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CASE REPORT Open Access

A case of AORPA in an adult presenting as secondary polycythemia

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Abstract

Background: Polycythemia refers to a condition of an abnormal increase in the red blood cell (RBC) mass. In men with a hematocrit > 60% or women with a hematocrit > 55%, there is 99% likelihood that the RBC mass is elevated. WHO uses a hemoglobin concentration > 16.5 g/dl in men or 16.0 g/dl in women to define an elevated RBC mass, in its criteria for the diagnosis of polycythemia vera. Congenital cardiac anomalies presenting as polycythemia are very rare in adults.

Case presentation: In this case report, the patient presented with non-specific complaint and had an incidental finding of secondary polycythemia, ultimately diagnosed as secondary to AORPA. Our patient, a 43-year-old male, presented to our emergency with focal onset seizures without impairment of consciousness involving left upper and lower limbs. On routine investigations, the patient was found to have grade III clubbing and hemoglobin of 19.8 g/dl with elevated Erythropoietin levels. Further workup of secondary polycythemia revealed anomalous origin of the right pulmonary artery (AORPA) from the ascending aorta with tortuous patent ductus arteriosus (PDA) and pulmonary hypertension. Secondary polycythemia, in our patient, was due to right to left shunt causing Eisenmenger phenomenon, following pulmonary hypertension, making it one of the rarest presentation in adults.

Conclusion: In the evaluation of secondary polycythemia, possibility of underlying asymptomatic cardiopulmonary shunt must be considered, though rare.

Keywords: Polycythemia, Secondary polycythemia, Eisenmenger phenomenon, AORPA, Cardiopulmonary shunt, Case report

Background

Polycythemia refers to a condition of an abnormal increase in the red blood cell (RBC) mass. In men with a hematocrit > 60% or women with a hematocrit > 55%, there is 99% likelihood that the RBC mass is elevated [1]. WHO uses a hemoglobin concentration > 16.5 g/dl in men or 16.0 g/dl in women to define a elevated RBC mass, in its criteria for the diagnosis of polycythemia vera [2]. Polycythemias can be differentiated as either primary or secondary polycythemias. In secondary polycythemia, elevation in hematocrit occurs most commonly as a response to chronic hypoxemia and is characterized by elevated erythropoietin (EPO) levels [3]. Congenital cardiac anomalies presenting as polycythemia are very rare in adults. In this case report, the patient presented with non-specific complaint and had an incidental finding of secondary polycythemia, ultimately diagnosed as secondary to anomalous origin of the right pulmonary artery (AORPA).

Case presentation

A 43-year-old male patient presented to our emergency with focal onset seizures without impairment of consciousness involving left upper and lower limbs. There was no prior history of headache, fever, and focal

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neurological deficit and no similar history in the past. During presentation, the patient had normal pulse rate, normal blood pressure, and saturation of 95% by pulse oximeter on room air. The patient was conscious and oriented. On general physical examination, grade III clubbing (Fig. 1) was seen without any pallor, cyanosis, lymphadenopathy, icterus, or pedal edema. Congestion of palpebral conjunctiva and oral mucosa was noted. On cardiovascular examination, loud S2 was heard, with normal S1 and no murmur. Respiratory system, per abdomen, and central nervous system examination was within normal limits. Non-contrast computed tomography (CT) head was done subsequently which showed calcified granuloma in the right parietal lobe. Further routine blood investigations were sent, which revealed hemoglobin of 19.8 g/dl, TLC of 14,000/cu.mm, platelet count of 40,000/cu.mm, and RBC count of 9.07 $\times$ 10^6 cu.mm. Biochemical parameters including renal function test, liver function tests, and serum electrolytes were normal. Seizures were attributed to calcified granuloma, probably secondary to past CNS *T. solium* infection, confirmed later on CEMRI of the brain, and the patient was started with anti-epileptics.

Repeated blood investigations showed hemoglobin in the range of 19–20 g/dl, with hematocrit in the range of 71–74%. With the finding of clubbing and repeated values of high hemoglobin, further workup for polycythemia was planned. History was reviewed with respect to personal, past, and drug history, according to which the patient had breathlessness on exertion for the past 1 year (NYHA class 2), without associated symptoms of chest pain, palpitation, or fatigue. The patient was a non-smoker, with no significant occupational exposure to smoke or fumes and no significant history in childhood. There was no history of snoring at night, no residence at high altitude or other co-morbidities including chronic renal disease, and no significant family history or drug intake. There was no history of loss of weight or loss of appetite. ECG was showing features of '*P*' *pulmonale*, without any ST/T changes. CXR showed decreased right lung field with mediastinal shift to right suggestive of hypoplastic right lung (Fig. 2). As measurement of RBC mass was not feasible, serum erythropoietin (EPO) levels were sent, and the patient was planned for 2D echocardiography. The patient underwent phlebotomy, after which the hemoglobin dropped to 16.8 g/dl, only to increase later to a baseline value of 19.8 g/dl. 2D echocardiography revealed features suspective of rheumatic mitral valve disease along with dilated right atrium and right ventricle with right ventricular systolic pressure of 100 mm Hg and severe tricuspid regurgitation, consistent with severe pulmonary hypertension. Subsequently, serum EPO levels were found to be elevated to 190 mIU/ml (4.30–29).

