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Session Information

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Abstract Information

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Title: SCREENING FOR ANEUPLOIDY USING MATERNAL BLOOD SPOTS IN THE FIRST AND SECOND TRIMESTER.

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Abstract Content

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Screening for aneuploidy using maternal blood spots in the first and second trimester. *S.B. Melancon¹, S. Demczuk¹, S. Zaor¹, M. Pelletier¹, D.A. Krantz², T.W. Hallahan², J.N. Macri².* 1) Procrea Genetic Services, Mont-Royal, PQ, Canada; 2) NTD Laboratories, Huntington Station, NY, USA.

Prenatal screening for Down syndrome (DS) and open neural tube defects (ONTD) using multiple maternal serum markers is now well established in the second trimester (T2) of pregnancy. Recent data suggest that maternal blood screening in the first trimester (T1) combined with an ultrasound (US) measurement of the fetal nuchal translucency (NT) would allow to reach around 90% sensitivity for DS at a 5% false positive rate. In collaboration with NTD Laboratories, PROCREA has been offering women from Eastern Canada, T1 and T2 maternal screening for DS and ONTD using dried blood spots on filter paper since 1999. NT measurements were selectively offered in T1 and US dating in T2. Before 13 6/7 weeks, PAPP-A and free b-hCG levels were coupled with fetal NT when available. Between 14 and 22 weeks, AFP and free b-hCG were assessed. Following positive screening results, couples were given the choice to follow up in a private or public clinical genetic setting. Until now, 20 640 pregnancies were monitored; 18 970 had T1 (92%) and 1 670 had T2. NT data were provided with 95,5% of T1 samples. There were 38 confirmed cases of DS, 36 in T1 and 2 in T2. In T1, 27/36 (75%) were at risk by blood markers alone, 24/33 (73%) by NT alone and 28/33 (85%) by

combined blood/NT results. In one T2 case of DS, blood markers were negative. Using combined blood/NT data in T1, 32 other confirmed aneuploidies were found to be at risk; trisomy 18 in 8, trisomy 13 in 3, Turner syndrome in 10, triploidy in 8 and rare chromosomal anomalies in 3 more. 3 cases of ONTD were found at US in T1. One trisomy 18 and two ONTD were detected in T2. Our results agree with recent combined T1 screening data in DS and T18. In addition, we confirmed the high sensitivity of combined T1 screening in cases of Turner syndrome and in the other less common aneuploidies as well.

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