Prenatal Screening for Down's Syndrome: Present and Future

One out of 650 babies is affected by Down's syndrome which is an inborn disease and occurs because of chromosomal abnormalities. Children affected by Down's syndrome will have different degrees of mental retardation and structural abnormalities. The risk for a fetus having Down's syndrome increases with the age of the mother.

Chorionic villus sampling (CVS) and amniocentesis are 2 invasive diagnostic prenatal tests for the detection of Down's syndrome. Although the two invasive tests are very accurate, they may cause miscarriage. Mothers may choose the safer alternatives to estimate the incidence: nuchal test, blood test and integrated test which combines nuchal test and blood test.

According to a research conducted by the Faculty of Medicine, HKU, the detection rates of nuchal test, blood test & integrated test are about 69%, 70% and 86% respectively. The research finding will serve as a basis for the Hospital Authority to design the prenatal screening for Down's syndrome, which will be implemented in the near future.

Additionally, the fetal nasal bone test by ultrasound is currently under study to determine its possibility and accuracy to estimate the risk of Down's syndrome, as the nasal bone of fetuses with Down's syndrome is found to be shorter than that of normal fetuses.