myocardial infarction, stroke, clinically significant arrhythmias, atrial fibrillation, arterial hypertension, dyslipidemia, nicotine addiction, diabetes, and malignancy, were recorded. Active neoplastic disease was defined as any malignancy diagnosed in the past 6 months, currently undergoing treatment, or showing recurrence or progression. All subjects underwent medical history, clinical examination, and initial transthoracic echocardiography. If needed, transesophageal echocardiography, cardiac computed tomography, or electrocardiography-gated computed tomography angiography was performed when specific aortic segments could not be visualized or exceeded 45 mm. Serum N-terminal pro-B-type natriuretic peptide (NT-proBNP) levels were measured when available. According to the decision of Pomeranian Medical University ethical committee no additional formal consent for this study was required. The registry was carried out under the patronage of the Valvular Heart Association of the Polish Cardiac Society.

Echocardiographic examination

Baseline clinical evaluation included M-mode, 2-dimentional (2D) echocardiography, and color Doppler, performed by experienced echocardiographers equipped with state-of-the-art technology following current guidelines [3]. Ventricular volume and ejection fraction were calculated using the 2-plane Simpson method, with 3-dimentional echocardiography used when

available. Regional wall motion abnormalities were noted. Bicuspid aortic valve disease was confirmed in the short axis view at the level of the aortic leaflets (Figure 1) [2]. Evaluation of aortic valve disease with regard to stenosis or regurgitation and 2D measurements of the aorta at various levels were performed according to the latest guidelines (Figure 2) [4, 5]. Additionally mitral and tricuspid valve defects were noted.

Clinical classification of bicuspid aortic valve disease

The BAV disease was categorized into 3 clinical subgroups: (i) **complex valvulo-aortopathy** characterized by concomitant or associated disorders (i.e., Turner syndrome, Loeys–Dietz syndrome, Shone complex, severe aortic coarctation, concomitant non-dilated cardiomyopathy) and/or by early/accelerated valve dysfunction and/or aortopathy; (ii) **typical valvulo-aortopathy** with progressive BAV dysfunction and/or aortic dilatation without other major associated disorders and (iii) **uncomplicated BAV** with mild or non-progressing valvulo-aortopathy without clinical manifestation [2].

Three BAV phenotypes were distinguished: (1) the fused BAV, (2) the 2-sinus BAV and (3) the partial-fusion BAV (Figure 1). **1.The fused BAV** was characterized by 2 of the 3 cusps appearing fused or joined within 3 distinguishable aortic sinuses, resulting in 2 functional cusps. Within the fused type 3 specific BAV phenotypes were differentiated: right–left cusp fusion, right–noncoronary cusp fusion and left–noncoronary cusp fusion. **2. The 2-sinus BAV** type was defined by the presence of 2 cusps of approximately equal size and shape, with each cusp occupying 180° of the annular circumference, and only 2 aortic sinuses, resulting in a 2-sinus/2-cusp valve. Two specific phenotypes of the 2-sinus BAV category were distinguished: latero-lateral (side-to-side) or antero-posterior (front and back) based on the short-axis base-of-the-heart plane. **3. The partial-fusion BAV** was recognized when a typical tricuspid aortic valve with 3 sinuses and 3 symmetrical cusps with a systolic triangular opening and commissural angles of 120 was present, yet at the base of one of the commissures <50% cusp fusion forming a small ‘mini-raphe’ was noted.

Definition of aortic dilatation and bicuspid aortic valve aortopathy

Three forms of aortic dilatation BAV were distinguished according to the latest the American Heart Association/American College of Cardiology 2022 comprehensive guideline for the diagnosis and management of aortic diseases [6]: (i) the ascending phenotype with dilatation preferentially located at the tubular ascending tract beyond the sino-tubular junction, (ii) the root phenotype with dilatation preferentially located at the root (sinuses of Valsalva) and (iii)

the extended phenotype with dilatation of the root, the ascending aorta and the arch (Figure 2). Reference normal values were adopted depending on gender with the cut-off values for aortic root 40 mm and 34 mm, ascending aorta 40 mm and 36 mm, and aortic arch 34 mm and 31mm for men and women respectively [5, 7].

Statistical analysis

Data analysis was performed using SAS version 9.4 (SAS Institute, Cary, NC, US). The normal distribution of all continuous variables was examined using the Kolmogorow–Smirnov test. Numerical variables with normal distribution are presented as mean with standard deviation, and skewed are expressed as median (interquartile range [IQR]). Categorical variables are reported as frequency and percentage and compared using the χ^2 Test for Equal Proportions or McNemar's Test. The proportion of categorical variables with the number of categories >2 was verified by general association of the Cochran–Mantel–Haenszel test. The differences between numerical variables were assessed by one-way ANOVA, with Tukey's post-hoc test for pairwise two-sided multiple comparison (normally distributed data) or, in the case of skewed distribution, with Kruskall–Wallis ANOVA with pairwise two-sided multiple comparison by DSFC method (Dwass, Steel, Critchlow-Fligner). A *P*-value <0.05 was considered statistically significant. No missing value imputation techniques were used.

RESULTS

The RE-BAV registry encompassed 814 patients with BAV, including 592 (72.7%) men. The mean (standard deviation [SD]) age was 50 (17.4) years. The most common symptoms reported were shortness of breath (54.1%) and chest pain (17.5%). The most common comorbid disease was hypertension (54.0%). 5.5% of patients had a history of cancer. Marfanoid features were present only in 9 (1.1%) participants. Clinical characteristics of the examined population is presented in Table 1.

Complex valvulo-aortopathy was present in 88 (10.8%) patients. The most common associated disorder was aortic coarctation found in 56 (7.1%) of the examined population. **Typical valvulo-aortopathy** with progressive BAV dysfunction and/or aortic dilatation without other major associated disorders was present in the majority of the examined population — 569 patients (69.9%), while **uncomplicated BAV** with mild or non-progressing valvulo-aortopathy without clinical manifestation was noted in 157 (19.3%).

