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ATRIOVENTRICULAR SEPTAL DEFECT: ETIOLOGY, PATHOGENESIS, CLASSIFICATION, MODERN DIAGNOSTIC METHODS, AND SURGICAL CORRECTION. LITERATURE REVIEW

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Abstract

Atrioventricular septal defect (AVSD) is a complex congenital heart defect characterized by abnormal endocardial cushion formation, leading to atrial and ventricular septal defects and atrioventricular valve anomalies. AVSD accounts for approximately 7% of all congenital heart defects and is frequently associated with Down syndrome. Without surgical intervention, patients with complete AVSD are at high risk of developing pulmonary hypertension and heart failure. Modern imaging techniques allow for precise anatomical assessment and optimization of surgical treatment. This study aims to investigate the etiology, pathogenesis, classification, modern diagnostic methods, and surgical correction techniques for AVSD.A review of publications from electronic databases PubMed, MEDLINE, Web of Science, Google Scholar, and an electronic library was conducted using relevant keywords over the past 10 years. A total of 50 sources were included, while duplicate publications and conference abstracts were excluded. Genetic and epigenetic factors, particularly mutations affecting endocardial cushion development, play a significant role in AVSD formation. Impaired cell migration leads to various AVSD subtypes. Modern radiological diagnostic techniques enable accurate defect evaluation. Surgical correction, including the double-patch and single-patch techniques, remains the primary treatment. In complex cases, palliative surgical approaches are employed. Modern diagnostic and surgical methods significantly improve the prognosis of AVSD patients. Genetic research facilitates early detection, while advanced surgical techniques reduce the risk of complications. A comprehensive approach integrating etiological research, pathogenetic modeling, precise diagnosis, and surgical interventions is key to the effective management of AVSD.

Keywords: *atrioventricular septal defect, etiology, pathogenesis, imaging diagnosis, surgical correction.*

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Резюме

АТРИОВЕНТРИКУЛЯРНЫЙ СЕПТАЛЬНЫЙ ДЕФЕКТ: ЭТИОЛОГИЯ, ПАТОГЕНЕЗ, КЛАССИФИКАЦИЯ, СОВРЕМЕННЫЕ МЕТОДЫ ДИАГНОСТИКИ И ХИРУРГИЧЕСКОЙ КОРРЕКЦИИ. ОБЗОР ЛИТЕРАТУРЫ

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Атриовентрикулярный септальный дефект (AVSD) — сложный врожденный порок сердца, характеризующийся нарушением формирования эндокардиальных валиков, что приводит к дефектам межпредсердной и межжелудочковой перегородок, а также аномалиям атриовентрикулярных клапанов. AVSD составляет 7% всех врожденных пороков сердца и часто встречается при синдроме Дауна. Без хирургического вмешательства пациенты с полной формой AVSD подвержены высокому риску легочной гипертензии и сердечной недостаточности. Современные методы визуализации позволяют точно оценить анатомию порока и оптимизировать хирургическое лечение. Целью исследования является изучение этиологии, патогенеза, классификации, современных методов диагностики и хирургической коррекции атриовентрикулярного септального дефекта. Проанализированы публикации в электронных базах данных PubMed, MEDLINE, Web of Science, Google Scholar и электронной библиотеке с использованием ключевых слов за последние 10 лет. В исследование были включены 50 источник, при этом дублирующиеся публикации и материалы конференций были исключены. В развитии AVSD значительную роль играют генетические и эпигенетические факторы, особенно мутации, влияющие на формирование эндокардиальных валиков. Нарушение миграции клеток приводит к различным формам AVSD. Современные методы лучевой диагностики позволяют точно оценить анатомию дефекта. Хирургическая коррекция, включая технику двойного и одинарного патча, является основным методом лечения. В сложных случаях применяется паллиативная хирургия. Современные диагностические и хирургические методы улучшают прогноз пациентов с AVSD. Генетические исследования способствуют раннему выявлению, а современные хирургические технологии снижают риск осложнений. Комплексный подход является ключевым в эффективном лечении AVSD.

Ключевые слова: *атриовентрикулярный септальный дефект, этиология, патогенез, визуальная диагностика, хирургическая коррекция.*

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Түйіндеме

АТРИОВЕНТРИКУЛЯРЛЫҚ СЕПТАЛЬДЫ АҚАУ: ЭТИОЛОГИЯСЫ, ПАТОГЕНЕЗІ, ЖІКТЕЛУІ, ЗАМАНАУИ ДИАГНОСТИКАСЫ ЖӘНЕ ХИРУРГИЯЛЫҚ ТҮЗЕТУ ӘДІСТЕРІ. ӘДЕБИЕТТІК ШОЛУ.

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Атриовентрикулярлық септальды ақау (AVSD) – эндокардиальды жастықшалардың дұрыс қалыптаспауынан жүрек аралық және қарынша аралық перделердің ақаулары мен атриовентрикулярлық қақпақшалардың аномалиялары дамитын күрделі туа біткен жүрек ақауы. AVSD барлық туа біткен жүрек ақауларының 7%-ын құрайды және көбінесе Даун синдромы бар науқастарда кездеседі. Хирургиялық араласусыз толық AVSD бар науқастар өкпе гипертензиясы мен жүрек жеткіліксіздігінің жоғары қаупіне ұшырайды. Заманауи визуализация

әдістері ақаудың анатомиясын дәл бағалауға және хирургиялық емдеуді оңтайландыруға мүмкіндік береді. Бұл зерттеудің мақсаты – AVSD-нің этиологиясын, патогенезін, жіктелуін, заманауи диагностика әдістері мен хирургиялық түзету тәсілдерін зерттеу. PubMed, MEDLINE, Web of Science, Google Scholar электрондық дерекқорларындағы соңғы 10 жылдағы жарияланымдарға шолу жасалды. Жалпы 50 дереккөз қамтылды, қайталанған басылымдар мен конференция материалдары алынып тасталды.

