CURRICULUM VITAE
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ACADEMIC APPOINTMENTS

2013-Present	Professor, The Jackson Laboratory for Genomic Medicine
2015-Present	Visiting Professor, EWHA Womans University, Seoul, South Korea
2013-2015	Visiting University Professor, Seoul National University, Seoul, South Korea
2008-2013	Associate Professor, Pathology, Harvard Medical School
2003-2008	Assistant Professor, Pathology, Harvard Medical School
2001-2003	Instructor, Pathology, Harvard Medical School

OTHER APPOINTMENTS/EMPLOYMENT/EXPERIENCE

2015-Present	Section Director, Clinical Cytogenetics Laboratory, The Jackson Laboratory for Genomic Medicine
2013-Present	Director, The Jackson Laboratory for Genomic Medicine
2011-Present	Honorary Professor, Chinese University of Hong Kong
2011-Present	Steering Committee, 1000 Genomes Project
2008-Present	Co-Chair, SV analysis group, 1000 Genomes Project
2006-Present	Associate Member, Broad Institute of Harvard and MIT
2010	Chair, Program Committee, American Society of Human Genetics
2009	Chair, Cytogenetics Core Advisory Board, NHGRI
2009-2013	Director, Molecular Genetic Research Unit, Brigham and Women's Hospital
2008-2013	Clinical Cytogeneticist, Pathology, Brigham and Women's Hospital
2007-2011	Honorary Associate Professor, Chinese University of Hong Kong
2006-2013	Director, DF/HCC Cytogenetics Core, Dana Farber/Harvard Cancer Center
2001-2008	Associate Clinical Cytogeneticist, Pathology, Brigham and Women's Hospital
2000-2006	Assistant Director, DF/HCC Cytogenetics Core, Dana Farber/Harvard Cancer Center
04/93-07/93	Invited Research Trainee, Johns Hopkins School of Medicine

EDUCATION

Postdoctoral Training

1998-2001	Research Fellow, Obstetrics, Gynecology and Reproductive Biology (Dr. Cynthia C. Morton), Harvard Medical School
1996-1998	Research Fellow, Molecular Cytogenetics (Prof. Malcolm Ferguson-Smith), Cambridge University

Other

1996	Ph.D., Medical Sciences, University of Alberta, Canada
1993	M.S., Experimental Pathology, University of Alberta, Canada
1990	B.S., Genetics, University of Alberta, Canada

PRINT AND ORAL SCHOLARSHIP

1. Tai DJ, Ragavendran A, Manavalan P, Stortchevoi A, Seabra CM, Erdin S, Collins RL, Blumenthal, I, Chen X, Shen Y, Sahin M, Chang C, Lee C, Guselle JF, Talkowski ME. Engineering microdeletions and microduplications by targeting segmental duplications with CRISPR. Nature Neuroscience. 2016 February.

2. Sudmant PH, Rausch T, Gardner EJ, Handsaker RE, Abyzov A, … , *Mills RE, *Gerstein M, *Bashir A, *Stegle O, *Devine SE, *Lee C, *Eichler EE, *Korbel JO. An integrated map of structural variation in 2,504 human genomes. Nature. 2015 Oct. 1;526 (7571) 75-81 **co-senior author*

3. 1000 Genomes Project Consortium. A global reference for human genetic variation. Nature. 2015 Oct. 1;526 (7571) 68-74

4. Park H, Cho S, Kim H, Na D, Han J, Chae J, Park C, Park O, Min S, Kang J, Choi B, Min J, Kown J, Suh Y, Kong S, Lee H, Liu E, Kim J, Kim S, Yang H, Lee C*. Genomic alteration in BCL2L1 and DLC1 contribute to drug sensitivity in gastric cancer. PNAS 2015 Sept.; 112 (40) 12492-12497

5. Brand H, Pillalamarri V, Collins RL, Eggert S, O'Dushlaine C, Braaten EB, Stone MR, Chambert K, Doty ND, Hanscom C, Rosenfeld JA, Ditmars H, Blais J, Mills R, Lee C, Gusella JF, McCarroll S, Smoller JW, Talkowski ME, Doyle AE. Cryptic and complex chromosomal aberrations in early-onset neuropsychiatric disorders. Am J Hum Genet. 2014 Oct 2;95(4):454-61. PMCID: PMC4185111

6. Longoni M, High FA, Russell MK, Kashani A, Tracy AA, Coletti CM, Hila R, Shamia A, Wells J, Ackerman KG, Wilson JM, Bult CJ, Lee C, Lage K, Pober BR, Donahoe PK. Molecular pathogenesis of congenital diaphragmatic hernia revealed by exome sequencing, developmental data, and bioinformatics. Proc Natl Acad Sci U S A. 2014 Aug 26;111(34):12450-5. PMCID: PMC4151769

7. Blackburn JS, Liu S, Wilder JL, Dobrinski KP, Lobbardi R, Moore FE, Martinez SA, Chen EY, Lee C, Langenau DM. Clonal evolution enhances leukemia-propagating cell frequency in T cell acute lymphoblastic leukemia through Akt/mTORC1 pathway. Cancer Cell. 2014 Mar 6. PMCID: PMC3992437

8. Silva AG, Krepischi AC, Torrezan GT, Capelli LP, Carraro DM, D'Angelo CS, Koiffmann CP, Zatz M, Naslavsky MS, Masotti C, Otto PA, Achatz MI, Mills RE, Lee C, Pearson PL, Rosenberg

C. Does germ-line deletion of the PIP gene constitute a widespread risk for cancer? Eur J Hum Genet. 2014 Mar;22(3):307-9. PMCID: PMC3925262

9. Korbel JO, Lee C. Genome assembly and haplotyping with Hi-C. Nat Biotechnol. 2013 Dec;31(12):1099-101.

10. Abyzov A, Iskow R, Gokcumen O, Radke DW, Balasubramanian S, Pei B, Habegger L; 1000 Genomes Project Consortium, *Lee C, Gerstein M. Analysis of variable retroduplications in human populations suggests coupling of retrotransposition to cell division. Genome Res. 2013 Dec;23(12):2042-52. PMCID: PMC3847774

11. Gokcumen O, Tischler V, Tica J, Zhu Q, Iskow RC, Lee E, Fritz MH, Langdon A, Stütz AM, Pavlidis P, Benes V, Mills RE, Park PJ, *Lee C, Korbel JO. Primate genome architecture influences structural variation mechanisms and functional consequences. Proc Natl Acad Sci U S A. 2013 Sep 24;110(39):15764-9. PMCID: PMC3785719

12. Chu JH, Rogers A, Ionita-Laza I, Darvishi K, Mills RE, Lee C, Raby BA. Copy number variation genotyping using family information. BMC Bioinformatics. 2013 May 9;14:157. PMCID: PMC3668900

13. Yang L, Luquette LJ, Gehlenborg N, Xi R, Haseley PS, Hsieh CH, Zhang C, Ren X, Protopopov A, Chin L, Kucherlapati R, Lee C, Park PJ. Diverse mechanisms of somatic structural variations in human cancer genomes. Cell. 2013 May 9;153(4):919-29. PMCID: PMC3704973

14. Howe K, Clark MD, Torroja CF, Torrance J, Berthelot C, Muffato M, Collins JE, Humphray S, McLaren K, Matthews L, McLaren S, Sealy I, Caccamo M, Churcher C, Scott C, Barrett JC, Koch R, Rauch GJ, White S, Chow W, Kilian B, Quintais LT, Guerra-Assunção JA, Zhou Y, Gu Y, Yen J, Vogel JH, Eyre T, Redmond S, Banerjee R, Chi J, Fu B, Langley E, Maguire SF, Laird GK, Lloyd D, Kenyon E, Donaldson S, Sehra H, Almeida-King J, Loveland J, Trevanion S, Jones M, Quail M, Willey D, Hunt A, Burton J, Sims S, McLay K, Plumb B, Davis J, Clee C, Oliver K, Clark R, Riddle C, Elliot D, Threadgold G, Harden G, Ware D, Begum S, Mortimore B, Kerry G, Heath P, Phillimore B, Tracey A, Corby N, Dunn M, Johnson C, Wood J, Clark S, Pelan S, Griffiths G, Smith M, Glithero R, Howden P, Barker N, Lloyd C, Stevens C, Harley J, Holt K, Panagiotidis G, Lovell J, Beasley H, Henderson C, Gordon D, Auger K, Wright D, Collins J, Raisen C, Dyer L, Leung K, Robertson L, Ambridge K, Leongamornlert D, McGuire S, Gilderthorp R, Griffiths C, Manthravadi D, Nichol S, Barker G, Whitehead S, Kay M, Brown J, Murnane C, Gray E, Humphries M, Sycamore N, Barker D, Saunders D, Wallis J, Babbage A, Hammond S, Mashreghi-Mohammadi M, Barr L, Martin S, Wray P, Ellington A, Matthews N, Ellwood M, Woodmansey R, Clark G, Cooper J, Tromans A, Grafham D, Skuce C, Pandian R, Andrews R, Harrison E, Kimberley A, Garnett J, Fosker N, Hall R, Garner P, Kelly D, Bird C, Palmer S, Gehring I, Berger A, Dooley CM, Ersan-Ürün Z, Eser C, Geiger H, Geisler M, Karotki L, Kirn A, Konantz J, Konantz M, Oberländer M, Rudolph-Geiger S, Teucke M, Lanz C, Raddatz G, Osoegawa K, Zhu B, Rapp A, Widaa S, Langford C, Yang F, Schuster SC, Carter NP, Harrow J, Ning Z, Herrero J, Searle SM, Enright A, Geisler R, Plasterk RH, Lee C, Westerfield M, de Jong PJ, Zon LI, Postlethwait JH, Nüsslein-Volhard C, Hubbard TJ, Roest Crollius H, Rogers J, Stemple DL. The zebrafish reference genome sequence and its relationship to the human genome. Nature. 2013 Apr 25;496(7446):498-503. PMCID: PMC3703927

