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Impracticality and inefficiency of first trimester population screening for birth defects. G.C. Cunningham, B. Currier and L. Feuchtbaum. California Department of Health Services, Berkeley, California.

Modeling the expectations of detection rate for all birth defects based on characteristics of prenatal care delivery in California, the reported detection rates and the logistics and practicalities of testing lead to the conclusion that second trimester screening is preferable. Cost per birth defect detected is lowest and detection rates are comparable with second trimester screening using multiple serum markers as compared to ultrasound with or without serum markers in the first trimester.

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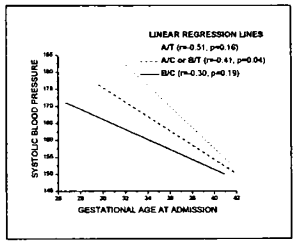
MTHFR mutation and a NOS3 polymorphism influence blood pressure characteristics in preeclampsia. S. Riskin-Mashiah[1], A. Pellicena[1], L.A. Hefler[1], C.B. Tempfer[1], A.R. Gregg[1,2]. [1]Dept. Ob/Gyn and [2]Human and Molecular Genetics, Baylor College of Medicine, Houston, TX.

Pathways involving nitric oxide (NO) and homocystine have been implicated in preeclampsia. A bi-allelic polymorphism within intron 4 (I-4) of the endothelial NO synthase gene (NOS3) and a missense mutation (C677T) in the methylenetetrahydrofolate reductase gene (MTHFR) have been independently associated with this disease. We have established an association between a NOS3 I-4 A allele and our preeclamptic women. In this study we evaluated the same women for an association with the MTHFR mutation. We also studied the combined effect of the NOS3 I-4 polymorphism and the MTHFR mutation on the clinical phenotype.

54 preeclamptic women (PET) and 37 control women (CTRL) were genotyped. Genotyping was performed by PCR (I-4) or PCR-RFLP analysis (MTHFR). PCR products were scored on a 1.5% (NOS3) and 3% (MTHFR) agarose gel. Women with at least one NOS3 I-4 A allele were combined for analysis otherwise women were scored as B. MTHFR scoring was either C (wildtype) or T (heterozygous). No woman homozygous for the MTHFR mutation was identified.

38.9% of PET women and 43.2% of CTRL women were MTHFR–T (p>.05). Mean gestational age at admission was the same for both groups. 9 women with PET were scored as A/T. These women had higher systolic blood pressure on admission, after adjusting for gestational age, compared to PET women scored as B/C (p=.05, ANCOVA). Women with PET scored as A/C or B/T had systolic blood pressure in between that observed among compound heterozygous women and those with wild type alleles at both loci (figure).



In this preliminary study, MTHFR C677T mutation was not associated with preeclampsia. Among women with preeclampsia, the compound heterozygous genotype, might be predictive of a more severe clinical course.