

Prenatal Microarray Logistics and Ordering

Kerry Shooner, MS, CGC
Laboratory Genetic Counselor
CCHMC

Logistics and Ordering Topics

- Ordering a Prenatal Microarray
- Timing of Prenatal Microarray
- What Microarrays Look Like
- Pre- and Post-Test Counseling Issues

Ordering a Prenatal Microarray

Possible Prenatal Indications:

- Suspicion of chromosome imbalance

This might include

- suggestive abnormal ultrasound findings
- advanced maternal age
- family history of chromosome imbalance
- family history of mental retardation or birth defects

- Defining known chromosome imbalance

Ordering a Prenatal Microarray

Specimens Needed:

1. FETAL

20 - 25 mls Amniotic Fluid (in sterile centrifuge tubes in sealed biohazard transport bag, leak-proof)

or

4 T-25 confluent flasks of cultured amniocytes

2. PARENTAL

5 mls peripheral blood in **EDTA tube** (purple top)

Ordering a Prenatal Microarray

Process:

1. Collect fetal and parental specimens
2. Order the test(s) desired:
 - Prenatal Test Requisition form (external orderers)
 - Order in ICIS or EPIC (CCHMC orderers)
3. Send samples to laboratory by rapid method
 - FedEx (room temp)
 - Courier pickup (local orderers) – call office to arrange

Ordering a Prenatal Microarray

List Price:

\$2972

plus culture charge, if applicable

Timing of Prenatal Microarray

Turn-Around Time

14 – 21 days

Consider the timing of when test results will be available, in context of your patient's pregnancy

Timing of Prenatal Microarray

- Issues that may increase TAT:
 - Quality of amniotic fluid
 - If not clear
 - Patient has polyhydramnios
 - Early (<16 wks) or late (>30 wks) gestation
 - Quantity of amniotic fluid
 - If less than 20-25 mls
 - Test order changes
 - Parental samples not promptly received

Microarray Platforms

1. **BAC** (Bacterial Artificial Chromosome)

DNA construct where the usual insert size is 150 kb, with a range from 100 to 300 kb.

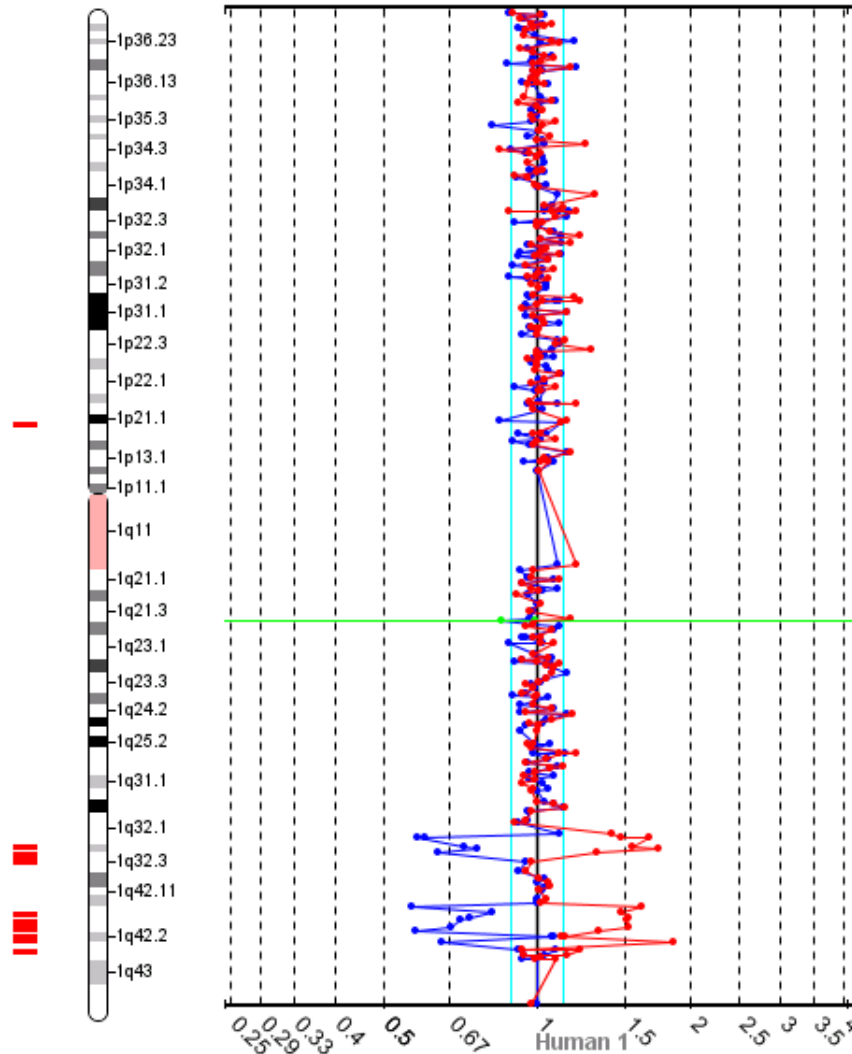
2. **OLIGO** (Oligonucleotides)

