

# GGA Prenatal SNP Microarray

GGA Prenatal SNP (single nucleotide polymorphism) microarray is designed with high density CNV+SNP probes (750K-2.69M probes) to detect small variations across the whole chromosome set. The detection rate is higher than karyotyping (G-banding) and traditional oligo-microarray.



## Primary prenatal diagnostic test for fetal ultrasound abnormalities and abnormal screening result

SNP microarray has been established as a useful diagnostic tool in pregnancy to detect fetal chromosomal abnormality. It could detect more chromosomal abnormalities than other existing testing methods and provides more comprehensive genetic information of the fetus.

Small chromosomal variations can lead to excess or shortage of genes within that region, thus resulting in a person with medical problems, such as abnormal growth, development, and abnormal function of the body's systems.

GGA Prenatal SNP Microarray with both CNV and SNP markers could identify genomic copy number changes and homozygosity, such as absence of heterozygosity (AOH), uniparental disomy (UPD), and triploidy, to help determine the genetic causes of abnormal findings and/or abnormal screening result.

## Types of Abnormalities Detectable by SNP microarray

	SNP microarray		Oligo/BAC array	Karyotyping
	HD array	750K array		
<b>Number of probes</b>	2.69M	750K	30-180K	
<b>Number of SNP markers</b>	750K	200K		
<b>FDA approved *</b>	✓			
<b>Identify Maternal cell Contamination</b>	✓	✓		
<b>Uniparental disomy (UPD) **</b> (both copies of a chromosome pair inherited from the same parent, associated with conditions such as Prader-Willi Syndrome, Angelman Syndrome, Beckwith - Wiedemann syndrome and etc.)	✓	✓		
<b>Triploidy</b> (69 chromosomes instead of the typical 46 chromosomes)	✓	✓		✓
<b>Microduplications/ microdeletions</b> (e.g. Prader-Willi Syndrome, Angelman Syndrome, DiGeorge Syndrome, Williams Syndrome, Duchenne Muscular Dystrophy and etc.)	✓	✓	✓	
<b>Numerical chromosomal abnormality</b> (e.g. Down Syndrome, Edwards Syndrome, Patau Syndrome, etc.)	✓	✓	✓	✓
<b>Balanced chromosomal structural rearrangement ***</b> (e.g. balanced translocation, balanced inversion)				✓

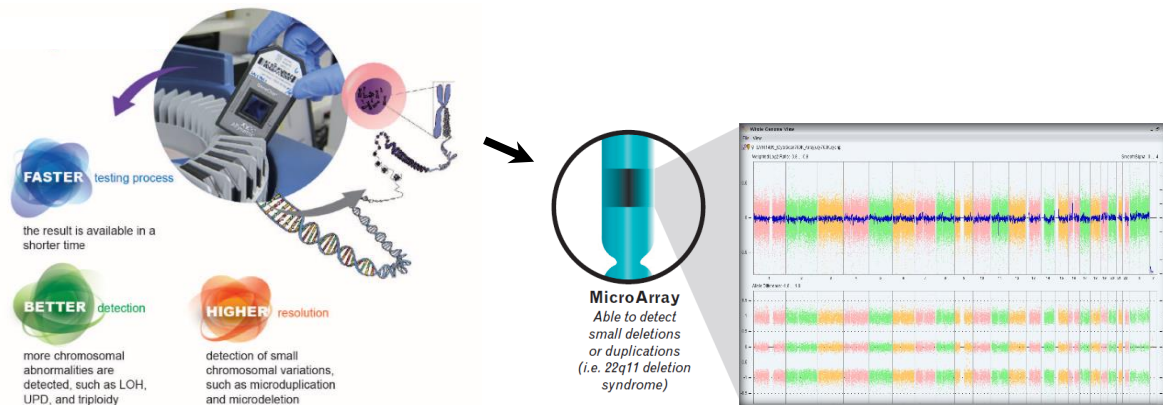
\* HD array is similar to FDA-approved chromosomal microarray (CytoScan Dx)

\*\* SNP microarray can detect uniparental isodisomy, but not uniparental heterodisomy.

\*\*\* Chromosomal balanced translocation, balanced inversion, and heterochromatic regions cannot be detected by SNP microarray. Therefore, a combination of SNP microarray and Karyotyping is suggested to further reduce the risks of delivering a baby with chromosomal abnormalities.

## Why choose GGA Prenatal SNP microarray

- Combines copy number markers with SNP markers at a high density to provide the highest resolution and coverage
- Able to detect more chromosomal abnormalities such as AOH, UPD and triploidy in one test
- Provide comprehensive genetic counseling support to clinicians
- CAP-accredited laboratory with over 12 years of experience in genetic testing
- GGA clinical studies yield a rich collection of genetic and clinical data on SNP microarray<sup>1-3</sup>



## When to consider Prenatal SNP microarray

The American College of Obstetricians and Gynecologists (ACOG) and the Society for Maternal-Fetal Medicine (SMFM) recommend that all pregnant women should be offered prenatal assessment for aneuploidy by screening or diagnostic testing regardless of maternal age or other risk factors.<sup>4</sup>

It is recommended for pregnant women who:

- desires definitive and comprehensive information about fetus
- have a family history of congenital anomalies or a previous pregnancy with a fetus with chromosomal abnormalities
- have abnormal prenatal findings from ultrasound scans, karyotype, and other screening tests (e.g. NIPT, First/Second-trimester maternal serum Down Syndrome screening)
- Non-invasive Prenatal Test (NIPT) showing no-call or inconclusive results due to low fetal fraction
- carries multiple gestations
- achieved pregnancy by Preimplantation Genetic Testing for Aneuploidy (PGT-A)

## Sample requirements

**Amniotic fluid:** 10-15mL

\*Other sample requirements (CVS or DNA) can be discuss further

Testing information

**Only DNA from amniotic fluid can totally represent the fetal DNA/conditions**

SNP microarray detects fetal DNA through amniotic fluid and diagnose the conditions. However, screening test such as NIPT detects cell free DNA which originates from placenta and may not be 100% representative of the fetal condition. Thus, SNP microarray is highly recommended for pregnant women with abnormal prenatal findings.

References:

1. J FORMOS MED ASSOC. Mar 2019, 118(3), 739-742
2. Pediatrics and Neonatology. (2020) 61, 343-345
3. Acta Obstet Gynecol Scand. 2020 Jun;99(6):775-782.
4. Obstet Gynecol 2016;127:e108-22.



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