

## GeneChip® Mapping Publications

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### Genotyping Technology:

1. Large-scale genotyping of complex DNA

Giulia C Kennedy, Hajime Matsuzaki, Shoulian Dong, Wei-min Liu, Jing Huang, Guoying Liu, Xing Su, Manqiu Cao, Wenwei Chen, Jane Zhang, Weiwei Liu, Geoffrey Yang, Xiaojun Di, Thomas Ryder, Zhijun He, Urvashi Surti, Michael S Phillips, Michael T Boyce-Jacino, Stephen PA Fodor, Keith W Jones

*Nature Biotechnology* 21, 1233 - 1237 (01 Oct 2003) Research

2. Algorithms for Large Scale Genotyping Microarrays

Wei-min Liu, Xiaojun Di, Geoffrey Yang, Hajime Matsuzaki, Jing Huang, Rui Mei, Thomas B. Ryder, Teresa A. Webster, Shoulian Dong, Guoying Liu, Keith W. Jones, Giulia C. Kennedy and David Kulp.. *Bioinformatics*. 2003 19: 2397-2403.

3. Parallel Genotyping of over 10,000 SNPs using a One Primer Assay on a High Density Oligonucleotide Array

Hajime Matsuzaki, Halina Loi, Shoulian Dong, Ya-Yu Tsai, Joy Fang, Jane Law, Xiaojun Di, Wei-Min Liu, Geoffrey Yang, Guoying Liu, Jing Huang, Giulia C. Kennedy, Thomas B. Ryder, Gregory Marcus, Sean Walsh, Mark D. Shriver, Jennifer M. Puck, Keith W. Jones and Rui Mei  
*Genome Research*. 14: 414-425, 2004

4. Genotyping over 100,000 SNPs on a Pair of Oligonucleotide Arrays

Hajime Matsuzaki, Shoulian Dong, Halina Loi, Xiaojun Di, Guoying Liu, Earl Hubbell, Jane Law, Tam Berntsen, Monica Chadha, Henry Hui, Geoffrey Yang, Teresa A. Webster, Simon Cawley, P. Sean Walsh, Keith W. Jones, and Rui Mei  
*Nature Methods* 1: 109 – 111, 2004

5. Genome-wide single-nucleotide polymorphism arrays demonstrate high fidelity of multiple displacement-based whole-genome amplification.

Tzvetkov MV, Becker C, Kulle B, Nurnberg P, Brockmoller J, Wojnowski L. *Electrophoresis*. 2005 Feb;26(3):710-5

6. Dynamic model based algorithms for screening and genotyping over 100K SNPs on oligonucleotide microarrays. Di X, Matsuzaki H, Webster TA, Hubbell E, Liu G, Dong S, Bartell D, Huang J, Chiles R, Yang G, Shen MM, Kulp D, Kennedy GC, Mei R, Jones KW, Cawley S.

*Bioinformatics*. 2005 May 1;21(9):1958-63. Epub 2005 Jan 18

7. ALOHOMORA: a tool for linkage analysis using 10K SNP array data.

Ruschendorf F, Nurnberg P. *Bioinformatics*. 2005 May 1;21(9):2123-5. Epub 2005 Jan 12.

8. Comparative linkage analysis and visualization of high-density oligonucleotide SNP array data.

Leykin I, Hao K, Cheng J, Meyer N, Pollak MR, Smith RJ, Wong WH, Rosenow C, Li C *BMC Genet*. 2005 Feb 15;6(1):7

9. Genome coverage and sequence fidelity of phi29 polymerase-based multiple strand

displacement whole genome amplification. Paez JG, Lin M, Beroukhim R, Lee JC, Zhao X,

Richter DJ, Gabriel S, Herman P, Sasaki H, Altshuler D, Li C, Meyerson M, Sellers WR. Nucleic Acids Res. 2004 May 18;32(9):e71.

10. PPC: an algorithm for accurate estimation of SNP allele frequencies in small equimolar pools of DNA using data from high density microarrays. Brohede J, Dunne R, McKay JD, Hannan GN. Nucleic Acids Res. 2005 Sep 30;33(17):e142.

11. Reliable high-throughput genotyping and loss-of-heterozygosity detection in formalin-fixed, paraffin-embedded tumors using single nucleotide polymorphism arrays. Lips EH, Dierssen JW, van Eijk R, Oosting J, Eilers PH, Tollenaar RA, de Graaf EJ, van't Slot R, Wijmenga C, Morreau H, van Wezel T. Cancer Res. 2005 Nov 15;65(22):10188-91.

## **Chromosomal Copy Number**

12. High-resolution analysis of DNA copy number using oligonucleotide microarrays. Bignell GR, Huang J, Greshock J, Watt S, Butler A, West S, Grigorova M, Jones KW, Wei W, Stratton MR, Futreal PA, Weber B, Shaperro MH, Wooster R. Genome Res. 2004 Feb;14(2):287-95

13. Genome-wide Loss of Heterozygosity Analysis from Laser Capture Microdissected Prostate Cancer Using Single Nucleotide Polymorphic Allele (SNP) Arrays and a Novel Bioinformatics Platform dChipSNP  
Marshall E. Lieberfarb, Ming Lin, Mirna Lechpammer, Cheng Li, David M. Tanenbaum, Phillip G. Febbo, Rene'e L. Wright, Judy Shim, Philip W. Kantoff, Massimo Loda, Matthew Meyerson, and William R. Sellers  
*Cancer Research* 63; 4781-4785, August 15, 2003

14. High-resolution single nucleotide polymorphism array and clustering analysis of loss of heterozygosity in human lung cancer cell lines.  
Janne, P.A., Li, C., Zhao, X., Girard, L., Chen, T.-H., Minna, J., Christiani, D.C., Johnson, B.E., Meyerson, M. . *Oncogene* 2004; April 8;23(15):2716-26.