Short sequences of nucleotides typically with twenty or fewer bases. Resolution 20-80 bp.

3. **SNP** (Single Nucleotide Polymorphism) – at CCHMC

DNA sequence variation occurring when a single nucleotide in genome differs between members of a species (or paired chromosomes in an individual).

BAC Array Data

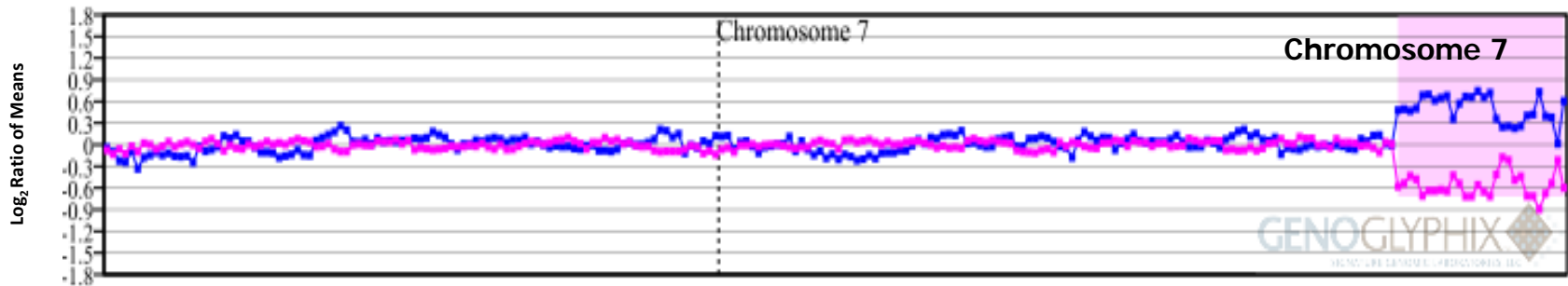


Complex rearrangement of DNA gains and losses on chromosome 1q

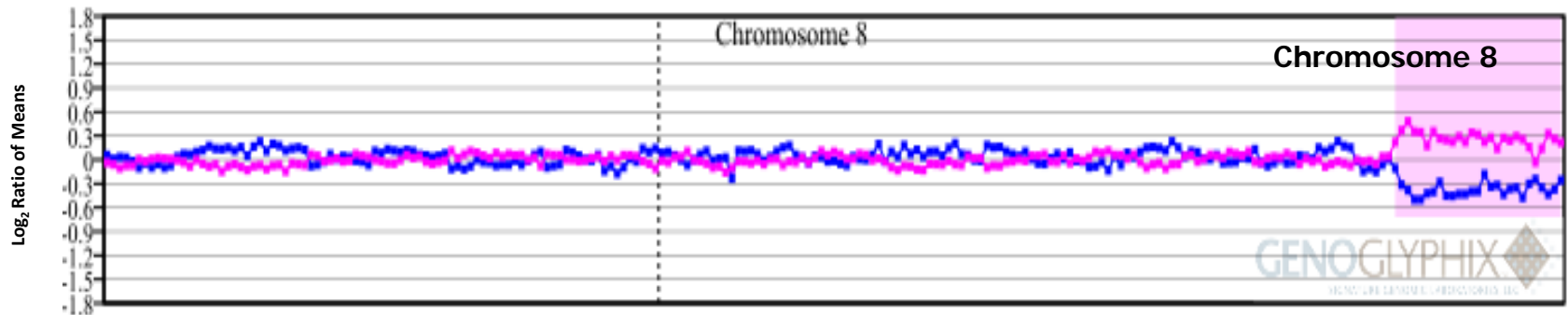
Signature Genomics

Oligo Array Data

7.7 Mb deletion 7q36.1q36.3 (includes *SHH* gene)

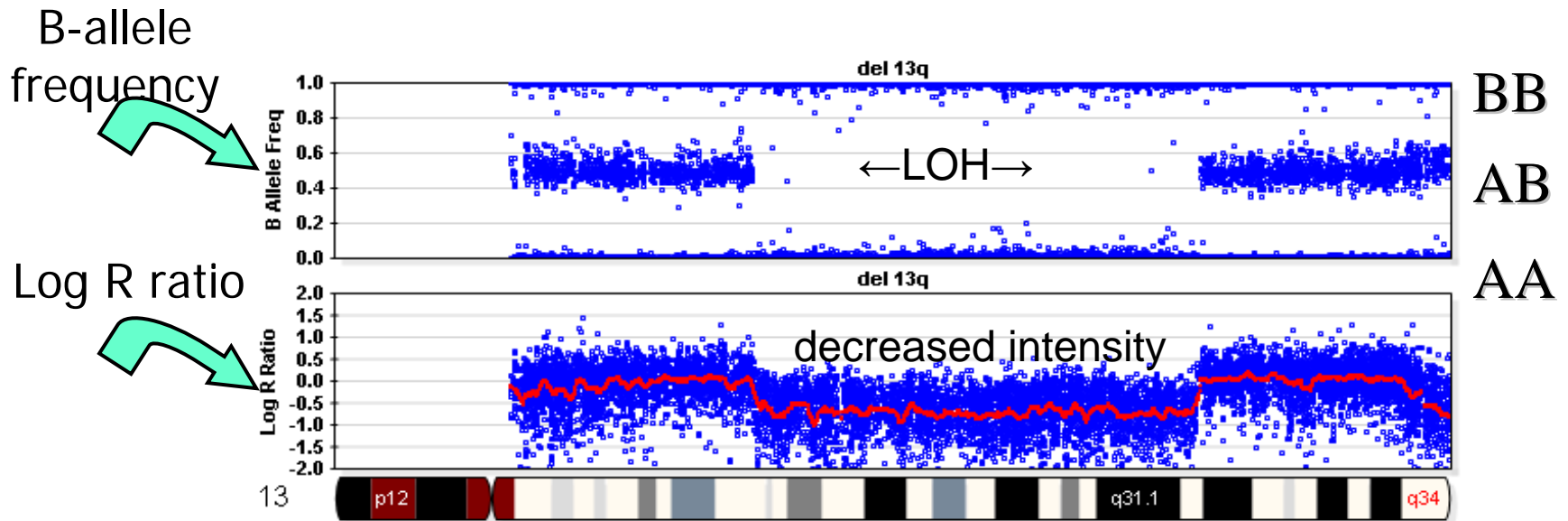


4.4 Mb gain 8q24.3



Signature Genomics

SNP Array Data



del(13)(q14.1q31.2) (46 Mb)

Illumina

Pre-Test Counseling for Prenatal Microarray

1. Explain limitations of the technology

- Is designed to detect gains or losses of DNA, not changes (mutations)
- May not pick up balanced rearrangements or mosaicism
- Abnormalities must be in an area covered by the array, and then must be of a certain size and/or gene content to be reported out

Pre-Test Counseling for Prenatal Microarray

2. Explain what the results might be

a) **“Normal”** – No Imbalance Detected

b) **Abnormal – Del/Dup with Known Significance**

Previous cases have had this abnormality and some clinical significance can be provided

c) **Abnormal – Del/Dup of Uncertain Significance**

- Ex: There are genes in the abnormal region that have no known relationship with disease; no patients reported in literature with this same abnormality
- Parents tested to aid interpretation

Post-Test Counseling for Prenatal Microarray

- Difficulty with clinical interpretation
 - Finding clinical information
 - Database of Genomic Variation and others
 - Pubmed
 - Previous cases tested
- Can create more uncertainty for families
- Can answer diagnosis questions

Post-Test Counseling for Prenatal Microarray

1. Explaining Results... reports are often complicated
2. Providing Resources

Handouts to explain patient results in patient-friendly language: 3 versions available on our website at:
www.cincinnatichildrens.org/cgh

Contact Information

Kerry Shooner, MS, CGC

513-636-6779

Kerry.Shooner@cchmc.org

Shelly Rudnick, MS, CGC

513-803-1767

Shelly.Rudnick@cchmc.org

Jennifer Ruschman, MS,
CGC

513-636-0110

Jennifer.Ruschman@cchmc.org

Kristen Sund, MS, PhD

513-803-1703

Kristen.Sund@cchmc.org

Website: www.cincinnatichildrens.org/cgh

