Toward a faster prenatal test for Down syndrome

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Scientists in California are reporting an advance toward rapid testing for pre-natal detection of Down syndrome and other birth defects that involve an abnormal number of chromosomes.

In a study scheduled for the Oct. 1, 2007 issue of ACS' journal, *Analytical Chemistry*, Stanford University bioengineering professor and Howard Hughes Medical Institute researcher Stephen R. Quake and his graduate student H. Christina Fan point out that most existing pre-natal tests depend on a technique termed karyotyping. It requires a two-week wait for anxious parents, while cells taken with amniocentesis or chorionic villus sampling are grown in laboratory culture and analyzed.

Laboratory studies with the new method produced accurate results within two hours. The test is a variation of the famed polymerase chain reaction (PCR) — the basis of the genetic engineering revolution — which produces thousands of identical copies of minute samples of DNA.

Using a technique known as the digital polymerase chain reaction, Quake and Fan replicated DNA from two cultures of cells growing in the laboratory. One consisted of a normal human cell line and the other had human cells with the Down variant. The digital PCR process allowed the researchers to count DNA molecules from the samples, substituting for the two-week cell culture process traditionally needed to produce enough DNA for karyotyping. With the precision derived from counting individual DNA molecules, researchers then were able to move ahead

without delay and determine which samples had the extra chromosome that indicates Down syndrome.

The digital PCR was performed in a commercially available microfluidic chip. The samples were loaded onto the chip, and then partitioned into thousands of chambers by microscopic mechanical valves. While PCR was performed, fluorescent material in the compartments containing individual DNA molecules lit up like an array of LEDs, while those without DNA did not glow. The technique enabled researchers to confirm the presence of abnormal chromosomes typical of Down syndrome with great accuracy.

Rapid testing alternatives already exist, but they are either too labor-intensive or not applicable to the whole population. “The technique we present in this paper can overcome these limitations. It is rapid and simple. We estimate that the entire procedure from sample collection to result readout would take only a few hours, substantially reducing the anxiety of the expectant parents,” Quake said.

The test is also potentially cheaper than other available methods and semi-automated, reducing the workload of lab personnel. And since the digital PCR technique is based on commercially available lab equipment, any interested physician could use it.

“We are confident that it will work on clinical samples of amniotic fluid or chorionic villus,” Fran said. The next step is to begin clinical trials to evaluate the sensitivity and specificity of the new test. The authors believe the new test could be available in as little as one year.

In addition, Quake cited the possibility that the method could lead to a blood test for Down syndrome. It would involve capturing fetal cells, which leak through the placenta and circulate in the mother’s blood, and analyzing their DNA for abnormalities with digital PCR. Other research

groups are also investigating a digital PCR-based Down syndrome test, he noted.

As a non-invasive test, it would be the safest approach to prenatal diagnosis of Down syndrome, since both amniocentesis and chorionic villus sampling pose risks to the fetus. Quake noted, however, that new techniques to separate the small fraction of fetal DNA in a mother's bloodstream must be developed before a blood test could be developed and tested.

Source: American Chemical Society

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