Bicuspid aortic valve phenotypes

The BAV phenotype was identified in 640 patients (Table 1). The fusion type was most common (508 cases, 79.4%), followed by the double-sinus type (101 cases, 15.8%), and partial-cusp fusion (31 cases, 4.8%) (P <0.001 for all comparisons) (Figure 3). Among the fusion phenotype, right-left fusion was most frequent (80.4%), followed by right-noncoronary (12.9%) and left-noncoronary cusp fusion (6.7%). Within the double-sinus type, the latero-lateral and antero-posterior phenotypes were equally common.

Differences between BAV phenotypes concerned patient age (P <0.001), atrial fibrillation (P <0.001), arterial hypertension (P = 0.034), nicotine addiction (P = 0.019), and NT-proBNP levels (P = 0.021). Patients with the 2-sinus type were the youngest, while those with the partial-fusion type were the oldest. Atrial fibrillation and arterial hypertension were most prevalent in the partial-fusion group, followed by the fused BAV and two-sinus types. Nicotine addiction was highest in the fused BAV group, while NT-proBNP levels were highest in the partial-fusion group. Cancer diagnoses were most common in the fused BAV type, although without significant differences between groups. Coronary artery disease was equally distributed among all BAV phenotypes.

Aortic dilatation was present in 302 (59.8%) patients with the fusion phenotype, 59 (58.4%) with two-sinus phenotype, and 17 (54.8%) with partial-fusion phenotype, with no significant differences. The type of aortic dilatation (ascending, root, or extended phenotype) did not vary among BAV phenotypes.

Echocardiographic examination

Transthoracic echocardiography was performed in all 814 subjects (Table 2). Additional imaging modalities were transesophageal echocardiography carried out in 19.3% and computed tomography in 22.5% of the population. The median duration of transthoracic echocardiography was 22 minutes (IQR 15–30). Slight hypertrophy of the left ventricular walls was observed, along with minor enlargement of the left atrium. Left ventricular ejection fraction was within normal limits, median (IQR): 60 (55–65)%. However global longitudinal strain was slightly depressed — mean (SD) –16.8% (3.7). Regional wall motion abnormalities were present in 11.9% of the population.

Significant (moderate/severe) aortic regurgitation was more common than stenosis (44.1% vs. 34.2%, respectively, McNemar's test p<0.001). Significant mitral regurgitation was more common than tricuspid regurgitation (13.1% vs. 9.5%, respectively, McNemar's test P = 0.003). Data presented in Figure 4.

Aortic dilatation was observed in 513 patients (63.6%), with the most common type being the extended phenotype (root and ascending aorta) in 212 patients (26.3%), followed by the ascending aorta phenotype in 196 (24.3%) and root type in 105 (13.0%) (P <0.001) (Table 2).

No aortic dissection was registered. After a heart-team decision, surgical intervention was scheduled in 139 patients (24.0%).

DISCUSSION

This registry offers a unique, large-scale overview of Polish patients with confirmed BAV, using the latest classification of BAV valvulo-aortopathy [2]. Improved noninvasive imaging, particularly echocardiography, has increased BAV diagnoses. Bicuspid aortic valve is a valvulo-aortopathy with diverse phenotypes, symptoms, and complications. Despite numerous studies, gaps remain in understanding BAV pathophysiology and clinical significance [8]. Most registries rely on small or retrospective samples with inconsistent classifications, complicating clinical practice, outcome prediction, and valve interventions [9–11].

This registry, based on voluntary participation of tertiary cardiac centers, ensured high data quality but does not allow estimation of the absolute prevalence of BAV in Poland. It focused on patients with heart defects or those qualified for intervention, leading to a relatively high average age, symptomatic cases in over half, and moderate-to-severe defects in over one-third. Bicuspid aortic valve patients are typically diagnosed and treated about a decade earlier than those with other aortic defect etiologies.

We analyzed BAV morphology and echocardiographic parameters in a large cohort of Polish adults from hospitals and outpatient clinics. Symptoms, comorbidities, valve dysfunction, aortic dilatation, and other pathologies were assessed. Valve phenotypes were classified using Michelena et al. criteria [2]. Transthoracic echocardiography was the primary imaging method, with transesophageal echocardiography or cardiac computed tomography performed in 20% of cases. All evaluations followed standardized protocols by expert echocardiographers.

Bicuspid aortic valve can be diagnosed at any age and in various clinical contexts. In our population (average age 50 years), BAV was more common in men (72.7%), consistent with previous data [12–14]. Similarly, in a younger cohort of 1135 children and adolescents, 67% of BAV cases were male [15].

Clinical presentations and comorbidities

Bicuspid aortic valve presentation can range from benign findings to severe complications like valve dysfunction, heart failure, or aortic aneurysm [8]. In our registry, the most common symptom was shortness of breath (54.1%), while 45.2% had no exercise limitations. Chest pain (17.5%) and syncope (5.6%) were also noted.

Comorbidities in BAV patients vary with age, often linked to genetic conditions in children and common diseases in adults. In our study, hypertension (54%), dyslipidemia (43.5%), coronary artery disease (16%), diabetes (10%), and atrial fibrillation (12.5%) were the most common, reflecting the cardiovascular risk profile of the studied Polish population. Other BAV registries report lower rates of hypertension (15%–38%), dyslipidemia (14%–30%), diabetes (6%–12%), and coronary artery disease (5%–14%) [13, 16–18].

Our registry uniquely categorizes BAV into three clinical-prognostic subgroups based on the latest expert consensus [2]. Most patients (69.9%) had typical valvulo-aortopathy with valve dysfunction and aortic dilatation, requiring long-term monitoring for risks like infective endocarditis, aortic dissection, and surgery. Uncomplicated BAV (present in 19.3%) is a mild, often incidental condition with non-progressing symptoms. Complex BAV with concomitant diseases, present only in 10.8%, included aortic coarctation as the most common associated pathology. This form of valvulo-aortopathy often requires surgical intervention at an earlier stage of life. Coexisting marfanoid features were rare (1.1%), consistent with previous reports [19].

