Unusual presentation of cyanotic congenital heart disease in adult

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Abstract:
A case report regarding cyanotic congenital heart disease - tetralogy of fallot presenting in the 4th decade of life, the patient is surviving till the age of 35 years without any surgical intervention. The important clinical manifestations were gingival and oral mucosal bleeding, fever and frequent chest infections. Interestingly in patients with tetralogy of fallot there is a seeming paradox between abnormal haemostasis on the one hand and a thrombotic predisposition in specific vascular beds on the other. This case report highlights a hemorrhagic tendency encountered in a case of an adult with tetralogy of fallot. Further investigations and imaging showed bronchiectasis of the left lower lobe along with bilateral fibrotic changes in both lungs which has been rarely reported in patients with oligemic lung fields so far. Uncorrected tetralogy of fallot presenting in the 4th decade of life is an uncommon entity, the development of bleeding tendencies and an altered coagulation profile with abnormalities in both the extrinsic and intrinsic coagulation pathway along with thrombocytopenia has been rarely reported.

Finally the association of tetralogy of fallot with bronchiectasis is still very rare.

Keyword: tetralogy of fallot, abnormal haemostasis, bronchiectasis.

Case history
Mr. A, 35 year old male, a known case of cyanotic congenital heart disease - tetralogy of fallot was admitted with the complaints of fever, productive cough for 1 week, shortness of breath grade 3, history of spontaneous gingival bleeding for past 3 days was present. History of cyanosis was present from infancy. No history of pedal edema and jaundice. On examination General examination revealed central cyanosis, hyperaemia, grade 3 pandigital clubbing present in both upper and lower limbs, intra oral examination revealed a cyanotic mucous membrane of lips, tongue and gingiva, fissured tongue and inflamed bleeding gingiva. His pulse rate was 102/mt regular, blood pressure was 110/60 mm Hg in the right upper limb, respiratory rate was 30/mt abdominothoracic type and temperature was normal. Cardiac examination revealed a grade
1 parasternal lift. On auscultation first heart sound was normal and second heart sound was single.
Grade 3/6 midsystolic murmur heard in left 3rd and 4th ics. Continuous murmur was heard over the precordium, back and interscapular region. Auscultation of respiratory system revealed bilateral coarse basal rales more on the left side. Per abdomen examination was normal. Central nervous system revealed no focal neurological deficit.

Investigations
The investigations done were hemoglobin – 15gm%. PCV – 40% peripheral smear study revealed microcytic hypochromic cells. RBC count – 4,50,000. Total count – 6680 DC P-69,L21,E2,M2,B0. Platelet count – 90000/cumm Urine analysis revealed trace of albumin, ketone bodies and sugar was negative. RBCs-4-6/hpf, epithelial cells-3-4/hpf. Blood sugar- 96mg%, Blood urea-24mg%, serum creatinine-0.7mg%. Liver function tests, serum electrolytes were normal. Blood group was A+, HIV, HbsAg and Anti HCV – negative. Blood widal, Microscopic agglutination test for leptospirosis – negative, blood and urine culture – revealed no growth. Sputum for acid fast bacilli – negative, sputum culture and sensitivity – was negative Prothrombin time (PT) – 114 s, normal 11-13 s (control PT 14.3), Activated partial thromboplastin time (aPTT) >130 s, N – 30-40 s, (control PTT 29.9), INR (International normalised ratio) – 11.5 Chest X ray – cor en sabot heart, bronchiectasis Left lower lobe with subtle fibrotic changes scattered in both lungs. Ultrasound abdomen was normal. Electrocardiography (ECG) – sinus rhythm, right axis deviation, tall R wave in V1 with loss of R wave in V2. Echocardiogram – large subaortic ventricular septal defect with bidirectional flow measuring 20 mm, over riding of aorta around 50%, pulmonary valve could not be visualised, normal LV systolic function, no pericardial effusion. Multiple aorto pulmonary collateral arteries were seen arising from the descending aorta. Echo features were suggestive of Tetralogy of fallot with pulmonary atresia. Computed tomography chest revealed bronchiectasis of left lower lobe with fibrotic changes lower lobes of both lungs

Course in the hospital
Patient was diagnosed as pulmonary atresia with ventricular septal defect with abnormal hemostasis, and left lower lobe bronchiectasis. Patient treated with inj.ceftriaxone 2gm iv bd and inj amikacin 500mg iv bd for 7 days and antipyretics for fever and respiratory tract infection. 12 units of fresh frozen plasma was transfused and vitamin K 10 mg im od for 3 days, T propanolol 40 mg 1 b d and multivitamin tablets given. Dental opinion was obtained – counselling on oral hygiene and oral prophylaxis was given. Patient was discharged at request 8 days later. On discharge gingival inflammation and bleeding subsided, patient was afebrile, PT - 41.0s (control 14.3) PTT – 62 s (control 29.9) and INR – 3.47.

