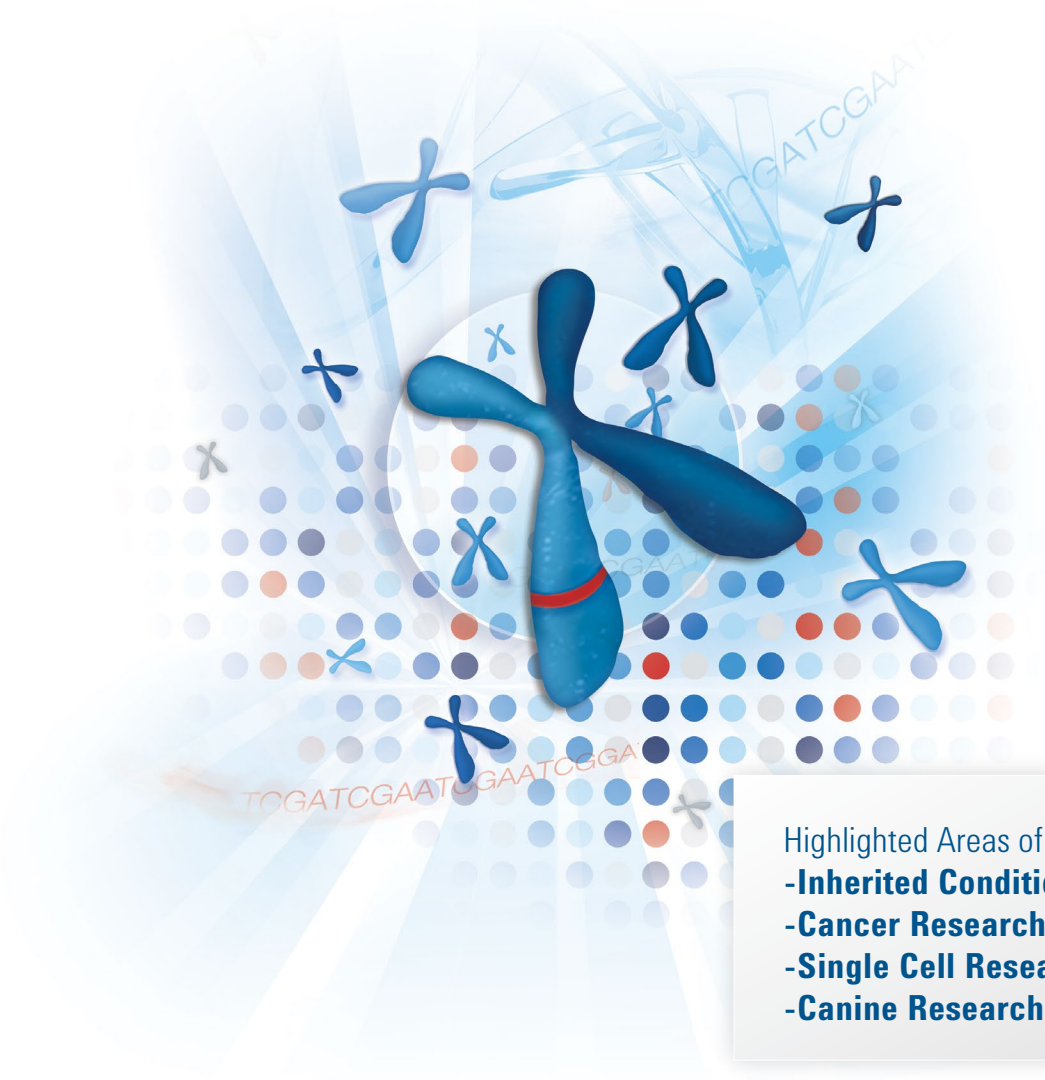
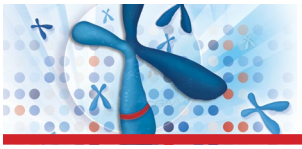
Publication List
CGH and CGH+SNP Microarrays

Revised June 2015



Highlighted Areas of Research
- **Inherited Condition Research**
- **Cancer Research**
- **Single Cell Research**
- **Canine Research**

