Case Series

Grown-up Congenital Heart (GUCH) Disease: Clinical challenges and role of electronic registries (GUCH & Heart Transplant)

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Abstract

Background: Adult Congenital Heart Disease (ACHD), also known as Grown Up Congenital Heart Disease, .(GUCH) is increasingly recognized due to improved survival from childhood, but its clinical presentation can be highly variable, often mimicking common adult cardiovascular conditions. Early recognition, appropriate management, and long-term follow-up are essential.

Case Presentation: We report three adult patients with congenital heart disease. The first involved a 43-year-old male with Ebstein anomaly, presenting with breathlessness, abdominal distension, palpitations, and swelling of both legs, complicated by atrial fibrillation and acute kidney injury. Echocardiography revealed pulmonary hypertension severe tricuspid regurgitation with right ventricular failure and preserved left ventricular function. He was managed with oxygen, IV diuretics, electrolyte correction, antiarrhythmics, and supportive care, resulting in clinical improvement. The second patient was a 57-year-old female with type 2 diabetes mellitus, presenting with chest pain mimicking unstable angina. ECG showed ST depression, while echocardiography and coronary angiography revealed normal epicardial coronaries, pulmonary hypertension and normal left ventricular function. She was managed with antiplatelets, statins, beta-blockers, proton pump inhibitors, and supportive therapy and discharged in stable condition. The third patient was a 37 years old female patient with large ASD(OS) and PAH admitted with chest pain and breathlessness. ECG showed biventricular hypertrophy with sinus tachycardia and chest –ray showed dense lung infiltrates. She was treated with bronchodilator and systemic steroids and BiPAP support discharged in stable condition.

Discussion: These patients illustrate the heterogeneous presentation of ACHD in adults, ranging from right heart failure and arrhythmias to angina-like symptoms without coronary obstruction. Management requires a multidisciplinary approach, including medical therapy, surgical intervention, and, when indicated, heart

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transplantation. Transplant registries are critical for tracking outcomes, guiding clinical decisions, and improving long-term survival in this population.

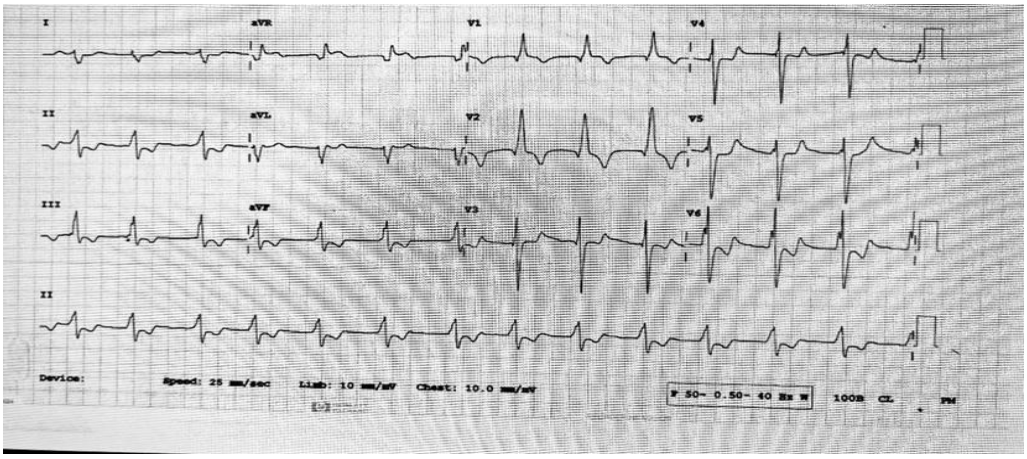
Conclusion: Adult congenital heart disease requires timely recognition, individualized management, and long-term follow-up. Participation in transplant registries enhances patient outcomes, supports research, and informs evidence-based strategies for managing rare ACHD presentations.

