

# Publications

## Recent publications on the Affymetrix® Genome-Wide Human SNP Array 6.0

### Cancer

#### 2011

Tabach Y., et al. Amplification of the 20q chromosomal arm occurs early in tumorigenic transformation and may initiate cancer. *PLoS One* **6**(1) (2011).

Yang J. J., et al. Ancestry and pharmacogenomics of relapse in acute lymphoblastic leukemia. *Nature Genetics* **43**(3):237-41 (2011).

Kim J. C., et al. Novel chemosensitive single-nucleotide polymorphism markers to targeted regimens in metastatic colorectal cancer. *Clinical Cancer Research* **17**(5):1200-9 (2011).

Stephens P. J., et al. Massive genomic rearrangement acquired in a single catastrophic event during cancer development. *Cell* **144**(1):27-40 (2011).

Pasaniuc B., et al. Enhanced statistical tests for GWAS in admixed populations: assessment using African Americans from CARE and a Breast Cancer Consortium. *PLoS Genetics* **7**(4) (2011).

#### 2010

Timmermann B., et al. Somatic mutation profiles of MSI and MSS colorectal cancer identified by whole exome next generation sequencing and bioinformatics analysis. *PLoS One* **5**(12) (2010).

Goya R., et al. SNVMix: predicting single nucleotide variants from next-generation sequencing of tumors. *Bioinformatics* **26**(6):730-6 (2010).

Gamazon E. R., et al. Chemotherapeutic drug susceptibility associated SNPs are enriched in expression quantitative trait loci. *Proceedings of the National Academy of Sciences of the United States of America* **107**(20):9287-92 (2010).

Kleppe M., et al. Deletion of the protein tyrosine phosphatase gene PTPN2 in T-cell acute lymphoblastic leukemia. *Nature Genetics* **42**(6) (2010).

Bignell G. R., et al. Signatures of mutation and selection in the cancer genome. *Nature* **463**(7283) (2010).

Liang Y., et al. Stem-like cancer cells are inducible by increasing genomic instability in cancer cells. *Journal of Biological Chemistry* **285**(7):4931-40 (2010).

Lee W., et al. The mutation spectrum revealed by paired genome sequences from a lung cancer patient. *Nature* **465**(7297):473-7 (2010).

Ivakhno S., et al. CNAanova: a new approach for finding recurrent copy number abnormalities in cancer SNP microarray data. *Bioinformatics* **26**(11):1395-402 (2010).

Mattison J., et al. Novel Candidate Cancer Genes Identified by a Large-Scale Cross-Species Comparative Oncogenomics Approach. *Cancer Research* **70**(3):883-95 (2010).

Restall I. J., et al. Induction of Premature Senescence by Hsp90 Inhibition in Small Cell Lung Cancer. *PLoS One* **5**(6) (2010).

Mao X., et al. Distinct Genomic Alterations in Prostate Cancers in Chinese and Western Populations Suggest Alternative Pathways of Prostate Carcinogenesis. *Cancer Research* **70**(13):5207-12 (2010).

Long J., et al. Identification of a Functional Genetic Variant at 16q12.1 for Breast Cancer Risk: Results from the Asia Breast Cancer Consortium. *PLoS Genetics* **6**(6) (2010).

Clark M. J., et al. U87MG Decoded: The Genomic Sequence of a Cytogenetically Aberrant Human Cancer Cell Line. *PLoS Genetics* **6**(1) (2010).

Ramakrishna M., et al. Identification of Candidate Growth Promoting Genes in Ovarian Cancer through Integrated Copy Number and Expression Analysis. *PLoS One* **5**(4) (2010).

Gorringe K. L., et al. Copy Number Analysis Identifies Novel Interactions Between Genomic Loci in Ovarian Cancer. *PLoS One* **5**(9) (2010).

Geiger T., et al. Proteomic changes resulting from gene copy number variations in cancer cells. *PLoS Genetics* **6**(9) (2010).

Gaudet, M. M., et al. Common Genetic Variants and Modification of Penetrance of BRCA2-Associated Breast Cancer. *PLoS Genetics* **6**(10) (2010).

Ramakrishna M., et al. Identification of candidate growth promoting genes in ovarian cancer through integrated copy number and expression analysis. *PLoS One* **5**(4) (2010).