Hence, further workup was planned to evaluate the causes of secondary polycythemia, and the patient was started on anti-platelets. ABG showed normal indices, though 6-min walk test had a fall in saturation of oxygen to >3%. Meth-hemoglobin and carboxy-hemoglobin assays were within normal limits. Trans-esophageal echocardiography was planned to confirm the rheumatic

Fig. 1 Clubbing of fingers

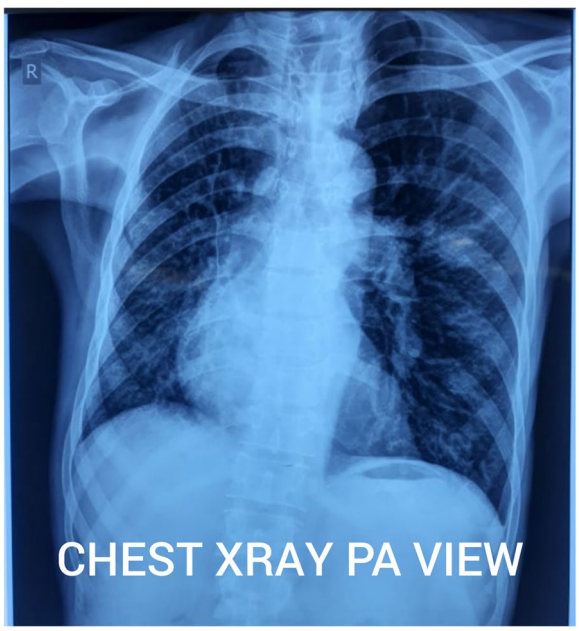

Fig. 2 Chest x-ray PA view

mitral valve findings seen in transthoracic echocardiography, which ruled out rheumatic heart disease. Ultrasonography of the abdomen was done, which showed heterogeneously hyperechoic wedge-shaped lesion in the spleen consistent with splenic infarct, which was attributed to polycythemia induced hypercoagulation. Having ruled out major lung pathology, cyanotic heart diseases, and obvious abdominal mass, finally, the possibility of cardiopulmonary vascular shunt was considered. CT cardiac with pulmonary angiogram was performed, which showed anomalous origin of the right pulmonary artery (AORPA) from the ascending aorta with tortuous patent ductus arteriosus (PDA) and pulmonary hypertension (Fig. 3 a, b).

