

**Signature Genomics**

## Points to Consider with Microarray Testing



Signature Genomics recognizes the health care provider's important role in discussing pertinent information about genetic testing with each patient or family<sup>1</sup>. As a courtesy to our ordering health care providers and patients, we offer the following **Points to Consider** when reviewing the benefits and limitations of microarray testing with patients and their families<sup>2</sup>.

1. Microarray Testing, also called *Microarray Analysis*, *Array-based Comparative Genomic Hybridization* or *array CGH*, is a new technology that evaluates chromosomes for gains or losses (duplications or deletions) of DNA segments that can cause birth defects and/or mental retardation.
2. Microarray analysis detects chromosome imbalances at a higher resolution than traditional chromosomal analysis, also called karyotyping. Typically, traditional karyotyping detects alterations at a resolution of 5 million basepairs or larger, whereas microarray testing will detect alterations that cannot be seen by traditional karyotyping, and sometimes may only be thousands of basepairs in size.
3. Microarray testing will not detect balanced chromosome rearrangements such as reciprocal translocations, Robertsonian translocations, inversions, and balanced insertions. These types of abnormalities may be detected by karyotyping.
4. Microarray testing is specifically designed to detect deletions and duplications of chromosome material and cannot detect all mutations in the examined chromosome regions. This technology will not detect sequence alterations or single base pair mutations, or abnormalities in other genes or loci not tested with this technology.
5. This technology was not designed to detect cases of mosaicism and its accuracy in detecting mosaicism is not well established.
6. Many of the well-characterized genetic conditions tested for by microarray testing are caused by more than one underlying genetic mechanism. Therefore, a normal result does not exclude a diagnosis of one or more of the syndromes assessed by the analysis.
7. A normal result does not rule out a chromosome change at an area of the genome not tested by the microarray.

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8. An abnormal test result may provide a clinical diagnosis, but many genetic syndromes have variability in clinical presentation and this technology cannot predict mildness or severity of a specific genetic condition.
9. Some test results may have unclear clinical significance, meaning that it is not clear whether the alteration detected causes a clinical abnormality in the patient. These cases may even require additional studies on the patient, or more rarely the parents, to assist with interpretation. Sometimes, even after family studies, it is impossible to confirm whether an abnormal test result causes or explains the clinical abnormalities in the patient tested.
10. Some test results may demonstrate an alteration that is also subsequently found in other “normal” family members. A possible explanation is that these alterations are normal human variants. However, the possibility that they somehow play a role in an abnormal clinical finding cannot be excluded. As more scientific and medical information is learned about the structure of human chromosomes, it is hoped that interpretation of these types of results will become clearer.
11. The SignatureChip and Signature PrenatalChip are whole genome arrays and therefore may identify genetic conditions not included in the representative list of Disorders Tested. The SignatureChip and Signature PrenatalChip may identify copy number changes associated with adult-onset conditions and/or cancer predisposition that are potentially unrelated to the patient’s current clinical findings, but may become apparent later in life. If there is a family history of a known or suspected genetic condition unrelated to the reason for testing, please contact the laboratory to discuss prior to sample submission.
12. For complete prenatal diagnosis, it is recommended that karyotyping be performed prior to, concurrent to, or following microarray testing analysis. Karyotyping will detect balanced chromosome rearrangements and in some cases can further characterize structural abnormalities detected by microarray testing.

For more information, including *Frequently Asked Questions for Physicians*, *Frequently Asked Questions for Patients* and *Frequently Asked Questions for Prenatal Testing*, please visit our website at [www.signaturegenomics.com](http://www.signaturegenomics.com), email [info@signaturegenomics.com](mailto:info@signaturegenomics.com) or call us directly at 1.877.SigChip (744.2447).

1 In accordance with Washington State Law RCW 7.70.050 and WAC 388-531-0050, providing patients with the information necessary for them to be able to give their informed consent for testing or treatment is the responsibility of the health care provider who has direct contact with the patient. Laboratory tests are ordered and prescribed by physicians so it is the physician, not the laboratory, that is required to obtain the patient’s informed consent for testing.

2 The contents of the *Points to Consider* are provided for informational purposes only and are not and should not be construed as medical advice, diagnosis, or treatment. Only a properly qualified physician can address specific questions regarding a patient’s health care needs. Individual inquiries about medical or healthcare issues should be addressed to appropriate healthcare professionals. Nothing contained in the *Points to Consider* should be used to replace or substitute for a patient’s personal physician’s advice.