Pleasance E. D., et al. A comprehensive catalogue of somatic mutations from a human cancer genome. *Nature* **463**(7278) (2010).

Andrews J., et al. Multi-Platform Whole-Genome Microarray Analyses Refine the Epigenetic Signature of Breast Cancer Metastasis with Gene Expression and Copy Number. *PLoS One* **5**(1) (2010).

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Zheng W., et al. Genome-wide association study identifies a new breast cancer susceptibility locus at 6q25.1. *Nature Genetics* **41**(3):324-8 (2009).

Maher C. A., et al. Transcriptome sequencing to detect gene fusions in cancer. *Nature* **458**(7234) (2009).

van Haaften G., et al. Somatic mutations of the histone H3K27 demethylase gene UTX in human cancer. *Nature Genetics* **41**(5):521-3 (2009).

Kanetsky P. A., et al. Common variation in KITLG and at 5q31.3 predisposes to testicular germ cell cancer. *Nature Genetics* **41**(7) (2009).

Legoffic A., et al. The reg4 Gene, Amplified in the Early Stages of Pancreatic Cancer Development, Is a Promising Therapeutic Target. *PLoS One* **4**(10) (2009).

Suh I., et al. Distinct loci on chromosome 1q21 and 6q22 predispose to familial nonmedullary thyroid cancer: A SNP array-based linkage analysis of 38 families. *Surgery* **146**(6):1073-80 (2009).

Soranzo N., et al. A genome-wide meta-analysis identifies 22 loci associated with eight hematological parameters in the HaemGen consortium. *Nature Genetics* **41**(11):1182-90 (2009).

Rudd M. K., et al. Segmental duplications mediate novel, clinically relevant chromosome rearrangements. *Human Molecular Genetics* **18**(16):2957-62 (2009).

Liu W. N., et al. Copy number analysis indicates monoclonal origin of lethal metastatic prostate cancer. *Nature Medicine* **15**(5):559-65 (2009).

Beeghly-Fadiel A., et al. Matrix metalloproteinase-2 polymorphisms and breast cancer susceptibility. *Cancer Epidemiology Biomarkers & Prevention* **18**(6):1770-6 (2009).

Kresse S. H., et al. LSAMP, a novel candidate tumor suppressor gene in human osteosarcomas, identified by array comparative genomic hybridization. *Genes, Chromosomes & Cancer* **48**(8):679-93 (2009).

Walter M. J., et al. Acquired copy number alterations in adult acute myeloid leukemia genomes. *Proceedings of the National Academy of Sciences of the United States of America* **106**(31):12950-5 (2009).

Maciejewski J. P., et al. Application of array-based whole genome scanning technologies as a cytogenetic tool in hematological malignancies. *British Journal of Haematology* **146**(5):479-88 (2009).

Belinsky M. G., et al. High density DNA array analysis reveals distinct genomic profiles in a subset of gastrointestinal stromal tumors. *Genes, Chromosomes & Cancer* **48**(10):886-96 (2009).

Gorringe K. L., et al. Are there any more ovarian tumor suppressor genes? A new perspective using ultra high-resolution copy number and loss of heterozygosity analysis. *Genes, Chromosomes & Cancer* **48**(10):931-42 (2009).

Loh M. L., et al. Mutations in CBL occur frequently in juvenile myelomonocytic leukemia. *Blood* **114**(9):1859-63 (2009).

Mardis E. R., et al. Recurring mutations found by sequencing an acute myeloid leukemia genome. *New England Journal of Medicine* **361**(11):1058-66 (2009).

Soh J., et al. Oncogene mutations, copy number gains and mutant allele specific imbalance (MASI) frequently occur together in tumor cells. *PLoS One* **4**(10):e7464 (2009).

Mullighan C. G., et al. Rearrangement of CRLF2 in B-progenitor- and Down syndrome-associated acute lymphoblastic leukemia. *Nature Genetics* **41**(11):1243-6 (2009).

Tiu R. V., et al. New lesions detected by single nucleotide polymorphism array-based chromosomal analysis have important clinical impact in acute myeloid leukemia. *Journal of Clinical Oncology* **27**(31):5219-26 (2009).