Agilent Technologies



Inherited Condition Research

1. Wu N, Ming X, Xiao J, Wu Z, Chen X, Shinawi M, Shen Y, Yu G, Liu J, Xie H, Gucev ZS, Liu S, Yang N, Al-Kateb H, Chen J, Zhang J, Hauser N, Zhang T, Tasic V, Liu P, Su X, Pan X, Liu C, Wang L, Shen J, Shen J, Chen Y, Zhang T, Zhang J, Choy KW, Wang J, Wang Q, Li S, Zhou W, Guo J, Wang Y, Zhang C, Zhao H, An Y, Zhao Y, Wang J, Liu Z, Zuo Y, Tian Y, Weng X, Sutton VR, Wang H, Ming Y, Kulkarni S, Zhong TP, Giampietro PF, Dunwoodie SL, Cheung SW, Zhang X, Jin L, Lupski JR, Qiu G, and Zhang F. **TBX6 null variants and a common hypomorphic allele in congenital scoliosis.** *N Engl J Med.* (2015) 372(4):341-50 ABSTRACT

2. Démurger F, Ichkou A, Mougou-Zerelli S, Le Merrer M, Goudefroye G, Delezoide AL, Quélin C, Manouvrier S, Baujat G, Fradin M, Pasquier L, Megarbané A, Faivre L, Baumann C, Nampoothiri S, Roume J, Isidor B, Lacombe D, Delrue MA, Mercier S, Philip N, Schaefer E, Holder M, Krause A, Laffargue F, Sinico M, Amram D, André G, Liquier A, Rossi M, Amiel J, Giuliano F, Boute O, Dieux-Coeslier A, Jacquemont ML, Afenjar A, Van Maldergem L, Lackmy-Port-Lis M, Vincent-Delorme C, Chauvet ML, Cormier-Daire V, Devisme L, Geneviève D, Munnich A, Viot G, Raoul O, Romana S, Gonzales M, Encha-Razavi F, Odent S, Vekemans M, and Attie-Bitach T. **New insights into genotype-phenotype correlation for GLI3 mutations.** *Eur J Hum Genet.* (2015) 23(1):92-102 ABSTRACT

3. Pons L, Cordier MP, Labalme A, Till M, Louvrier C, Schluth-Bolard C, Lesca G, Edery P, and Sanlaville D. **A new syndrome of intellectual disability with dysmorphism due to TBL1XR1 deletion.** *Am J Med Genet A.* (2015) 167A(1):164-8. ABSTRACT

4. van Haelst MM, Monroe GR, Duran K, van Binsbergen E, Breur JM, Giltay JC, van Haaften G. **Further confirmation of the MED13L haploinsufficiency syndrome.** *Eur J Hum Genet.* (2015) 23(1):135-8. ABSTRACT

5. Lombardo B, Zarrilli F, Ceglia C, Vitale A, Keller S, Sarchiapone M, Carli V, Stuppia L, Chiariotti L, Castaldo G, and Pastore L. **Two novel genomic rearrangements identified in suicide subjects using a-CGH array.** *Clin Chem Lab Med.* (2015) ABSTRACT

6. Nishimoto HK, Ha K, Jones JR, Dwivedi A, Cho H-M, Layman LC, and Kim H-G. **The historical Coffin–Lowry syndrome family revisited: identification of two novel mutations of RPS6KA3 in three male patients.** *Am. J. Med. Genet.* (2014) 164A:2172–2179 ABSTRACT

7. Olson H, Shen Y, Avallone J, Sheidley BR, Pinsky R, Bergin AM, Berry GT, Duffy FH, Eksioglu Y, Harris DJ, Hisama FM, Ho E, Irons M, Jacobsen CM, James P, Kothare S, Khwaja O, Lipton J, Loddenkemper T, Markowitz J, Maski K, Megerian T, Neilan E, Raffalli PC, Robbins M, Roberts A, Roe E, Rollins C, Sahin M, Sarco D, Schonwald A, Smith SE, Soul J, Stoler JM, Takeoka M, Tan W, Torres AR, Tsai P, Urion DK, Weissman L, Wolff R, Wu B, Miller DT, and Poduri A. **Copy number variation plays an important role in clinical epilepsy.** *Ann. Neurol.* (2014) 75:943–958 ABSTRACT