AVSD қалыптасуында генетикалық және эпигенетикалық факторлар, әсіресе эндокардиальды жастықшалардың дамуына әсер ететін мутациялар, маңызды рөл атқарады. Эмбрионалды жасушалардың миграциясының бұзылуы ақаудың әртүрлі түрлерінің дамуына әкеледі. Заманауи радиологиялық диагностика әдістері ақаудың анатомиялық ерекшеліктерін дәл анықтауға мүмкіндік береді. Қос патч және бір патч әдістерін қамтитын хирургиялық түзету – AVSD емдеудің негізгі тәсілі болып табылады. Күрделі жағдайларда паллиативті хирургия қолданылады. Заманауи диагностикалық және хирургиялық әдістер AVSD бар науқастардың болжамын едәуір жақсартады. Генетикалық зерттеулер ақаудың ерте анықталуына ықпал етеді, ал жетілдірілген хирургиялық технологиялар асқыну қаупін төмендетеді. Этиологиялық зерттеу, патогенезді модельдеу, дәл диагностика және хирургиялық араласу әдістерін біріктіретін кешенді көзқарас AVSD басқарудағы негізгі стратегия болып табылады.

Түйінді сөздер: *атриовентрикулярлық септальды ақау, этиология, патогенез, визуалды диагностика, хирургиялық түзету.*

Дәйексөз үшін:

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Introduction

Atrioventricular septal defect (AVSD) is a significant congenital heart anomaly that arises from improper development of the endocardial cushions during embryogenesis. It is characterized by a common atrioventricular junction, defects in the atrial and ventricular septa, and abnormalities in the atrioventricular valves. AVSD is classified into partial, intermediate, and complete forms based on the extent of septal involvement and the structural abnormalities of the valves. This condition represents approximately 7% of all congenital heart defects and has a notably higher prevalence in individuals with Down syndrome, accounting for 40–50% of cases in this population [34, 32, 10, 31, 29, 13].

Clinically, AVSD poses significant challenges due to its impact on hemodynamics. Left-to-right shunting of blood through septal defects, atrioventricular valve regurgitation, and increased pulmonary blood flow contribute to progressive heart failure and pulmonary hypertension if untreated. Moreover, the spectrum of clinical manifestations varies widely, ranging from asymptomatic cases to severe presentations in infancy [32, 25, 43, 13, 16].

Radiological imaging plays a crucial role in diagnosing and evaluating AVSD. Echocardiography remains the first-line modality, providing detailed visualization of the common atrioventricular junction and associated anomalies. However, advances in cardiac computed tomography (CT) have enhanced diagnostic precision, offering high-resolution, three-dimensional reconstructions for complex cases. Notable radiological features, such as the "gooseneck sign," serve as key indicators in imaging studies [24, 43, 19, 3, 7].

Surgical correction is the cornerstone of AVSD management, with early intervention recommended to prevent irreversible pulmonary vascular disease. Techniques such as double-patch and modified single-patch repairs are commonly employed, each with distinct advantages. Recent studies have demonstrated comparable long-term survival and event-free outcomes between these techniques, underscoring the importance of individualized surgical planning [9, 23, 4, 20].

Despite advancements in treatment, challenges persist, including postoperative complications such as atrioventricular valve regurgitation and arrhythmias. The development of minimally invasive approaches and refined imaging technologies continues to improve outcomes and expand treatment options for AVSD [1, 2, 44, 30, 12, 20].

Given the significant clinical burden and evolving management strategies, this review aims to provide a comprehensive synthesis of current knowledge on AVSD. Emphasis is placed on the role of advanced imaging modalities in diagnosis, the outcomes of various surgical techniques, and emerging therapeutic approaches to optimize care for affected individuals.

The purpose of this review was to explore the etiology, pathogenesis, methods for diagnosing atrioventricular septal defect (AVSD), focusing on improving screening sensitivity and diagnostic accuracy, as well as optimizing surgical correction strategies to enhance patient outcomes.

Search strategy. A review of publications from electronic databases PubMed, MEDLINE, Web of Science, Google Scholar, and an electronic library was conducted using relevant keywords over the past 10 years. A total of 51 sources were included, while duplicate publications and conference abstracts were excluded.

Discussion

Epidemiology of Atrioventricular Septal Defect (AVSD)

1. Prevalence of AVSD

AVSD is one of the most common forms of congenital heart defects, accounting for approximately 7% of all congenital heart anomalies. The global prevalence is around 2 per 10,000 live births, emphasizing the

significance of this condition in pediatric cardiology [34, 50, 8, 48, 16, 4]. A particularly high prevalence is observed in patients with Down syndrome, where AVSD is detected in 40–50% of cases, making this defect a key marker in this genetic condition [13].

2. Global Variability

A meta-analysis of data from 1970 to 2017 revealed significant geographic heterogeneity in the prevalence of congenital heart defects, including AVSD. According to studies conducted in various regions, Asia demonstrates the highest incidence rate—9,342 cases per 1,000 live births, surpassing Europe and North America, where diagnostic and registration practices are more comprehensive. In Africa, however, the lowest prevalence rates have been recorded—2,315 cases per 1,000, likely due to limited diagnostic capabilities and restricted access to healthcare services [50, 41, 5, 28, 33].

3. Gender Differences

According to epidemiological studies, AVSD occurs at a similar frequency in males and females, except for certain subtypes of the defect, where minor gender differences have been noted. However, these data require further confirmation in larger study populations [13].

4. Age and Clinical Presentation

AVSD is most commonly diagnosed within the first few months of life due to severe hemodynamic disturbances. In adult patients, the defect is rarely detected, usually in the form of partial AVSD, such as ostium primum defects or isolated valvular abnormalities [37].

Without surgical intervention, children with AVSD face a high risk of developing pulmonary hypertension, heart failure, and early mortality [12, 4].

5. Risk Factors and Associations

● **Genetic Factors**: Down syndrome remains the most significant association with AVSD, increasing the likelihood of this defect by 200 times compared to the general population [34, 50, 24, 15, 13, 4].

● **Teratogenic Factors**: Exposure to teratogens, including maternal diabetes and intrauterine alcohol exposure, has also been linked to an increased risk of AVSD [34, 50, 24, 15, 33].

● **Regional Characteristics**: The availability of medical care and early diagnosis significantly influence the detection rate of the defect, particularly in developing countries [34, 50, 24, 15, 27, 33].

6. Trends in Incidence Over Time

Meta-analysis data indicate an increase in diagnosed cases of AVSD from 1995 to 2017. This trend is attributed to advancements in imaging techniques, such as echocardiography and CT, and the earlier detection of less severe forms of the defect, including partial AVSD [35, 42, 33, 3].