Valvulopathy

Valvular dysfunction is common in BAV, with 33% developing significant aortic valve disease [20]. Bicuspid aortic valve is thought to have an autosomal dominant inheritance linked to defective heart development genes [21]. Dysfunction varies by fusion type, with regurgitation reported in 32%–43% and stenosis in 38%–62% in prior studies [13, 22]. In our cohort, regurgitation (44%) was more common than stenosis (34%), likely due to the high prevalence of right-left cusp fusion (80.4%), which predisposes to regurgitation. Right-noncoronary cusp fusion, less frequent in our cohort, is more prone to stenosis and surgery [22]. Ethnicity may also influence these patterns, as right-left cusp fusion is more common in Europeans than Asians (44.2% vs. 26.8%; P <0.001) [17]. Understanding interethnic differences in BAV morphology and function is essential for optimizing global transcatheter aortic valve replacement strategies, as severe aortic stenosis remains the leading cause of aortic valve interventions [8, 23].

In our population, left and right ventricular dimensions were normal, but the left atrium was enlarged, and mild left ventricular hypertrophy was noted, consistent with other registries [13, 16, 18]. The peak aortic gradient was mildly elevated (21 mm Hg), and left ventricular ejection fraction was normal — mean (SD) 57.7% (10.3), which is in line with other studies [13, 18]. However, global longitudinal strain was slightly reduced — mean (SD) –16.8% (3.7), suggesting potential left ventricular overload due to altered BAV-related hemodynamics.

The study confirmed that fused-type anomalies are predominant, occurring in almost 80% of cases, though slightly lower than the 90%–95% reported in other studies, likely due to classification methods or ethnic differences [8]. Among fused BAV types, left-right coronary leaflet fusion was most common (80.4%), followed by right-noncoronary fusion, with left-noncoronary fusion being the least frequent (6.7%). The 2-sinus phenotype, present in 16% of cases, showed equal distribution of its subtypes (50% each). Patients with 2-sinus BAV type were younger and had fewer comorbidities, including lower rates of atrial fibrillation, nicotine addiction, and arterial hypertension. As noted in the Italian REBECCA registry, the 2-sinus form, though morphologically severe, is linked to fewer surgeries due to the younger population [13]. Its prevalence in our study aligns with French data [16] but is higher than in the International BAV Consortium study [8], likely reflecting ethnic and genetic diversity. Globally, the 2-sinus type is more common in Europeans, while the fusion type predominates in Asians [24]. The oldest age group was observed in fused BAV patients (average age 57), representing 4.8% of anomalies. This group had higher rates of comorbidities, including diabetes, hypertension, nicotine addiction, and the highest NT-proBNP levels, reflecting their age. Tachyarrhythmias, particularly atrial fibrillation, were more common in partial-fusion BAV, affecting 36.7% of patients. This rare subtype was identified more frequently in our registry due to advanced imaging techniques like high-resolution echocardiography and cardiac computed tomography. Previously, partial-fusion BAV was mainly recognized during valve or aortic surgeries [25–27].

Aortopathy

Bicuspid aortic valve aortopathy is common and heterogeneous, increasing the risk of aortic aneurysms and dissection. Altered hemodynamics and valve-related changes contribute to aortopathy, even in normally functioning valves, leading to asymmetric dilatation and wall stress, promoting aneurysm formation [28–30]. BAV aortopathy can occur independently of valvular dysfunction. The latest classification identifies three types of aortic dilatation [2], with

the frequency described at 70% for the ascending, 20% for the root, and 10% for the extended (10%) phenotype [31, 32].

In our cohort, aortic dilatation occurred in 64% of cases, with the extended phenotype being most frequent (26%), followed by the ascending (24%) and root (13%). In comparison, Kong et al. [17] in a study on almost 2 thousand patients reported aortic dilatation in 45% of cases, with the extended (14%) and root phenotypes (12%) predominating. Differences in prevalence likely stem from varying definitions. The root phenotype affects the root with mild ascending dilatation, while the ascending phenotype involves the ascending aorta with mild root dilation. Our study measured absolute aortic dimensions using sex-specific cut-offs, without accounting for relative dilatation.

Limitations

Our registry, based on voluntary participation of tertiary cardiac centers, ensured high data quality but is not an epidemiological study and therefore provides no data on BAV prevalence in the Polish population. It lacks patient follow-up data, which is being collected for a future publication. Information on family history, pharmacotherapy, and outpatient versus hospitalized status was not included. While dyslipidemia data were provided, information on detailed lipid distribution, which could offer insights into its role in BAV-related calcific aortic stenosis, is lacking [33].

CONCLUSIONS

The first large-scale registry of BAV using the latest classification provides insights into the morphology of BAV-associated valvular and aortic phenotypes in the adult Polish population. The most common BAV phenotype was typical valvulo-artopathy, with the most frequent valvular and aortic malformation being the right-left fusion and extended aortic dilatation (root and ascending aorta), respectively. This spectrum is similar to the findings reported in other population and ethnicities, with only slight differences in the frequency of specific variants. Our study provides a basis for further research exploring the associations between BAV phenotypes and disease presentation.

Article information

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Table 1. Clinical characteristics of patients with specific BAV phenotypes

Variable	n = 814	Fused BAV[a] n = 508	2-sinus BAV[a] n = 101	Partial-fusion BAV[a] n = 31	*P*-overall test
Age, years, mean (SD)	50.0 (17.4)	53.0 (16.8)	46.3 (15.4)	57.2 (16.8)	**<0.001**[1]
Male, n (%)	592 (72.7)	383 (75.5)	73 (72.3)	22 (71.0)	0.70
Weight, kg, mean (SD)	81.0 (16.7)	82.1 (16.5)	80.2 (17.0)	86.9 (21.4)	0.17
Height, cm, mean (SD)	173 (9.7)	174 (9.3)	173 (10.5)	173 (9.9)	0.83
BMI, kg/m^2, mean (SD)	26.8 (4.7)	27.0 (4.6)	26.5 (4.2)	28.9 (6.1)	0.06
Marfanoid features, n (%)	9 (1.1)	6 (1.2)	1 (1.0)	1 (3.4)	0.56
Heart rate, beats/min, median (IQR)	70 (65 – 77)	70 (64 – 78)	70 (65 - 77)	70 (65 – 85)	0.08
SBP, mm Hg, mean (SD)	128.0 (15.9)	129.5 (16,1)	126.8 (15,8)	124.2 (14,1)	0.09
DBP, mm Hg, mean (SD)	77.0 (11.0)	77.3 (10,7)	75.9 (13,5)	74.1 (11,7)	0.21
Symptoms					
Dyspnea at rest, n (%)	431 (54.1)	276 (55.5)	48 (48.0)	22 (71.0)	0.07
NYHA: III, IV, n (%)	68 (8.6)	53 (10.7)	5 (5.0)	5 (16.1)	0.12
Chest pain , n (%)	137 (17.5)	84 (17.4)	22 (22.4)	5 (16.7)	0.48
Syncope, n (%)	44 (5.6)	21 (4.3)	6 (6.1)	1 (3.3)	0.72
Concomitant diseases					
Coronary artery disease, n (%)	126 (16.0)	85 (17.4)	15 (15.6)	7 (22.6)	0.67
Myocardial infarction, n (%)	53 (6.7)	31 (6.3)	8 (8.2)	4 (12.9)	0.33
Stroke, n (%)	26 (3.4)	14 (2.9)	4 (4.3)	0 (0)	0.49