15. Gokcumen O, Zhu Q, Mulder LC, Iskow RC, Austermann C, Scharer CD, Raj T, Boss JM, Sunyaev S, Price A, Stranger B, Simon V, *Lee C. Balancing selection on a regulatory region exhibiting ancient variation that predates human-neandertal divergence. PLoS Genet. 2013 Apr;9(4):e1003404. PMCID: PMC3623772

16. Longoni M, Lage K, Russell MK, Loscertales M, Abdul-Rahman OA, Baynam G, Bleyl SB, Brady PD, Breckpot J, Chen CP, Devriendt K, Gillessen-Kaesbach G, Grix AW, Noonan KM, Park JH, Tracy AA, Lee C, Donahoe PK, Pober BR. Congenital diaphragmatic hernia interval on chromosome 8p23.1 characterized by genetics and protein interaction networks. Am J Med Genet A. 2012 Dec;158A(12):3148-58. PMCID: PMC3761361

17. 1000 Genomes Project Consortium, Abecasis GR, Auton A, Brooks LD, DePristo MA, Durbin RM, Handsaker RE, Kang HM, Marth GT, McVean GA. An integrated map of genetic variation from 1,092 human genomes. Nature. 2012 Nov 1;491(7422):56-65. PMCID: PMC3498066

18. Lee E, Iskow R, Yang L, Gokcumen O, Haseley P, Luquette LJ 3rd, Lohr JG, Harris CC, Ding L, Wilson RK, Wheeler DA, Gibbs RA, Kucherlapati R, Lee C, Kharchenko PV, Park PJ; Cancer Genome Atlas Research Network. Landscape of somatic retrotransposition in human cancers. Science. 2012 Aug 24;337(6097):967-71. PMC3656569

19. Iskow RC, Gokcumen O, Abyzov A, Malukiewicz J, Zhu Q, Sukumar AT, Pai AA, Mills RE, Habegger L, Cusanovich DA, Rubel MA, Perry GH, Gerstein M, Stone AC, Gilad Y, *Lee C. Regulatory element copy number differences shape primate expression profiles. Proc Natl Acad Sci U S A. 2012 Jul 31;109(31):12656-61. PMCID: PMC3411951

20. Perry GH, Xue Y, Smith RS, Meyer WK, Calışkan M, Yanez-Cuna O, Lee AS, Gutiérrez-Arcelus M, Ober C, Hollox EJ, Tyler-Smith C, *Lee C. Evolutionary genetics of the human Rh blood group system. Hum Genet. 2012 Jul;131(7):1205-16. PMCID: PMC3378649

21. Iskow RC, Gokcumen O, *Lee C. Exploring the role of copy number variants in human adaptation. Trends Genet. 2012 Jun;28(6):245-57. PMCID: PMC3533238

22. Setlur SR, *Lee C. Tumor archaeology reveals that mutations love company. Cell. 2012 May 25;149(5):959-61.

23. Demichelis F, Setlur SR, Banerjee S, Chakravarty D, Chen JY, Chen CX, Huang J, Beltran H, Oldridge DA, Kitabayashi N, Stenzel B, Schaefer G, Horninger W, Bektic J, Chinnaiyan AM, Goldenberg S, Siddiqui J, Regan MM, Kearney M, Soong TD, Rickman DS, Elemento O, Wei JT, Scherr DS, Sanda MA, Bartsch G, Lee C, Klocker H, Rubin MA. Identification of functionally active, low frequency copy number variants at 15q21.3 and 12q21.31 associated with prostate cancer risk. Proc Natl Acad Sci U S A. 2012 Apr 24;109(17):6686-91. PMCID: PMC3340033

24. Chen Z, Cheng K, Walton Z, Wang Y, Ebi H, Shimamura T, Liu Y, Tupper T, Ouyang J, Li J, Gao P, Woo MS, Xu C, Yanagita M, Altabef A, Wang S, Lee C, Nakada Y, Peña CG, Sun Y, Franchetti Y, Yao C, Saur A, Cameron MD, Nishino M, Hayes DN, Wilkerson MD, Roberts PJ, Lee CB, Bardeesy N, Butaney M, Chirieac LR, Costa DB, Jackman D, Sharpless NE, Castrillon DH, Demetri GD, Jänne PA, Pandolfi PP, Cantley LC, Kung AL, Engelman JA, Wong KK. A murine

lung cancer co-clinical trial identifies genetic modifiers of therapeutic response. Nature. 2012 Mar 18;483(7391):613-7. PMCID: PMC3385933

25. Chiang C, Jacobsen JC, Ernst C, Hanscom C, Heilbut A, Blumenthal I, Mills RE, Kirby A, Lindgren AM, Rudiger SR, McLaughlan CJ, Bawden CS, Reid SJ, Faull RL, Snell RG, Hall IM, Shen Y, Ohsumi TK, Borowsky ML, Daly MJ, Lee C, Morton CC, MacDonald ME, Gusella JF, Talkowski ME. Complex reorganization and predominant non-homologous repair following chromosomal breakage in karyotypically balanced germline rearrangements and transgenic integration. Nat Genet. 2012 Mar 4;44(4):390-7, S1. PMCID: PMC3340016

26. Brown KH, Dobrinski KP, Lee AS, Gokcumen O, Mills RE, Shi X, Chong WW, Chen JY, Yoo P, David S, Peterson SM, Raj T, Choy KW, Stranger BE, Williamson RE, Zon LI, Freeman JL, *Lee C. Extensive genetic diversity and substructuring among zebrafish strains revealed through copy number variant analysis. Proc Natl Acad Sci U S A. 2012 Jan 10;109(2):529-34. PMCID: PMC3258620

27. Frazer JK, Batchelor LA, Bradley DF, Brown KH, Dobrinski KP, Lee C, Trede NS. Genomic amplification of an endogenous retrovirus in zebrafish T-cell malignancies. Adv Hematol. 2012;2012:627920. PMCID: PMC3382231

28. Rudner LA, Brown KH, Dobrinski KP, Bradley DF, Garcia MI, Smith AC, Downie JM, Meeker ND, Look AT, Downing JR, Gutierrez A, Mullighan CG, Schiffman JD, Lee C, Trede NS, Frazer JK. Shared acquired genomic changes in zebrafish and human T-ALL. Oncogene. 2011 Oct 13;30(41):4289-96.

29. Ju YS, Kim JI, Kim S, Hong D, Park H, Shin JY, Lee S, Lee WC, Kim S, Yu SB, Park SS, Seo SH, Yun JY, Kim HJ, Lee DS, Yavartanoo M, Kang HP, Gokcumen O, Govindaraju DR, Jung JH, Chong H, Yang KS, Kim H, Lee C, Seo JS. Extensive genomic and transcriptional diversity identified through massively parallel DNA and RNA sequencing of eighteen Korean individuals. Nat Genet. 2011 Jul 3;43(8):745-52.