Discussion
Tetralogy of Fallot was first described by Dane, Nichols and Stenson 1. In 1888 a French physician. Etienne Fallot separated it from other forms of cyanotic heart diseases, hence the name Tetralogy of Fallot 1. Its overall incidence is 10% of all forms of congenital heart diseases; males are more commonly affected than females 2. Bleeding tendency in patients with cyanotic heart disease has been known for at least 50 years 4. Various type of coagulation abnormalities including thrombocytopenia, factor deficiency, fibrinolysis and disseminated intravascular coagulation, defective clot retraction,
prolonged thrombin and partial thromboplastin time, hypofibrinogenaemia, accelerated fibrinolysis, excess of fibrin degradation products (FDPs) and platelet function defects have all been described separately or in various combinations in these patients. The underlying mechanisms in these abnormal results are reduced synthesis of coagulation factors. Our patient’s abnormal hemostasis was a result of deficient clotting factors, which was corrected by transfusion of fresh frozen plasma. In patients with CCHD, platelets have both qualitative and quantitative abnormalities. Low platelet counts as observed in out patient are attributed to either shortened half life of the platelets or decreased production of platelets as megakaryocytes escape fragmentation in lungs owing to a right-to-left shunt. There is also a deficit in the platelet adhesion receptor glycoprotein lb, which can also contribute to hemostatic complications. Qualitative platelet defects associated with CCHD include abnormal aggregation of platelets in response to adenosine diphosphates, epinephrine and collagen, which is directly related to the degree of polycythemia.

Chronic hypoxemia in cyanotic CHD results in secondary erythrocytosis due to increased erythropoietin production. The commonly used term polycythemia is a misnomer because white cell counts are normal and platelet counts are normal to decreased. Red blood cell precursor may replace platelet stem cells in the bone marrow, leading to a thrombocytopenia, bleeding tendency and iron deficiency anaemia. Similar findings were observed in this patient, total WBC count -6680 and platelet count – 90000/cu mm. Cyanotic patients with erythrocytosis may have compensated or decompensated hematocrits. Hemostasis is abnormal in cyanotic CHD, due in part to the increased blood volume and engorged capillaries, abnormalities in platelet function and sensitivity to aspirin or nonsteroidal anti-inflammatory agents, and abnormalities of the extrinsic and intrinsic coagulation system. In this patient abnormalities in both intrinsic and extrinsic pathways was observed in view of the prolonged PT and aPTT. Gingival bleeding is seen because of due to this. There is higher incidence of caries and periodontal disease activity because of poor oral hygiene and lack of dental attention. In a recent study by arslan et al in 2011 where 49 children with congenital heart disease were evaluated for abnormalities in coagulation 16% were found to have a prolongation of PT, 10% had a prolonged aPTT and low fibrinogen level was found in 13%. Interestingly a prolonged PT and aPTT was found in this patient. Usually congenital heart defects with left to right shunt are associated with plethoric lung fields where the incidence of respiratory tract infections is high. But in cyanotic congenital heart disease with oligemic lung fields the incidence of respiratory tract infection is usually low. This patient with Tetralogy of Fallot presented in adulthood with complaints of breathlessness on exertion and frequent respiratory infections which is an unusual combination. Clinical examination and relevant investigations revealed association of Tetralogy of Fallot with bronchiectasis. There are a few instances of tetralogy of fallot associated with bronchiectasis. Review of literature reveals that conditions of the heart which produce oligemia of lung seem to favour infections with tuberculosis. Coronarobronchial artery fistula is a rare vascular anomaly secondary to the enlargement of the pre-existing vascular anastomosis between coronary and bronchial arteries. It probably occurs when there is a persistent disturbance of the pressure equilibrium in
volving either the coronary, bronchial or pulmonary circulation. CBF is associated with congenital heart diseases (CHD) like Tetralogy of Fallot (TOF) and pulmonary artery hypoplasia where there is a drop in the pressure in the pulmonary bed. Localized bronchiectasis is the most common associated condition among the CBF patients. The bronchiectasis involvement of the left lower lobe seems to be commonly associated with the development of CBF. Interestingly this patient had multiple aorto pulmonary collateral arteries and bronchiectatic changes primarily involving the left lower lobe. In that report by Lin et al., a patient with bronchiectasis and Tetralogy of Fallot developed clinical and radiographic evidence of pneumonia following endotracheal intubation for corrective surgery, and the causative organism was reported to be Capnocytophaga Ochracea. Capnocytophaga has been recognized as an opportunistic pathogen causing systemic infections in immunocompromised individuals with granulocytopenia and oral ulceration. The genus Capnocytophaga is comprised of fastidious, capnophilic, fusiform gram-negative bacilli has been recognized as an opportunistic pathogen causing systemic infections.

Conclusion
Uncorrected Tetralogy of Fallot is an uncommon entity. For unoperated patients with Fallot’s tetralogy of all degrees of severity, 11% are alive at age 20 years, 6% at 30 years and 3% at age 40 years. It is also interesting to note that the patient survived till this age without any medical or surgical treatment. A bleeding tendency has been recognised in patients with cyanotic heart disease. There is a seeming paradox between abnormal haemostasis on the one hand and a thrombotic predisposition in specific vascular beds on the other. Interestingly in literature the association between Tetralogy of Fallot and bronchiectasis is very rarely found.

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