ATYPICAL PRESENTATION OF PERIVENTRICULAR HAEMORRHAGIC INFARCTION IN THE PRESENCE OF THROMBOPHILIA

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Introduction: Periventricular haemorrhagic infarction (PVHI) is a complication of preterm birth associated with cardiorespiratory instability. The role of thrombophilia as a possible additional risk factor in infants with an atypical presentation when PVHI was of presumed antenatal onset or > 96 h after birth in the absence of a preceding sudden deterioration, has not been investigated extensively.

Objective: To report thrombophilic mutations in preterm infants with an atypical presentation of PVHI.

Methods: From January 2005 - December 2010 data of infants (gestational age 28.7 to 36.7 weeks, median 30.6 weeks; birth weight 580 to 3090 gram, median 1300 gram) who developed an atypical PVHI were collected. Mutation analysis of the factor V Leiden (G1691A), prothrombin (G20210A) gene and C677T and A1298C polymorphisms in the MTHFR gene was performed, as well as the plasma lipoprotein(a) and homocysteine levels.

Results: Twenty-one preterm infants presented with an atypical PVHI. Among these infants a heterozygous factor V Leiden mutation was found in seven (33%) infants and their mothers (population incidence is 3-5%), in one concomitant with a heterozygous prothrombin gene mutation and one had an isolated heterozygous prothrombin gene mutation. C677T or A1298C polymorphism in the MTHFR gene was present in all these 8 infants. In two siblings, a mutation in *COL4A1* gene was found.

Conclusions: An atypical presentation of PVHI in preterm infants is often associated with gene mutations, especially the factor V Leiden mutation.