30. Pinto D, Darvishi K, Shi X, Rajan D, Rigler D, Fitzgerald T, Lionel AC, Thiruvahindrapuram B, Macdonald JR, Mills R, Prasad A, Noonan K, Gribble S, Prigmore E, Donahoe PK, Smith RS, Park JH, Hurles ME, Carter NP, Lee C, Scherer SW, Feuk L. Comprehensive assessment of array-based platforms and calling algorithms for detection of copy number variants. Nat Biotechnol. 2011 May 8;29(6):512-20. PMCID: PMC3270583

31. Mills RE, Walter K, Stewart C, Handsaker RE, Chen K, Alkan C, Abyzov A, Yoon SC, Ye K, Cheetham RK, Chinwalla A, Conrad DF, Fu Y, Grubert F, Hajirasouliha I, Hormozdiari F, Iakoucheva LM, Iqbal Z, Kang S, Kidd JM, Konkel MK, Korn J, Khurana E, Kural D, Lam HY, Leng J, Li R, Li Y, Lin CY, Luo R, Mu XJ, Nemesh J, Peckham HE, Rausch T, Scally A, Shi X, Stromberg MP, Stütz AM, Urban AE, Walker JA, Wu J, Zhang Y, Zhang ZD, Batzer MA, Ding L, Marth GT, McVean G, Sebat J, Snyder M, Wang J, Ye K, *Eichler EE, *Gerstein MB, *Hurles ME, *Lee C, *McCarroll SA, *Korbel JO; 1000 Genomes Project. Mapping copy number variation by population-scale genome sequencing. Nature. 2011 Feb 3;470(7332):59-65. PMCID: PMC3077050 **co-senior author*

32. Yu L, Reader JC, Chen C, Zhao XF, Ha JS, Lee C, York T, Gojo I, Baer MR, Ning Y. Activation of a novel palmitoyltransferase ZDHHC14 in acute biphenotypic leukemia and subsets of acute myeloid leukemia. Leukemia. 2011 Feb;25(2):367-71.

33. Dobrinski KP, Brown KH, Freeman JL, *Lee C. Molecular cytogenetic methodologies and a BAC probe panel resource for genomic analyses in the zebrafish. Methods Cell Biol. 2011;104:237-57.

34. Gokcumen O, Babb PL, Iskow RC, Zhu Q, Shi X, Mills RE, Ionita-Laza I, Vallender EJ, Clark AG, Johnson WE, *Lee C. Refinement of primate copy number variation hotspots identifies candidate genomic regions evolving under positive selection. Genome Biol. 2011;12(5):R52. PMCID: PMC3219974

35. Ordulu Z, Dal Cin P, Chong WW, Choy KW, Lee C, Muto MG, Quade BJ, Morton CC. Disseminated peritoneal leiomyomatosis after laparoscopic supracervical hysterectomy with characteristic molecular cytogenetic findings of uterine leiomyoma. Genes Chromosomes Cancer. 2010 Dec;49(12):1152-60. PMCID: PMC2955189

36. Setlur SR, Ihm C, Tchinda J, Shams S, Werner L, Cho EK, Thompson C, Phillips K, Rassenti LZ, Kipps TJ, Neuberg D, Freedman AS, Lee C, Brown JR. Comparison of familial and sporadic chronic lymphocytic leukaemia using high resolution array comparative genomic hybridization. Br J Haematol. 2010 Nov;151(4):336-45. PMCID: PMC3584328

37. 1000 Genomes Project Consortium, Abecasis GR, Altshuler D, Auton A, Brooks LD, Durbin RM, Gibbs RA, Hurles ME, McVean GA. A map of human genome variation from population-scale sequencing. Nature. 2010 Oct 28;467(7319):1061-73. PMCID: PMC3042601

38. Ligon AH, Morton CC, Bieber FR, Fletcher JA, Giersch AB, Kantarci S, Leach N, Lee C, Sandstrom M, Weremowicz S, Xiao S, Dal Cin P. Reporting of diagnostic cytogenetic results. Curr Protoc Hum Genet. 2010 Oct; Appendix 1:1D.1-23.

39. International HapMap 3 Consortium, Altshuler DM, Gibbs RA, Peltonen L, Altshuler DM, Gibbs RA, Peltonen L, Dermitzakis E, Schaffner SF, Yu F, Peltonen L, Dermitzakis E, Bonnen PE, Altshuler DM, Gibbs RA, de Bakker PI, Deloukas P, Gabriel SB, Gwilliam R, Hunt S, Inouye M, Jia X, Palotie A, Parkin M, Whittaker P, Yu F, Chang K, Hawes A, Lewis LR, Ren Y, Wheeler D, Gibbs RA, Muzny DM, Barnes C, Darvishi K, Hurles M, Korn JM, Kristiansson K, Lee C, McCarrol SA, Nemesh J, Dermitzakis E, Keinan A, Montgomery SB, Pollack S, Price AL, Soranzo N, Bonnen PE, Gibbs RA, Gonzaga-Jauregui C, Keinan A, Price AL, Yu F, Anttila V, Brodeur W, Daly MJ, Leslie S, McVean G, Moutsianas L, Nguyen H, Schaffner SF, Zhang Q, Ghori MJ, McGinnis R, McLaren W, Pollack S, Price AL, Schaffner SF, Takeuchi F, Grossman SR, Shlyakhter I, Hostetter EB, Sabeti PC, Adebamowo CA, Foster MW, Gordon DR, Licinio J, Manca MC, Marshall PA, Matsuda I, Ngare D, Wang VO, Reddy D, Rotimi CN, Royal CD, Sharp RR, Zeng C, Brooks LD, McEwen JE. Integrating common and rare genetic variation in diverse human populations. Nature. 2010 Sep 2;467(7311):52-8. PMCID: PMC3173859

40. *Lee C. The future of prenatal cytogenetic diagnostics: a personal perspective. Prenat Diagn. 2010 Jul;30(7):706-9. PMCID: PMC2893288

41. Sholl LM, Xiao Y, Joshi V, Yeap BY, Cioffredi LA, Jackman DM, Lee C, Jänne PA, Lindeman NI. EGFR mutation is a better predictor of response to tyrosine kinase inhibitors in non-small cell lung carcinoma than FISH, CISH, and immunohistochemistry. Am J Clin Pathol. 2010 Jun;133(6):922-34. PMCID: PMC3156055

42. Miller DT, Adam MP, Aradhya S, Biesecker LG, Brothman AR, Carter NP, Church DM, Crolla JA, Eichler EE, Epstein CJ, Faucett WA, Feuk L, Friedman JM, Hamosh A, Jackson L, Kaminsky EB, Kok K, Krantz ID, Kuhn RM, Lee C, Ostell JM, Rosenberg C, Scherer SW, Spinner NB, Stavropoulos DJ, Tepperberg JH, Thorland EC, Vermeesch JR, Waggoner DJ, Watson MS, Martin CL, Ledbetter DH. Consensus statement: chromosomal microarray is a first-tier clinical diagnostic test for individuals with developmental disabilities or congenital anomalies. Am J Hum Genet. 2010 May 14;86(5):749-64. PMCID: PMC2869000

43. Park H, Kim JI, Ju YS, Gokcumen O, Mills RE, Kim S, Lee S, Suh D, Hong D, Kang HP, Yoo YJ, Shin JY, Kim HJ, Yavartanoo M, Chang YW, Ha JS, Chong W, Hwang GR, Darvishi K, Kim H, Yang SJ, Yang KS, Kim H, Hurles ME, Scherer SW, Carter NP, Tyler-Smith C, *Lee C, Seo JS. Discovery of common Asian copy number variants using integrated high-resolution array CGH and massively parallel DNA sequencing. Nat Genet. 2010 May;42(5):400-5. PMCID: PMC3329635

44. Conrad DF, Bird C, Blackburne B, Lindsay S, Mamanova L, Lee C, Turner DJ, Hurles ME. Mutation spectrum revealed by breakpoint sequencing of human germline CNVs. Nat Genet. 2010 May;42(5):385-91. PMCID: PMC3428939

45. Ercan D, Zejnullahu K, Yonesaka K, Xiao Y, Capelletti M, Rogers A, Lifshits E, Brown A, Lee C, Christensen JG, Kwiatkowski DJ, Engelman JA, Jänne PA. Amplification of EGFR T790M causes resistance to an irreversible EGFR inhibitor. Oncogene. 2010 Apr 22;29(16):2346-56. PMCID: PMC2859699

46. Wellcome Trust Case Control Consortium, Craddock N, Hurles ME, Cardin N, Pearson RD, Plagnol V, Robson S, Vukcevic D, Barnes C, Conrad DF, Giannoulatou E, Holmes C, Marchini JL, Stirrups K, Tobin MD, Wain LV, Yau C, Aerts J, Ahmad T, Andrews TD, Arbury H, Attwood A, et al. Genome-wide association study of CNVs in 16,000 cases of eight common diseases and 3,000 shared controls. Nature. 2010 Apr 1;464(7289):713-20. PMCID: PMC2892339

47. Conrad DF, Pinto D, Redon R, Feuk L, Gokcumen O, Zhang Y, Aerts J, Andrews TD, Barnes C, Campbell P, Fitzgerald T, Hu M, Ihm CH, Kristiansson K, Macarthur DG, Macdonald JR, Onyiah I, Pang AW, Robson S, Stirrups K, Valsesia A, Walter K, Wei J; Wellcome Trust Case Control Consortium, *Tyler-Smith C, *Carter NP, *Lee C, *Scherer SW, *Hurles ME. Origins and functional impact of copy number variation in the human genome. Nature. 2010 Apr 1;464(7289):704-12. PMCID: PMC3330748 **co-senior author*

48. *Lee C, Scherer SW. The clinical context of copy number variation in the human genome. Expert Rev Mol Med. 2010 Mar 9;12:e8.

