592 CONTINGENCY BASED PROVISION OF A GENETIC SONOGRAM FOR WOMEN RECEIVING SECOND TRIMESTER MATERNAL SERUM DOWN SYNDROME SCREENING

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OBJECTIVE: To determine to what extent the referral of more women for the second trimester genetic sonogram (GS) would improve the net performance of second trimester screening for Down syndrome.

STUDY DESIGN: Second trimester maternal serum quadruple test results (MS-AFP, uE3, hCG, and INH-A) were computer simulated for a population of women with maternal ages seen in the US pregnancy population in 2000. Those women with risks greater than a specified level had their risk modified by a GS that, by itself, had a 53.1% detection rate (DR) and 14.3% false-positive rate (FPR) (Smith-Bindman et al; Prenat Diagn. 2007; 27;535-44). Final risks were classified as positive or negative on the basis of a second trimester cut-off of 1:270 or 1:190. Efficacy of this protocol was compared with serum screening alone, and with combined serum and GS screening for all women.

RESULTS: When increasing numbers of women receive a genetic sonogram, the net detection rate for the combined serum and ultrasound protocol is improved and can exceed that obtained by serum screening alone (Table). The FPR of this combined screen is lower than serum screening alone, regardless of the number of women receiving the genetic sonogram.

CONCLUSION: Increasing the number of women who receive the genetic sonogram can improve the detection of affected pregnancies without creating unacceptable false-positive rates.

GS indication	Women needing GS (%)	Amnio 1:270 DR, FPR (%)	Amnio 1:190 DR, FPR (%)
No sonogram	0	82.8, 6.4	79.2, 4.7
>1:190	4.9	76.1, 3.9	74.2, 2.9
>1:270	6.6	73.3, 4.1	76.4, 3.2
>1:350	8.2	79.8, 4.3	77.7, 3.4
>1:430	9.5	80.9, 4.5	79.0, 3.6
>1:510	11.0	81.8, 4.7	79.8, 3.8
No cut-off	100.0	85.1, 5.7	81.2, 4.2

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593 FIRST TRIMESTER T13/18 SCREENING USING MATERNAL DRIED BLOOD FREE BETA HCG, PAPP-A AND NT

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OBJECTIVE: To determine the effectiveness of first trimester T13/18 screening using the combined protocol of free Beta hCG, PAPP-A, and NT in a large dataset.

STUDY DESIGN: Free Beta and PAPP-A MoM values from 66 cases of T13/18 were analyzed. Biochemical screening results were modeled based on the distribution of the 66 T13/18 cases and previously published data on 10,000 unaffected pregnancies. NT screening results were modeled using published T13/18 NT MoM distribution parameters from the Fetal Medicine Foundation. All modeling was based on the distribution of live births in the USA in year 2000.

RESULTS: The median MoM was 0.215 (95% CI: 0.185, 0.315) and 0.32 (95% CI: 0.265,0.385) for free Beta and PAPP-A, respectively. The correlation between free Beta and PAPP-A was 0.3295. There was no significant trend with gestational age for either free Beta (P=0.094) or PAPP-A (P=0.483). The overall screening performance with biochemistry alone was similar to previously published data from the FMF. The false positive rate and detection rate was 1.7% and 82% in this study compared with 1.9% and 84% based on FMF distributions. Adding in NT to the biochemistry results in this study increased detection rate to 94% and reduced the false positive rate to 0.3%.

CONCLUSION: We confirm the published estimates of 0.3% FP rate and 95% detection rate of T13/18 with first trimester free Beta, PAPP-A and NT. Patients undergoing such screening gain the advantage of potential early diagnosis with a very small chance of requiring CVS for diagnostic testing.

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594 PRENATAL COCAINE EXPOSURE AND AUDITORY THRESHOLD

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OBJECTIVE: Background: Prenatal drug & alcohol exposures have been associated with adverse childhood outcomes including auditory dysfunction which in turn can impact child development.

OBJECTIVE: To examine the relation between prenatal cocaine exposure and hearing deficit.

STUDY DESIGN: Substance use was assessed prospectively for urban African American pregnant women. At each prenatal visit, women were interviewed regarding alcohol and drug use during the preceding 2 weeks. Prenatal alcohol exposure level was expressed as oz. of absolute alcohol/day. Cocaine exposure was dichotomized as heavy and persistent user vs. no or "some," with heavy defined as cocaine 2-3 times/week at the initial prenatal visit or at least once/week throughout pregnancy. Classification as persistent required a positive infant or maternal urine drug screen at delivery. Children's auditory threshold was assessed at 7-year follow-up. If children could not hear a tone at 30dB at any of four frequencies (500, 1000, 2000, or 4000), auditory function was considered deficient. Analysis: In a multiple regression, all covariates and prenatal cigarette, marijuana and heroin exposure were entered in the 1st step (if á <.10). Prenatal alcohol and cocaine exposure were entered simultaneously in the 2nd step.

RESULTS: Data were available for 516 children (49% girls). After controlling for multiple covariates, adding prenatal alcohol and cocaine exposure to the model accounted for 5% additional variance. However, a relation was seen only between heavy and/or persisten prenatal cocaine exposure (β=.15, p<.01) and higher auditory threshold. Prenatal alcohol exposure was not significantly related (β=−.06, NS).

CONCLUSION: We have previously demonstrated deficits in language development related to prenatal cocaine. Understanding how cocaine impacts both hearing and language development is critical for developing intervention strategies.

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595 MATERNAL PEROXISOME PROLIFERATOR-ACTIVATOR RECEPTOR ALPHA (PPARA) GENE POLYMORPHISM, RACE/ETHNICITY AND WEIGHT GAIN DURING PREGNANCY

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OBJECTIVE: PPARa is a nuclear receptor that regulates expression of genes involved in lipid metabolism and the transport of fatty acids across placental membranes. A single nucleotide polymorphism (SNP) at position 484 in exon 5 of the PPARa gene results in increased PPARa production. We hypothesized that possession of the PPARa polymorphism would be a risk factor for increased maternal and/or neonatal weight gain during pregnancy.

STUDY DESIGN: 481 women who delivered a singleton baby at NYHQ between August 2005 and April 2007 were enrolled. Buccal swabs were obtained from mothers and newborns shortly after delivery and tested for the PPARa SNP by polymerase chain reaction and endonuclease digestion. Clinical data were obtained after completion of all testing.

RESULTS: The PPARa SNP was much more common in Hispanic women (10 of 108, 9.3%) than in White, Black, Asian and East Indian women (5 of 349, 1.4%) (p = .0004). Among the Hispanics, the polymorphism was present in 10 of 71 (14.1%) women from the Caribbean as opposed to none of 37 from Mexico or Central or South America (p = .0146). The median weight at delivery of Hispanic women with the polymorphism (88.7 kg) was greater than that of other Hispanic women (77.4 kg) (p = .0352). In contrast, there were no differences in median pre-pregnancy weight between Hispanic women who were positive (63.0 kg) or negative (63.9 kg) for this polymorphism. Neonatal birthweights and rates of preterm birth were unrelated to the PPARa genotype.

CONCLUSION: The PPARa SNP is more common in Hispanic women from the Caribbean than in women from other racial/ethnic groups and is associated with an elevated weight gain during pregnancy. Determination of the PPARa genotype in Hispanic women may be useful in focusing nutritional advise during pregnancy.

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