Genetics in Medicine
Official Journal of the American College of Medical Genetics

NEWS RELEASE

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Next-Generation Sequencing to Detect Chromosome Abnormalities

BETHESDA, MD—February 8, 2012 | Identifying fetuses with an abnormal numbers of chromosomes using next-generation DNA sequencing could potentially reduce invasive diagnostic procedures and related fetal losses, suggests a study published online this week in *Genetics in Medicine*, the official peer-reviewed journal of the American College of Medical Genetics. Among high-risk pregnancies, sequencing circulating cell-free DNA was found to detect nearly all cases of Down syndrome, Edwards syndrome (trisomy 18), and Patau syndrome (trisomy 13), at a low false-positive rate.

Most ultrasound and biochemical screening programs can detect trisomy 18 and/or trisomy 13 cases at a rate of 60% or higher. Next-generation sequencing of circulating cell-free DNA in maternal plasma has been shown previously to identify nearly all Down syndrome pregnancies. Glenn Palomaki and colleagues have now addressed whether the technique could also be used to identify trisomy 18 and trisomy 13 by testing samples from a cohort of 4,664 pregnant women. For trisomy 18, the detection rate was 100% (59/59) with a false positive rate of 0.28%, and for trisomy 13, the detection rate was 91.7% (11/12) with a false positive rate of 0.97%. By modifying the cut-off defining a positive test for trisomy 18 and trisomy 13, the overall detection rate for all three aneuploidies was 98.9% (280/283) with a false positive rate as low as 0.1% (2/1688). In 0.9% of pregnancies (17/1988, including three trisomy 18 pregnancies) no interpretation could be made.

The authors suggest that if next-generation sequencing is implemented as the next step after a positive screening result, there will be far fewer false positives among high-risk women, and a lower risk of losing an unaffected pregnancy due to unnecessary invasive diagnostic testing.

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About the American College of Medical Genetics and ACMG Foundation

Founded in 1991, the American College of Medical Genetics (www.acmg.net) advances the practice of medical genetics by providing education, resources and a voice for more than 1400 biochemical, clinical, cytogenetic, medical and molecular geneticists, genetic counselors and other healthcare professionals committed to the practice of medical genetics and genomics. ACMG's activities include the development of laboratory and practice standards and guidelines, advocating for quality genetic services in healthcare and in public health, and promoting the development of methods to diagnose, treat and prevent genetic disease. *Genetics in Medicine*, published monthly, is the official ACMG peer-reviewed journal. ACMG's website (www.acmg.net) offers a variety of resources including Policy Statements, Practice Guidelines, Educational Resources, and a Find a Geneticist tool. The educational and public health programs of the American College of Medical Genetics are dependent upon charitable gifts from corporations, foundations, and individuals. The ACMG Foundation (www.acmgfoundation.org), a 501(c)(3) nonprofit organization, is a community of supporters and contributors who understand the importance of medical genetics and genomics and genetic counseling in healthcare. Established in 1992, the ACMG Foundation supports the American College of Medical Genetics' mission to "translate genes into health" by raising funds to promote the profession of medical genetics and genomics to medical students, to support the development of practice guidelines for practicing physicians, to advance the aware-