49. Turke AB, Zejnullahu K, Wu YL, Song Y, Dias-Santagata D, Lifshits E, Toschi L, Rogers A, Mok T, Sequist L, Lindeman NI, Murphy C, Akhavanfard S, Yeap BY, Xiao Y, Capelletti M, Iafrate AJ, Lee C, Christensen JG, Engelman JA, Jänne PA. Preexistence and clonal selection of MET

amplification in EGFR mutant NSCLC. Cancer Cell. 2010 Jan 19;17(1):77-88. PMCID: PMC2980857

50. Pang AW, MacDonald JR, Pinto D, Wei J, Rafiq MA, Conrad DF, Park H, Hurles ME, Lee C, Venter JC, Kirkness EF, Levy S, Feuk L, Scherer SW. Towards a comprehensive structural variation map of an individual human genome. Genome Biol. 2010;11(5):R52. PMCID: PMC2898065

51. Setlur SR, Chen CX, Hossain RR, Ha JS, Van Doren VE, Stenzel B, Steiner E, Oldridge D, Kitabayashi N, Banerjee S, Chen JY, Schäfer G, Horninger W, Lee C, Rubin MA, Klocker H, Demichelis F. Genetic variation of genes involved in dihydrotestosterone metabolism and the risk of prostate cancer. Cancer Epidemiol Biomarkers Prev. 2010 Jan;19(1):229-39.

52. Rhodes J, Amsterdam A, Sanda T, Moreau LA, McKenna K, Heinrichs S, Ganem NJ, Ho KW, Neuberg DS, Johnston A, Ahn Y, Kutok JL, Hromas R, Wray J, Lee C, Murphy C, Radtke I, Downing JR, Fleming MD, MacConaill LE, Amatruda JF, Gutierrez A, Galinsky I, Stone RM, Ross EA, Pellman DS, Kanki JP, Look AT. Emi1 maintains genomic integrity during zebrafish embryogenesis and cooperates with p53 in tumor suppression. Mol Cell Biol. 2009 Nov;29(21):5911-22. PMCID: PMC2772726

53. Gökçümen O, Lee C. Copy number variants (CNVs) in primate species using array-based comparative genomic hybridization. Methods. 2009 Sep;49(1):18-25. PMCID: PMC2796453

54. Kim JI, Ju YS, Park H, Kim S, Lee S, Yi JH, Mudge J, Miller NA, Hong D, Bell CJ, Kim HS, Chung IS, Lee WC, Lee JS, Seo SH, Yun JY, Woo HN, Lee H, Suh D, Lee S, Kim HJ, Yavartanoo M, Kwak M, Zheng Y, Lee MK, Park H, Kim JY, Gokcumen O, Mills RE, Zaranek AW, Thakuria J, Wu X, Kim RW, Huntley JJ, Luo S, Schroth GP, Wu TD, Kim H, Yang KS, Park WY, Kim H, Church GM, Lee C, Kingsmore SF, Seo JS. A highly annotated whole-genome sequence of a Korean individual. Nature. 2009 Aug 20;460(7258):1011-5. PMCID: PMC2860965

55. Demichelis F, Setlur SR, Beroukhim R, Perner S, Korbel JO, Lafargue CJ, Pflueger D, Pina C, Hofer MD, Sboner A, Svensson MA, Rickman DS, Urban A, Snyder M, Meyerson M, Lee C, Gerstein MB, Kuefer R, Rubin MA. Distinct genomic aberrations associated with ERG rearranged prostate cancer. Genes Chrom Cancer. 2009; Apr;48(4):366-80. PMCID: PMC2674964

56. Freeman JL, Ceol C, Feng H, Langenau DM, Belair C, Stern HM, Song A, Paw BH, Look AT, Zhou Y, Zon LI, *Lee C. Construction and application of a zebrafish array comparative genomic hybridization platform. Genes Chromosomes Cancer. 2009 Feb;48(2):155-70. PMCID: PMC2605212

57. Ionita-Laza I, Rogers AJ, Lange C, Raby BA, *Lee C. Genetic association analysis of copy-number variation (CNV) in human disease pathogenesis. Genomics. 2009 Jan;93(1):22-6. PMCID: PMC2631358

58. Perry GH, Yang F, Marques-Bonet T, Murphy C, Fitzgerald T, Lee AS, Hyland C, Stone AC, Hurles ME, Tyler-Smith C, Eichler EE, Carter NP, Lee C, Redon R. Copy number variation and evolution in humans and chimpanzees. Genome Res. 2008 Nov;18(11):1698-710. PMCID: PMC2577862

59. Korn JM, Kuruvilla FG, McCarroll SA, Wysoker A, Nemesh J, Cawley S, Hubbell E, Veitch J, Collins PJ, Darvishi K, Lee C, Nizzari MM, Gabriel SB, Purcell S, Daly MJ, Altshuler D. Integrated genotype calling and association analysis of SNPs, common copy number polymorphisms and rare CNVs. Nat Genet. 2008 Oct;40(10):1253-60. PMCID: PMC2756534

60. Xue Y, Sun D, Daly A, Yang F, Zhou X, Zhao M, Huang N, Zerjal T, Lee C, Carter NP, Hurles ME, Tyler-Smith C. Adaptive evolution of UGT2B17 copy-number variation. Am J Hum Genet. 2008 Sep;83(3):337-46. PMCID: PMC2556428

61. Koivunen JP, Mermel C, Zejnullahu K, Murphy C, Lifshits E, Holmes AJ, Choi HG, Kim J, Chiang D, Thomas R, Lee J, Richards WG, Sugarbaker DJ, Ducko C, Lindeman N, Marcoux JP, Engelman JA, Gray NS, Lee C, Meyerson M, Jänne PA. EML4-ALK fusion gene and efficacy of an ALK kinase inhibitor in lung cancer. Clin Cancer Res. 2008 Jul 1;14(13):4275-83. PMCID: PMC3025451

62. Van Vlierberghe P, van Grotel M, Tchinda J, Lee C, Beverloo HB, van der Spek PJ, Stubbs A, Cools J, Nagata K, Fornerod M, Buijs-Gladdines J, Horstmann M, van Wering ER, Soulier J, Pieters R, Meijerink JP. The recurrent SET-NUP214 fusion as a new HOXA activation mechanism in pediatric T-cell acute lymphoblastic leukemia. Blood. 2008 May 1;111(9):4668-80. PMCID: PMC2343598

63. Lee AS, Gutiérrez-Arcelus M, Perry GH, Vallender EJ, Johnson WE, Miller GM, Korbel JO, *Lee C. Analysis of copy number variation in the rhesus macaque genome identifies candidate loci for evolutionary and human disease studies. Hum Mol Genet. 2008 Apr 15;17(8):1127-36.

64. Jalali A, Aldinger KA, Chary A, McLone DG, Bowman RM, Le LC, Jardine P, Newbury-Ecob R, Mallick A, Jafari N, Russell EJ, Curran J, Nguyen P, Ouahchi K, Lee C, Dobyns WB, Millen KJ, Pina-Neto JM, Kessler JA, Bassuk AG. Linkage to chromosome 2q36.1 in autosomal dominant Dandy-Walker malformation with occipital cephalocele and evidence for genetic heterogeneity. Hum Genet. 2008 Apr;123(3):237-45. PMCID: PMC2822644

65. Ionita-Laza I, Perry GH, Raby BA, Klanderman B, Lee C, Laird NM, Weiss ST, Lange C. On the analysis of copy-number variations in genome-wide association studies: a translation of the family-based association test. Genet Epidemiol. 2008 Apr;32(3):273-84.

66. Perry GH, Ben-Dor A, Tsalenko A, Sampas N, Rodriguez-Revenga L, Tran CW, Scheffer A, Steinfeld I, Tsang P, Yamada NA, Park HS, Kim JI, Seo JS, Yakhini Z, Laderman S, Bruhn L, *Lee C. The fine-scale and complex architecture of human copy-number variation. Am J Hum Genet. 2008 Mar;82(3):685-95. PMCID: PMC2661628

67. *Lee C, Morton CC. Structural genomic variation and personalized medicine. N Engl J Med. 2008 Feb 14;358(7):740-1.

