

Latest Diagnosis of Down's Syndrome Polymerase Chain Reaction (PCR) Test

Down's syndrome affects one in 650 newborns. The new polymerase chain reaction (PCR) test can confirm if a fetus has Down's syndrome or not in only one to two days. PCR test may replace the conventional karyotyping that keeps to-be mothers and fathers waiting for three weeks.

Down's syndrome is an inborn disease and occurs because of chromosomal abnormalities. The risk of having Down's syndrome increases with the age of the mother.

The traditional karyotyping makes an organized profile of chromosomes. It takes two to three weeks from amniocentesis to having karyotyping results. Karyotyping also requires experienced technicians of cytogenetics with years of training to make an organized profile of chromosomes.

On the other hand, the new PCR test takes only one to two days, and the analysis of 30 to 40 samples can be performed by machines at the same time. Developed in 1990s, the accuracy of PCR test is now 100%.

The Department of Obstetrics and Gynaecology, Faculty of Medicine, HKU is conducting a study to investigate whether the rapid PCR test, which offers shorter waiting time for the result, could help to reduce pregnant women's anxiety.