Keywords: Adult congenital heart disease; Ebstein anomaly; Right heart failure; Heart transplantation; Transplant registry; Arrhythmia

1.1. Patient 1

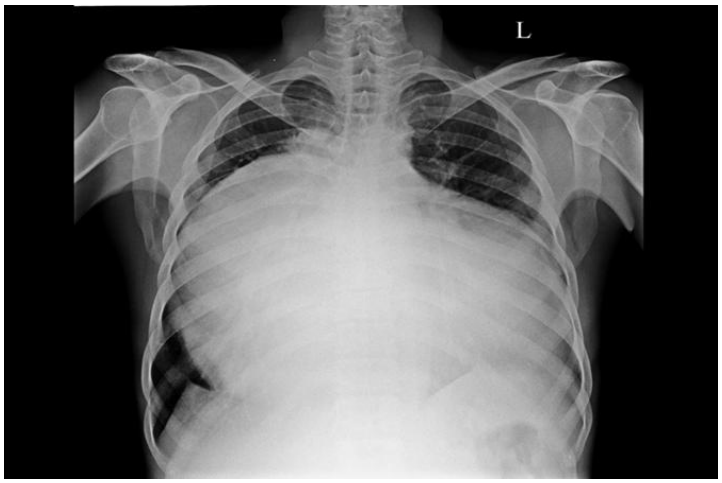
A 43-year-old male, euglycemic and normotensive, with a known history of congenital acyanotic heart disease (Ebstein's anomaly), presented with breathlessness, abdominal distension, and palpitations for 4–5 days, along with cold, cough, and expectoration for 2 days. He also reported breathlessness on exertion and swelling of both legs. The patient was known to have Diabetes Mellitus, Hypertension, and Dyslipidemia,

On examination, he was conscious and oriented, dyspneic with an SpO_2 of 93%, and tachycardic (HR 130 bpm). ECG showed Right Axis Deviation (RAD), Right Ventricular Hypertrophy (RVH) and a junctional (nodal) rhythm.



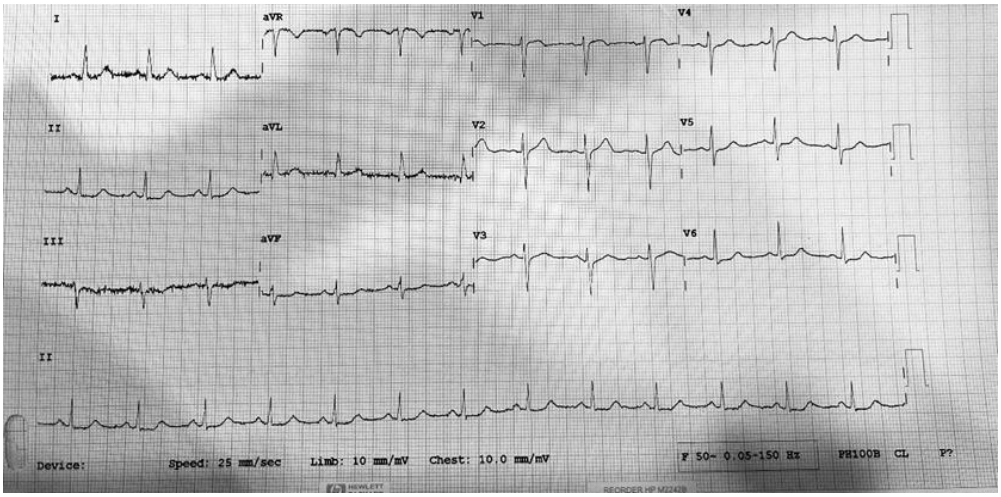
Echocardiography revealed a grossly dilated right atrium, atrialized right ventricle, 65 mm apical displacement of the tricuspid valve, no regional wall motion abnormality (RWMA), good left ventricular function (LVF), severe unrestricted tricuspid regurgitation, right ventricular dysfunction, and a dilated inferior vena cava (IVC).

Laboratory investigations revealed haemoglobin 11.9 g/dL, monocyte 12.4%, lymphocyte 12.2%, ESR 50mm/hr, platelet 87000 cells/cu.mm, GGT 102U/L, AST 36.8U/L, Indirect bilirubin 1.10mg/dl, direct bilirubin 0.94mg/dl. Urine examination revealed 1-2 pus cells. Initially potassium was 6.92mmol/L reduced to 3.3mmol/L with potassium binder. Creatinine was rising from 0.93mg/dl to 3.45mg/dl indicative of acute kidney injury. X ray revealed severe cardiomegaly.