68. O'Neil J, Tchinda J, Gutierrez A, Moreau L, Maser RS, Wong KK, Li W, McKenna K, Liu XS, Feng B, Neuberg D, Silverman L, DeAngelo DJ, Kutok JL, Rothstein R, DePinho RA, Chin L, *Lee C, Look AT. Alu elements mediate MYB gene tandem duplication in human T-ALL. J Exp Med. 2007 Dec 24;204(13):3059-66. PMCID: PMC2150982

69. Perry GH, Dominy NJ, Claw KG, Lee AS, Fiegler H, Redon R, Werner J, Villanea FA, Mountain JL, Misra R, Carter NP, *Lee C, *Stone AC. Diet and the evolution of human amylase gene copy number variation. Nat Genet. 2007 Oct;39(10):1256-60. PMCID: PMC2377015 **co-senior author*

70. Rodriguez-Revenga L, Mila M, Rosenberg C, Lamb A, *Lee C. Structural variation in the human genome: the impact of copy number variants on clinical diagnosis. Genet Med. 2007 Sep;9(9):600-6.

71. Demichelis F, Fall K, Perner S, Andrén O, Schmidt F, Setlur SR, Hoshida Y, Mosquera JM, Pawitan Y, Lee C, Adami HO, Mucci LA, Kantoff PW, Andersson SO, Chinnaiyan AM, Johansson JE, Rubin MA. TMPRSS2:ERG gene fusion associated with lethal prostate cancer in a watchful waiting cohort. Oncogene. 2007 Jul 5;26(31):4596-9.

72. *Lee C, Iafrate AJ, Brothman AR. Copy number variations and clinical cytogenetic diagnosis of constitutional disorders. Nat Genet. 2007 Jul;39(7 Suppl):S48-54.

73. Scherer SW, Lee C, Birney E, Altshuler DM, Eichler EE, Carter NP, Hurles ME, Feuk L. Challenges and standards in integrating surveys of structural variation. Nat Genet. 2007 Jul;39(7 Suppl):S7-15. PMCID: PMC2698291

74. Freeman JL, Adeniyi A, Banerjee R, Dallaire S, Maguire SF, Chi J, Ng BL, Zepeda C, Scott CE, Humphray S, Rogers J, Zhou Y, Zon LI, Carter NP, Yang F, *Lee C. Definition of the zebrafish genome using flow cytometry and cytogenetic mapping. BMC Genomics. 2007 Jun 27;8:195. PMCID: PMC1925092

75. Engelman JA, Zejnullahu K, Mitsudomi T, Song Y, Hyland C, Park JO, Lindeman N, Gale CM, Zhao X, Christensen J, Kosaka T, Holmes AJ, Rogers AM, Cappuzzo F, Mok T, Lee C, Johnson BE, Cantley LC, Jänne PA. MET amplification leads to gefitinib resistance in lung cancer by activating ERBB3 signaling. Science. 2007 May 18;316(5827):1039-43.

76. Human Genome Structural Variation Working Group, Eichler EE, Nickerson DA, Altshuler D, Bowcock AM, Brooks LD, Carter NP, Church DM, Felsenfeld A, Guyer M, Lee C, Lupski JR, Mullikin JC, Pritchard JK, Sebat J, Sherry ST, Smith D, Valle D, Waterston RH. Completing the map of human genetic variation. Nature. 2007 May 10;447(7141):161-5. PMCID: PMC2685471

77. Parker-Katiraee L, Carson AR, Yamada T, Arnaud P, Feil R, Abu-Amero SN, Moore GE, Kaneda M, Perry GH, Stone AC, Lee C, Meguro-Horike M, Sasaki H, Kobayashi K, Nakabayashi K, Scherer SW. Identification of the imprinted KLF14 transcription factor undergoing human-specific accelerated evolution. PLoS Genet. 2007 May 4;3(5):e65. PMCID: PMC1865561

78. Mertz KD, Setlur SR, Dhanasekaran SM, Demichelis F, Perner S, Tomlins S, Tchinda J, Laxman B, Vessella RL, Beroukhim R, Lee C, Chinnaiyan AM, Rubin MA. Molecular characterization of TMPRSS2-ERG gene fusion in the NCI-H660 prostate cancer cell line: a new perspective for an old model. Neoplasia. 2007 Mar;9(3):200-6. PMCID: PMC1838578

79. Stranger BE, Forrest MS, Dunning M, Ingle CE, Beazley C, Thorne N, Redon R, Bird CP, de Grassi A, Lee C, Tyler-Smith C, Carter N, Scherer SW, Tavaré S, Deloukas P, Hurles ME,

Dermitzakis ET. Relative impact of nucleotide and copy number variation on gene expression phenotypes. Science. 2007 Feb 9;315(5813):848-53. PMCID: PMC2665772

80. Shepard JL, Amatruda JF, Finkelstein D, Ziai J, Finley KR, Stern HM, Chiang K, Hersey C, Barut B, Freeman JL, Lee C, Glickman JN, Kutok JL, Aster JC, Zon LI. A mutation in separase causes genome instability and increased susceptibility to epithelial cancer. Genes Dev. 2007 Jan 1;21(1):55-9. PMCID: PMC1759900

81. Khaja R, Zhang J, MacDonald JR, He Y, Joseph-George AM, Wei J, Rafiq MA, Qian C, Shago M, Pantano L, Aburatani H, Jones K, Redon R, Hurles M, Armengol L, Estivill X, Mural RJ, Lee C, Scherer SW, Feuk L. Genome assembly comparison identifies structural variants in the human genome. Nat Genet. 2006 Dec;38(12):1413-8. PMCID: PMC2674632

82. Komura D, Shen F, Ishikawa S, Fitch KR, Chen W, Zhang J, Liu G, Ihara S, Nakamura H, Hurles ME, Lee C, Scherer SW, Jones KW, Shapero MH, Huang J, Aburatani H. Genome-wide detection of human copy number variations using high-density DNA oligonucleotide arrays. Genome Res. 2006 Dec;16(12):1575-84. PMCID: PMC1665641

83. Fiegler H, Redon R, Andrews D, Scott C, Andrews R, Carder C, Clark R, Dovey O, Ellis P, Feuk L, French L, Hunt P, Kalaitzopoulos D, Larkin J, Montgomery L, Perry GH, Plumb BW, Porter K, Rigby RE, Rigler D, Valsesia A, Langford C, Humphray SJ, Scherer SW, Lee C, Hurles ME, Carter NP. Accurate and reliable high-throughput detection of copy number variation in the human genome. Genome Res. 2006 Dec;16(12):1566-74. PMCID: PMC1665640

84. Kim MY, Yim SH, Kwon MS, Kim TM, Shin SH, Kang HM, Lee C, Chung YJ. Recurrent genomic alterations with impact on survival in colorectal cancer identified by genome-wide array comparative genomic hybridization. Gastroenterology. 2006 Dec;131(6):1913-24.

85. Redon R, Ishikawa S, Fitch KR, Feuk L, Perry GH, Andrews TD, Fiegler H, Shapero MH, Carson AR, Chen W, Cho EK, Dallaire S, Freeman JL, González JR, Gratacòs M, Huang J, Kalaitzopoulos D, Komura D, MacDonald JR, Marshall CR, Mei R, Montgomery L, Nishimura K, Okamura K, Shen F, Somerville MJ, Tchinda J, Valsesia A, Woodwark C, Yang F, Zhang J, Zerjal T, Zhang J, Armengol L, Conrad DF, Estivill X, *Tyler-Smith C, *Carter NP, Aburatani H, *Lee C, *Jones KW, *Scherer SW, *Hurles ME. Global variation in copy number in the human genome. Nature. 2006 Nov 23;444(7118):444-54. PMCID: PMC2669898 **co-senior author*

86. Van Vlierberghe P, van Grotel M, Beverloo HB, Lee C, Helgason T, Buijs-Gladdines J, Passier M, van Wering ER, Veerman AJ, Kamps WA, Meijerink JP, Pieters R. The cryptic chromosomal deletion del(11)(p12p13) as a new activation mechanism of LMO2 in pediatric T-cell acute lymphoblastic leukemia. Blood. 2006 Nov 15;108(10):3520-9.

87. Tchinda J, *Lee C. Detecting copy number variation in the human genome using comparative genomic hybridization. Biotechniques. 2006 Oct;41(4):385, 387, 389 passim.

88. Perner S, Demichelis F, Beroukhim R, Schmidt FH, Mosquera JM, Setlur S, Tchinda J, Tomlins SA, Hofer MD, Pienta KG, Kuefer R, Vessella R, Sun XW, Meyerson M, Lee C, Sellers WR, Chinnaiyan AM, Rubin MA. TMPRSS2:ERG fusion-associated deletions provide insight into the heterogeneity of prostate cancer. Cancer Res. 2006 Sep 1;66(17):8337-41.

