



Don't settle for limited data

Data shows Illumina next-generation sequencing is the NIPT technology of choice

PUBLISHED SUPPORT OF NEXT GENERATION SEQUENCING (NGS)

Noninvasive prenatal testing (NIPT) provides accurate information about major chromosome abnormalities in a fetus as early as 10 weeks gestation using a single maternal blood draw.

- Next-generation sequencing (NGS) is the most-published method for performing NIPT^{1,2}
- 99.7% of NIPT samples in published studies were run on Illumina NGS technology (Table 1)

Table 1: 99.7% of NIPT Samples Run on Illumina NGS Systems

Test (Company)	platform	Platform provider	Number of published samples		
			Illumina NGS	Ion Proton NGS	Affymetrix array
Illumiscreen prenatal test (Illumina)	NGS	Illumina	114,163	0	0
MaterniT21 PLUS Test (Sequenom)	NGS	Illumina	302,544	0	0
NIFTY Test (BGI)	NGS	Illumina	174,145	0	0
Panorama Prenatal Screen (Natera)	NGS	Illumina	56,229	0	0
Other LDTs	NGS	Illumina	14,655	0	0
ONA® Test (Premaitha)	NGS	Ion Proton	0	684	0
Harmony Prenatal Test (Ariosa)*	NGS	Affymetrix	72,114	0	1,677
Total	NGS		733,850	684	1,677

A PubMed search for "cell-free, DNA, prenatal," "noninvasive prenatal testing," and "noninvasive prenatal screening" was performed on July 25, 2016. All validation and clinical studies using unique samples were included, where a current clinical NIPT provider performed sample analysis. Case studies, review articles, and studies published in a language other than English were excluded. A total of 76 published studies were surveyed. Data calculations on fi Illumina, Inc. 2016. NGS = next-generation sequencing; either whole-genome or targeted LDTs = laboratory developed tests.

References

- The American College of Obstetricians and Gynecologists. Committee Opinion: Cell-free DNA screening for fetal aneuploidy. www.acog.org/Resources-And-Publications/Committee-Opinions/Committee-on-Genetics/Cell-free-DNA-Screening-for-Fetal-Aneuploidy. Published June 26, 2015. Accessed August 5, 2015.
- 2. Benn P, Borell A, Chiu R, et al. Position statement from the Aneuploidy Screening Committee on behalf of the Board of the International Society for Prenatal Diagnosis. *Prenat Diagn.*, 2013;33:622-629. doi: 10.1002/pd.4139.

^{*} In 2014, Ariosa switched from sequencing to arrays for clinical samples despite limited published data on this platform.