Conversely, the incidence of severe cases, such as left ventricular outflow tract obstructions, has decreased, likely due to improvements in prenatal diagnostics and medical decision-making, including termination of pregnancy in cases of severe fetal anomalies [45, 17, 33, 12].

7. Importance of Epidemiological Studies

Epidemiological data play a crucial role in planning healthcare services and resource allocation for the treatment of AVSD. Improving access to modern diagnostic technologies in developing countries could significantly impact morbidity and mortality rates associated with this defect [34, 50, 24, 15, 33].

Pathogenesis of Atrioventricular Septal Defect (AVSD)

1. Key Mechanisms of Pathogenesis

AVSD develops due to abnormal embryonic formation of endocardial cushions, which play a crucial role in separating the common atrioventricular canal into two distinct atrioventricular orifices. These cushions contribute to the development of the atrioventricular valves, atrial septum, and ventricular septum. Disruptions in this process lead to various anomalies, including:

● The formation of a common atrioventricular junction.

● The presence of atrial and/or ventricular septal defects.

● Valve anomalies, including fusion, cleft formation, or underdevelopment of valve leaflets 15, 31, 29, 13].

2. Role of Endocardial Cushions

Endocardial cushions are mesenchymal structures that form due to the epithelial-mesenchymal transition (EMT) of endocardial cells. These cells migrate, proliferate, and fuse to create the structural framework of the heart. Disruptions at any stage of this process - such as deficient migration or improper differentiation - result in the characteristic malformations observed in AVSD [42, 31, 29].

3. Embryonic Development of the Heart

● **Weeks 4–5**: Formation of the primary heart tube, division of the atria and ventricles, and emergence of endocardial cushions.

● **Weeks 6–7**: Fusion of the endocardial cushions and initiation of atrioventricular septation.

● **Weeks 7–8**: Completion of atrial and ventricular septation and formation of the atrioventricular valves [29, 12].

In AVSD, the fusion of cushions and septation process is impaired, leading to:

● Deficiency of mesenchymal cells.

● Misalignment of valve leaflets.

● Formation of a common valve instead of two distinct mitral and tricuspid valves.

4. Genetic Factors

● Chromosomal Abnormalities: The most significant risk factor for AVSD is trisomy 21 (Down syndrome). Patients with Down syndrome exhibit defects in signaling pathways, such as the Notch pathway, which is crucial for endocardial cushion development [13].

● Genetic Mutations: Mutations in genes such as GATA4, NKX2-5, and CRELD1 contribute to the pathogenesis of AVSD. These genes regulate cell migration, proliferation, and apoptosis during heart development [31, 16].

5. Hemodynamic Abnormalities

● Blood Shunting: In the presence of atrial and/or ventricular septal defects, blood is shunted from the left heart chambers to the right, causing pulmonary overcirculation and increasing the risk of pulmonary hypertension [13, 12].

● Valve Dysfunction: Insufficiency of the common atrioventricular valve leads to regurgitation and volume overload of the ventricles, contributing to the progression of heart failure [37, 20].

6. Role of Radiological Findings in Understanding Pathogenesis

Radiological techniques, such as CT and echocardiography, allow visualization of key pathogenetic features of AVSD, including:

● The common valve and its abnormal orientation.

● Atrial and ventricular septal defects.

● Right heart hypertrophy due to volume overload [3, 7].

7. Influence of Environmental and Epigenetic Factors

● Maternal Factors: Conditions such as maternal diabetes, intrauterine alcohol exposure, and certain medications (e.g., lithium) increase the risk of AVSD.

● Epigenetic Changes: DNA methylation abnormalities and microRNA dysregulation affect the expression of genes involved in endocardial cushion development [33, 12].

8. Future Perspectives in AVSD Pathogenesis Research

Current research is focused on:

● Identifying novel genes and molecular pathways involved in endocardial cushion development.

● Exploring regenerative medicine approaches, such as stem cell therapy, to correct defects at the embryonic development stage 16, 4].

Modern Radiological Methods for Diagnosing Atrioventricular Septal Defect (AVSD)

Radiological diagnosis of atrioventricular septal defect (AVSD) plays a key role in preoperative planning, evaluating the hemodynamic consequences of the defect, and monitoring the patient's postoperative condition. Modern imaging techniques include echocardiography (ECHO), computed tomography (CT), magnetic resonance imaging (MRI), and angiography, each of which offers specific capabilities for detailed assessment of heart anatomy.

Echocardiography in AVSD Diagnosis

Echocardiography remains the primary diagnostic method for AVSD, providing real-time anatomical and hemodynamic information. Modern echocardiographic techniques include two-dimensional (2D), three-dimensional (3D) echocardiography, and prenatal fetal echocardiography.

● Two-dimensional echocardiography (2D-ECHO) helps identify the common atrioventricular valve, assess the size of atrial and ventricular septal defects, and determine the degree of regurgitation. The study by *Singh P. et al.* (2010) demonstrated that 2D-ECHO has high accuracy in visualizing valvular structures, particularly in young children [2, 27, 11, 40, 44, 46].

● Three-dimensional echocardiography (3D-ECHO) provides more detailed volumetric images, which are particularly useful for evaluating the atrioventricular valve and planning surgical intervention. According to Jegatheeswaran A. et al. (2010), 3D-ECHO significantly improves the diagnosis of valve leaflet clefts and residual defects after surgery [26].

● Transthoracic echocardiography (TTE) is the first-line diagnostic tool for AVSD suspicion. *Atik E. et al.* (2019) reported that TTE is effective in detecting primary atrial septal defects, valvular anomalies, and the degree of atrioventricular valve regurgitation [6, 12].

● Transesophageal echocardiography (TEE) allows for a more detailed visualization of valvular anatomy and adjacent structures, especially in complex cases. *Wang G. et al.* (2020) highlighted that 3D-TEE enhances the assessment of valve anatomy and function, which is critical for preoperative planning [12, 20, 47].

● Prenatal echocardiography, particularly the FINE (Fetal Intelligent Navigation Echocardiography) method, has demonstrated high efficacy in detecting AVSD in fetuses. *Veronese P. et al.* (2023) noted that this technique allows for highly accurate automatic visualization of standard cardiac views and congenital defects [46].

Computed Tomography in AVSD Diagnosis

Computed tomography (CT) plays a crucial role in diagnosing complex anatomical variants of AVSD. Unlike echocardiography, CT provides high-resolution three-dimensional images, which are particularly useful for preoperative surgical planning.