89. Freeman JL, Perry GH, Feuk L, Redon R, McCarroll SA, Altshuler DM, Aburatani H, Jones KW, Tyler-Smith C, Hurles ME, Carter NP, Scherer SW, *Lee C. Copy number variation: new insights in genome diversity. Genome Res. 2006 Aug;16(8):949-61.

90. van Vlierberghe P, Meijerink JP, Lee C, Ferrando AA, Look AT, van Wering ER, Beverloo HB, Aster JC, Pieters R. A new recurrent 9q34 duplication in pediatric T-cell acute lymphoblastic leukemia. Leukemia. 2006 Jul;20(7):1245-53.

91. Perry GH, Tchinda J, McGrath SD, Zhang J, Picker SR, Cáceres AM, Iafrate AJ, Tyler-Smith C, Scherer SW, Eichler EE, Stone AC, *Lee C. Hotspots for copy number variation in chimpanzees and humans. Proc Natl Acad Sci U S A. 2006 May 23;103(21):8006-11. PMCID: PMC1472420

92. Yan CT, Kaushal D, Murphy M, Zhang Y, Datta A, Chen C, Monroe B, Mostoslavsky G, Coakley K, Gao Y, Mills KD, Fazeli AP, Tepsuporn S, Hall G, Mulligan R, Fox E, Bronson R, De Girolami U, Lee C, Alt FW. XRCC4 suppresses medulloblastomas with recurrent translocations in p53-deficient mice. Proc Natl Acad Sci U S A. 2006 May 9;103(19):7378-83. PMCID: PMC1464348

93. Nadeau JH, *Lee C. Genetics: copies count. Nature. 2006 Feb 16;439(7078):798-9.

94. Cho EK, Tchinda J, Freeman JL, Chung YJ, Cai WW, *Lee C. Array-based comparative genomic hybridization and copy number variation in cancer research. Cytogenet Genome Res. 2006;115(3-4):262-72.

95. Ouahchi K, Lindeman N, *Lee C. Copy number variants and pharmacogenomics. Pharmacogenomics. 2006 Jan;7(1):25-9.

96. McCarroll SA, Hadnott TN, Perry GH, Sabeti PC, Zody MC, Barrett JC, Dallaire S, Gabriel SB, Lee C, Daly MJ, Altshuler DM; International HapMap Consortium. Common deletion polymorphisms in the human genome. Nat Genet. 2006 Jan;38(1):86-92.

97. Kantarci S, Casavant D, Prada C, Russell M, Byrne J, Haug LW, Jennings R, Manning S, Blaise F, Boyd TK, Fryns JP, Holmes LB, Donahoe PK, Lee C, Kimonis V, Pober BR. Findings from aCGH in patients with congenital diaphragmatic hernia (CDH): a possible locus for Fryns syndrome. Am J Med Genet A. 2006 Jan 1;140(1):17-23. PMCID: PMC2891730

98. Tomlins SA, Rhodes DR, Perner S, Dhanasekaran SM, Mehra R, Sun XW, Varambally S, Cao X, Tchinda J, Kuefer R, Lee C, Montie JE, Shah RB, Pienta KJ, Rubin MA, Chinnaiyan AM. Recurrent fusion of TMPRSS2 and ETS transcription factor genes in prostate cancer. Science. 2005 Oct 28;310(5748):644-8.

99. Shepard JL, Amatruda JF, Stern HM, Subramanian A, Finkelstein D, Ziai J, Finley KR, Pfaff KL, Hersey C, Zhou Y, Barut B, Freedman M, Lee C, Spitsbergen J, Neuberg D, Weber G, Golub TR, Glickman JN, Kutok JL, Aster JC, Zon LI. A zebrafish bmyb mutation causes genome instability and increased cancer susceptibility. Proc Natl Acad Sci U S A. 2005 Sep 13;102(37):13194-9. PMCID: PMC1198999

100. Garraway LA, Widlund HR, Rubin MA, Getz G, Berger AJ, Ramaswamy S, Beroukhim R, Milner DA, Granter SR, Du J, Lee C, Wagner SN, Li C, Golub TR, Rimm DL, Meyerson ML, Fisher DE, Sellers WR. Integrative genomic analyses identify MITF as a lineage survival oncogene amplified in malignant melanoma. Nature. 2005 Jul 7;436(7047):117-22.

101. *Lee C. Vive la difference! Nat Genet. 2005 Jul;37(7):660-1.

102. Jeon Y, Bekiranov S, Karnani N, Kapranov P, Ghosh S, MacAlpine D, Lee C, Hwang DS, Gingeras TR, Dutta A. Temporal profile of replication of human chromosomes. Proc Natl Acad Sci U S A. 2005 May 3;102(18):6419-24. PMCID: PMC1088349

103. Klaassens M, van Dooren M, Eussen HJ, Douben H, den Dekker AT, Lee C, Donahoe PK, Galjaard RJ, Goemaere N, de Krijger RR, Wouters C, Wauters J, Oostra BA, Tibboel D, de Klein A. Congenital diaphragmatic hernia and chromosome 15q26: determination of a candidate region by use of fluorescent in situ hybridization and array-based comparative genomic hybridization. Am J Hum Genet. 2005 May;76(5):877-82. PMCID: PMC1199376

104. Patton EE, Widlund HR, Kutok JL, Kopani KR, Amatruda JF, Murphey RD, Berghmans S, Mayhall EA, Traver D, Fletcher CD, Aster JC, Granter SR, Look AT, Lee C, Fisher DE, Zon LI. BRAF mutations are sufficient to promote nevi formation and cooperate with p53 in the genesis of melanoma. Curr Biol. 2005 Feb 8;15(3):249-54.

105. Ligon AH, Morton CC, Bieber FR, Fletcher JA, Giersch AB, Lee C, Sandstrom M, Weremowicz S, Xiao S, Dal Cin P. Reporting of diagnostic cytogenetic results. Curr Protoc Hum Genet. 2004 Nov;Appendix 1:Appendix 1D.

106. Roberts AE, Irons MB, Kimonis VE, Mulliken JB, Morton CC, *Lee C, Listewnik M. Description of a case of distal 2p trisomy by array-based comparative genomic hybridization: a high resolution genome-wide investigation for chromosomal aneuploidy in a single assay. Am J Med Genet A. 2004 Oct 1;130A(2):204-7.

107. Iafrate AJ, Feuk L, Rivera MN, Listewnik ML, Donahoe PK, Qi Y, Scherer SW, *Lee C. Detection of large-scale variation in the human genome. Nat Genet. 2004 Sep;36(9):949-51.

108. Poss KD, Nechiporuk A, Stringer KF, Lee C, Keating MT. Germ cell aneuploidy in zebrafish with mutations in the mitotic checkpoint gene mps1. Genes Dev. 2004 Jul 1;18(13):1527-32. PMCID: PMC443515

109. Rooney S, Sekiguchi J, Whitlow S, Eckersdorff M, Manis JP, Lee C, Ferguson DO, Alt FW. Artemis and p53 cooperate to suppress oncogenic N-myc amplification in progenitor B cells. Proc Natl Acad Sci U S A. 2004 Feb 24;101(8):2410-5. PMCID: PMC356964

110. *Lee C, Smith A. Molecular cytogenetic methodologies and a bacterial artificial chromosome (BAC) probe panel resource for genomic analyses in zebrafish. Methods Cell Biol. 2004;77:241-54.

111. Masumori N, Tsuchiya K, Tu WH, Lee C, Kasper S, Tsukamoto T, Shappell SB, Matusik RJ. An allograft model of androgen independent prostatic neuroendocrine carcinoma derived from a large probasin promoter-T antigen transgenic mouse line. J Urol. 2004 Jan;171(1):439-42.

112. Wilkinson K, Velloso ER, Lopes LF, Lee C, Aster JC, Shipp MA, Aguiar RC. Cloning of the t(1;5)(q23;q33) in a myeloproliferative disorder associated with eosinophilia: involvement of PDGFRB and response to imatinib. Blood. 2003 Dec 1;102(12):4187-90.

113. Bassing CH, Suh H, Ferguson DO, Chua KF, Manis J, Eckersdorff M, Gleason M, Bronson R, Lee C, Alt FW. Histone H2AX: a dosage-dependent suppressor of oncogenic translocations and tumors. Cell. 2003 Aug 8;114(3):359-70.