X Ray Chest showing severe cardiomegaly USG abdomen showed congestive hepatomegaly, mild ascites, and a tiny left renal cortical cyst. The patient developed Atrial Fibrillation, which was successfully reverted to normal sinus rhythm pharmacologically with Amiodarone. He was managed with oxygen support, IV diuretics, electrolyte correction, nephroprotective therapy, and other supportive measures. He was diagnosed with congenital acyanotic heart disease (Ebstein's anomaly) with right heart failure and good left ventricular function. The patient improved with treatment and was discharged in a stable condition.

1.2. Patient 2



A 57-year-old female, a known case of type 2 diabetes mellitus, presented with recurrent chest pain for 2 days. She was initially managed at an outside hospital and referred for further evaluation. On examination, she was conscious and oriented. ECG was unremarkable.

Laboratory investigations revealed that haemoglobin 11.9 g/dL, lymphocytes 41.5%, and eosinophils 0.3%, Lymphocytes 41.5%, ESR 44mm/hr., Urine examination revealed pus cells 1-2. Echocardiography showed no regional wall motion abnormality, good left ventricular function, grade I diastolic dysfunction, trivial mitral regurgitation, and a small perimembranous ventricular septal defect with left-to-right shunt, along with severe pulmonary arterial hypertension. She was diagnosed with coronary artery disease – unstable angina with preserved left ventricular function and normal sinus rhythm, along with congenital acyanotic heart disease (small peri membranous VSD with left-to-right shunt).

After obtaining informed consent, she underwent coronary angiography on 29 January 2026, which revealed normal epicardial coronary arteries. The procedure was uneventful, and she remained hemodynamically stable post-procedure. A gastroenterology consultation was obtained for epigastric pain, and appropriate management was carried out. She was treated with antiplatelets, statins, beta-blockers, proton pump inhibitors, and supportive measures.

Her symptoms gradually improved, she was ambulating well, shifted to the ward, and subsequently discharged in a stable condition with diagnoses of coronary artery disease – unstable angina, type 2 diabetes mellitus, corpus-predominant gastritis, congenital acyanotic heart disease (small perimembranous VSD with left-to-right shunt), severe pulmonary arterial hypertension, and grade I left ventricular diastolic dysfunction.

1.3. Patient 3

A 37-year-old female, a known case of obstructive airway disease and atrial septal defect with severe pulmonary arterial hypertension was admitted with pain over the left side of chest, for 10 days and cough with expectoration for 3 days (Yellowish sputum) along with depression.

On examination she was tachypneic, and hypoxic, ABG results revealed type 2 respiratory failure. Spo2: 77% with 4 litres O2 / 92% with 10 litres. H/o orthopnea (+), grade II-III MMRC and breathlessness. Her X-ray, ECG and Echo showed remarkable evidence of her ongoing condition as follows.

