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Adult Cardiology & Clinical Cardiovascular Medicine • Poster Presentation

ABSTRACT AND SECONDARY ERYTHROCYTOSIS IN A 49-YEAR-OLD MAN: A RARE CASE OF SURVIVAL INTO THE FIFTH DECADE

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Background: Tetralogy of Fallot (TOF) is the most common cyanotic congenital heart disease, usually repaired in infancy or early childhood. Survival into the fifth decade without correction is exceedingly rare. Unrepaired adults face chronic hypoxaemia, secondary erythrocytosis, arrhythmias, thromboembolism, and sudden cardiac death, with syncope signaling particularly high risk.

Case Presentation: A 49-year-old male truck driver presented with fatigability, progressive bilateral leg swelling, weight loss, central cyanosis, and a syncopal episode while driving. He described lifelong cyanosis and exertional limitation but had never undergone cardiac evaluation. Examination showed central cyanosis, clubbing, pedal oedema, a harsh ejection systolic murmur, and an S4 gallop. Haemoglobin was 18.1 g/dL and packed cell volume 57%, consistent with secondary erythrocytosis. Electrocardiography showed right axis deviation, right ventricular hypertrophy, biatrial enlargement, and anteroseptal ST–T changes. Echocardiography confirmed unrepaired TOF with a large malaligned ventricular septal defect, ~50% aortic override, severe infundibular right ventricular outflow obstruction, marked right ventricular hypertrophy, biatrial enlargement, and preserved left ventricular function (LVEF 69%). He was started on heart failure therapy and referred for cardiothoracic surgical evaluation.

Conclusion: Survival with unrepaired TOF into the fifth decade reflects a favourable balance between outflow obstruction and pulmonary blood flow. Although late repair is feasible, it carries higher operative mortality and reduced long-term survival compared with the general population. This case highlights syncope as a warning sign of severe obstruction or malignant arrhythmia, and stresses maintaining suspicion for congenital heart disease in cyanotic adults, with echocardiography as the cornerstone of diagnosis and timely referral.

Keywords: Tetralogy of Fallot; unrepaired congenital heart disease; secondary erythrocytosis; syncope; adult congenital heart disease; sudden cardiac death