114. Paw BH, Davidson AJ, Zhou Y, Li R, Pratt SJ, Lee C, Trede NS, Brownlie A, Donovan A, Liao EC, Ziai JM, Drejer AH, Guo W, Kim CH, Gwynn B, Peters LL, Chernova MN, Alper SL, Zapata A, Wickramasinghe SN, Lee MJ, Lux SE, Fritz A, Postlethwait JH, Zon LI. Cell-specific mitotic defect and dyserythropoiesis associated with erythroid band 3 deficiency. Nat Genet. 2003 May;34(1):59-64.

115. Vaziri C, Saxena S, Jeon Y, Lee C, Murata K, Machida Y, Wagle N, Hwang DS, Dutta A. A p53-dependent checkpoint pathway prevents rereplication. Mol Cell. 2003 Apr;11(4):997-1008. Erratum in: Mol Cell. 2003 May;11(5):1415.

116. Picker JD, Cox GF, Fan YS, Fowler DJ, Weremowicz S, Morton CC, *Lee C. Multicolor karyotypic interpretation of a heterochromatin-associated marker chromosome in a dysmorphic girl with developmental delay. Am J Med Genet. 2002 Jul 15;110(4):393-6.

117. Zhu C, Mills KD, Ferguson DO, Lee C, Manis J, Fleming J, Gao Y, Morton CC, Alt FW. Unrepaired DNA breaks in p53-deficient cells lead to oncogenic gene amplification subsequent to translocations. Cell. 2002 Jun 28;109(7):811-21.

118. *Lee C, Lemyre E, Miron PM, Morton CC. Multicolor fluorescence in situ hybridization in clinical cytogenetic diagnostics. Curr Opin Pediatr. 2001 Dec;13(6):550-5.

119. Sharpless NE, Ferguson DO, O'Hagan RC, Castrillon DH, Lee C, Farazi PA, Alson S, Fleming J, Morton CC, Frank K, Chin L, Alt FW, DePinho RA. Impaired nonhomologous end-joining provokes soft tissue sarcomas harboring chromosomal translocations, amplifications, and deletions. Mol Cell. 2001 Dec;8(6):1187-96.

120. Robertson NG, Resendes BL, Lin JS, Lee C, Aster JC, Adams JC, Morton CC. Inner ear localization of mRNA and protein products of COCH, mutated in the sensorineural deafness and vestibular disorder, DFNA9. Hum Mol Genet. 2001 Oct 15;10(22):2493-500.

121. Qin X, Miwa T, Aktas H, Gao M, Lee C, Qian YM, Morton CC, Shahsafaei A, Song WC, Halperin JA. Genomic structure, functional comparison, and tissue distribution of mouse Cd59a and Cd59b. Mamm Genome. 2001 Aug;12(8):582-9.

122. Alagramam KN, Yuan H, Kuehn MH, Murcia CL, Wayne S, Srisailpathy CR, Lowry RB, Knaus R, Van Laer L, Bernier FP, Schwartz S, Lee C, Morton CC, Mullins RF, Ramesh A, Van Camp G, Hageman GS, Woychik RP, Smith RJ. Mutations in the novel protocadherin PCDH15 cause Usher syndrome type 1F. Hum Mol Genet. 2001 Aug 1;10(16):1709-18. Erratum in: Hum Mol Genet 2001 Oct 15;10(22):2603.

123. Lee C, Rens W, Yang F. Multicolor Fluorescence In Situ Hybridization (FISH) approaches for simultaneous analysis of the entire human genome. Curr Protoc Hum Genet. 2001 May; Chapter 4:Unit 4.9.

124. Lee C, Fowler DJ, Lemyre E, Sandstrom MM, Holmes LB, Morton CC. Prenatal diagnosis and molecular cytogenetics in a case of partial trisomy 14 and monosomy 21. Am J Med Genet. 2001 May 1;100(3):246-50.

125. *Lee C, Murray MF, Miron PM, Marsden D, Irons M, Wilkins-Haug LE, Morton CC. Clinical picture: Multicolour karyotyping. Lancet. 2001 Apr 21;357(9264):1240.

126. Lee C, Gisselsson D, Jin C, Nordgren A, Ferguson DO, Blennow E, Fletcher JA, Morton CC. Limitations of chromosome classification by multicolor karyotyping. Am J Hum Genet. 2001 Apr;68(4):1043-7. PMCID: PMC1275623

127. Hong YK, Kim DH, Beletskii A, Lee C, Memili E, Strauss WM. Development of two bacterial artificial chromosome shuttle vectors for a recombination-based cloning and regulated expression of large genes in mammalian cells. Anal Biochem. 2001 Apr 1;291(1):142-8.

128. Lee C, Critcher R, Zhang JG, Mills W, Farr CJ. Distribution of gamma satellite DNA on the human X and Y chromosomes suggests that it is not required for mitotic centromere function. Chromosoma. 2000 Sep;109(6):381-9.

129. Guo L, Schreiber TH, Weremowicz S, Morton CC, Lee C, Zhou J. Identification and characterization of a novel polycystin family member, polycystin-L2, in mouse and human: sequence, expression, alternative splicing, and chromosomal localization. Genomics. 2000 Mar 15;64(3):241-51.

130. Li YC, Lee C, Hseu TH, Li SY, Lin CC. Direct visualization of the genomic distribution and organization of two cervid centromeric satellite DNA families. Cytogenet Cell Genet. 2000;89(3-4):192-8.

131. Li YC, Lee C, Sanoudou D, Hseu TH, Li SY, Lin CC. Interstitial colocalization of two cervid satellite DNAs involved in the genesis of the Indian muntjac karyotype. Chromosome Res. 2000;8(5):363-73.

132. Mills W, Critcher R, Lee C, Farr CJ. Generation of an approximately 2.4 Mb human X centromere-based minichromosome by targeted telomere-associated chromosome fragmentation in DT40. Hum Mol Genet. 1999 May;8(5):751-61.

133. Lee C, Stanyon R, Lin CC, Ferguson-Smith MA. Conservation of human gamma-X centromeric satellite DNA among primates with an autosomal localization in certain Old World monkeys. Chromosome Res. 1999;7(1):43-7.

134. *Lee C, Griffin DK, O'Brien PC, Yang F, Lin CC, Ferguson-Smith MA. Defining the anatomy of the Rangifer tarandus sex chromosomes. Chromosoma. 1998 Mar;107(1):61-9.

135. Lee C, Wevrick R, Fisher RB, Ferguson-Smith MA, Lin CC. Human centromeric DNAs. Hum Genet. 1997 Sep;100(3-4):291-304.

136. Lee C, Court DR, Cho C, Haslett JL, Lin CC. Higher-order organization of subrepeats and the evolution of cervid satellite I DNA. J Mol Evol. 1997 Mar;44(3):327-35.

137. Lee C, Lin CC. Conservation of a 31-bp bovine subrepeat in centromeric satellite DNA monomers of Cervus elaphus and other cervid species. Chromosome Res. 1996 Sep;4(6):427-35.

138. Lee C, Li X, Jabs EW, Court D, Lin CC. Human gamma X satellite DNA: an X chromosome specific centromeric DNA sequence. Chromosoma. 1995 Nov;104(2):103-12.

139. Lee C, Ritchie DB, Lin CC. A tandemly repetitive, centromeric DNA sequence from the Canadian woodland caribou (Rangifer tarandus caribou): its conservation and evolution in several deer species. Chromosome Res. 1994 Jul;2(4):293-306.

140. Lin CC, Sasi R, Lee C, Fan YS, Court D. Isolation and identification of a novel tandemly repeated DNA sequence in the centromeric region of human chromosome 8. Chromosoma. 1993 May;102(5):333-9.

141. Vamvakopoulos NC, Griffin CA, Hawkins AL, Lee C, Chrousos GP, Jabs EW. Mapping the intron-containing human hsp90 alpha (HSPCAL4) gene to chromosome band 14q32. Cytogenet Cell Genet. 1993;64(3-4):224-6.

142. Lee C, Sasi R, Lin CC. Interstitial localization of telomeric DNA sequences in the Indian muntjac chromosomes: further evidence for tandem chromosome fusions in the karyotypic evolution of the Asian muntjacs. Cytogenet Cell Genet. 1993;63(3):156-9.

143. Fan YS, Sasi R, Lee C, Winter JS, Waterman MR, Lin CC. Localization of the human CYP17 gene (cytochrome P450(17 alpha)) to 10q24.3 by fluorescence in situ hybridization and simultaneous chromosome banding. Genomics. 1992 Dec;14(4):1110-1.

144. Fan YS, Sasi R, Lee C, Court D, Lin CC. Mapping of 50 cosmid clones isolated from a flow-sorted human X chromosome library by fluorescence in situ hybridization. Genomics. 1992 Oct;14(2):542-5.