On 13/3/26

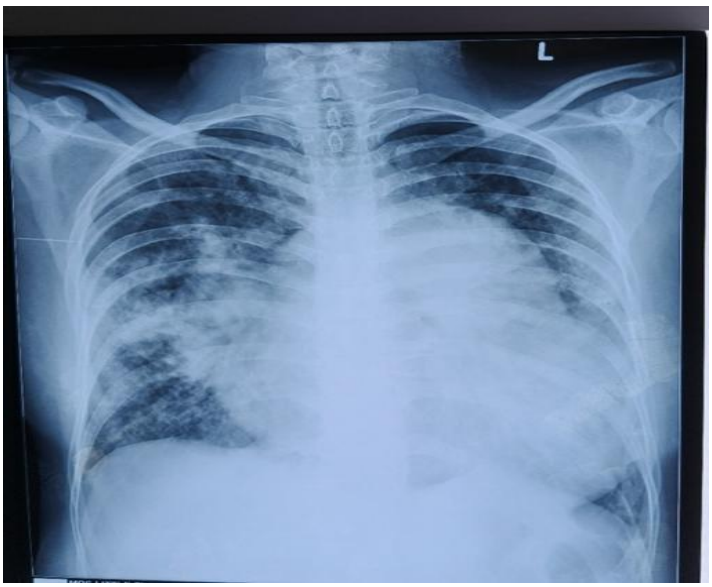


Fig (1): X ray showed dense bilateral lung infiltrates

On 18/03/26

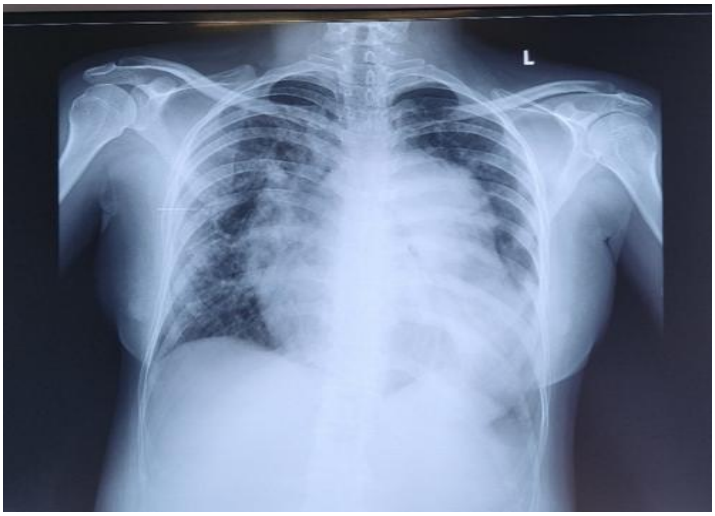


Fig (2): X ray showed reduced bilateral lung infiltrates

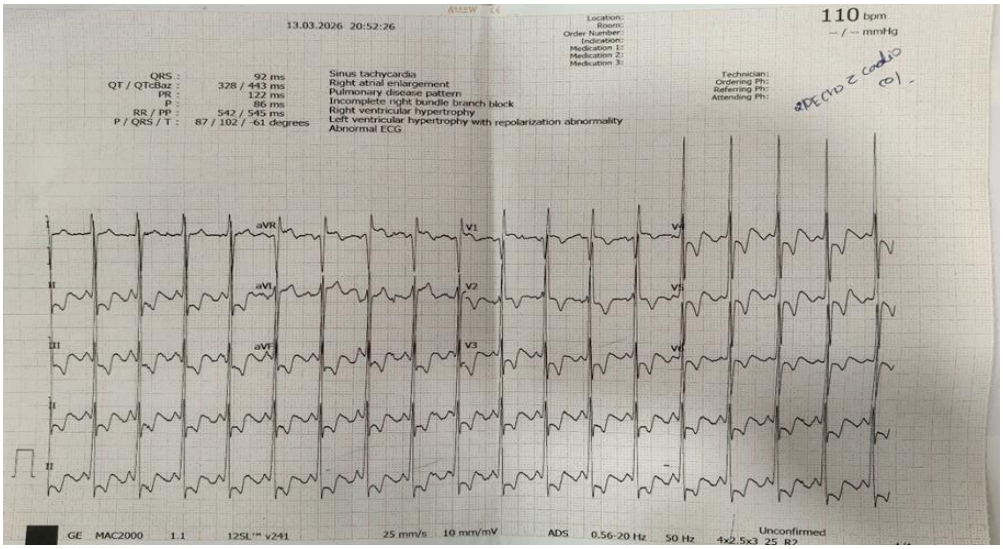


Fig (3): ECG showed Biventricular Hypertrophy

Lab investigation revealed: C Reactive Protein (CRP) - 236.9 Hb – 10.31, and elevated AST - 5l0 U/L, and ALT – 597 U/L.

A transthoracic echocardiogram performed on 14 March 2026 revealed a large ostium secundum atrial septal defect measuring 32 mm with a bidirectional shunt, the left ventricle demonstrated preserved systolic function with an ejection fraction of 61% and no regional wall motion abnormalities. In contrast, the right ventricle was dilated with evidence of systolic dysfunction, as indicated by a reduced tricuspid annular plane systolic excursion (TAPSE) of 15 mm. Severe tricuspid regurgitation was noted, likely secondary to right ventricular dilatation and annular enlargement.

