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## Non-invasive prenatal testing (NIPT) a boon for expectant mothers

Aug 2014 , Pank Jit Sin



The incidence of fetal deformities is expected to increase due to the higher age of pregnancy. Medical Tribune interviewed Dr. Mahesh Choolani, non-executive director, Innovations Exchange (INEX), Singapore, about the importance of prenatal testing, in particular the role of non-invasive prenatal testing (NIPT).

**Q: Compared to traditional amniocentesis and CVS tests, how much safer are non-invasive prenatal tests?**

A: The non-invasive prenatal test poses absolutely no physical risk to mother or baby as it analyses cell-free fetal DNA from just 10ml of maternal blood. On the other hand, invasive prenatal tests such as amniocentesis and CVS carry a risk of up to 1% of miscarriage.

**Q: Would you recommend NIPT be carried out on all pregnant women or only on a subset with specific conditions?**

A: NIPT is recommended for women who have the following conditions – high-risk result for first trimester screen; women of maternal age of 35 years and above; women with a previous pregnancy affected by trisomy; and women with a family history of trisomy. We follow the guidelines of the American College of Obstetricians and Gynecologists, which recommends NIPT for women with high-risk pregnancies.

**Q: How does the iGeneScreen (a NIPT test kit licensed to INEX) test achieve such a high degree of accuracy?**

A: The iGeneScreen is considered one of the more accurate tests, with detection rates of over 99% for Down Syndrome (trisomy 21), Edwards Syndrome (trisomy 18) and Patau Syndrome (trisomy 13).

iGeneScreen is based on a next-generation sequencing technology called massively parallel sequencing (MPS), which is able to perform up to 5 million reads to achieve the highest accuracy.

It is also important to note that unlike other NIPT tests which are based on small sample sizes, iGeneScreen is the only test that has been validated on an all-comer population of 11,105 pregnant women, the largest study for any NIPT to date.

The study was conducted on an actual screening population at large, instead of extrapolated from a highly selected sample

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set with matched controls. Therefore, the positive predictive value (PPV) and negative predictive value (NPV) of the trisomies tested are more likely to be a true representation of the test performance.

**Q: Where can patients get access to this test?**

A: In Malaysia, we have partnerships with major labs and hospitals, so patients can ask their doctors about access to the test. iGeneScreen is also available at Quantum Diagnostics. Our Singapore lab facility will be opening in early 2015, so patients will be able to receive faster turnaround time on their reports.

**Q: Apart from iGenescreen, what other NIPTs are there?**

A: There are several other tests from the US, but samples have to be sent to the US for processing. iGeneScreen samples are processed in Asia.



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