Ventricular arrhythmia, n (%)	60 (7.7)	39 (8.1)	3 (3.1)	4 (13.3)	0.11
Atrial fibrillation, n (%)	98 (12.5)	71 (14.7)	6 (6.2)	11 (36.7)	**<0.001[2]**
Other SVT, n (%)	85 (10.9)	50 (10.4)	12 (12.8)	7 (23.3)	0.09
Arterial hypertension, n (%)	422 (54.0)	281 (58.3)	43 (44.8)	20 (64.5)	**0.034[3]**
Dyslipidemia, n (%)	334 (43.5)	227 (48.1)	33 (35.5)	16 (53.3)	0.062
Diabetes, n (%)	80 (10.3)	58 (12.1)	5 (5.3)	6 (20.0)	0.051
Nicotine addiction, n (%)	157 (20.7)	124 (26.4)	13 (13.7)	5 (16.7)	**0.019[4]**
Neoplasm, n (%)	43 (5.5)	38 (7.9)	3 (3.1)	0 (0)	0.07
Active neoplasm, n (%)	15 (1.9)	12 (2.5)	2 (2.1)	0 (0)	0.67
NT-proBNP, pg/ml., median (IQR)	183 (58 – 871)	250 (73– 1151)	382 (169 - 1320)	1999 (435 – 4300)	**0.021[5]**
Clinical type of valvulo-aortopathy BAV disease					
Complex valvulo-aortopathy, n (%)	88 (10.8)	30 (5.9)	7 (6.9)	1 (3.2)	0.75
Typical valvulo-aortopathy, n (%)	569 (69.9)	381 (75.0)	68 (67.3)	23 (74.2)	0.28
Uncomplicated BAV, n (%)	157 (19.3)	97 (19.1)	26 (25.7)	7 (22.6)	0.30
Aortic coarctation, n (%)	56 (7.1)	13 (2.6)	4 (4.0)	0 (0)	0.48

Data presented as mean (standard deviation), median (interquartile range) or counts and percentage

[a]Group of 640 patients with specified BAV subtypes. [1]Fused BAV vs. 2-sinus BAV: P <0.001, 2-sinus vs. partial fusion: P = 0.004. [2]Fused BAV vs. 2-sinus BAV: P = 0.02, fused vs. partial fusion: P = 0.004, 2-sinus vs. partial fusion: P <0.001. [3]Fused BAV vs. 2-sinus BAV: P = 0.015, 2-sinus vs. partial fusion: P = 0.056. [4]Fused BAV vs. 2-sinus BAV: P = 0.01. [5]Fused BAV vs. partial fusion: P = 0.023, 2-sinus BAV vs. partial fusion: P = 0.12

Abbreviations: BAV, bicuspid aortic valve; BMI, body mass index; DBP, diastolic blood pressure; NTproBNP, N-terminal prohormone of brain natriuretic peptide; n, number of patients; NYHA, New York Heart Association; SBP, systolic blood pressure; SD, standard deviation; SVT, supraventricular tachycardia

Table 2. Echocardiographic data of the studied population (n = 814)

