Latest study finds BGI's NIFTY test performs best in noninvasive prenatal screening

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Lab (Test Name)		%	True positive	False positive
Sequenom (MaterniT21, SafeT21)	121	44%	98 (81%)	23 (19%)
Ariosa (Harmony)	25	9%	18 (72%)	7 (28%)
Natera (Panorama)	40	14%	32 (80%)	8 (20%)
Illumina (Verifi)	44	16%	35 (80%)	9 (20%)
BGI (NIFTY)	47	17%	41 (87%)	6 (13%)
Integrated Genetics (Informaseq)	1	<1%	0	1 (100%)
Total	**278**	**95%**	**224**	**54**
unknown	**16**	**5%**		
Total	**294**			

On April 1, 2015, researchers from the Baylor College of Medicine, the Chinese University of Hong Kong and the University of Hong Kong have published a study titled Accurate Description of DNA-Based Noninvasive Prenatal Screening in *The New England Journal of Medicine*. The study found that BGI's NIFTY test performed better than all other NIPT tests examined.

In the study, researchers evaluated the performance of noninvasive prenatal screening in a multicenter cohort of women who had received positive results from the screening and were referred for invasive prenatal diagnostic testing to confirm the presence of fetal aneuploidy. They compared the results of the noninvasive prenatal screening reported by Sequenom, BGI, Illumina, Natera, Ariosa Diagnostics and Integrated Genetics.

Of 307 samples that were reported positive by noninvasive prenatal screening, 238 of the 294 cases (81%) that were later found to have a nonmosaic karyotype were correctly detected. However, 9% of the women who received positive screening results for trisomy 21, 23% for trisomy 18, 46% for trisomy 13, 62% for monosomy X, and 17% for XXX, XXY, or XYY abnormalities were carrying fetuses with normal karyotypes. In addition, another fetus was found to have trisomy 21 and one fetus was found to have monosomy X after routine second-trimester ultrasonography showed fetal abnormalities, although previous results of noninvasive prenatal screening were normal.

Of the 15 false positive cases that were identified at the Chinese University of Hong Kong, 8 (5 with trisomy 21, 1 with trisomy 18, and 2 with trisomy 13) are known to have resulted in the live birth of a newborn with no apparent abnormalities as determined through examination by a pediatrician.

Researchers described that intrinsic biologic factors such as somatic mosaicism, confined placental mosaicism, and maternal copy-number imbalance are true sources of false positive and false negative results of noninvasive prenatal screening. According to the study, BGI's NIFTY Test had a true positive of 87% and false positive of 13%. Among the six laboratories, BGI demonstrated the best test performance.

Through collecting a small sample of peripheral blood from the pregnant woman, NIFTY is able to extract cell free fetal DNA. Utilizing next generation high throughput sequencing technology combined with bioinformatics analysis, NIFTY provides a highly accurate risk rate of occurrence for chromosomal aneuploidy in the fetus. As a non-invasive prenatal test, NIFTY avoids the risk of miscarriage associated with invasive diagnostic procedures such as amniocentesis, which carries a 1% risk of miscarriage.

As of April 2015, NIFTY is offered by more than 2000 healthcare providers in more than 60 countries including England, Australia, Spain, Singapore, Israel, the Czech Republic, Turkey, Thailand and China, with nearly 500,000 test samples processed so far with an accuracy rate of more than 99%.

Considering the intrinsic biologic factors,

researchers suggested following the recommendation by the American College of Obstetricians and Gynecologists and the Society for Maternal-Fetal Medicine, positive findings on noninvasive prenatal screening must be followed by invasive prenatal diagnostic testing before any irreversible decisions are made.

More information: Accurate Description of DNA-Based Noninvasive Prenatal Screening , www.nejm.org/doi/full/10.1056/NEJMc1412222

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