8. Vincent M, Collet C, Verloes A, Lambert L, Herlin C, Blanchet C, Sanchez E, Drunat S, Vigneron J, Laplanche JL, Puechberty J, Sarda P, and Geneviève D. **Large deletions encompassing the TCOF1 and CAMK2A genes are responsible for Treacher Collins syndrome with intellectual disability.** *Eur J Hum Genet.* (2014) 22(1):52-6 ABSTRACT

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10. Rogers AJ, Chu JH, Darvishi K, Ionita-Laza I, Lehmann H, Mills R, Lee C, and Raby BA. **Copy number variation prevalence in known asthma genes and their impact on asthma susceptibility.** *Clin. and Exp. Allergy.* (2013) 43:455–62 ABSTRACT

Cancer Research

1. Wanjala J, Taylor BS, Chapinski C, Hieronymus H, Wongvipat J, Chen Y, Nanjangud GJ, Schultz N, Xie Y, Liu S, Lu W, Yang Q, Sander C, Chen Z, Sawyers CL, and Carver BS. **Identifying actionable targets through integrative analyses of GEM model and human prostate cancer genomic profiling.** *Mol Cancer Ther.* (2015) 14(1):278-88 ABSTRACT

2. Ilie M, Nunes M, Blot L, Hofman V, Long-Mira E, Butori C, Selva E, Merino-Trigo A, Vénissac N, Mouroux J, Vrignaud P, and Hofman P. **Setting up a wide panel of patient-derived tumor xenografts of non-small cell lung cancer by improving the preanalytical steps.** *Cancer Med.* (2015) 4(2):201-11 ABSTRACT

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4. Verbeke SL, de Jong D, Bertoni F, Sciot R, Antonescu CR, Szuhai K, and Bovée JV. **Array CGH analysis identifies two distinct subgroups of primary angiosarcoma of bone.** *Genes Chromosomes Cancer.* (2015) 54(2):72-81 ABSTRACT

5. Choy E, MacConaill LE, Cote GM, Le LP, Shen JK, Nielsen GP, Lafrate AJ, Garraway LA, Hornicek FJ, and Duan Z. **Genotyping cancer-associated genes in chordoma identifies mutations in oncogenes and areas of chromosomal loss involving CDKN2A, PTEN, and SMARCB1.** *PLoS ONE* (2014) 9(7): e101283. doi:10.1371/ journal.pone.0101283 ABSTRACT

6. Conconi D, Panzeri E, Redaelli S, Bovo G, Viganò P, Strada G, Dalprà L, and Bentivegna A. **Chromosomal imbalances in human bladder urothelial carcinoma: similarities and differences between biopsy samples and cancer stem-like cells.** *BMC Cancer* (2014) 14:646 ABSTRACT

7. Jang SH, Park JW, Kim HR, Seong JK, and Kim HK. **ADRM1 gene amplification is a candidate driver for metastatic gastric cancers.** *Clin. Exp. Metastasis.* (2014) 31:727–733 ABSTRACT

8. Sun I, Li M, Huang X, Xu J, Gao Z, and Liu C. **High-resolution genome-wide analysis identified recurrent genetic alterations in NK/T-cell lymphoma, nasal type, which are associated with disease progression.** *Med. Oncol.* (2014) 31:71 ABSTRACT

9. Camps J, Pitt JJ, Emons G, Hummon AB, Case CM, Grade M, Jones TL, Nguyen QT, Ghadimi BM, Beissbarth T, Difilippantonio MJ, Caplen NJ, and Ried T. **Genetic amplification of the NOTCH modulator LNX2 upregulates the WNT/β- Catenin pathway in colorectal cancer.** *Cancer Res.* (2013) 73: 2003–2013 ABSTRACT

10. Errami Y, Brim H, Oumouna-Benachour K, Oumouna M, Naura AS, Kim H, Davis CJ, Kim JG,Ashktorab H, Fallon K, Xu M, Zhang J, Del Valle L, and Boulares AH. **ICAD deficiency in human colon cancer and predisposition to colon tumorigenesis: Linkage to apoptosis resistance and genomic instability.** *PLoS ONE* (2013) 8: e57871. doi:10.1371/journal. pone.0057871 ABSTRACT

