

# GGA Postnatal SNP Microarray

GGA Postnatal 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.



## The FIRST-TIER clinical diagnostic test for Individuals with Developmental Disabilities or Congenital Anomalies

The World Health Organization (WHO) estimates that 6-8% of children suffer from developmental delay globally. Approximately 60-80% of children with intellectual disability do not have significantly abnormal features and are therefore difficult to diagnose. SNP microarray has been established as a useful diagnostic tool for investigation of genetic causes in children with abnormal development such as unexplained developmental delays, intellectual disability, multiple congenital anomalies, and/or autism spectrum disorders.

GGA Postnatal 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 in children more effectively and give them the most appropriate care they need and deserve.

## Types of Abnormalities Detectable by SNP microarray

	SNP microarray		Array CGH (Oligo/BAC)	Karyotyping
	HD array	750K array		
<b>Diagnostic resolution *</b>	Highest	High	Medium	Low
<b>Number of probes</b>	2.69M	750K	30-180K	
<b>Number of SNP markers</b>	750K	200K		
<b>FDA approved **</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)				✓
<b>Coverage of ClinGen</b>	100%	100%		
<b>Coverage of OMIM Morbid genes</b>	98%	83%		

\* Structural rearrangement and low-level mosaicism of the chromosome are excluded

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

\*\*\* Uniparental heterodisomy is excluded

# Finding the underlying genetic causes of clinical abnormalities can help with:



**BETTER** medical care



**FASTER** diagnosis

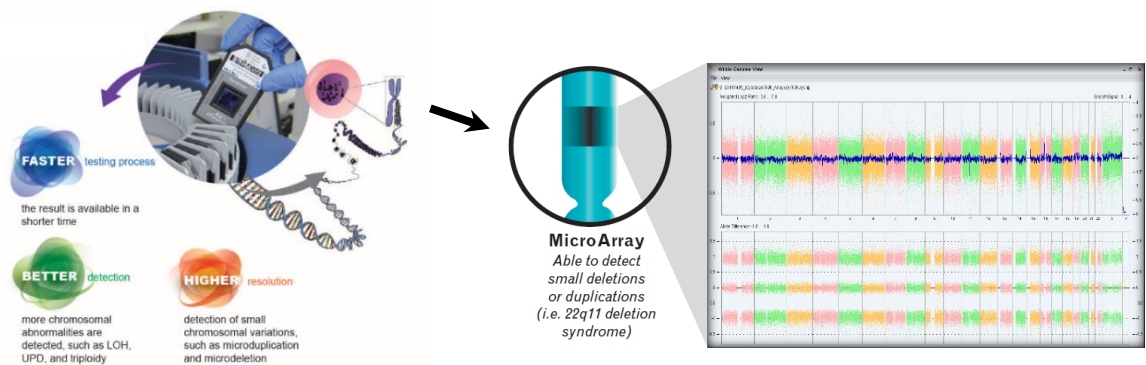


**BETTER** family planning

- Understand the disease prognosis and comorbidity information
- Early intervention
- Alter medical management
- End diagnostic odyssey and search for answers
- Alleviate anxiety and psychological burden due to uncertainty
- Clarify recurrence risk for future pregnancy
- Adjust reproductive plans
- More genetic testing options

## Why choose GGA Postnatal 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 Postnatal SNP microarray

- Unexplained developmental delays
- Unexplained intellectual disabilities
- Autism spectrum disorders
- Congenital defects/dysmorphism
- Other congenital anomalies with unknown cause

## Sample requirement

Blood: 3-5 mL

**Early intervention golden period: 10 times more effective!**

Early intervention for children with developmental delay can effectively improve prognosis and development. The average treatment outcome with early intervention before 3 years of age is 10 times better than late intervention!

**Helpful Tips**

### 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.



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