Pantaleo M. A., et al. Insulin-like growth factor 1 receptor expression in wild-type GISTs: A potential novel therapeutic target. *International Journal of Cancer* **125**(12):2991-4 (2009).

Popova T., et al. Genome Alteration Print (GAP): A tool to visualize and mine complex cancer genomic profiles obtained by SNP arrays. *Genome Biology* **10**(11):R128 Epub (2009).

Pos, Z., et al. Genomic scale analysis of racial impact on response to IFN-alpha. *Proceedings of the National Academy of Sciences of the United States of America* **107**(2):803-8 (2009).

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Ley T. J., et al. DNA sequencing of a cytogenetically normal acute myeloid leukaemia genome. *Nature* **456**(7218):66-72 (2008).

Ziegler A., et al. Biostatistical aspects of genome-wide association studies. *Biometrical Journal* **50**(1):8-28 (2008).

Mullighan C. G., et al. Genomic analysis of the clonal origins of relapsed acute lymphoblastic leukemia. *Science* **322**(5906):1377-80 (2008).

Lee Y. S., et al. CGcgh: a tool for molecular karyotyping using DNA microarray-based comparative genomic hybridization (array-CGH). *Journal of Biomedical Science* **15**(6):687-96 (2008).

## CNS disorders

### 2011

Shaikh T. H., et al. Genes and biological processes commonly disrupted in rare and heterogeneous developmental delay syndromes. *Human Molecular Genetics* **20**(5):880-93 (2011).

Caliskan M., et al. Exome sequencing reveals a novel mutation for autosomal recessive non-syndromic mental retardation in the TECR gene on chromosome 19p13. *Human Molecular Genetics* **20**(7):1285-9 (2011).

Klitten L. L., et al. Duplication of MAOA, MAOB, and NDP in a patient with mental retardation and epilepsy. *European Journal of Human Genetics* **19**(1):1-2 (2011).

Castelo-Branco P., et al. Neural Tumor-Initiating Cells Have Distinct Telomere Maintenance and Can be Safely Targeted for Telomerase Inhibition. *Clinical Cancer Research* **17**(1):111-21 (2011).

Ou Z., et al. Observation and prediction of recurrent human translocations mediated by NAHR between nonhomologous chromosomes. *Genome Research* **21**(1):33-46 (2011).

Phadke S. R., et al. Report of Two Brothers With Short Stature, Microcephaly, Mental Retardation, and Retinoschisis-A New Mental Retardation Syndrome? *American Journal of Medical Genetics Part A* **155A**(1):9-13 (2011).

Grasshoff U., et al. De novo MECP2 duplication in two females with random X-inactivation and moderate mental retardation. *European Journal of Human Genetics* **19**(5):507-12 (2011).

Bakircioglu M., et al. The Essential Role of Centrosomal NDE1 in Human Cerebral Cortex Neurogenesis. *American Journal of Human Genetics* **88**(5):523-35 (2011).

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Leon E., et al. Mosaic Down Syndrome in a Patient With Low-Level Mosaicism Detected by Microarray. *American Journal of Medical Genetics Part A* **152A**(12):3154-6 (2010).

Bartsch O., et al. Four Unrelated Patients With Lubs X-Linked Mental Retardation Syndrome and Different Xq28 Duplications. *American Journal of Medical Genetics Part A* **152A**(2):305-12 (2010).

Fernandez B. A., et al. Phenotypic spectrum associated with de novo and inherited deletions and duplications at 16p11.2 in individuals ascertained for diagnosis of autism spectrum disorder. *Journal of Medical Genetics* **47**(3):195-203 (2010).

Eggermann T., et al. Identification of a 21q22 duplication in a Silver-Russell syndrome patient further narrows down the Down syndrome critical region. *American Journal of Medical Genetics Part A* **152A**(2):356-9 (2010).

Girirajan S., et al. A recurrent 16p12.1 microdeletion supports a two-hit model for severe developmental delay. *Nature Genetics* **42**(3):203-9 (2010).

Ellinor P. T., et al. Common variants in KCNN3 are associated with lone atrial fibrillation. *Nature Genetics* **42**(3):240-4 (2010).

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Berkel S., et al. Mutations in the SHANK2 synaptic scaffolding gene in autism spectrum disorder and mental retardation. *Nature Genetics* **42**(6):489-91 (2010).