● Multislice CT (MSCT) is used for detailed assessment of septal defects, atrioventricular valves, and major vessels. The study by *Yoshitake S. et al.* (2020) confirmed that phase-contrast CT accurately determines the location of the atrioventricular conduction system, reducing the risk of conduction system injury during surgery [1, 36, 40, 49].

● The study by *Balpande N. et al.* (2022) demonstrated that CT can detect the "gooseneck sign" characteristic of AVSD, a feature previously identified only in angiographic studies. This finding underscores the importance of CT imaging in visualizing endocardial cushion defects [1, 2, 3, 7, 9, 23].

● *Nunes M. et al.* (2021) found that modern low-dose CT protocols minimize radiation exposure while maintaining high image quality. The authors emphasized that CT is particularly useful for assessing the anatomy of major vessels, determining defect size, and clarifying valve apparatus status [7, 9,13, 28].

● High-speed dual spiral CT reduces radiation exposure, making it the preferred imaging modality for children. *Goo H.W. et al.* (2018) demonstrated that ECG-synchronized CT enhances the accuracy of coronary vessel imaging and minimizes motion artifacts [22, 36, 38].

Magnetic Resonance Imaging in AVSD Diagnosis

Cardiac MRI is an indispensable diagnostic tool for AVSD, especially when echocardiographic visualization is limited.

● The study by *Elders B. et al.* (2019) demonstrated that 4D flow MRI allows for a detailed assessment of hemodynamic parameters, including wall shear stress of the aorta, which can change in AVSD patients after surgical correction. These findings highlight the importance of MRI in long-term patient monitoring [6, 16, 27, 48].

● *Schumacher K. et al.* (2023) showed that one of the key postoperative complications of AVSD repair is residual atrioventricular valve regurgitation, making MRI the preferred method for long-term monitoring of such patients [27, 38, 49].

● Contrast-enhanced MRI allows for the detection of myocardial fibrosis, which is crucial for prognosis in AVSD patients. Alsoufi B. et al. (2019) also noted that MRI plays a critical role in evaluating residual shunts and valvular dysfunctions following AVSD surgical correction [4, 16, 27].

Angiography and Cardiac Catheterization

Cardiac catheterization and angiography are used in complex anatomical forms of AVSD, particularly when

precise measurements of pulmonary artery pressure and cardiac chamber pressures are required.

● *Nicolay S. et al.* (2015) emphasized that angiography remains the "gold standard" for intracardiac shunt assessment and is crucial for determining pulmonary vascular resistance before surgery [23, 30, 33].

● The study by Deri & English (2018) confirmed that angiography is an essential tool for evaluating pulmonary vascular resistance before surgery, particularly in cases with high pulmonary hypertension risk [30, 18].

● Modern non-invasive techniques, such as CT angiography (CTA), provide high-quality cardiovascular imaging without the need for invasive catheterization. The study by *Chen S.J. et al.* (2017) compared ECG-synchronized CTA with traditional angiography, demonstrating that CTA offers high diagnostic accuracy with reduced radiation exposure [30, 36, 48].

Modern radiological methods significantly enhance the diagnostic accuracy, surgical planning, and postoperative monitoring of AVSD:

● Echocardiography (2D, 3D, TEE, fetal) remains the primary diagnostic tool, offering real-time hemodynamic and structural assessment.

● CT imaging (MSCT, phase-contrast, ECG-synchronized) is crucial for detailed anatomical assessment and preoperative planning.

● MRI is essential for functional evaluation, long-term postoperative monitoring, and fibrosis detection.

● Angiography and cardiac catheterization remain vital in complex cases, particularly for hemodynamic assessment and surgical decision-making.

Advances in imaging techniques have improved AVSD detection rates, reduced the need for invasive procedures, and contributed to better surgical outcomes and long-term patient management.

Table 1.

Comparative analysis of methods.

Method	Advantages	Limitations
Echocardiography (2D, 3D, TEE, fetal)	Accessibility, non-invasiveness, real-time hemodynamic assessment	Limited image quality in obese patients or those with post-surgical changes
Computed Tomography (MSCT, CTA)	High spatial resolution, ability to generate 3D reconstructions	Radiation exposure, need for contrast administration
MRI (4D flow, contrast-enhanced)	High tissue contrast, functional assessment capabilities	Long scan duration, contraindications for patients with metallic implants
Angiography	Gold standard for shunt evaluation, direct hemodynamic measurement possible	Invasiveness, risk of complications

Each radiological diagnostic method for AVSD has its own specific characteristics. The use of modern radiological techniques significantly improves the detection of this pathology, facilitates surgical planning, and ensures postoperative patient monitoring. Echocardiography remains the primary diagnostic method, while CT and MRI play a crucial role in refining anatomical details, functional assessment, and predicting surgical outcomes. Angiography continues to be used in complex cases and prior to surgical intervention. The combination of these methods provides a comprehensive approach to AVSD diagnosis and treatment.

Recent studies (*Jegatheeswaran A. et al., Singh P. et al., Yoshitake S. et al., Schumacher et al.)* confirm that a multidisciplinary approach to radiological AVSD diagnosis significantly increases diagnostic accuracy and improves patient prognosis [18, 49, 22, 26, 46, 40].

Classification of Atrioventricular Septal Defect (AVSD)

Atrioventricular septal defects (AVSD) represent a heterogeneous group of congenital heart defects that vary in anatomical configuration, presence, and severity of septal and valvular abnormalities. Based on anatomical characteristics, AVSD can be classified into complete, partial, and intermediate forms.

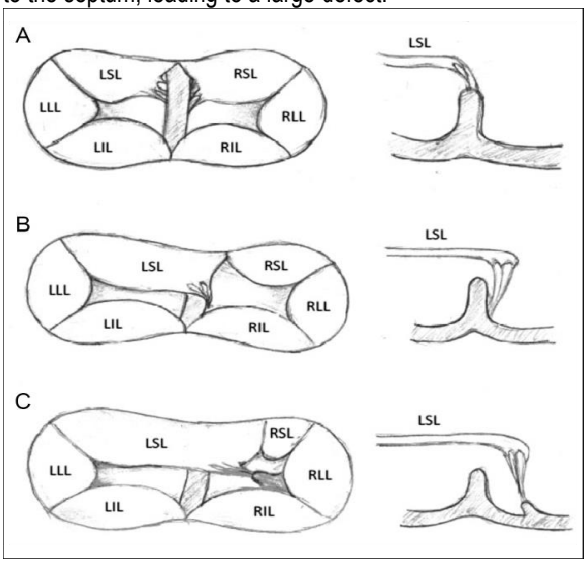
Rastelli Classification

The Rastelli classification is based on the attachment of the superior bridging leaflet to the interventricular septum and distinguishes three types of AVSD:

● Type A – The bridging leaflet attaches to the crest of the interventricular septum, creating a minimal defect.

