Complete genome sequence can be ID'd from amniotic fluid

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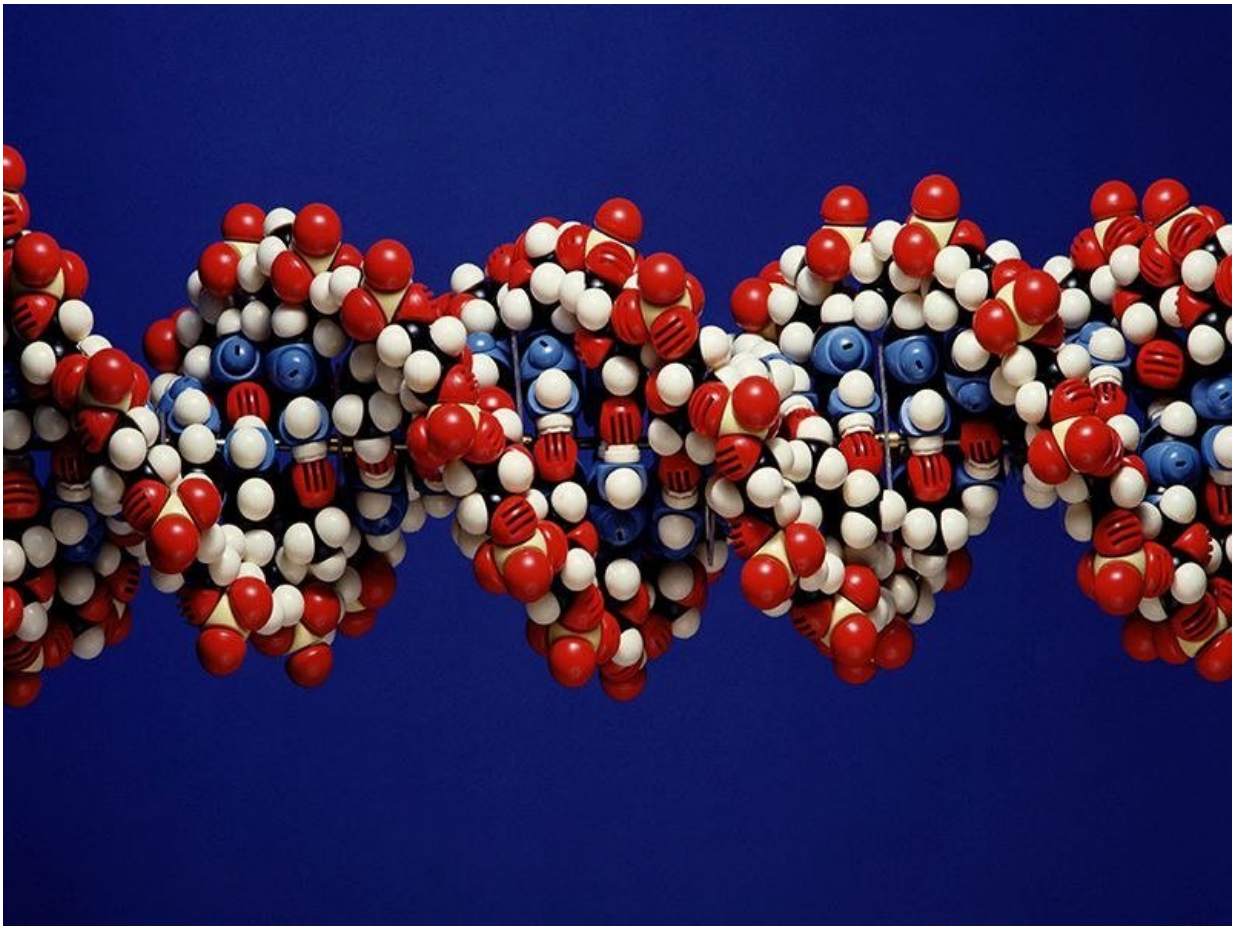

(HealthDay)—The complete genome sequence of fetuses can be

elucidated from amniotic fluid, according to a study published online March 15 in *Clinical Chemistry*.

Qing Mao, from Complete Genomics Inc., in San Jose, Calif., and colleagues isolated cellular DNA and cell-free DNA (cfDNA) from the cell pellet of 31 amniocenteses and conducted sequencing to approximately 50× genome coverage. Long fragment read libraries were generated from DNA isolated from cells from a subset of samples, and sequencing was performed to approximately 100× genome coverage.

The researchers found that concordance of variant calls exceeded 96 percent between the two DNA sources and with parental libraries. Two fetal genomes had potentially detrimental variants in chromodomain helicase DNA binding protein 8 and low-density lipoprotein receptor-related protein 1; variations of these have been linked to autism spectrum disorder and keratosis pilaris atrophicans, respectively. For a variety of diseases, drug sensitivities and carrier information of fetuses were discovered.

"We were able to elucidate the complete genome sequence of 31 fetuses from amniotic fluid and demonstrate that the cfDNA or DNA from the cell pellet can be analyzed with little difference in quality," the authors write. "We believe that current technologies could analyze this material in a highly accurate and complete manner and that analyses like these should be considered for addition to current amniocentesis procedures."

Several authors disclosed financial ties to Complete Genomics Inc.

More information: Abstract
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