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Simultaneous Acute Arterial and Venous Cerebral Thrombosis and Acute Upper Limb Thrombotic Ischemia due to Combined Uncommon Hereditary Factors for Thrombophilia in Young Adult

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Introduction

Thrombophilia is an important medical problem that carries a great morbidity and mortality especially in young patients and great laboratory workup is usually needed to detect the underlying etiology. Several factors have been claimed to be responsible for thrombosis, some of them are common and others are not, the acquired causes represent the majority while hereditary causes are the minority. The case here is an example for a serious presentation of acute thrombosis that develop simultaneously in several arteries and veins in a previously healthy young adult who showed excellent recovery after treatment.

Case Report

Thirty seven years old male working as accountant, married with two offspring, cigarette smoker for last five years, not known to be diabetic or hypertensive with no previous history of cardiac disease developed transient ischemic attacks in the form of amarousis fugaes on the right eye that resolves spontaneously within minutes, 2 weeks later he developed acute left sided hemiplegia with hemianathesia with upper motor neuron facial and hypoglossal nerve affection. He also suffered bulbar manifestation with weak palatal and pharyngeal reflexes bilaterally, this was associated with severe agonizing headache, vomiting and blurring of vision. At the same time he developed pain and numbness in right hand. On examination he was fully conscious, oriented, irritable and crying from headache. Pulse on left side was 80/min regular, right upper limb was cold, cyanosed with a very weak radial, ulnar and brachial pulse. BP was 160/100 mmHg on left side, right carotid pulse was weak with no bruit. Right lower limb was pale distally with absent popliteal and anterior tibial artery pulsation and a weak posterior tibial pulse. His brother gave a history of MI, DVT and pulmonary embolism at age of 45 years. Urgent CT brain showed brain edema and fundus showed papilledema, ECG, echocardiography, blood glucose, kidney function, ESR and immune profile were normal. Cholesterol was high 232 mg/dL, LDL 170 mg/dL, HDL 34 mg/dL & TG 217 mg/dL. Urgent duplex scanning revealed complete occlusion of right ICA intracranially, right axillary artery severe stenosis (86%) by an acute translucent thrombus with a jet flow at 165 cm/sec, Complete occlusion of left IJV by an acute thrombus and occlusion of right popliteal artery, ATA and proximal segment of PTA by a chronic thrombus with collateral refilling of distal segment of PTA with damped monophasic flow. Urgent MRI, MRA, MRV were done, MRI revealed right basal ganglia infarction, brain edema and multiple ischemic lesions in both hemispheres and brain stem. MRA and MRV showed complete ICA occlusion on the right side, occluded left transverse sinus, sigmoid sinus and IJN. Patient was urgently treated with full dose enoxaparin, aspirin, statin, carbonic anhydrase inhibitor, analgesics and dehydrating measure. Thrombophilia screening revealed a homozygous DD ACE gene abnormality and heterozygous 4 G/5 G allele plasminogen activator inhibitor-1, his brother showed the same abnormalities. Two weeks after treatment patient improved, headache resolved, right axillary artery showed subtotal recanalization with small thrombotic residue reducing lumen by only 10% and right upper limb became warm with adequately felt radial and ulnar with pressure 110 mmHg compared to 120 mmHg on the left side, left IJV re-canalized completely (100%) with no thrombotic residue, neurologically patient became ambulant with grade 4 power in lower limb and 2-3 in upper limb, can perform activity of daily living and most of instrumental activity of daily living independently and he started to return work (Figure 1).

Conclusion

This is a very rare case of acute simultaneous arterial and venous cerebral thrombosis together with acute upper limb thrombosis and chronic lower limb thrombosis in a previously healthy young adult proved to have two rare hereditary factors for thrombophilia that was also recorded in his brother.

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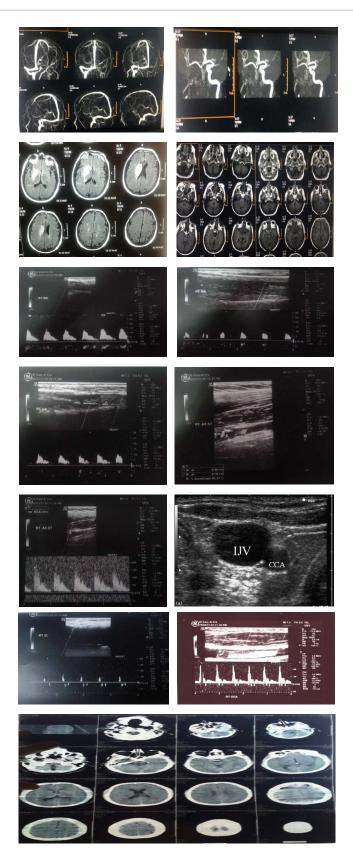


Figure 1: Serious presentation of acute thrombosis.