15. An integrated view of copy number and allelic alterations in the cancer genome using single nucleotide polymorphism arrays Zhao X, Li C, Paez JG, Chin K, Janne PA, Chen TH, Girard L, Minna J, Christiani D, Leo C, Gray JW, Sellers WR, Meyerson M *Cancer Res.* 2004 May 1;64(9):3060-71.

16. Whole Genome DNA Copy Number Changes Identified by High Density Oligonucleotide Arrays.  
Huang J., Wei W., Zhang J., Liu G., Bignell G.R., Stratton M.R., Futreal P.A., Wooster R., Jones K.W. and Shaperro M. H. *Human Genomics*, 2004, May Vol. 1(4) 287-299

17. Allelic imbalance analysis by high-density single-nucleotide polymorphic allele (SNP) array with whole genome amplified DNA  
Kwong-Kwok Wong, Yvonne T. M. Tsang, Jianhe Shen, Rita S. Cheng, Yi-Mieng Chang, Tsz-Kwong Man and Ching C. Lau  
*Nucleic Acids Research*, 2004, May 17 Vol. 32(9)

18. Concurrent analysis of loss of heterozygosity (LOH) and copy number abnormality (CNA) for oral premalignancy progression using the Affymetrix 10K SNP mapping array. Zhou, X., Mok, S. C., Chen, Z., Li, Y. & Wong, D. T. *Hum Genet* 115, 327-30 (2004).
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21. Molecular karyotyping using an SNP array for genomewide genotyping. Rauch A, Ruschendorf F, Huang J, Trautmann U, Becker C, Thiel C, Jones KW, Reis A, Nurnberg P. *J Med Genet.* 2004 Dec;41(12):916-22.
22. Allelic dosage analysis with genotyping microarrays  
Shumpei Ishikawa, Daisuke Komura, Shingo Tsuji, Kunihiro Nishimura, Shogo Yamamoto, Binaya Panda, Jing Huang, Masashi Fukayama, Keith W. Jones, Hiroyuki Aburatani  
*Biochemical and Biophysical Research Communications* 333 (2005) 1309–1314
23. High-density single nucleotide polymorphism array defines novel stage and location-dependent allelic imbalances in human bladder tumors  
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Xiaojun Zhao, Barbara A. Weir, Thomas LaFramboise, Ming Lin, Rameen Beroukhim, Levi Garraway, Javad Beheshti, Jeffrey C. Lee, Katsuhiko Naoki, William G. Richards, David Sugarbaker, Fei Chen, Mark A. Rubin, Pasi A. Janne, Luc Girard, John Minna, David Christiani, Cheng Li, William R. Sellers, and Matthew Meyerson  
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25. Integrative genomic analyses identify MITF as a lineage survival oncogene amplified in malignant melanoma  
Levi A. Garraway, Hans R. Widlund, Mark A. Rubin, Gad Getz, Aaron J. Berger, Sridhar Ramaswamy, Rameen Beroukhim, Danny A. Milner, Scott R. Granter, Jinyan Du, Charles Lee, Stephan N. Wagner, Cheng Li, Todd R. Golub, David L. Rimm, Matthew L. Meyerson, David E. Fisher & William R. Sellers  
*Nature.* 2005 Jul 7;436(7047):117-22.
26. A Robust Algorithm for Copy Number Detection Using High-Density Oligonucleotide Single Nucleotide Polymorphism Genotyping Arrays  
Yasuhito Nannya, Masashi Sanada, Kumi Nakazaki, Noriko Hosoya, Lili Wang, Akira Hangaishi, Mineo Kurokawa, Shigeru Chiba, Dione K. Bailey, Giulia C. Kennedy, and Seishi Ogawa  
*Cancer Res* 2005 65: 6071-6079
27. Genome-wide single nucleotide polymorphism analysis reveals frequent partial uniparental disomy due to somatic recombination in acute myeloid leukemias

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J. A.E. Irving, L. Bloodworth, N. P. Bown, M. C. Case, L. A. Hogarth, and A. G. Hall *Cancer Res* 65, 3053-3058, 2005.

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## **Linkage**

37. Affymetrix GeneChip system: moving from research to the clinic. Ragoussis J, Elvidge G. *Expert Rev Mol Diagn.* 2006 Mar;6(2):145-52. Review.

38. Highly multiplexed molecular inversion probe genotyping: over 10,000 targeted SNPs genotyped in a single tube assay. Hardenbol P, Yu F, Belmont J, Mackenzie J, Bruckner C, Brundage T, Boudreau A, Chow S, Eberle J, Erbilgin A, Falkowski M, Fitzgerald R, Ghose S, Iartchouk O, Jain M, Karlin-Neumann G, Lu X, Miao X, Moore B, Moorhead M, Namsaraev E, Pasternak S, Prakash E, Tran K, Wang Z, Jones HB, Davis RW, Willis TD, Gibbs RA. *Genome Res.* 2005 Feb;15(2):269-75.
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