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Pluzhnikov A., et al. Spoiling the whole bunch: quality control aimed at preserving the integrity of high-throughput genotyping. *American Journal of Human Genetics* **87**(1):123-8 (2010).

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Donahoe P. K., et al. Genetic tools and algorithms for gene discovery in major congenital anomalies. *Birth Defects Research Part A, Clinical and Molecular Teratology* **85**(1):6-12 (2009).

Johnson C., et al. Convergent genome wide association results for bipolar disorder and substance dependence. *American Journal of Medical Genetics Part B: Neuropsychiatric Genetics* **150B**(2):182-90 (2009).

Zhang D., et al. Singleton deletions throughout the genome increase risk of bipolar disorder. *Molecular Psychiatry* **14**(4):376-80 (2009).

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Mahon P. B., et al. Genome-wide linkage and follow-up association study of postpartum mood symptoms. *American Journal of Psychiatry* **166**(11):1229-37 (2009).

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Stone J. L., *et al.* Rare chromosomal deletions and duplications increase risk of schizophrenia. *Nature* **455**(7210):237-41 (2008).

Mick E., *et al.* Genome-wide association study of response to methylphenidate in 187 children with attention-deficit/hyperactivity disorder. *American Journal of Medical Genetics Part B: Neuropsychiatric Genetics* **147B**(8):1412-8 (2008).

## **Cardiovascular**

### **2011**

Fox E. R., *et al.* Association of genetic variation with systolic and diastolic blood pressure among African Americans: the Candidate Gene Association Resource study. *Human Molecular Genetics* **20**(11):2273-84 (2011).

Zhu X. F., *et al.* Combined admixture mapping and association analysis identifies a novel blood pressure genetic locus on 5p13: contributions from the CARE consortium. *Human Molecular Genetics* **20**(11):2285-95 (2011).

O'Seaghdha C. M., *et al.* The MYH9/APOL1 region and chronic kidney disease in European-Americans. *Human Molecular Genetics* **20**(12):2450-6 (2011).

Kato N., *et al.* Meta-analysis of genome-wide association studies identifies common variants associated with blood pressure variation in east Asians. *Nature Genetics* **43**(6) (2011).

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Ellinor P. T., *et al.* Common variants in KCNN3 are associated with lone atrial fibrillation. *Nature Genetics* **42**(3) (2010).

Arking D. E., *et al.* Genome-Wide Association Study Identifies GPC5 as a Novel Genetic Locus Protective against Sudden Cardiac Arrest. *PLoS One* **5**(3) (2010).

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Stewart A. F. R., *et al.* Kinesin Family Member 6 Variant Trp719Arg Does Not Associate With Angiographically Defined Coronary Artery Disease in the Ottawa Heart Genomics Study. *Journal of the American College of Cardiology* **53**(16):1471-2 (2009).

Erdmann J., *et al.* New susceptibility locus for coronary artery disease on chromosome 3q22.3. *Nature Genetics* **41**(3):280-2 (2009).

Kathiresan S., *et al.* Genome-wide association of early-onset myocardial infarction with single nucleotide polymorphisms and copy number variants. *Nature Genetics* **41**(3):334-41 (2009).

Pfeufer A., *et al.* Common variants at ten loci modulate the QT interval duration in the QTSCD Study. *Nature Genetics* **41**(4):407-14 (2009).

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Ikram M. A., *et al.* Genomewide Association Studies of Stroke. *New England Journal of Medicine* **360**(17):1718-28 (2009).

Shuldiner A. R., et al. Association of Cytochrome P450 2C19 Genotype With the Antiplatelet Effect and Clinical Efficacy of Clopidogrel Therapy. *Jama-Journal of the American Medical Association* **302**(8):849-58 (2009).

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Ganesh S. K., et al. Multiple loci influence erythrocyte phenotypes in the CHARGE Consortium. *Nature Genetics* **41**(11) (2009).

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## Epidemiology and population studies

### 2011

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Ou Z., et al. Observation and prediction of recurrent human translocations mediated by NAHR between nonhomologous chromosomes. *Genome Research* **21**(1):33-46 (2011).

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Li Y., et al. Low-coverage sequencing: Implications for design of complex trait association studies. *Genome Research* **21**(6):940-51 (2011).

