MTHFR GENE (C677T) HETEROZYGOUS MUTATION AND RECURRENT PREGNANCY LOSSES

T. Barbakadze, M. Janelidze, N. Janelidze, L. Chkonia, T. Charkviani, N. Kochiashvili, N. Pirckhelani

Objectives: It is well known, that MTHFR gene homozygous mutation is the cause of elevated homocysteine level, but clinical cases shows, that MTHFR gene heterozygous mutation is expressed as the high level of homocysteine and it is very frequent cause of recurrent pregnancy losses.

Methods: Under observation is patient 29 years old during her 4th pregnancy, who before admission to our clinic had one physiological delivery and after this two recurrent pregnancy losses. Along with the physical examination patient undergo analysis: determination of Homocystein level in the blood, detection of MTHFR, FV Leiden and Prothrombin gene mutation PCR diagnosis, screening for APS, cytogenetic analysis of couple karyotype. Depending on condition of the patient during treatment we are using - Femibion, Clexan, Cardiomagnyl, Detralex.

Result: Patient B.S. 29 years old with one physiological delivery and after this two pregnancy losses on 7-8th and 20-21th weeks of gestation. Neither the present internal partition of uterus, endometrial polyps nor deformation of the uterus cavity has not been identified. APS was not detected. FV Leiden, Prothrombin and MTHFR(A1298C) genes mutation were excluded. MTHFR gene (C677T) heterozygous mutation was detected. Homocysteine level we determined on the 7-8th weeks of gestation and the result was 11 umol/l (N- less than 9,9 umol/l); On the 20th weeks of gestation and the result was 4,5 umol/l (N-less than umol/l). From 5th weeks of pregnancy she is taking Femibion 1 and after 12th weeks of gestation Femibion 2, according the coagulation she is treated by Clexan, Cardiomagnyl and after 20th weeks of gestation she is taking Detralex. Now the day she is 30th weeks of gestation and pregnancy is progressing according gestational age. Conclusion: Case report shows, that MTHFR gene (C677T) heterozygous mutation is cause of slightly elevated Homocystein level. Administration of Femibion 1 and Femibion 2, which contain 200 mkg L-5-MTHF(Metafolin), for correction of Homocystein level, gives very good clinical result. So women affected by MTHFR heterozygous mutation can be benefit from this.

REFERENCES

Ben L. MTHFR Mutations with low Homocysteine a Problem? Yes, 2011, MTHFR.net
Van Beynum L et a.: eur Heart J 2010 ; 21(4) : 464-71