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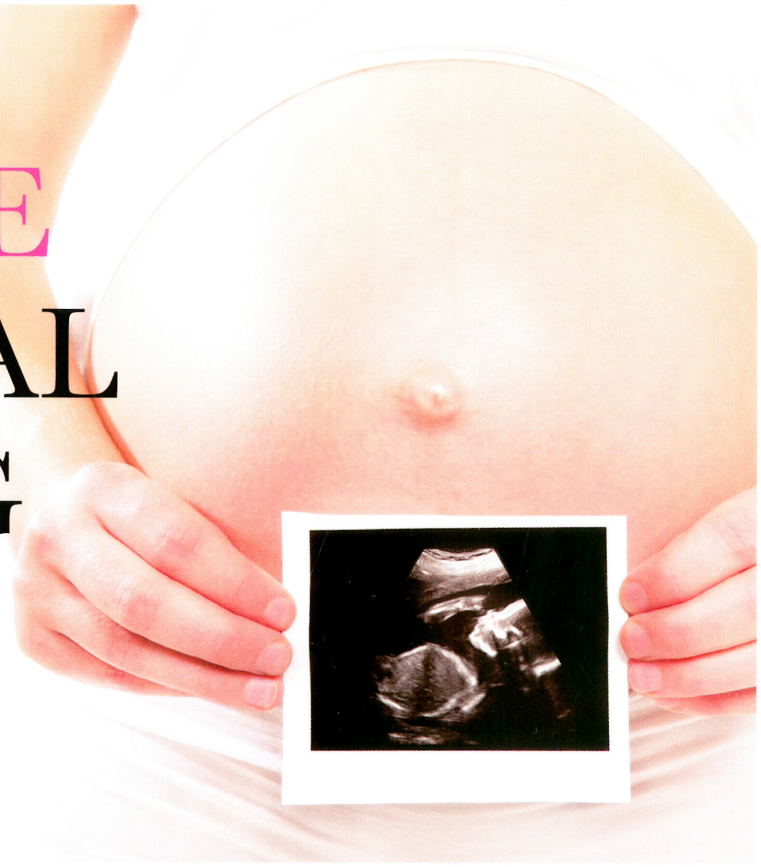
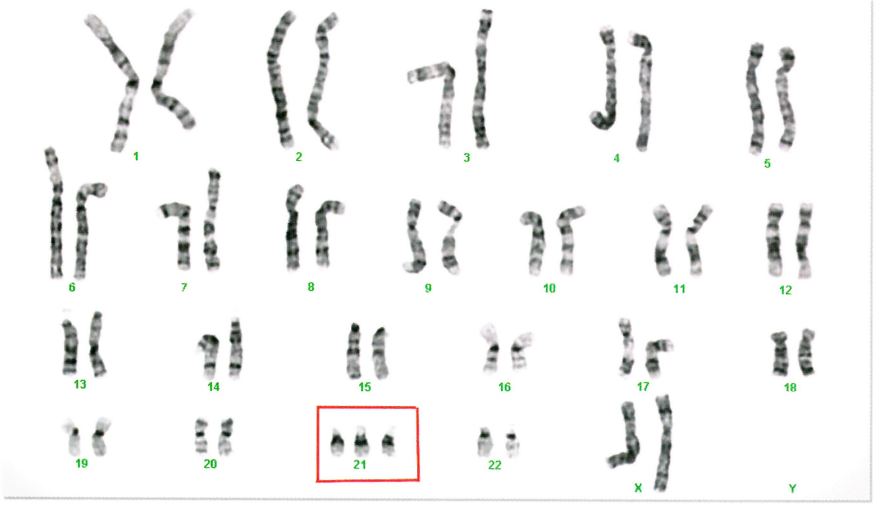
NON-INVASIVE PRENATAL TESTING (NIPT)

DAWN OF A NEW ERA



The normal human genome consists of 23 pairs of chromosomes and forms the basis for all human cell functions. The presence of incorrect number of chromosomes within the cell (known as aneuploidy) can lead to miscarriages, which are very common, occurring in 15% of pregnancies. 1 in every 300 live-births is aneuploidy. An example is Down syndrome which is caused by an extra chromosome 21 (trisomy 21).

Traditionally, **DIAGNOSIS** of aneuploidy can be made prenatally by amniocentesis or chorionic villous sampling (CVS). These procedures involve aspiration of liquor from the amniotic sac or aspiration of cells from the placenta respectively. However, both of these procedures pose risks to the mother and foetus. Most significantly, there is a risk of miscarriage of about 1 in 300 of such procedures.



▲ Extra chromosome 21 in Down syndrome

Risk-free prenatal **SCREENING** for Down syndrome can be done by a blood test from the expectant woman and ultrasound measurement of the skin fold behind the foetal neck (nuchal translucency). An example is the OSCAR test. There is no increased risk of miscarriage from the test itself.

However, one must note that **SCREENING** tests are only risk estimations and not diagnostic. They can only detect risk of Down syndrome up to 90%. Furthermore, about 5% of foetuses deemed high risk by these screening tests are actually normal.