Multiplicity of hereditary thrombophilic factors inherited from both parents results in child catastrophe

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Here we present a rare case of hereditary thrombophilia due to transfer of multiple hereditary thrombophilic factors from father and mother with lethal outcome.

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Introduction

Hereditary thrombophilia is an important medical problem that carries a great morbidity and mortality risk, and great laboratory workup is usually needed to detect the underlying etiology. Several hereditary factors have been claimed to be responsible for thrombosis; some of them are common, whereas others are not [1].

Case report

A 22-year-old woman who was married to her cousin presented with pain and swelling of left lower limb 4 months after her first normal vaginal delivery. She sought medical advice and was diagnosed as having acute left femoropopliteal deep venous thrombosis. She was treated with enoxaparin and then warfarin for 1 year. She presented to our hospital to reassess her limb, as swelling did not resolve completely. Clinically there was swelling of the left calf (5 cm more than right). Duplex examination revealed remnants of old adherent thrombi at femoropopliteal vein with postphlebitic changes. Detailed history revealed loss of her first baby owing to bilateral upper and lower limb acute arterial thrombosis. She was kept on intrauterine device (IUD) for contraception waiting counseling about further pregnancies.

Her child was born by normal vaginal delivery with normal physical and mental functions, and she passed a healthy period with normal milestones and activity for the first 4 months. Then the child started to develop bluish discoloration of fingers and toes and was crying all the time. Urgent medical consultation in emergency room (ER) revealed rapid pulse, normal blood pressure, normal color of lips and tongue, and normal heart, chest, and abdominal examination. All digits were cold and bluish, with absent radial, ulnar, and tibial arteries.

The child was urgently admitted to ICU and was investigated by basic laboratory workup, blood gases, ECG, echocardiography, and arterial duplex scanning. Results revealed normal arterial oxygen saturation (98%), sinus tachycardia, normal echocardiographic study, and bilateral acute thrombotic occlusion of radial, ulnar, and tibial arteries together with the palmer and planter arches.

A blood sample was sent to test all hereditary factors of thrombophilia, and she was treated with intravenous heparin infusion; however, thrombosis extends proximally along limbs, and the cerebral arteries started to be occluded. She became comatose and had respiratory seizure, and all measures to resuscitate failed.

Table 1 Inherited factors of thrombophilia in the studied child & parents

Factors	Mother	Father	Child
Factor V (G1691A)	Heterozygous		Heterozygous
Factor V (H 1299R)	Heterozygous		Heterozygous
PAI-1 (4G/5G)	Heterozygous	Homozygous	Heterozygous
MTHFR (A1298C)	Homozygous	Heterozygous	Heterozygous
ACE I/D	Heterozygous	Homozygous	Heterozygous
Apo E 112TGC (Cys)	Homozygous		Homozygous

ACE, angiotensin converting enzyme.

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Conclusion

This child developed widespread acute arterial thrombosis secondary to the presence of six inherited factors of thrombophilia. The disease was very aggressive and terminated child life. Parents were also found to have multiple risk factors (mother has six and father has three); in addition, the mother started to develop venous occlusion. They contacted center of preventive medicine for counseling for risk in further pregnancies and was informed that risk of transmission of abnormal factors of thrombophilia to the next baby is high and they decided to postpone pregnancy and to take enough time to think (Table 1).

Discussion

This case represents a very rare case of medical catastrophe of unprovoked multiple simultaneous upper and lower limb arterial thrombosis in a child with progression of thrombosis proximally and also occlusion of the cerebral arteries despite the use of anticoagulants. The thrombosis was attributed to the presence of six inherited factors for thrombophilia from both parents. Moreover, the mother developed lower limb venous thrombosis with incomplete recanalization after 1 year treatment with anticoagulants. Although venous thrombosis is much more common than arterial thrombosis even in presence of hereditary factors, this child had only extensive arterial thrombosis. The mother was found to have also six abnormal hereditary factors for thrombophilia, but the only presentation was postpartum deep venous thrombosis and she did not develop any arterial thrombosis. On the contrary, the father is harboring three abnormal hereditary factors for thrombophilia and was healthy with neither arterial nor venous thrombotic event. So presentation of thrombosis vary from patient to patient even with presence of similar hereditary risk factors for thrombosis, some patients remain asymptomatic, whereas others develop venous thrombosis and/or arterial thrombosis.Previous studies reported high incidence of stroke in children with factor V Leiden mutation [1–3], whereas others showed that homozygous angiotensin converting enzyme (ACE) abnormality also increases venous thrombosis [4].

This case scenario clarifies the importance of premarital screening program for hereditary thrombophilia especially in presence of positive consanguinity and if results are positive, and counseling at the preventive medical center should be obtained.

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Conflicts of interest

There are no conflicts of interest.

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