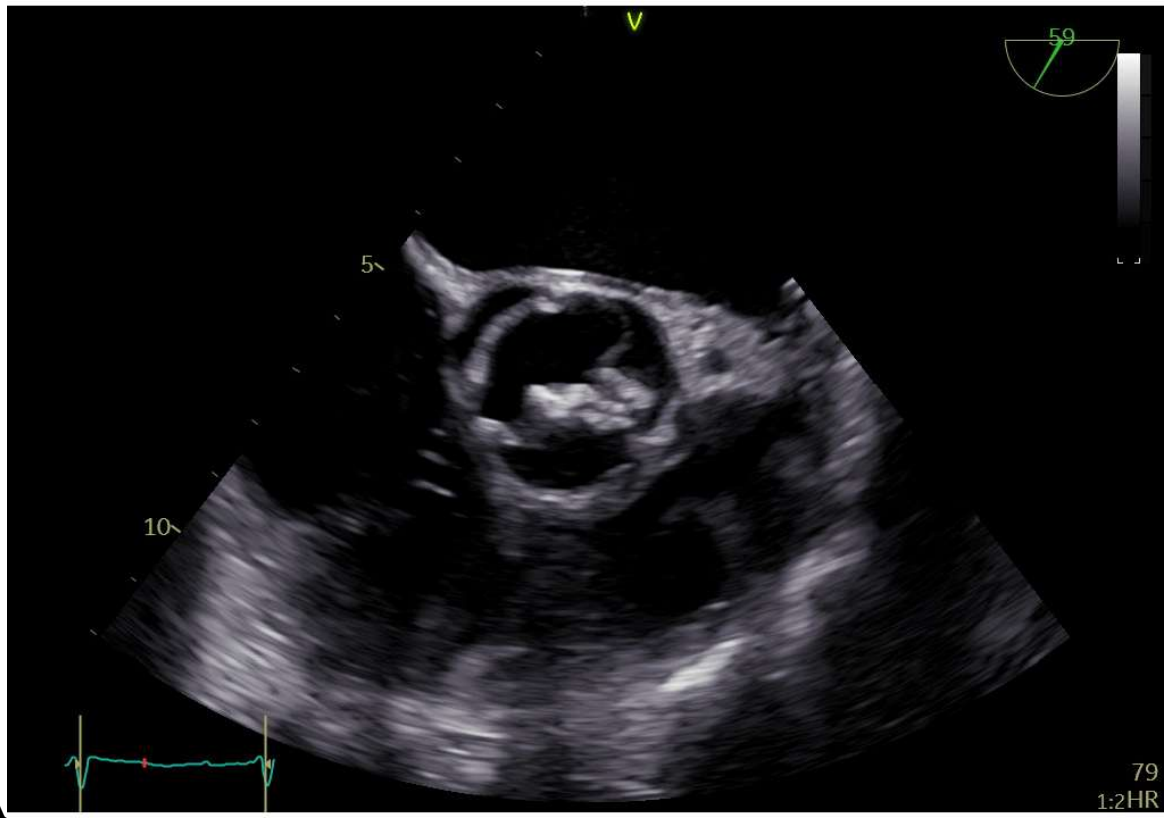
Variable	**Statistics**
Regional wall motion abnormalities, n (%)	96 (11.9)
LVESD, mm, mean (SD)	35.3 (8.6)
LVEDD, mm, mean (SD)	51.8 (8.3)
Interventricular septum, mm, mean (SD)	11.8 (2.5)
Posterior wall, mm, mean (SD)	10.7 (2.1)
Left atrium — AP dimension, mm, mean (SD)	38.5 (7.6)
Left atrial volume, ml/m^2, median (IQR)	39 (23.0–65.9)
Ejection fraction, 2D, %, median (IQR)	60 (55–65)
LVESV, ml, median (IQR)	60 (37–91)
LVEDV, ml, median (IQR)	126 (90–180)
GLS, %, mean (SD)	–16.8 (3.7)
LVOT, mm, mean (SD)	23.6 (3.4)
VTI LVOT, cm, mean (SD)	22.2 (7.4)
Tricuspid regurgitation V_{max}, m/s, mean (SD)	2.50 (0.55)
TAPSE, mm, mean (SD)	23.4 (4.3)
Aortic bulb, mm, mean (SD)	37.7 (5.9)
Sino-tubular junction, mm, mean (SD)	32.3 (5.8)
Ascending aorta, mm, mean (SD)	39.0 (7.5)
Aortic arch, mm, mean (SD)	28.8 (5.7)
Descending aorta, mm, mean (SD)	22.9 (4.2)
Aortic ring, mm, (SD)	25.2 (3.7)
Aortic dilatation, n (%)	513 (63.6)
Aortopathy form	
Ascending phenotype, n (%)	196 (24.3)
Root phenotype, n (%)	105 (13.0)
Extended phenotype, n (%)	212 (26.3)
Aortic valve V_{max}, m/s	2.6 (1.2)
Aortic valve $Grad_{max}$, mm Hg, median (IQR)	21 (11.6 – 50.0)
Aortic valve $Grad_{mean}$, mm Hg, median (IQR)	13.0 (6.5 - 34.0)

Aortic valve area, cm^2, median (IQR)	1.25 (0.9- 2.0)
CT — performance, n (%)	183 (22.5)
TEE — performance, n (%)	157 (19.3)
TTE examination time, min, median (IQR)	22 (15 – 30)
3D LVEF, n (%)	60 (7,37)

Data presented as mean (standard deviation), median (interquartile range) or counts and percentage

*Group of 640 patients with specified BAV subtypes

Abbreviations: AP, antero-posterior; CT, computed tomography; GLS, global longitudinal strain; $Grad_{max}$, maximal gradient; $Grad_{mean}$, mean gradient; LVEDD, left ventricular end-diastolic diameter; LVESD, left ventricular end-systolic diameter; LVEDV, left ventricular end-diastolic volume; LVESV, left ventricular end-systolic volume; LVOT, left ventricular outflow tract; n, number of patients; TAPSE, tricuspid annular plane systolic excursion; TEE, transoephageal echocardiography; TTE, transthoracic echocardiography; V_{max} maximal velocity; VTI, velocity time integral; 3D, 3 dimensional echocardiography



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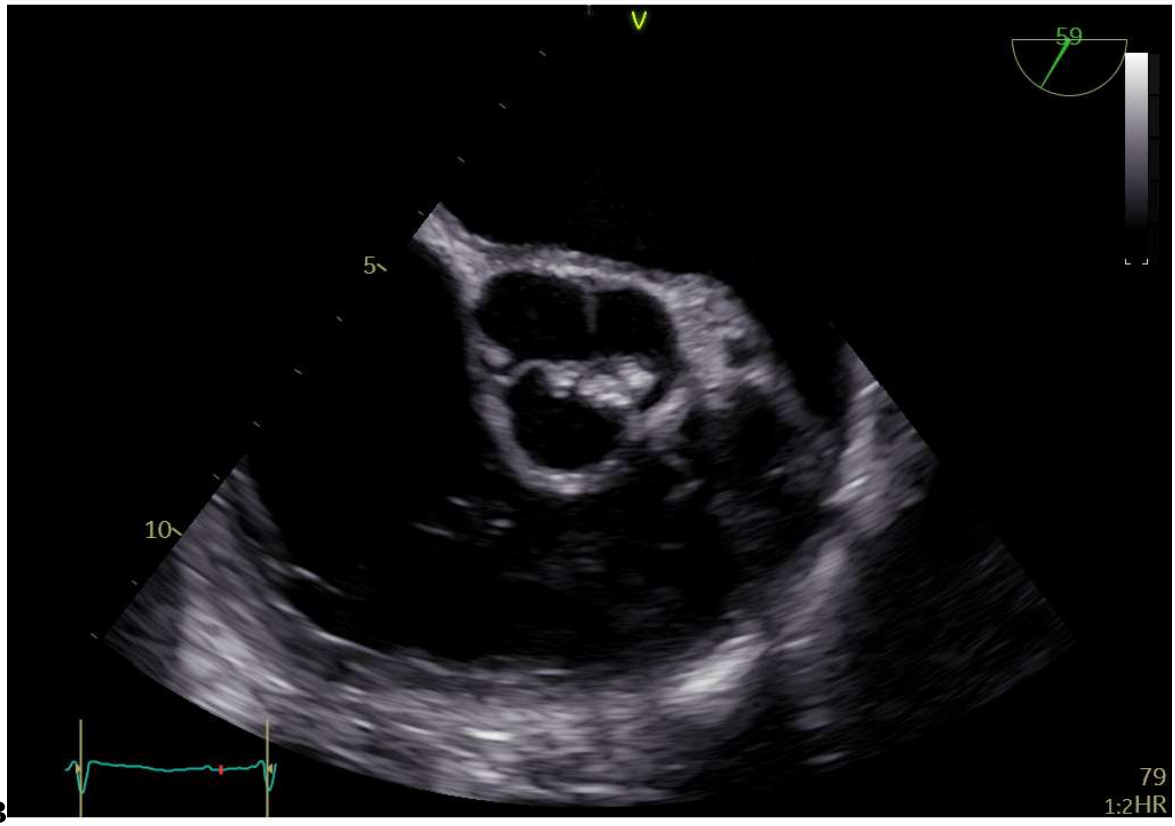