During hospitalization, the patient showed gradual clinical improvement with resolution of hypoxia and stabilization of respiratory parameters. psychiatric evaluation was also undertaken due to associated depressive symptoms, and she was initiated on antidepressant therapy.

A notable aspect of this case is the absence of definitive surgical or percutaneous intervention for over a decade, with the patient being managed solely by medical therapy. Prolonged reliance on medical management alone, as seen in this patient, likely contributed to the evolution of severe pulmonary hypertension, right ventricular dysfunction, and bidirectional shunting, rendering the defect inoperable.

2. Discussion

Ebstein anomaly is a rare congenital heart defect characterized by apical displacement of the tricuspid valve leaflets, resulting in atrialization of a portion of the right ventricle and varying degrees of tricuspid regurgitation. The severity of displacement and regurgitation determines the clinical presentation, which can range from asymptomatic cases detected incidentally to severe right heart failure in adulthood.

Adults with Ebstein anomaly may remain asymptomatic for years, but common presentations include fatigue, exertional dyspnea, palpitations, and exercise intolerance. Some patients develop peripheral edema, ascites, or hepatomegaly as signs of right heart failure. Arrhythmias, particularly atrial fibrillation, atrial flutter, or supraventricular tachycardia, are frequent and may lead to syncope or sudden cardiac events. Cyanosis is typically mild in adults, especially in acyanotic forms. Medical therapy is mainly supportive and indicated in symptomatic patients or those with mild-to-moderate diseases.

Diuretics are used to manage fluid overload, while antiarrhythmic medications such as amiodarone or beta-blockers help control atrial arrhythmia. Anticoagulation is recommended in patients with atrial fibrillation to prevent thromboembolism. Managing comorbidities such as diabetes, hypertension, or coronary artery disease is essential to reduce the risk of worsening cardiac function. Surgery is indicated in adults with severe tricuspid regurgitation, symptomatic right heart failure, or uncontrolled arrhythmias. Tricuspid valve repair is preferred when feasible, as it preserves native valve function. Valve replacement (mechanical or bioprosthetic) is considered when repair is not possible.

Concomitant procedures may include Maze procedure for arrhythmia correction or closure of an atrial septal defect if present. Timing of surgery is crucial, as early intervention can prevent irreversible right ventricular dysfunction.

3. Heart Transplantation

Heart transplantation is reserved for patients with severe, refractory right ventricular failure, inoperable anatomy, or failed previous surgical repairs. Pre-transplant evaluation requires multidisciplinary assessment, including cardiology, nephrology, and anesthesia teams. Post-transplant, patients require lifelong immunosuppression and follow-up, and outcomes are significantly improved when managed at experienced transplant centers.

4. Importance of Transplant Registry

We initiated our registry with three cases of grown-up congenital heart disease (GUCH). Among these, one patient diagnosed with ventricular septal defect (VSD) was identified as a candidate for defect closure. Another patient with Ebstein's anomaly demonstrated disease progression warranting consideration for advanced therapies, including potential heart transplantation, and is currently under close longitudinal follow-up. The third patient, presenting with a large atrial septal defect (ASD) accompanied by a bidirectional shunt, was enrolled in the heart failure–heart transplant (HF-HT) registry at Kauvery

Hospital, Vadapalani. These cases highlight the effectiveness of our structured follow-up system in facilitating timely referral and access to advanced therapeutic interventions.

5. Conclusion

Adult congenital heart disease presents with a wide range of manifestations, from mild symptoms to severe heart failure and arrhythmia, often resembling common adult cardiac conditions. The cases described highlight this variability, including Ebstein anomaly with severe tricuspid regurgitation and arrhythmias, and angina-like symptoms with normal coronary arteries, underscoring diagnostic challenges.

Management requires a multidisciplinary approach with appropriate medical therapy, timely intervention, and consideration of heart transplantation in advanced cases. Transplant registries are essential for monitoring outcomes, guiding evidence-based care, enabling research, and ensuring standardized follow-up and improved survival.

Overall, early recognition, individualized management, and active registry participation are key to optimizing outcomes and advancing care in adult congenital heart disease.

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