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First Trimester Down Syndrome Screening in Twins: Moving From "Pseudo-Risk" to Real Risk. *D.A. Krantz¹, T.W. Hallahan¹, E. Orlandi², C. Rossi², J.W. Larsen³, P.D. Buchanan⁴, V. Klein⁵, J.N. Macri¹.* 1) NTD Laboratories, Huntington Stat, NY; 2) Centro di Diagnosi Prenatale, Palermo, Italy; 3) The George Washington Univ. Medical Center, Washington, DC; 4) GeneCare Medical Genetics Center, Chapel Hill, NC; 5) North Shore Univ., Manhasset, NY.

Objective: To develop a first-trimester Down syndrome screening method for twin pregnancies that factors in chorionicity along with the two nuchal translucency values and the free beta hCG and PAPP-A levels to calculate risks for each individual twin fetus, the risk that at least 1 twin might be affected and the risk that both twins might be affected and to determine if biochemical markers are necessary for this method. Methods: A series of 212 unaffected twin fetuses were used to develop reference data for the method. A Monte Carlo simulation of 100,000 sets of unaffected, discordant for Down syndrome and concordant for Down syndrome twins was performed. Results: In monochorionic pregnancies, at a fixed 5% false positive rate, the detection rate increased from 84% to 94% when biochemistry was included in addition to nuchal translucency while at a fixed 80% detection rate, the false positive rate was reduced from 3.0% to 0.5% by adding in biochemistry. In dichorionic pregnancies, in a population in which 2/3 of twins were dizygotic, the detection rate increased from 78% to 83% for discordant cases of Down syndrome and from 82% to 92% in concordant cases of Down syndrome when biochemistry was included in addition to nuchal translucency while at a fixed 80% detection rate for discordant cases, the false positive rate was reduced from 6.4% to 3.5% after adding in biochemistry. Results were similar for populations with a different percentage of twins that were dizygotic. Conclusions: This new method of screening for Down syndrome in twin pregnancy provides clinicians and patients with additional information compared to the current "pseudo-risk" screening method. Based on the data in this study the method is effective and the use of biochemistry can increase detection and/or reduce false positives compared to ultrasound alone.