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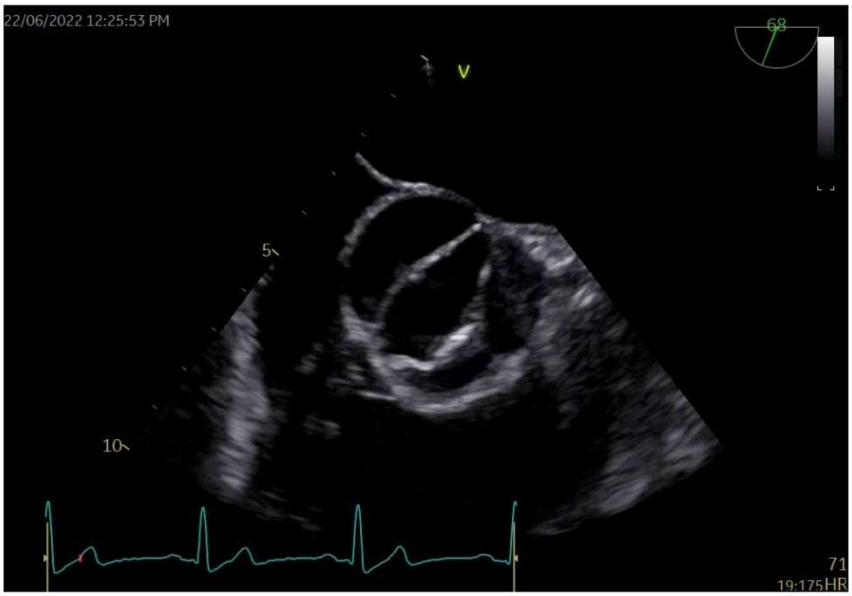

