Thalassemia intermedia, inherited thrombophilia, and intrauterine growth restriction

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A 26-year-old splenectomised, nulliparous woman with beta-thalassemia intermedia presented after a spontaneous pregnancy. Her personal and family history of thrombosis was negative. She was started on folic acid and calcium supplements, prophylactic anticoagulation, and given regular transfusions. At 16 weeks of gestation she developed superficial vein thrombosis in her left leg and was given a therapeutic dose of subcutaneous low molecular weight heparin (LMWH). Further investigations showed homozygosity for C677T 5,10-methylenetetrahydrolfolate reductase (MTHFR) mutation.

In the third trimester an ultrasonographic scan revealed intrauterine growth restriction (IUGR). At 35 weeks of gestation she gave birth to a healthy neonate weighing 1915 g (below the 5th percentile for this gestational age). Subcutaneous LMWH was reintroduced after delivery.

Beta-thalassemia is a congenital anemia characterized by either partial (intermedia, TI) or complete (major, TM) deficiency in the production of beta-globin chains, which determines the clinical outcome [1]. TI is also associated with gestational complications that are mainly attributed to maternal anemia [1,2].

There is no agreement on the overall management of thromboembolic disease, as well as gestational complications of women with TI [1,2]. Prophylactic anticoagulation did not protect the patient from venous thrombosis, while therapeutic anticoagulation and regular transfusions did not prevent the development of IUGR.

It is unclear whether homozygosity for C677T MTHFR mutation contributed to IUGR development. It is likely that administration of a standard dose of folate, in a patient with increased requirements due to chronic anemia, was inadequate to overcome the genetic defect resulting in hyperhomocysteinemia. Hyperhomocysteinemia is a risk factor for placental abruption and pre-eclampsia, due to placental vasculopathy [3]. Although hyperhomocysteinemia does not increase the risk of IUGR, in the presence of other thrombophilic factors it may contribute to its development.

Pregnant women with TI should have a thorough assessment in order to determine additional thrombophilic factors and to individualize their treatment accordingly.

References

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