● Type B – The leaflet extends across the interventricular septum and attaches to an abnormal papillary muscle structure in the right ventricle.

● Type C – The leaflet remains free and does not attach to the septum, leading to a large defect.



This classification was first detailed in the study by *Singh P. et al.* (2010) in the journal *Echocardiography*, highlighting its importance in preoperative planning and patient prognosis [40, 46].

Balanced and Unbalanced Forms of AVSD

AVSD can also be classified based on the balance between the right and left ventricles. This classification is particularly important for determining surgical strategy.

● Balanced AVSD (bAVSD) – The common atrioventricular (AV) valve is positioned to allow relatively equal ventricular filling.

● Unbalanced AVSD (uAVSD) – The septum is shifted toward one ventricle, causing hypoplasia of the affected ventricle and hemodynamic disturbances.

The study by Jegatheeswaran A. et al. (2010) demonstrated that unbalanced forms of AVSD significantly increase the risk of surgical intervention and are associated with higher mortality [26].

Classification Based on Blood Shunting Levels

Another classification method for AVSD is based on the level of pathological blood shunting:

● Atrial-level defect (ostium primum defect) – Blood shunts from the left atrium to the right atrium.

● Ventricular-level defect – Blood shunts through a hole in the interventricular septum.

● Combined defect – Shunting occurs at both the atrial and ventricular levels.

This classification is used to clarify the clinical presentation of AVSD and guide treatment strategies [47].

Anatomical Variations

There are also anatomical subtypes of AVSD that may be associated with other congenital anomalies, such as hypoplastic left heart syndrome, tetralogy of Fallot, and transposition of the great arteries. These features must be considered when selecting a surgical strategy, as highlighted in the study by *Jegatheeswaran A. et al.* (2010).

Thus, the classification of AVSD plays a crucial role in diagnosis, prognosis, and the selection of surgical tactics. Based on echocardiography, CT, and MRI data, the most optimal approach for each patient can be determined.

Clinical Presentation of Atrioventricular Septal Defect

The clinical presentation of atrioventricular septal defect (AVSD) varies depending on the type of defect, degree of blood shunting, functional state of the valves, and presence of associated anomalies. In newborns and young children, AVSD most commonly presents with heart failure symptoms due to significant left-to-right shunting and pulmonary volume overload. According to Rigby M. (2021), these symptoms include tachypnea, poor weight gain, feeding difficulties, and pronounced fatigue with minimal activity [29, 37].

Patients with complete AVSD exhibit more severe symptoms due to substantial volume overload and early development of pulmonary hypertension. Clinicians frequently observe cardiomegaly on physical examination, which is confirmed by chest radiography findings, as reported by *Almarzooq Z. et al.* (2019) [3, 37]. Additionally, auscultation may reveal a systolic murmur caused by regurgitation through the common atrioventricular valve.

In contrast, partial AVSD may remain asymptomatic during childhood and be diagnosed only in adulthood, particularly upon the development of pulmonary hypertension or progressive valvular dysfunction. *Balpande N. et al.* (2022) emphasize that such patients may lack significant symptoms in early stages, but later develop exertional dyspnea, arrhythmias, and signs of right ventricular failure [3, 7].

The clinical course of AVSD is closely linked to the presence of associated syndromes. In patients with Down syndrome, AVSD is detected in 40–50% of cases, and the condition is often complicated by concurrent cardiac and extracardiac anomalies. *Hua Z. et al.* (2020) note that these patients have a higher incidence of progressive pulmonary hypertension, which is attributed to abnormal pulmonary vascular development [47].

Surgical Techniques for the Correction of Atrioventricular Septal Defect

Surgical correction of atrioventricular septal defect (AVSD) is the standard treatment aimed at eliminating anatomical defects, restoring normal hemodynamics, and preventing the progression of pulmonary hypertension. The choice of surgical technique depends on the type of AVSD, the patient's age, defect size, and associated pathologies. The primary approach is biventricular repair, which involves closure of atrial and ventricular septal defects as well as reconstruction of the atrioventricular valve. According to *Fong L.S. et al.* (2019), two main techniques are most commonly used: double patch repair (Double Patch) and modified single patch repair (Modified Single Patch) [4,20].

The double patch technique involves the use of two separate patches to close the atrial and ventricular septal defects, ensuring precise anatomical reconstruction of the septa and valves. This approach is preferred for patients with large defects and complex valve anatomy. The study by *Wang G. et al.* (2020) emphasizes that double patch repair provides long-term structural stability and minimizes the risk of residual defects [20,47].

In contrast, the modified single patch technique involves using one patch to close both the atrial and ventricular septal defects. This method is less invasive and is associated with reduced cardiopulmonary bypass time, making it preferable for infants and high-risk surgical patients. However, as noted by *Alsoufi B. et al.* (2020), the single patch technique may be less effective for large defects and cases with significant valvular insufficiency [4, 16].

For patients with a functionally single ventricle, where complete repair is not feasible, palliative surgical strategies are implemented. These cases require a staged approach, including shunt procedures (e.g., Blalock-Taussig shunt) to improve pulmonary blood flow and the Fontan procedure, which directs venous blood flow to the pulmonary arteries, bypassing the right heart. The study by *Nunes M. et al.* (2021) highlights that staged palliative procedures remain a viable option for patients with severe morphological abnormalities that preclude biventricular repair [7, 13].

Postoperative follow-up is critical for evaluating the effectiveness of surgical repair and detecting complications. The most common postoperative issues include residual septal defects, atrioventricular valve regurgitation, and arrhythmias. These complications often require additional surgical or medical management. According to *Balpande N. et al.* (2022), modern imaging techniques, such as 3D echocardiography and CT, allow for early detection and intervention in postoperative complications [39, 3].

