

Expert Predicts Routine First-Trimester Screening

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SAN FRANCISCO — First-trimester screening for aneuploidy may soon become the standard of care, Robert H. Ball, M.D., predicted at a meeting on antepartum and intrapartum management sponsored by the University of California, San Francisco.

There is sufficient evidence to support the incorporation of first-trimester testing into prenatal practice in the United States, and the focus now should be on the most effective way to implement this, according to a consensus statement produced at a

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workshop sponsored by the National Institute of Child Health and Human Development in December 2004.

At least seven studies now have shown that detection of aneuploidy can be im-

proved by analyzing a combination of risk factors in the first trimester: maternal age, nuchal translucency measured on ultrasound, and serum testing for β -HCG and PAPP-A.

Women at risk may be offered more definitive testing earlier in pregnancy with this approach, or results of negative first-trimester screens can be integrated with second-trimester serum screening for improved aneuploidy detection in the second trimester.

"This is a big deal, no doubt about it," said Dr. Ball, a perinatologist at UCSF Children's Hospital. Most nuchal translucency screens will be done in prenatal diagnostic centers, not in every ob.gyn.'s office, he said.

Dr. Ball presented data at the meeting from the First and Second Trimester Evaluation of Risk for Aneuploidy (FASTER) trial, in which he was an investigator. The results have been accepted for publication in the *New England Journal of Medicine*.

The study obtained follow-up information for 37,002 births, from an impressive 97% of the women who enrolled and underwent first- and second-trimester screening. Aneuploidy detection rates improved when first-trimester serum screening was added to nuchal translucency measurement, compared with using nuchal translucency alone to assess risk.

Detection improved further when these first-trimester results were integrated with second-trimester serum screening.

The study's size provided the power to analyze subgroups, including results based on the gestational week of screening. The best time for first-trimester screening was week 11: When screening was performed at that time, detection rates were 73%

with nuchal translucency alone and 93% with the combined nuchal translucency/serum screen. After integrating the combined first-trimester screen with second-trimester screening, the detection rate was 98%.

First trimester screening may pose scheduling problems, he predicted. "That's sort of a logistical nightmare, if you think about it," because physicians will be scrambling to get patients into prenatal diagnostic centers before the 11-

week window passes, he said during the meeting.

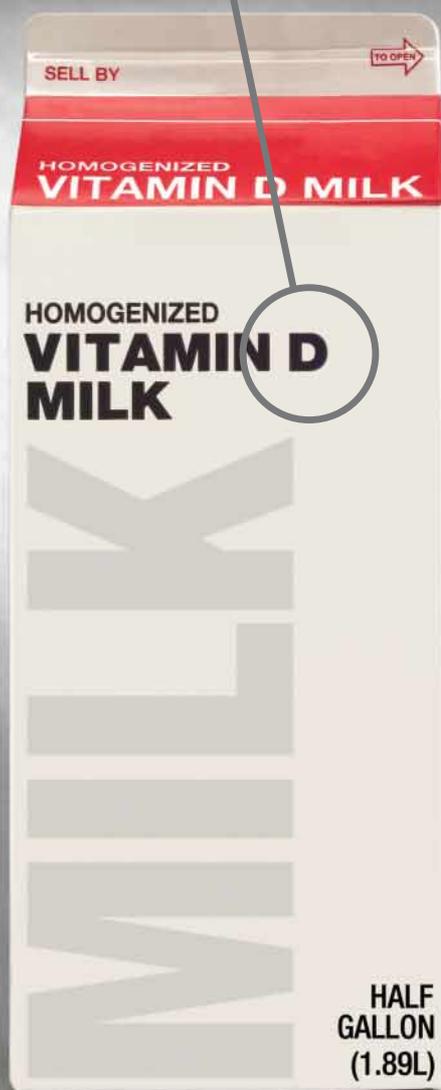
"I can imagine people suing because they didn't get in for their nuchal translucency screening and they have a Down syndrome kid" that might have been detected earlier, Dr. Ball added.

In hopes that the integrated first- and second-trimester screening strategies might obviate the need for genetic sonograms, investigators analyzed the FASTER data with and without genetic

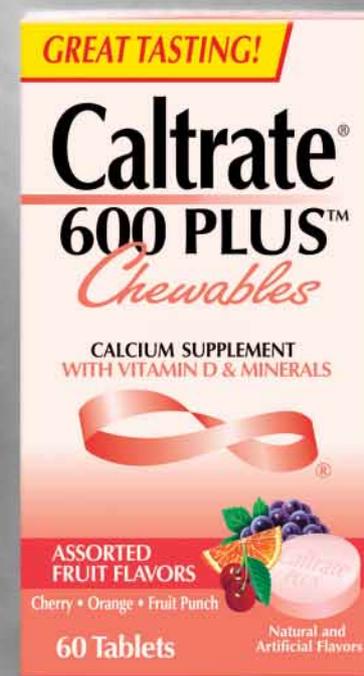
sonogram results. They found that the likelihood of aneuploidy greatly increased with identification of at least one genetic marker on ultrasound. Genetic ultrasound results improved the overall 84% detection rate for aneuploidy using first-trimester screening alone and decreased the false-positive rate.

"Much to my chagrin, having a genetic sonogram after you've had a bucket load of other tests is actually going to be useful," Dr. Ball said. ■

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References: 1. Office of the Surgeon General. *Bone Health and Osteoporosis: A Surgeon General's Report: What Is Bone Disease?* Available at: www.surgeongeneral.gov/library/bonehealth/factsheet1.html. Accessed November 19, 2004. Page 3. 2. *Bone Health and Osteoporosis: A Report of the Surgeon General*. Washington, DC: Office of the Surgeon General, US Dept of Health and Human Services; 2004:12.

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