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"Genetic Pap" Screen for Down Syndrome May Replace Current Tests

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March 28, 2005 (Los Angeles) — A simple, inexpensive, and noninvasive procedure may be able to replace the screening techniques currently used to diagnose Down syndrome (DS) prenatally, according to a presentation given here by Moshe D. Fejgin, MD, at the 52nd annual meeting of the Society for Gynecologic Investigation.

"A simple, early screening test is in the making which is going to be easier and be very attractive to the practitioner," Dr. Fejgin told Medscape. Amniocentesis and chorionic villi sampling (CVS), the current gold standards for diagnosing chromosomal abnormalities prenatally, are invasive and expensive, and they may compromise the viability of the pregnancy. In addition, given a false-negative rate of 20%, the mother may continue a pregnancy assuming that her baby is genetically healthy, when in fact the child has DS.

The limitations of current testing stimulated research into an alternative method that would be just as accurate but less invasive and less expensive. The new method is similar to use of a Papanicolau test to diagnose cervical cancer in nonpregnant women. The physician collects trophoblast cells from the external cervical opening of pregnant women using a cytobrush.

The technique relies on fetal trophoblast cells that are shed into the cervix. "We have a window of opportunity between five and 12 weeks," Dr. Fejgin told meeting attendees. "We can never find fetal cells in the cervix after 12 weeks' gestation." Dr. Fejgin is a professor at Tel Aviv University in Israel.

Cells were centrifuged and slide-mounted. Immunohistochemistry was performed using a panel of antitrophoblast antibodies. The stained cells were identified and scored. Investigators applied fluorescence in situ hybridization (FISH) for chromosomes X and Y to determine sex first. On a second run with the same cells, FISH probes for trisomy 21 were applied to fetal cells from group 1, and probes for trisomies 13 and 18 were applied to fetal cells from group 2.

This fetal Pap test technique is a superior approach to testing fetal cells in maternal blood, which Dr. Fejgin described to meeting attendees as a technique that "has been the future for the last 25 years and doesn't mature."

Dr. Fejgin reported data that he and his colleagues collected through December 2004. The method was first tested in samples taken from women who chose elective termination of their pregnancies (group 1). Eighty-seven percent (252 of 290) of group 1 samples contained the trophoblast cells necessary to run the test. The test correctly identified sex (determined from a placental sample) in 236 (94%) of 252 samples. The method was then used to screen older women scheduled for CVS

or amniocentesis (group 2). The investigators identified the necessary trophoblast cells in 164 (86%) of 190 of group 2 samples. The test determined sex correctly in 146 (89%) of 164.

As of March 1, 2005, 110 women completed pregnancies, and 100 of them delivered healthy babies. The technique identified two cases of trisomy 21 and 2 mosaics (one Turner syndrome, one Klinefelter syndrome). Of the 10 nonhealthy pregnancies, four resulted in missed abortions, two were terminated in light of DS diagnosed using the fetal Pap test, two were terminated in light of ultrasound results, there was one fetal death due to a cord accident, and one child showed signs of developmental delay at six months of age.

Dr. Fejgin told Medscape that his team plans to start clinical trials in the U.S. later this year and that he expects the test kit to become available within the next 18 months. Dr. Fejgin summed up the main advantage of this new technique for Medscape: "You can do this early, at five to six weeks' gestation — before you can see the fetus you can get fetal cells."

The study was sponsored by the Meir Medical Center in Kfar-Saba, Israel. Dr. Fejgin reports no conflicts of interest.

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Reviewed by Gary D. Vogin, MD

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