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Case Report Open Access

Atypical Complications of Huntington Chorea Disease

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Abstract

Neurodegenerative diseases are defined as hereditary or acquired conditions which are characterized by progressive nervous system dysfunction. They include diseases such as Alzheimer's Disease and other dementias, Huntington's Disease, and so on. We report a case of 59-years-old man with Huntington Chorea admitted to our department for dyspnoea in massive pulmonary embolism due to a migration of a part of thrombus from deep venous thrombosis situs. He had recent history of pneumonia and severe hypomobility, and the thrombophilic screening showed a deficiency of C protein and S protein. While the diagnostic workup had been done, we discovered severe coronary disease, patent foramen ovale and paradoxical embolism.

Keywords: Thrombosis; Pulmonary embolism; Patent foramen ovale; Atrial septal defect

Introduction

Foramen ovale, also called Foramen Botalli or Ostium Secundum of Born, is an anatomical interatrial communication which links the right atrium to the left atrium in the fetal heart [1]. Histologically, it is a fibromuscolar flapping tissue which crescent in the limbus of the fossa ovalis. Following the birth, when there is an increased blood return to the left atrium because the lungs have expanded, the septum primum should normally closed against the left side of the limbus of the fossa ovalis. The Patent Foramen Ovale (PFO) is an atrial septal defect: It is one of the most common intra-cardiac anomaly. Its incidence is 10-15% of the normal population and 27% of the autopsy. It was the first congenital cardiac defect to be repaired by using cardiopulmonary bypass. In approximately 25% of individuals, the foramen ovale remains patent for life. In these persons there is a potential condition of paradoxical embolism of air, thrombotic materials and fat [1].

Paradoxical Embolism (PDE) is defined as a coexistence of pulmonary embolism and systemic arterial embolism [2]. The incidence of paradoxical emboli is relatively low: Fewer than 2% of all cases of systemic arterial emboli have been described. PFO has been reported to be an important risk factor for PDE because of right-to-left-shunting. PDE is frequently associated with cryptogenic stroke as well as peripheral embolism, brain abscess, and decompression sickness in underwater divers. Other clinical manifestations of PDE that have been described include renal infarction, retina artery occlusion and myocardial infarction [3-9].

Case Report

A 59-years-old man with a medical history of Huntington Chorea, recent pneumonia, no family history of cardiovascular disease, was admitted with progressive dyspnoea and a chest pain that had started one day before. On the physical examination, his vital signs showed a pulse rate of 95 bpm, arterial blood pressure of 70/40 mmHg, respiratory rate of 24 breaths/min, and an oxygen saturation of 88%, breathing room air. He also had jugular venous distension and regular but distant heart sounds, with neither murmur nor rubs on cardiac auscultation. The lab test showed deficiency of coagulation proteins C and S, the other thrombophilia tests were negative. Chest X-ray showed severe cardiomegaly. ECG showed low voltage QRS, with ischemic changes (ST depression and T inversion in V1-V2-V3). CT diagnosed a massive pulmonary embolism with intraluminal filling defects of pulmonary arteries (Figure 1) which causes a sub-occlusion of right pulmonary, a low density area at the superior splenic tip and a suspected intraluminal filling defect of the left common femoral artery. The CT scan cardiac images detected PFO and thrombus in the atria (Figure 2). A 2D-Transthoracic Echocardiography (TTE) identified a big fluttering thrombus trapped in the foramen ovale which obstructed the left ventricular outflow tract, signs of paradoxical movement of atrial septum, moderate right heart stain (PAPs 70 mmHg), normal left ventricular volume and kinesis. Selective coronary angiography diagnosed a critical stenosis of descending anterior artery. An intraoperative Transesophageal Echocardiography (ETE) revealed a significant PFO with biatrial vermicular floating thrombus causing massive left ventricular outflow tract obstruction. The patient underwent sternotomy, performed a central cannulation for cardiopulmonary bypass, drainage was facilitated by minimal anti-Trendelenburg positioning and snaring of cavas. The RA was opened, we closed directly the PFO and we did a single bypass graft with internal thoracic artery. The postoperative period was no complicated. At the second postoperative day, it was positioned an IVC filter. The patient did a CT before discharging which showed a few residual intraluminal filling defects in the each side of lobar bronchus levels and segmental bronchus levels. The patient was discharged at the 10th postoperative day, the dismissed transthoracic echocardiography showed a normal volume and kinesis of right and left ventricles (pulmonary acceleration time 92 m.sec, E/A=1, PAPs 35 mmHg), a correct closure of foramen ovale without any shunt. The complete thrombophilic screening was reported in Table 1.