Surgical treatment of AVSD has significantly reduced mortality and improved the quality of life for patients. However, long-term outcomes depend on the accuracy of preoperative diagnostics, the choice of surgical technique, and the effectiveness of postoperative monitoring. Advancements in surgical techniques and anesthetic

management continue to improve the prognosis for patients with this complex congenital heart defect.

Conclusions

Atrioventricular septal defect (AVSD) represents one of the most significant groups of congenital heart defects, characterized by complex anatomy and variability in clinical presentation. Modern data highlight its high prevalence in patients with Down syndrome and its substantial contribution to the overall structure of congenital cardiac anomalies. The diversity of AVSD forms, ranging from partial to complete defects, requires precise diagnosis and an individualized treatment approach. Imaging modalities, including echocardiography, computed tomography, and magnetic resonance imaging, serve as essential tools for assessing the anatomical and functional characteristics of the defect. Echocardiography remains the gold standard for diagnosis, while CT and MRI provide additional data for preoperative planning and postoperative monitoring. The clinical presentation of AVSD varies from an asymptomatic course to severe manifestations of heart failure and pulmonary hypertension. Partial AVSD often remains undetected until adulthood, whereas complete forms present with early symptoms requiring urgent intervention.

Surgical treatment is the primary method of AVSD correction, aiming to restore normal hemodynamics and prevent complications. Modern techniques, such as double and modified single patch repair, have demonstrated high efficacy and safety. In complex cases, including single-ventricle physiology, staged palliative procedures such as shunting and the Fontan procedure are utilized. Long-term treatment outcomes depend on early diagnosis, the selection of an appropriate surgical technique, and meticulous postoperative monitoring. Advanced imaging methods enable the timely detection of complications, such as residual defects and valvular regurgitation, and facilitate appropriate intervention. The importance of a multidisciplinary approach in managing patients with AVSD continues to grow, contributing to improved quality of life and reduced mortality.

This article provides a comprehensive review of the epidemiology, clinical presentation, diagnostic approaches, and surgical treatment methods for AVSD, emphasizing the significance of an individualized approach for each patient. [4, 20, 47, 18, 26]

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References:

1.*Alhawri K.A., Mcmahon C.J., Alrih M.M., Alzein Y., Khan A.A., Mohammed S.K., Alalwi K.S., Walsh K.P., Kenny D.P., McGuinness J.G., Nolke L., Redmond J.M.* Atrioventricular septal defect and tetralogy of Fallot - A single tertiary center experience: A retrospective review. Annals of pediatric cardiology, 2019. *12*(2), 103–109. https://doi.org/10.4103/apc.APC_87_18

2.*Alizadehasl A., Amini-Salehi E., Jebelli S.F. H., Hosseini K., Aliabadi A.Y., Yazzaf R., Nobakht S.* Presentation of mitral valve cleft with concurrent atrial septal defect and ventricular septal defect detected by three-dimensional transesophageal echocardiography: a case report. Journal of medical case reports, 2024.18(1), 387. https://doi.org/10.1186/s13256-024-04704-y

3.*Almarzooq Z., Patel B., McGregor P.C., Aragam J.* Partial Atrioventricular Canal Defect With an Anomalous Left Circumflex Coronary Artery in an Elderly Veteran. JACC. Case reports, 2019. 1(3), 291–296. https://doi.org/10.1016/j.jaccas.2019.08.005

4.*Alsoufi B., McCracken C., Kanter K., Shashidharan S., Border W., Kogon B.* Outcomes of Multistage Palliation of Infants With Single Ventricle and Atrioventricular Septal Defect. World journal for pediatric & congenital heart surgery, 2020. 11(1), 39–48. https://doi.org/10.1177/2150135119885890

5.*Andersen N.D., Turek J.W.* Echocardiographic Assessment of Complete Atrioventricular Canal Defects: A Balancing Act. Seminars in thoracic and cardiovascular surgery, 2019. 31(1), 87–88. https://doi.org/10.1053/j.semtcvs.2018.11.005

6.*Atik E., Binotto M.A., & Barreto A.C.* Case 6/2019 - Total Atrioventricular Septal Defect, 12 Years After Operative Correction, No Residual Defects. Arquivos brasileiros de cardiologia, 2019. 13(5), 999–1001. https://doi.org/10.5935/abc.20190192

7.*Balpande N., Bodhey N.K., Singh S.* Cardiac Computed Tomography Pictorial Equivalent of Angiographic Gooseneck Sign in a Case of Atrioventricular Septal Defect with Cor Triatriatum Sinister. Heart views: the official journal of the Gulf Heart Association, 2022. 23(4), 235–239. https://doi.org/10.4103/heartviews.heartviews_47_22

8.*Bhattarai P., Karki M.* Role of ECG in the Accidental Finding of an Atrioventricular Septal Defect in an Asymptomatic Patient Undergoing Cosmetic Surgery. Cureus, 2024. 16(1), e52406. https://doi.org/10.7759/cureus.52406

9.*Boutayeb A.* Complete Atrioventricular Canal Defect: Towards a More Physiological Repair. Heart, lung & circulation, 2018. 27(3), e4–e6. https://doi.org/10.1016/j.hlc.2017.05.136

10. *Burkhart H.M., Anderson H.N.* Commentary: Complete atrioventricular septal defect in young infants: The advantages of early repair. The Journal of thoracic and cardiovascular surgery, 2021. 161(6), 2155–2156. https://doi.org/10.1016/j.jtcvs.2020.09.091

11. *Chen X.F., Jiang F., Li L., Chen Y., Chen X., Jiang, Y.Y., Xiang L., Ma X.J.* Application of low-dose dual-source computed tomography angiography in children with complex congenital heart disease. Experimental and therapeutic medicine, 2017. 14(2), 1177–1183. https://doi.org/10.3892/etm.2017.4591

12. *Dawary M.A., Alshamdin F.D., Alkhalaf L.H., Alkhamis A.O., Khouqeer F.A.* Outcomes of surgical repair of complete atrioventricular canal defect in patients younger than 2 years of age. Annals of Saudi medicine, 2019. *39*(6), 422–425. https://doi.org/10.5144/0256-4947.2019.422

13. *De Oliveira Nunes M., Witt D.R., Casey S.A., Rigsby C.K., Hlavacek A.M., Chowdhury S.M., Nicol, E. D., Semple, T., Lesser J.R., Storey K.M., Kunz M.S., Schmidt*