11. Hosein AN, Song S, McCart Reed AE, Jayanthan J, Reid LE, Kutasovic JR, Cummings MC, Waddell N, Lakhani SR, Chenevix-Trench G, and Simpson PT. **Evaluating the repair of DNA derived from formalin-fixed paraffin-embedded tissues prior to genomic profiling by SNP–CGH analysis.** *Lab Investigation* (2013) 93: 701–710 ABSTRACT

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13. Pan CC, and Epstein JI. **Common chromosomal aberrations detected by array comparative genomic hybridization in specialized stromal tumors of the prostate.** *Modern Pathology* (2013) 26: 1536–1543 ABSTRACT

14. Petrini I, Wang Y, Zucali PA, Lee HS, Pham T, Voeller D, Meltzer PS, and Giaccone G. **Copy number aberrations of genes regulating normal thymus development in thymic epithelial tumors.** *Clin. Cancer Res.* (2013) 19: 1960–1971 ABSTRACT

15. Salaverria I, Martin-Guerrero I, Burkhardt B, Kreuz M, Zenz T, Oschlies I, Arnold N, Baudis M, Bens S, Garcıa-Orad A, Lisfeld J, Schwaenen C, Szczepanowski M, Wessendorf S, Pfreundschuh M, Trumper L, Klapper W, and Siebert R. **High resolution copy number analysis of IRF4 translocation-positive diffuse large B-cell and follicular lymphomas.** *Genes Chromosomes Cancer.* (2013) 52:150–155 ABSTRACT

16. Spaepen M, Xavier NE, Sagaert X, Hertogh GD, Beert E, Wimmer K, Matthijs G, Legius E, and Brems H. **EPCAM germline and somatic rearrangements in lynch syndrome: identification of a novel 3'EPCAM deletion.** *Genes Chromosomes Cancer.* (2013) 52: 845–54 ABSTRACT

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19. Czyż ZT, Hoffmann M, Schlimok G, Polzer B, and Klein CA. **Reliable single cell array CGH for clinical samples.** *PLoS One.* (2014) Jan 21;9(1):e85907 ABSTRACT

20. Pećina-Šlaus N, Gotovac K, Kafka A, Tomas D, and Borovečki F. **Genetic changes observed in a case of adult pilocytic astrocytoma revealed by array CGH analysis.** *Mol Cytogenet.* (2014) 23;7(1):95 ABSTRACT

Single Cell Research

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Stem Cell Research

1. Baronchelli S, Bentivegna A, Redaelli S, Riva G, Butta V, Paoletta L, Isimbaldi G, Miozzo M, Tabano S, Daga A, Marubbi D, Cattaneo M, Biunno I, and Dalprà L. **Delineating the cytogenomic and epigenomic landscapes of glioma stem cell lines.** (2013) *PLoS ONE* 8(2): e57462. doi:10.1371/journal. pone.0057462 ABSTRACT

2. Estrada JC, Torres Y, Benguría A, Dopazo A, Roche E, Carrera-Quintanar L, Pérez RA, Enríquez JA, Torres R, Ramírez JC, Samper E, and Bernad A. **Human mesenchymal stem cell-replicative senescence and oxidative stress are closely linked to aneuploidy.** *Cell Death and Disease* (2013) 4, e691; doi:10.1038/ cddis.2013.211 ABSTRACT

3. Guo CW, Kawakatsu M, Idemitsu M, Urata Y, Goto S, Ono Y, Hamano K, and Li T-S. **Culture under low physiological oxygen conditions improves the stemness and quality of induced pluripotent stem cells.** *J Cell. Physio.* (2013) 228:2159–66. ABSTRACT

Canine Research

1. Poorman K, Borst L, Moroff S, Roy S, Labelle P, Motsinger-Reif A, and Breen M. **Comparative cytogenetic characterization of primary canine melanocytic lesions using array CGH and fluorescence in situ hybridization.** *Chromosome Res.* (2014) Dec 16 ABSTRACT

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