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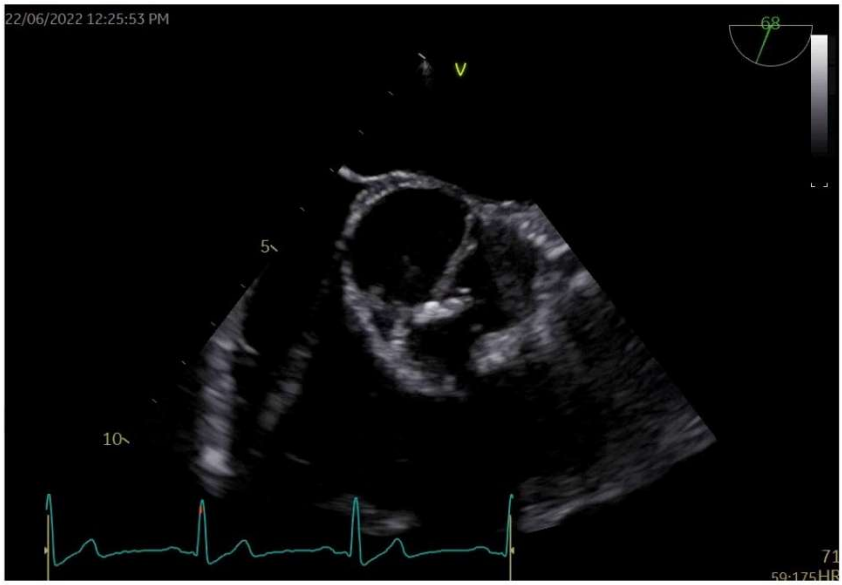
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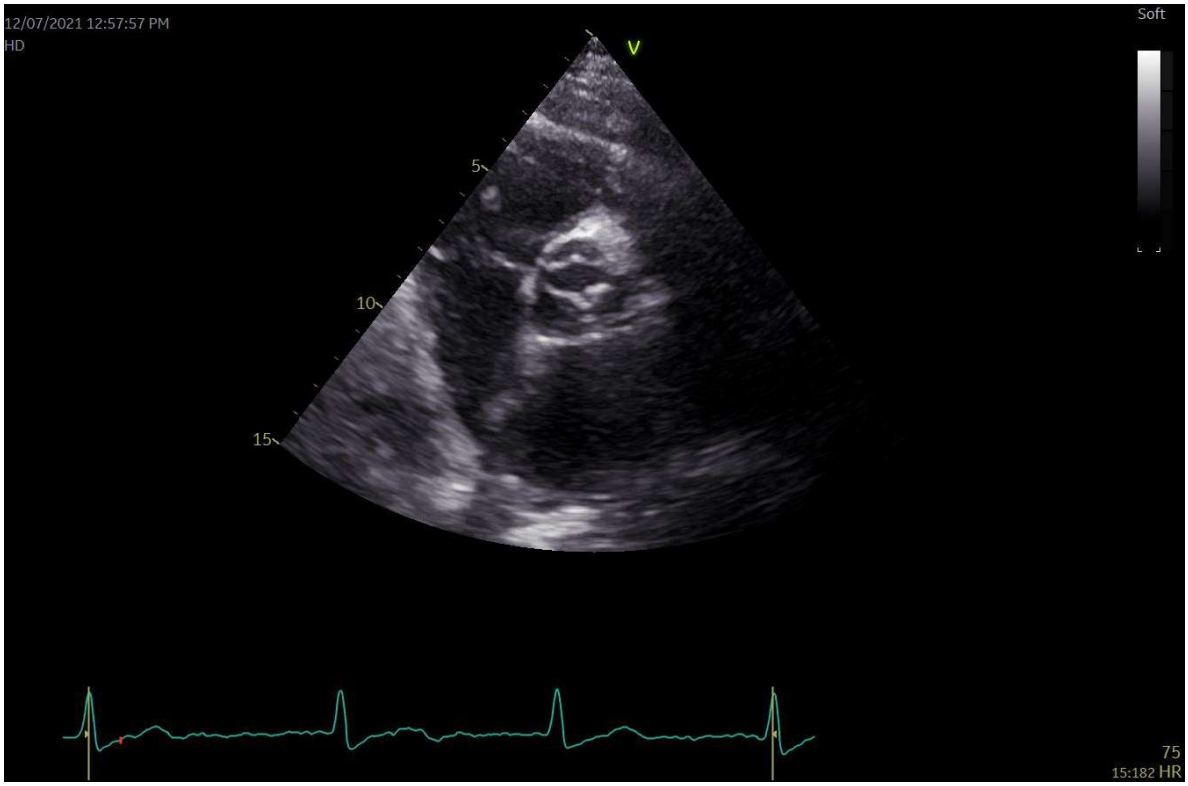
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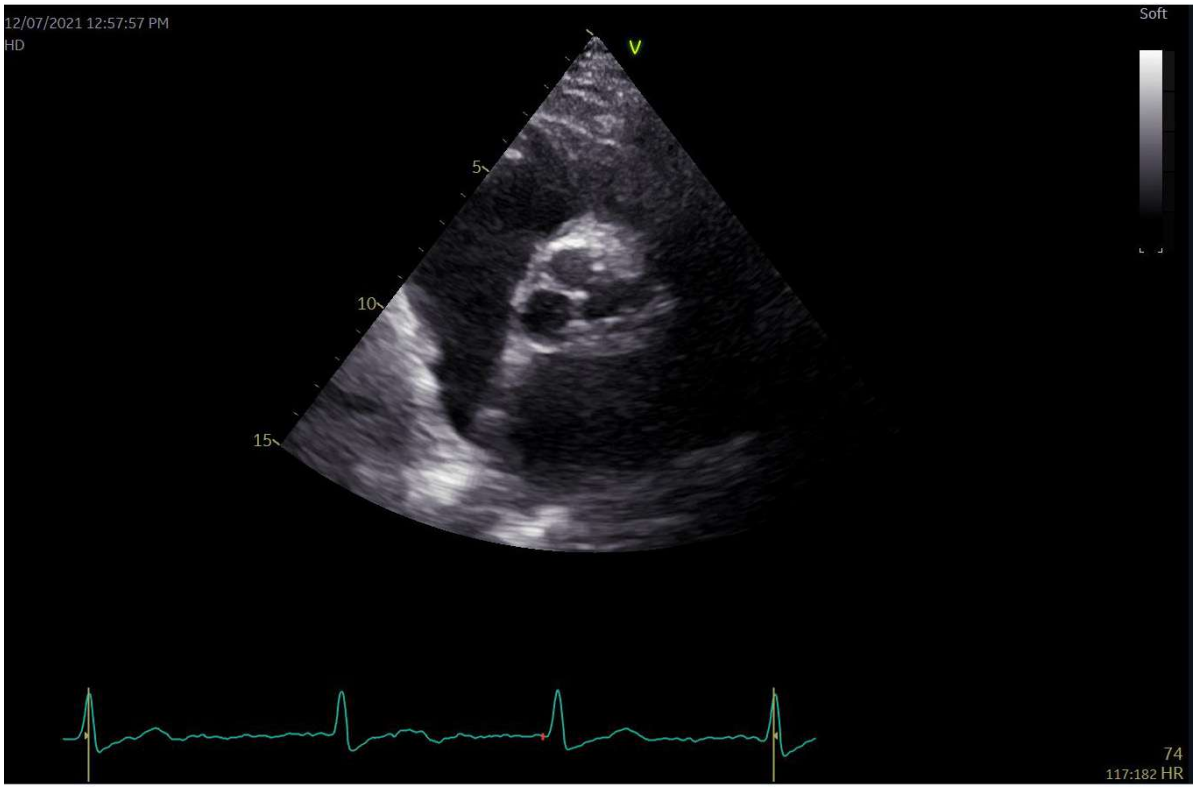

F

Figure 1. Examples of bicuspid aortic valve (BAV) phenotypes. **A.** Fused BAV: right-left aortic cusp fusion in systole; TEE. **B.** Fused BAV: right-left aortic cusp fusion in diastole; TEE. **C.** Two-sinus BAV: antero-posterior type in systole; TEE. **D.** Two-sinus BAV: antero-posterior type in diastole; TEE. **E.** Partial-fusion BAV: partial left-noncoronary cusp fusion in systole; TTE. **F.** Partial-fusion BAV: partial left-noncoronary cusp fusion in diastole; TTE

