Down syndrome test breakthrough 'on the horizon'

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A less risky non-invasive procedure based on maternal DNA to diagnose Down syndrome could be generally available to pregnant women by 2013, a Cyprus researcher said Tuesday.

"Down's Syndrome, or Trisomy 21, is the most common cause of mental retardation with an incidence of one in 600 births," Philippos Patsalis of the Institute of Neurology and Genetics in Nicosia told reporters.

"This method can apply to all pregnancies not only those at risk," he added, describing it as "one of the most important milestones in the history of the institute."

He said the cheaper and quicker method of blood sampling rather than collecting fluid from the womb will encourage more couples to take the test and therefore slowly eradicate the disease.

"There is no cure, so this is a test for couples who want to know and prevent it, they are the ones who must decide on the fate of the pregnancy."

Invasive procedures currently used for prenatal diagnosis -- in the 16th week of pregnancy -- pose a one percent risk of foetal loss.

The diagnosis is therefore only made available to high risk women, which fails to catch all cases.

"Over the last few years, scientists have been looking for a new, non-invasive method, offered to all pregnancies and not associated with any risk of miscarriage," the 48-year-old doctor said.

The preliminary report, published in the journal Nature Medicine, is the latest of several recent studies that indicate scientists can detect Down's syndrome through simple DNA blood tests.

Patsalis said the new test would be faster as it could take less than five days to obtain results, while not requiring specialised or complex lab equipment or know-how.

"The test can be easily introduced into every genetic diagnostic lab in the world," he said.

Down's syndrome is caused by having an extra copy of chromosome 21 and the risk increases as a woman gets older.

Currently, pregnant women receive blood tests and ultrasound to find out if the foetus is at risk before invasive action is taken for a definite diagnosis.

The latest research involved scientists in Cyprus, Greece and Britain using a sample of women with Greek ethnic origin.

A larger scale clinical study using 1,000 women of different ethnic origin -- to test the current 100 percent accuracy levels -- is now being prepared to take place in Europe, Japan, Australia and the United States.

Patsalis said his team is also researching modifying the method so it can be used to test for other genetic diseases such as cystic fibrosis.

In the trial, Patsalis' team correctly diagnosed 14 cases where there were

extra copies of chromosome 21, and 26 normal cases.

There is no such commercial test available to pregnant women.

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