Publicationlist

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Selected recent publications using CytoScan® Cytogenetics Solution

Wang J. C., *et al.* Mosaic isochromosome 15q and maternal uniparental isodisomy for chromosome 15 in a patient with morbid obesity and variant pws-like phenotype. *American Journal of Medical Genetics Part A* **161A**(7):1695–1701 (2013).

"The oligonucleotide-SNP array analysis did not reveal any significant copy number alterations genome-wide. However, SNP data revealed allelic homozygosity (hmz) for the entire chromosome 15 in a mosaic pattern, suggestive of an apparently mosaic uniparental isodisomy for entire chromosome 15q."

Kearney H. M., *et al*. Diagnostic Implications of Excessive Homozygosity Detected by SNP-Based Microarrays: Consanguinity, Uniparental Disomy, and Recessive Single-Gene Mutations. *Clinics in Laboratory Medicine* **31**(4):595–613 (2011).

Brown N., *et al*. 5q31.3 Microdeletion syndrome: Clinical and molecular characterization of two further cases. *American Journal of Medical Genetics Part A* **9999**:1–5 (2013).

Wapner R. J., *et al.* Chromosomal microarray versus karyotyping for prenatal diagnosis. *New England Journal of Medicine* **367**(23):2175–2184 (2012).

"...a post hoc review determined that had the SNP data been analyzed, the triploid cases would have been detected. We therefore suggest that arrays used for prenatal testing should contain SNP probes that can reliably identify triploidy."

Mason-Suares H., *et al*. Density matters: comparison of array platforms for detection of copy-number variation and copy-neutral abnormalities. *Genetics in Medicine* doi:10.1038/gim.2013.36 (2013).

"The low-density array called absence-of-heterozygosity regions not confirmed by the other platforms and also overestimated the length of true absence-of-heterozygosity regions. Furthermore, the low- and mid-density platforms failed to detect some small absence-of-heterozygosity regions that were identified by the high-density platform."

Simons A., *et al.* Genome-wide arrays in routine diagnostics of hematological malignancies. *Human Mutation* **33**(6):941–948 (2012).

Huh P., *et al.* Different characteristics Identified by Single Nucleotide Polymorphism Array Analysis in Leukemia Suggest the Need for Different Application Strategies Depending on Disease Category. *Genes Chromosomes Cancer* **52**(1):44–55 (2013).

Pei J., *et al.* Chromothripsis in a case of TP53-deficient chronic lymphocytic leukemia. *Leukemia Research Reports* **1**(1):4–6 (2012).

"Although FISH analysis is now widely used for the cytogenetic assessment of CLL, other approaches such as oligonucleotide-based array comparative genomic hybridization and single nucleotide polymorphism (SNP) gene chips show comparable results but also assess all chromosomal regions rather than the current standard clinical practice of identifying alterations with probes targeting only 4–5 chromosomal sites."

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Choi S. M., *et al*. Near-haploid B lymphoblastic leukemia with an apparent hyperdiploid karyotype: the critical role of SNP analysis in establishing proper diagnosis. *Journal of Hematopathology* doi:10.1007/s12308-013-0189-5 (2013).

Setoodeh R., *et al.* Double-hit mantle cell lymphoma with MYC gene rearrangement or amplification: a report of four cases and review of the literature. *International Journal of Clinical and Experimental Pathology* **6**(2):155–167 (2013).

Selected recent publications on SNP-array tools

Wierenga K. J., *et al*. A clinical evaluation tool for SNP arrays, especially for autosomal recessive conditions in offspring of consanguineous parents. *Genetics in Medicine* **15**(5):354–360 (2013).

Schroeder C., *et al*. UPDtool: a tool for detection of iso- and heterodisomy in parent-child trios using SNP microarrays. *Bioinformatics*. **29**(12):1562–1564 (2013).

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