C.W., Stanberry L.I., Han. B.K. Multi-institution assessment of the use and risk of cardiovascular computed tomography in pediatric patients with congenital heart disease. Journal of cardiovascular computed tomography, 2021. 15(5), 441–448. https://doi.org/10.1016/j.jcct.2021.01.003

14. *Deri A., English K.* Educational series in congenital heart disease: Echocardiographic assessment of left to right shunts: atrial septal defect, ventricular septal defect, atrioventricular septal defect, patent arterial duct. *Echo research and practice*, 2018. 5(1), R1–R16. https://doi.org/10.1530/ERP-17-0062

15. *Digilio M.C., Pugnalon F., De Luca A., Calcagni G., Baban A., Dentici M.L., Versacci P., Dallapiccola B., Tartaglia M., Marino B.* Atrioventricular canal defect and genetic syndromes: The unifying role of sonic hedgehog. Clinical genetics, 2019. 95(2), 268–276. https://doi.org/10.1111/cge.13375

16. *Elders B., Westenberg J., van den Boogaard P., Calkoen E., Blom N., Kroft L., de Roos A., & Roest A.* Altered ascending aortic wall shear stress in patients with corrected atrioventricular septal defect: a comprehensive cardiovascular magnetic resonance and 4D flow MRI evaluation. Cardiology in the young, 2019. 29(5), 637–642. https://doi.org/10.1017/S1047951119000374

17. *Ezzeldin D.A., Hafez M.S., Mansour A.* Multidetector computed tomography for patients with congenital heart disease: a multi-center registry from Africa and Middle East; patients' characteristics and procedural safety. The Egyptian heart journal : (EHJ) : official bulletin of the Egyptian Society of Cardiology, 2021. *73*(1), 90. https://doi.org/10.1186/s43044-021-00217-x

18. *Espinola-Zavaleta N., Muñoz-Castellanos L., Kuri-Nivón M., Keirns C.* Understanding atrioventricular septal defect: anatomoechocardiographic correlation. Cardiovascular ultrasound, 2008. 6, 33. https://doi.org/10.1186/1476-7120-6-33

19. *Fernando R.J., Johnson S.D., Carter T.* Partial Atrioventricular Canal Defect. Anesthesiology, 2019. 131(4), 896–897. https://doi.org/10.1097/ALN.0000000000002795

20. *Fong L.S., Betts K., Bell D., Konstantinov I.E., Nicholson I.A., Winlaw D.S., Orr Y., Australian CAVSD Study Group* Complete atrioventricular septal defect repair in Australia: Results over 25 years. *The Journal of thoracic and cardiovascular surgery*, 2020. 159(3), 1014–1025.e8. https://doi.org/10.1016/j.jtcvs.2019.08.005

21. *Gedikbaşı A., Oztarhan K., Yıldırım G., Gül A., Asar-Canaz E., Ceylan Y.* Atrioventricular and ventricular septal defects; topographical analysis and impact of associated cardiac and extracardiac findings and postpartum outcome. Journal of the Turkish German Gynecological Association, 2010. 11(1), 8–15.

22. *Goo H.W.* Image Quality and Radiation Dose of High-Pitch Dual-Source Spiral Cardiothoracic Computed Tomography in Young Children with Congenital Heart Disease: Comparison of Non-Electrocardiography Synchronization and Prospective Electrocardiography Triggering. Korean journal of radiology, 2018. 19(6), 1031–1041. https://doi.org/10.3348/kjr.2018.19.6.1031

23. *Gosnell J., Pietila T., Samuel B.P., Kurup H.K., Haw, M.P., Vettukattil, J.J.* Integration of Computed Tomography and Three-Dimensional Echocardiography for Hybrid Three-Dimensional Printing in Congenital Heart Disease. Journal of digital imaging, 2016. 29(6), 665–669. https://doi.org/10.1007/s10278-016-9879-8

24. *He C., Zhou Y., Tang S.S., Luo L.H., Feng K.* Completed atrioventricular block induced by atrial septal defect occluder unfolding: A case report. World journal of clinical cases, 2020. 8(22), 5715–5721. https://doi.org/10.12998/wjcc.v8.i22.5715

25. *Husain A.* Commentary: To band or not to band - is that really the question? Journal of Thoracic and Cardiovascular Surgery, 2021. 161(5), 1603–1604. https://doi.org/10.1016/j.jtcvs.2020.08.087

26. *Jegatheeswaran A., Pizarro C., Caldarone C.A., Cohen M.S., Baffa J.M., Gremmels D.B., Mertens L., Morell, V.O., Williams W.G., Blackstone E.H., McCrindle B.W., Overman D.M.* Echocardiographic definition and surgical decision-making in unbalanced atrioventricular septal defect: a Congenital Heart Surgeons' Society multiinstitutional study. Circulation, 2010. 122(11 Suppl), S209–S215. https://doi.org/10.1161/CIRCULATIONAHA.109.925636

27. *Jones A.L., White B.R., Ghosh R.M., Mondal A., Ampah S., Ho D.Y., Whitehead K., Harris M.A., Biko D.M., Partington S., Fuller S., Cohen M.S., Fogel M.A.* Cardiac magnetic resonance predictors for successful primary biventricular repair of unbalanced complete common atrioventricular canal. Cardiology in the young, 2024. 34(2), 387–394. https://doi.org/10.1017/S1047951123001786

28. *Kantor B., Nagel E., Schoenhagen P., Barkhausen, J., Gerber T.C.* Coronary computed tomography and magnetic resonance imaging. Current problems in cardiology, 2009. 34(4), 145–217. https://doi.org/10.1016/j.cpcardiol.2008.12.002

29. *Kaski J.P., Wolfenden J., Josen M., Daubeney P.E., Shinebourne E.A.* Can atrioventricular septal defects exist with intact septal structures? Heart (British Cardiac Society), 2006. 92(6), 832–835. https://doi.org/10.1136/hrt.2005.069278

30. *Linask K.K., Han M., Bravo-Valenzuela N.J.* Changes in vitelline and utero-placental hemodynamics: implications for cardiovascular development. Frontiers in physiology, 2014. 5, 390. https://doi.org/10.3389/fphys.2014.00390

31. *Liu Y., Chen S., Zühlke L., Black G.C., Choy M.K., Li, N., Keavney B.D.* Global birth prevalence of congenital heart defects 1970-2017: updated systematic review and meta-analysis of 260 studies. International journal of epidemiology, 2019. 48(2), 455–463. https://doi.org/10.1093/ije/dyz009

