Methylenetetrahydrofolate reductase C677T gene mutation prevalence and its contribution with other thrombophilic factors in pediatric cases with portal vein thrombosis

Absract

The relatively common mutation C677T of the methylenetetrahydrofolate reductase (MTFHR) gene has been implicated to contribute, with other factors, in the pathogenesis of vascular thrombosis. **Aim of Work:** To assess the prevalence of MTFHR C677T gene mutation and other thrombophilic factors as risk factors in the development of portal vein thrombosis (PVT) in the Egyptian children. **Patients and Methods:** Forty children with PVT were enrolled in the study, in addition to 20 age- and sex-matched controls. Molecular studies of MTFHR C677T gene mutation, as well as prothrombin G20210A and factor V Leiden (FVL) were carried out. Assays for Protein C, protein S, antithrombin III and activated protein C resistance (APCR) were performed. **Results:** Twenty-seven patients had detectable hereditary thrombophilia (67.5%); four (10%) had MTFHR C677T gene mutation (2 of them were homozygous), 12 (30%) had FVL mutation, 11(27.5%) had protein C deficiency, 6 (15%) had prothrombin G20210A mutation, one (2.5%) had antithrombin III deficiency and none had protein S deficiency. Five children had more than one defect. **Conclusion:** Involvement of MTFHR C677T gene mutation in the pathogenesis of thrombophilic PVT can only be considered if it is homozygous and/or associated with other risk factors as FVL mutation. However FVL mutation still represents the commonest hereditary thrombophilia associated with PVT.