Discussion

The neurodegenerative genetic disorder of Huntington chorea affects muscle coordination and leads to mental decline and behavioural

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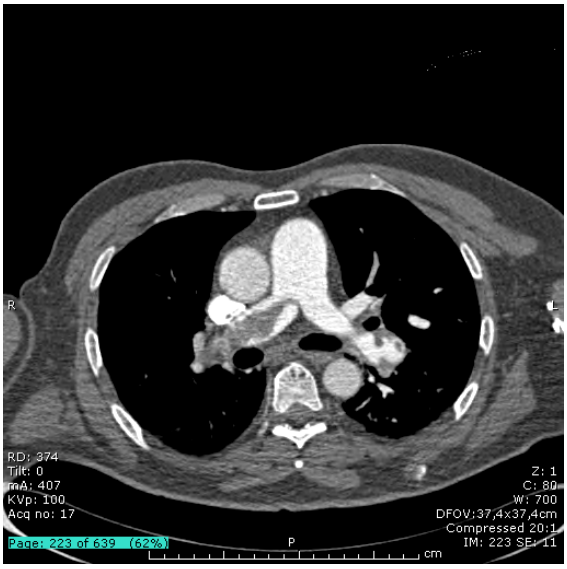

Figure 1: CT scan short-axis documented massive pulmonary embolism with intraluminal filling defects of pulmonary arteries which causes a sub-occlusion of right pulmonary.

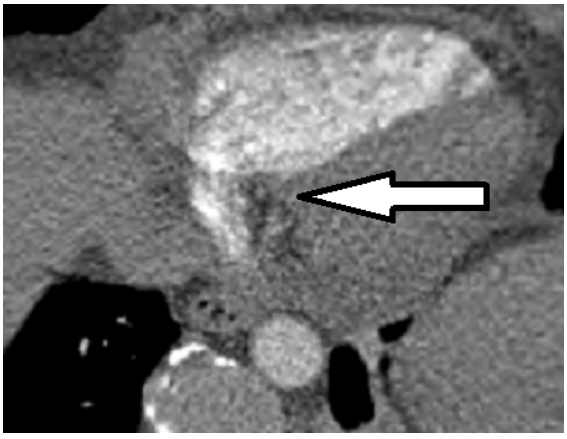

Figure 2: CT scan short-axis showed smoked flow in right atrium and the presence of thrombus (arrow).

Thrombophilia Screening	
Primary thromhophilic condition	**Value**
MTHFR mutation	Not carried
F V Leiden mutation	Not carried
Activated partial thromboplastin time (aPTT)	Normal, 32.5 second
Prothrombin time (PT)	Normal, 12.1 second
Antithrombin	Normal, 79%
Protein C	Deficiency, 40%
Protein S	Deficiency, 55%
Fibrinogen	Normal, 0.2 g/L
Secondary thrombophilic condition	
Immobility	Yes
Infection	Yes
Drug	Yes

Table 1: Thrombophilia screening of the patient.

symptoms. Symptoms of the disease can vary between individuals and affected members of the same family, but usually progress predictably [10-12]. The most frequent life-threatening complications related to this disease are pneumonia and heart disease. Additionally, HD patients have higher incidence of choking and respiratory complications, gastrointestinal diseases (such as cancer of the pancreas), and suicide than the non-HD population [13].

The correlation between HD patient and hypercoagulability is not well described in the scientific literature.

According to Shrivastava et al., Platelets are evinced as a systemic tool in a variety of disorders, including neurodegenerative diseases [14]. Recently Behari and Shrivastava [15], highlight a suspicious role of platelets in the pathophysiology of various neurodegenerative diseases such as Huntington Chorea. They also discussed "how platelets epitomize ultrastructural, morphological, biochemical and molecular changes" and may increase the risk of hypercoagulability.

The scientific literature is lack of paper describing the relationship of Huntington chorea (neurodegenerative disease) and hypercoagulability.

Our case report is an example of multifactorial conditions causing acquired thrombophilic assessment:

1. Drug collateral effects: Tetrabenazine thearapy caused sedation, somnolence and hypotension;
2. The increasing motor deterioration due to Chorea;
3. The hypercoagulability predisposition causes by hypomobility, recent infection (pneumonia) and deficiency of coagulation proteins C and S;

As the results of the hemodynamic changes causing by multifactorial conditions, the pulmonary thromboembolism and the acute events in unknown cardiac diseases (critical coronary disease and patent foramen ovale) have induced respiratory and cardiovascular instability.

According to our experience, the neurodegenerative disease patients may do cardiovascular and thrombophilic screening because their complex therapy and comorbidities in order to decrease life-threatening events and increase their survival.

Conclusion

Our case report is unusual association with unknown cardiac disorders (critical coronary disease and patent foramen ovale) and acquired thrombophilic status (deficiency of C protein, deficiency of S protein) in neurodegenerative disease patient (HD). The early diagnostic workup and the periodic pharmacological check in these patients may reduce comorbidities and recovery hence enhances survival of them.

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