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Vogler C., et al. Microarray-Based Maps of Copy-Number Variant Regions in European and Sub-Saharan Populations. *PLoS One* **5**(12) (2010).

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Rudd M. K., et al. Segmental duplications mediate novel, clinically relevant chromosome rearrangements. *Human Molecular Genetics* **18**(16):2957-62 (2009).

Price A. L., et al. The Impact of Divergence Time on the Nature of Population Structure: An Example from Iceland. *PLoS Genetics* **5**(6) (2009).

Xing J. C., et al. Fine-scaled human genetic structure revealed by SNP microarrays. *Genome Research* **19**(5):815-25 (2009).

Wain L. V., et al. Genomic copy number variation, human health, and disease. *Lancet* **374**(9686):340-50 (2009).

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Reich D., et al. Reconstructing Indian population history. *Nature* **461**(7263) (2009).

Teo Y. Y., et al. Singapore Genome Variation Project: A haplotype map of three Southeast Asian populations *Genome Research* **19**(11):2154-62 (2009).

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Ziegler A., et al. Biostatistical aspects of genome-wide association studies. *Biometrical Journal* **50**(1):8-28 (2008).

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## **Rare disorders**

## **2011**

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- Thie C., et al. NEK1 Mutations Cause Short-Rib Polydactyly Syndrome Type Majewski. *American Journal of Human Genetics* **88**(1):106-14 (2011).
- White S., et al. Copy Number Variation in Patients with Disorders of Sex Development Due to 46, XY Gonadal Dysgenesis. *PLoS One* **6**(3) (2011).
- Phadke S. R., et al. Report of Two Brothers With Short Stature, Microcephaly, Mental Retardation, and Retinoschisis-A New Mental Retardation Syndrome? *American Journal of Medical Genetics Part A* **155A**(1):9-13 (2011).
- Kayserili H., et al. A Novel Homozygous COL11A2 Deletion Causes a C-Terminal Protein Truncation With Incomplete mRNA Decay in a Turkish Patient. *American Journal of Medical Genetics Part A* **155A**(1):180-5 (2011).
- Sutton E., et al. Identification of SOX3 as an XX male sex reversal gene in mice and humans. *Journal of Clinical Investigation* **121**(1):328-41 (2011).
- Cox J. J., et al. A SOX9 Duplication and Familial 46,XX Developmental Testicular Disorder. *New England Journal of Medicine* **364**(1):91-3 (2011).
- Papenhausen P., et al. UPD Detection Using Homozygosity Profiling With a SNP Genotyping Microarray. *American Journal of Medical Genetics Part A* **155A**(4):757-68 (2011).
- Fabbro S., et al. Homozygosity mapping with SNP arrays confirms 3p21 as a recessive locus for gray platelet syndrome and narrows the interval significantly. *Blood* **117**(12):3430-4 (2011).
- Lynch S. A., et al. The 12q14 microdeletion syndrome: six new cases confirming the role of HMGA2 in growth. *European Journal of Human Genetics* **19**(5):534-9 (2011).
- Kariminejad A., et al. Pericentric inversion of chromosome 18 in parents leading to a phenotypically normal child with segmental uniparental disomy 18. *European Journal of Human Genetics* **19**(5):555-60 (2011).
- Bowen M. E., et al. Loss-of-Function Mutations in PTPN11 Cause Metachondromatosis, but Not Ollier Disease or Maffucci Syndrome. *PLoS Genetics* **7**(4) (2011).
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- ## 2010
- Eggermann T., et al. Identification of a 21q22 duplication in a Silver-Russell syndrome patient further narrows down the Down syndrome critical region. *American Journal of Medical Genetics Part A* **152A**(2):356-9 (2010).
- Latour P., et al. A Major Determinant for Binding and Aminoacylation of tRNA(Ala) in Cytoplasmic Alanyl-tRNA Synthetase Is Mutated in Dominant Axonal Charcot-Marie-Tooth Disease. *American Journal of Human Genetics* **86**(1):77-82 (2010).
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- Zou Y. S., et al. A Complex Maternal Rearrangement Results in a Pure 10.8 Mb Duplication of the 5q13.1-q14.1 Region in an Affected Son. *American Journal of Medical Genetics Part A* **152A**(2):498-503 (2010).

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