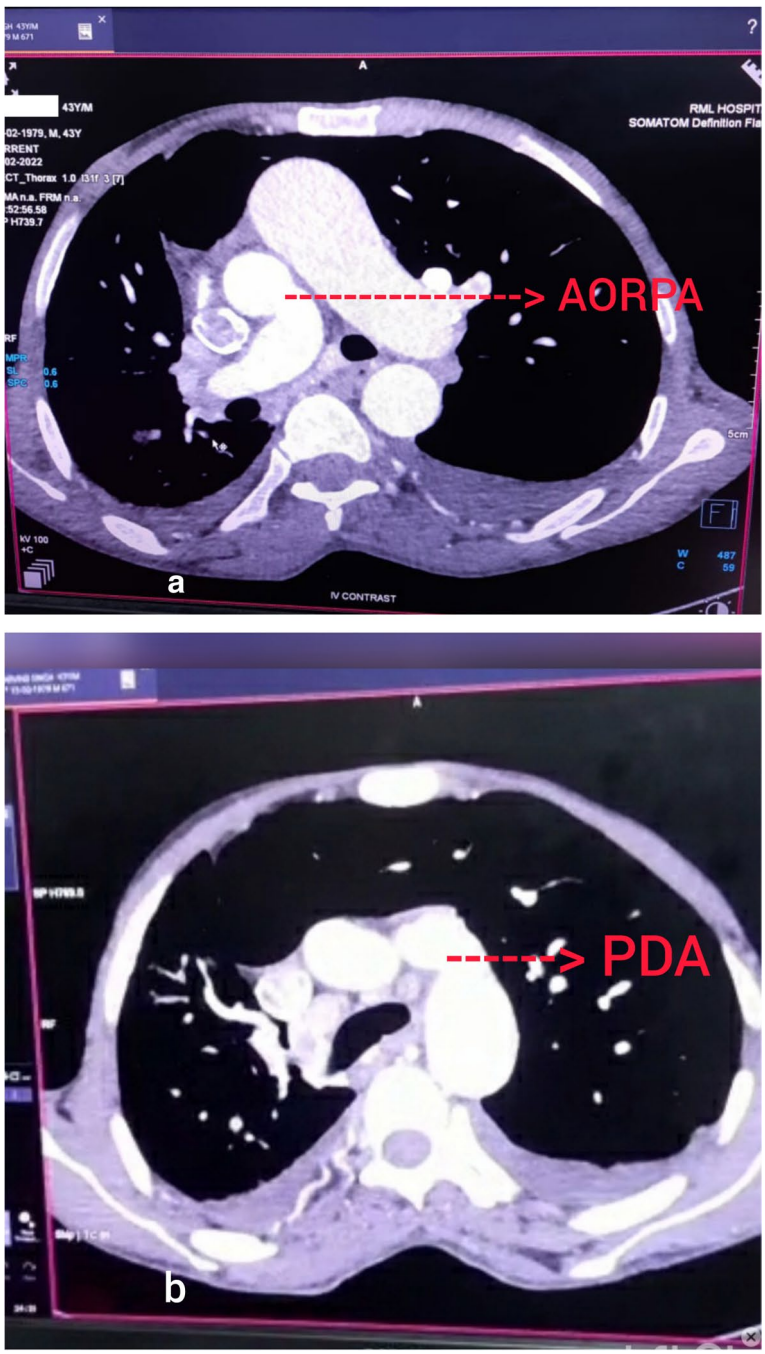

Fig. 3 CT cardiac with pulmonary angiogram. **a** AORPA **b** PDA

Secondary polycythemia, in our patient, was due to right to left shunt causing Eisenmenger phenomenon, following pulmonary hypertension, making it one of the rarest presentation in adults. The patient was planned for regular phlebotomy and is under follow-up. Following phlebotomy, patient hemoglobin dropped to 15 g/dl.

Discussion

COPD, congenital heart disease, and severe pulmonary hypertension are found to be the most common etiologies of secondary polycythemia; though exact data regarding the epidemiology are lacking [4]. Secondary polycythemia associated with cyanotic congenital heart disease is a physiological and desirable response to chronic hypoxia [5]. The presence of the right pulmonary artery originating from the ascending aorta, referred to as an anomalous origin of the right pulmonary artery from the ascending aorta (AORPA), is a rare congenital malformation accounting for 0.1% of congenital heart disease [6]. Ninety-five percent of AORPA cases are diagnosed during infancy, and only 5% are reported in adults. The AORPA is usually diagnosed with other congenital cardiac anomalies; the most common of which is patent ductus arteriosus (PDA), which was noted in our patient also [7]. AORPA has been classified as type IIb under the classification of aortopulmonary windows by Berry et al. and is prone to Eisenmenger syndrome if left uncorrected [8].

In a case report by Haywood et al., cardiac catheterization in an adult AORPA showed diminished blood oxygen saturation of 80% in left ventricle and 78% in the arch of aorta. This finding shows that right to left shunting through PDA was significant in causing desaturation [7]. There is an inverse relationship between oxygen saturation and red blood cell count: the lower the oxygen saturation, the higher the red blood cell count, thus leading to polycythemia. Due to the chronic slow progressive hypoxemia, patients with Eisenmenger syndrome are prone to have secondary polycythemia, and even have a decreased amount of platelets, as seen in our case [9].

The anomaly created 2 separate blood circuits to each lung—the left lung receiving all of systemic blood volume from the right ventricle and the right lung receiving oxygenated blood from the left ventricle. This lifelong systemic flow to the left lung and the transmission of systemic pressure from the left ventricle to right lung caused pulmonary hypertension [7]. Patients with AORPA commonly present with dyspnea and cyanosis with cardiac murmur during auscultation and can present lately with complications like pulmonary hypertension or heart failure, though our patient was relatively asymptomatic and was diagnosed during the workup of incidental secondary polycythemia.

There are reports of aortopulmonary window with Eisenmenger syndrome causing polycythemia, with initial presentation related to cardiac pathology [10, 11]. Mortality rate of patients with AORPA was found to exceed 80% during the first year of life, making the chances of survival to adulthood less likely [12]. CT cardiac angiogram is considered the gold standard of diagnosis and surgical intervention being the main treatment of AORPA [6].

Conclusion

AORPA, being a rare congenital anomaly, has much lesser prevalence among adults, making our presentation one of the rarest disease but presenting as a relatively common scenario. Our patient had features of severe pulmonary hypertension, making the chances of surgical correction impossible. The patient was planned for regular phlebotomy, to prevent complications of erythrocytosis, though controversial [13] and is under follow-up. Thus, in the evaluation of secondary polycythemia, the possibility of underlying asymptomatic cardiopulmonary shunt must be considered, though rare.

Abbreviations

AORPA: Anomalous origin of the right pulmonary artery; CT: Computed tomography; CXR: Chest x-ray; ECG: Electro-cardiograph; EPO: Erythropoietin; PDA: Patent ductus arteriosus; RBC: Red blood cells; WHO: World Health Organization.

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Authors' contributions

All authors contributed in the management of the patient and in preparation, proof-reading, and submission of the article. The authors read and approved the final manuscript.

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Availability of data and materials

Not applicable.

Declarations

Ethics approval and consent to participate

Written informed consent was obtained to participate. Ethical approval not applicable for case reports as per institutional committee. Proper written consent from the patient has been obtained in the patient's own language.

Consent for publication

Written informed consent to publish this case was obtained from the patient.

Competing interests

The authors declare that they have no competing interests.

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