DNA sequencing of pap smears non-invasive alternative to prenatal genetic screening

Targeted DNA sequencing of fetal cells from Papanicolaou (Pap) smears may offer an improved and earlier route to prenatal genetic screening, according to a report published [Nov. 2].

The novel strategy, which analyzes DNA from trophoblasts that have been shed into the endocervical canal (ECC), correctly distinguished fetal DNA in 20 consecutive samples.

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The three types of prenatal screening tests currently in widespread use have limitations: invasiveness and use later in pregnancy (8 to 20 or more weeks) for amniocentesis and chorionic villus sampling (CVS) and low fetal fraction for cell-free fetal DNA testing….

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Teamed with detection of genetic markers that span the genome, the approach is "a straightforward alternative that uses a Pap smear to capture intact fetal trophoblast cells in numbers sufficient for next-generation sequencing as early as 5 weeks of gestation," the investigators write.

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Pap smear–based prenatal testing offers several potential advantages. A woman's body mass index does not compromise Pap screening results…[James Byrne, MD, chair of the Department of Obstetrics and Gynecology at the Santa Clara Valley Medical Center] lists other advantages. "Reliable testing can be obtained earlier in pregnancy. It is noninvasive, which would provide reassurance to many patients…."

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