Abbreviations: TEE, transoesophageal examination; TTE, transthoracic examination

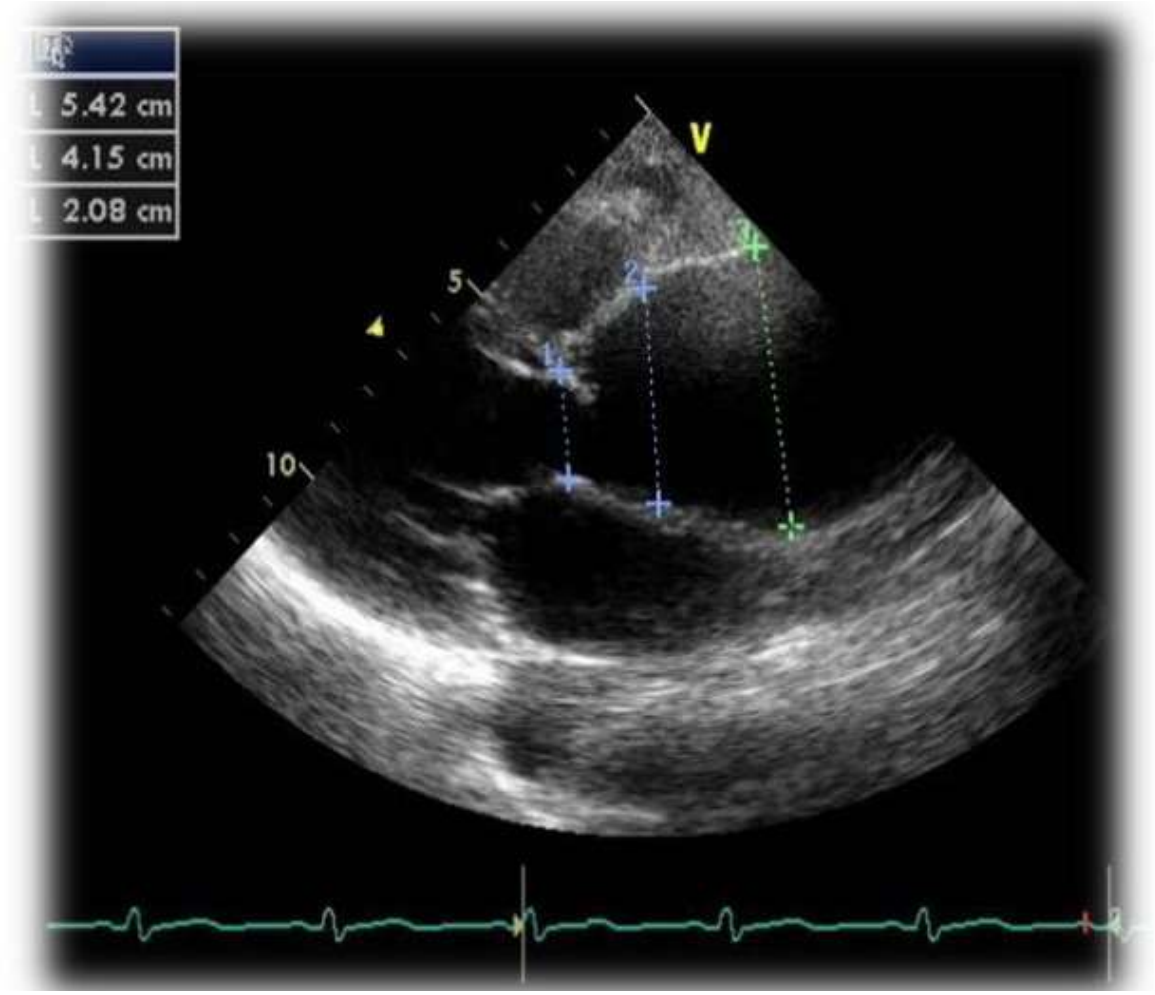

Figure 2. Transthoracic 2-dimensional parasternal long axis view of the extended phenotype of aortic dilatation. Measurements of the left ventricular outflow tract (1), aortic root (2) and ascending aorta (3). The sino-tubular junction is undistinguishable in this case

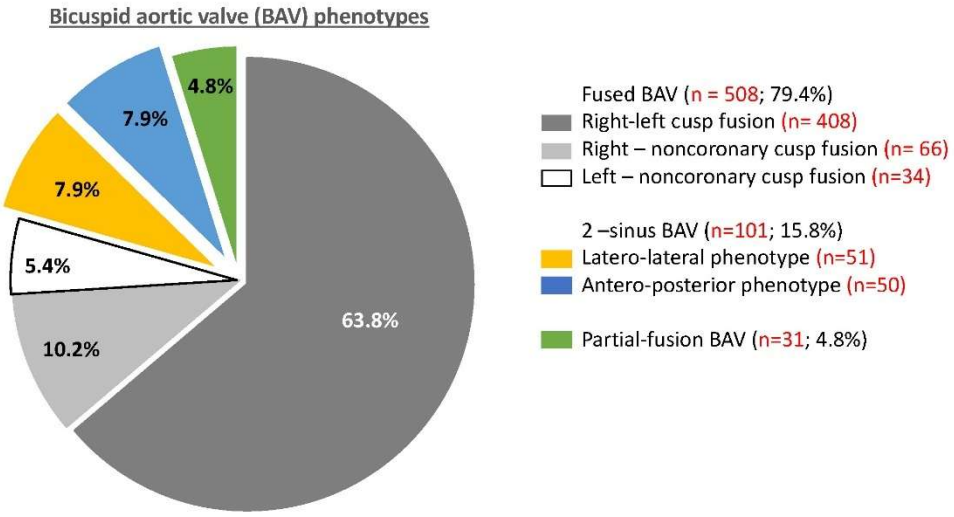

Figure 3. Schematic representation of the frequency bicuspid aortic valve (BAV) types. The graph presents the relative frequency of each BAV type in relation to the 640 patients with specified BAV types

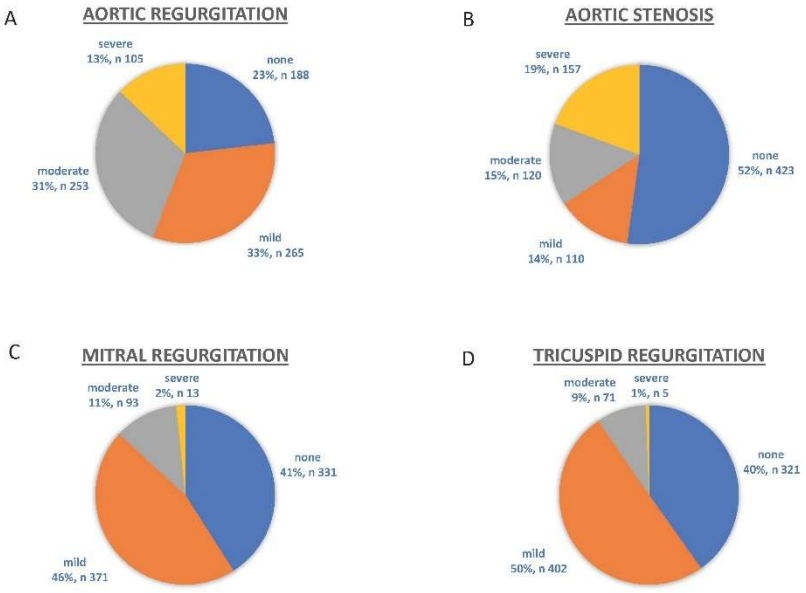

Figure 4. Schematic representation of the frequency and degree of valve diseases in the examined population. **A.** Aortic regurgitation. **B.** Aortic stenosis. **C.** Mitral regurgitation. **D.** Tricuspid regurgitation