NON-INVASIVE PRENATAL TESTING

IT IS ONLY NATURAL TO WORRY ABOUT ONE'S CHILD-TO-BE WHEN ONE IS PREGNANT. AFTER ALL, WHICH PARENT would not want the best for their children? Fortunately, there is now a wide array of tests designed to inform and reassure the expecting mother. Known as prenatal tests, these tests allow the parents to identify characteristics of the foetus (such as age, sex, size and physical features) and health problems that the baby may have or may develop soon after birth.

There are two common diagnostic prenatal tests performed today: amniocentesis and chorionic villus sampling (CVS), both of which are invasive procedures where a needle is inserted into the pregnant woman's womb and a small amount of amniotic fluid or placental tissue (trophoblast) is extracted respectively. Both of these tests are designed to test for fetal trisomies, where a problem in cellular division of the embryo causes the foetus to gain an additional set of chromosomes.

Depending on where the error takes place and which chromosome is duplicated, a trisomy could cause chromosomal conditions such as Down Syndrome (trisomy 21), Edwards Syndrome (trisomy 18) and Patau Syndrome (trisomy 13). Both amniocentesis and CVS are thus diagnostic tests performed during pregnancy to allow the mother to determine definitively if her child has any of the above conditions.

However, due to the invasive nature of these procedures, there is a chance that the baby could miscarry as a result of the test. Because the chance of a miscarriage is apporximately 1%, many mothers will often opt not to have the procedure performed for fear that they may lose the baby. This is, of course, an entirely valid and reasonable fear.

Enter non-invasive prenatal testing, a procedure that no longer requires inserting a needle into the womb and risking the baby's health. Instead, blood is simply drawn from the expecting mother and then sent to a laboratory. There, small fragments of DNA that have entered the mother's bloodstream from the foetus are analysed through a bioinformatics procedure to determine if the baby has any of the three most common trisomies mentioned above.

This is a new form of prenatal testing that has only been available in this year, thanks to advances made in genomic sequencing. As the entire procedure is only a blood test, and non-invasive, there is absolutely no risk to either mother or child. The test is also highly sensitive, with a 99% detection rate; in comparison, the next most sensitive non-invasive prenatal test for Down Syndrome - the nuchal translucency test/first trimester screen – has a detection rate of only 84%.

The Choolani Clinic is currently the only clinic in Singapore offering non-invasive prenatal testing (NIPT). The first test was performed in May 2012. ■

PROF MAHESH CHOOLANI | Obstetrician & Gynaecologist
MBBS, MMed (O&G), FAMS, FRCOG, FRANZCOG, Ph.D

The Choolani Clinic
38 Irrawaddy Road, #11-59 Mount Elizabeth Novena Tel: 6570 2000
www.choolaniclinic.com