32. *Murakami A.N., Baston G.G., Cardoso M.R.R., Marchi C.H., Croti U.A.* Rare Association Between Atrioventricular Septal Defect and Partial Anomalous Pulmonary Venous Connection. Brazilian journal of cardiovascular surgery, 2019. 34(6), 765–768. https://doi.org/10.21470/1678-9741-2019-0203

33. *Nicolay S., Salgado R.A., Shivalkar B., Van Herck P.L., Vrints C., Parizel P.M.* CT imaging features of atrioventricular shunts: what the radiologist must know. Insights into imaging, 2016. 7(1), 119–129. https://doi.org/10.1007/s13244-015-0452-7

34. *Pugnaloni F., Digilio M.C., Putotto C., De Luca E., Marino B., Versacci, P.* Genetics of atrioventricular canal defects. Italian journal of pediatrics, 2020. 46(1), 61. https://doi.org/10.1186/s13052-020-00825-4

35. *Pugnaloni F., Versacci P., Marino B., & Digilio M.C.* Atrioventricular canal defect is the classic congenital heart disease in Bardet-Biedl syndrome. Annals of human genetics, 2021. 85(3-4), 101–102. https://doi.org/10.1111/ahg.12413

36. *Rajiah P., Schoenhagen P.* The role of computed tomography in pre-procedural planning of cardiovascular surgery and intervention. Insights into imaging, 2013. 4(5), 671–689. https://doi.org/10.1007/s13244-013-0270-8

37. *Rigby M.* Atrioventricular Septal Defect: What Is in a Name? Journal of cardiovascular development and disease, 2021. 8(2), 19. https://doi.org/10.3390/jcdd8020019

38. *Schumacher K., Marin Cuartas M., Meier S., Aydin, M.I., Borger M.A., Dähnert I., Kostelka M., Vollroth M.* Long-term results following atrioventricular septal defect repair. Journal of cardiothoracic surgery, 2023. 18(1), 250. https://doi.org/10.1186/s13019-023-02355-6

39. *Schusterova I., Jakubova M., Vachalcova M., Sieradzka K., Gibarty C., Poruban T., Gasparovic I., Artemiou P.* Three-dimensional transesophageal echocardiography in diagnosis of intermediate atrioventricular septal defect in the adult: case report and literature review. Journal of cardiothoracic surgery, 2021. 16(1), 209. https://doi.org/10.1186/s13019-021-01596-7

40. *Singh P., Mehta A., Nanda N.C.* Live/real time three-dimensional transthoracic echocardiographic findings in an adult with complete atrioventricular septal defect. Echocardiography (Mount Kisco, N.Y.), 2010. 27(1), 87–90. https://doi.org/10.1111/j.1540-8175.2009.01129.x

41. *Sofian A., Rusdiana R., Rasyid A.M.A., & Agustian A.H.* Surgical repair of partial atrioventricular septal defect (pAVSD) in adulthood: A rare case report. International journal of surgery case reports, 2025. 131, 111378. https://doi.org/10.1016/j.ijscr.2025.111378

42. *Song L., Ling Y.,* Repair of partial atrioventricular canal defect in adult patients: two-year follow-up outcomes of a retrospective study. Journal of cardiothoracic surgery, 2019. *14*(1), 106. https://doi.org/10.1186/s13019-019-0931-x

43. *St-Onge S., Amin S., Cianciulli A., Jolley M.A., Drouin S.* 3D Echocardiogram Visualization: A New Method Based on "Focus + Context". Proceedings of SPIE--the International Society for Optical Engineering, 2024.12929, 129290T. https://doi.org/10.1117/12.3006214

44. *Surkova E., West C., Flick C., Ilagan L., Gatzoulis, M.A., Senior R., Li W.* Added value of three-dimensional transthoracic echocardiography in assessment of an adult patient with atrioventricular septal defect. Echocardiography (Mount Kisco, N.Y.), 2019. 36(4), 809–812. https://doi.org/10.1111/echo.14300

45. *Vereczkey A., Kósa Z., Csáky-Szunyogh M., & Czeizel A.E.* Isolated atrioventricular canal defects: birth outcomes and risk factors: a population-based Hungarian case-control study, 1980-1996. Birth defects research. Part A, Clinical and molecular teratology, 2013.97(4), 217–224. https://doi.org/10.1002/bdra.23124

46. *Veronese P., Guariento A., Cattapan C., Fedrigo M., Gervasi M.T., Angelini A., Riva A., Vida V.* Prenatal Diagnosis and Fetopsy Validation of Complete Atrioventricular Septal Defects Using the Fetal Intelligent Navigation Echocardiography Method. Diagnostics (Basel, Switzerland), 2023. 13(3), 456. https://doi.org/10.3390/diagnostics13030456

47. *Wang G., Ma K., Pang K., Hua Z., Zhang S., Qi, L., Yang Y., Feng Z., Mao F., Zhang H., Li S.* Modified Single Repair Technique for Complete Atrioventricular Septal Defect: A Propensity Score Matching Analysis. Pediatric cardiology, 2020. 41(3), 615–623. https://doi.org/10.1007/s00246-020-02292-4

48. *Yang J.C., Lin M.T., Jaw F.S., Chen S.J., Wang J.K., Shih T.T., Wu M.H., Li Y.W.* Trends in the utilization of computed tomography and cardiac catheterization among children with congenital heart disease. Journal of the Formosan Medical Association = Taiwan yi zhi, 2015. 114(11), 1061–1068. https://doi.org/10.1016/j.jfma.2014.08.004

49. *Yoshitake S., Kaneko Y., Morita K., Hoshino M., Oshima Y., Takahashi M., Anderson R.H., SPring* 8 Cardiovascular Structure Analyzing Research Group Visualization and quantification of the atrioventricular conduction axis in hearts with ventricular septal defect using phase contrast computed tomography. The Journal of thoracic and cardiovascular surgery, 2020. 160(2), 490–496. https://doi.org/10.1016/j.jtcvs.2020.02.036

50. *Zhang F., Qi L., Zhao M., Han S., Zhang H., Wang G.* Global research landscape on the genetics of congenital heart disease: A bibliometric and visualized analysis via VOSviewer and CiteSpace. Medicine, 2024.103(43), e40261. https://doi.org/10.1097/MD.0000000000040261

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