Books/Book Chapters

1. Dobrinski KP, Brown KH, Freeman JL, *Lee C. Molecular cytogenetic methodologies and a BAC probe panel resource for genomic analyses in the zebrafish. In: Detrich HW, Westerfield M, Zon LI, editors. *Zebrafish*, Fourth Edition. Cellular & Developmental Biology and Genetics, Genomics & Informatics; *Methods Cell Biol*. 104: 237-57, 2011.

2. Ligon AH, Morton CC, Bieber FR, Fletcher JA, Giersch AB, Kantarci S, Leach N, Lee C, Sandstrom M, Weremowicz S, Xiao S, Dal Cin P. Reporting of Diagnostic Cytogenetic Results. In: Dracopoli NC, Haines JL, Korf BR, Morton CC, Seidman CE, Seidman JG, Smith DR, editors. *Current Protocols in Human Genetics*. New York: John Wiley and Sons. p. A.1D.1-23, 2010.

3. *Lee C, Palmer D, Freeman JL, Brown KH. Molecular cytogenetic methodologies and a second generation BAC probe panel resource for zebrafish genomic analysis. In: The Zebrafish: 3rd Edition Genetics, Genomics and Informatics. Detrich HW, Zon LI & Westerfield M (eds) Elsevier Academic Press, London.

4. Morton CC, *Lee C. Cytogenetics in Reproduction. In: Strauss JF, Barbieri R, editors. *Yen and Jaffe's Reproductive Endocrinology, Sixth Edition*. New York: Elsevier. p. 31.1-11, 2009.

5. Smith RS, Gutierrez-Arcelus M, Tran CW, Park S, Couter CJ, *Lee C. Structural diversity in the human genome and its impact on disease susceptibility. In: *Encyclopedia of Life Sciences* (ELS). Chichester: John Wiley & Sons, Ltd. p. 1-12, 2008.

6. Ligon AH, Morton CC, Bieber FR, Fletcher JA, Giersch AB, Lee C, Sandstrom M, Weremowicz S, Xiao S, Dal Cin P. Reporting of Diagnostic Cytogenetic Results. In: Dracopoli NC, Haines JL, Korf BR, Morton CC, Seidman CE, Seidman JG, Smith DR, editors. *Current Protocols in Human Genetics*. New York: John Wiley and Sons. p. A.1D.1-28, 2004.

7. *Lee C, Smith A. Molecular cytogenetic methodologies and a BAC probe panel resource for genomic analyses in the zebrafish. In: Detrich HW, Westerfield M, Zon LI, editors. *Zebrafish*, Second Edition. Cellular & Developmental Biology and Genetics, Genomics & Informatics; 2004.

8. *Lee C, Rens W, Yang F. Multicolor fluorescence in situ hybridization (FISH) approaches for simultaneous analysis of the entire human genome. In: Dracopoli NC, Haines JL, Korf BR, Morton CC, Seidman CE, Seidman JG, Smith DR, editors. *Current Protocols in Human Genetics*. New York: John Wiley and Sons. p. 4.9.1-11, 2000.

Non Print Materials

1. Lee C. Widespread structural variations in the human genome. 2007, In He, M. (ed.), Using bioinformatics in the exploration of genetic diversity: Fundamentals and recent advances, The Biomedical & Life Sciences Collection, Henry Stewart Talks Ltd, London.

Theses

1. Lee C. Two mammalian centromeric satellite DNA families [Ph.D. dissertation]. Edmonton (Alberta): University of Alberta; 1996.

2. Lee C. Tandemly repetitive DNA in the karyotypic and phylogenetic evolution of *Cervidae* species [M.Sc. dissertation]. Edmonton (Alberta): University of Alberta; 1993.

Invited Presentations

2016 Seminar, Boston University, Boston, MA
2015 Seminar, Hudson Alpha Institute for Biotechnology, Huntsville, AL
2015 Lecture, Seoul National Cancer Hospital Symposium, Seoul, South Korea
2015 Lecture**,** Seoul International Symposium of Surgical Oncology, Seoul, South Korea

2015 Seminar, Connecticut Children's Medical Center Pediatric Translational Seminar Series, Farmington, CT
2014 Seminar, Department of Computational Biology and Bioinformatics, Yale University
2014 Seminar, University of Connecticut Health Center Institute for Systems Genomics Annual Networking Workshop, Storrs, CT
2014 Lecture, Medical University of Białystok, Poland, Bialystock, Poland
2014 Lecture, Korean Endocrine Society Meeting, Moojoo, South Korea
2013 Plenary Presentation, 9th European Cytogenetics Conference, Dublin, Ireland
2013 Plenary Presentation, 19th International Chromosome Conference, University of Bologna, Italy
2013 Laboratory Medicine and Pathology Grand Rounds, University of Minnesota, Minneapolis, MN
2013 Annual Meeting for the Society for Molecular Biology and Evolution / Platform Presentation Chicago, IL
2013 Seminar, University of Bialystok, Bialystok, Poland
2013 Plenary Lecture, Annual Meeting for the Association for Molecular Pathology, Phoenix, AZ
2013 Plenary Presentation, New England Regional Genetics Group Annual Meeting
2013 Lecture, NERGG (New England Regional Genetics Group) Annual Meeting, Portsmouth, NH
2012 Plenary Presentation, Personalized Genomic Medicine Meeting, Seoul, South Korea
2012 Plenary Presentation, Human Genome Organization (HUGO) Annual Meeting, Sydney, Australia
2012 Seminar, Karolinska Institute, Stockholm, Sweden
2012 Plenary Presentation, Chromosome-Centric Human Proteome Project (C-HPP) Workshop, Beijing, China
2012 Seminar, Peking Medical College, Beijing, China
2012 Seminar, Frontiers in Genomics – National University of Mexico – Cuernevaca, Cuernevaca, Mexico
2012 Plenary Presentation, Annual Meeting of the Asian Clinical Oncology Society, Seoul, South Korea
2012 Plenary Presentation, Human Genome Variation Annual Meeting, Shanghai, China
2012 Seminar, Albert Einstein College of Medicine, Montefiore University Hospital, Department of Pathology
2012 Plenary Presentation, 53rd Annual Short Course on Medical and Experimental Mammalian Genetics, The Jackson Laboratory, Bar Harbor, ME
2012 Plenary Lecture, Meeting of the North Carolina Cytogenetics Group, Renaissance Asheville Hotel, Asheville, NC
2012 Keynote Address, Pathology Research Day, Medical University of South Carolina, Charleston, SC
2012 Seminar, Pathology Grand Rounds, University of Rochester, Rochester, NY
2012 Seminar, Center for Human Genetics, Katholic University of Leuven, Leuven, Belgium
2012 Plenary Presentation, Annual Genetics Retreat, Harvard Medical School
2012 Plenary Presentation, Harvard Catalyst Course – Genetic Literacy, Harvard Medical School
2011 Plenary Presentation, Human Genome Organization (HUGO) Annual Meeting, Dubai, United Arab Emirates
2011 Plenary Presentation, Korean Academy of Science and Technology Meeting, Seoul, South Korea
2011 Plenary Presentation, 18th International Chromosome Conference, University of Manchester, Manchester, UK
2011 Plenary Presentation, Recent Advances in Clinical Genetics using High Throughput Technologies, Chinese University of Hong Kong, Hong Kong

2011 Plenary Presentation, Keystone Symposia – Functional Consequences of Structural Variation in the Genome, Sheraton Steamboat Resort, Steamboat Springs, CO
2011 Plenary Presentation, International Standardization Cytogenomic Array Meeting, Emory University, Atlanta, GA
2010 Seminar, University of Melbourne, Murdoch Childrens Research Institute, Melbourne, VIC, Australia
2010 Plenary Presentation, 4th Asia Pacific Nutrigenomics Conference, Auckland, New Zealand
2010 Plenary Presentation, 15th International Conference on Prenatal Diagnosis, Amsterdam, Netherlands
2010 Plenary Presentation, Annual Meeting of the Korean Biological Sciences Society, Seoul, South Korea
2010 Plenary Presentation, AnEUploidy Workshop, Split, Croatia
2010 Keynote Address, George Brumley Jr. Lecture, Duke University Medical School, Department of Pediatrics, Durham, NC
2010 Plenary Presentation, 51st Annual Short course on Medical and Experimental Mammalian Genetics, The Jackson Laboratory, Bar Harbor, ME
2010 Plenary Presentation, Institute of Genomic Medicine at UCSD, University of California at San Diego
2010 Seminar, Biological and Biomedical Sciences Program, Harvard Medical School
2009 Plenary Presentation (Selected abstract), European Society of Human Genetics Annual Meeting, European Society of Human Genetics, Vienna, Austria
2009 Seminar, Kyung Hee University, Seoul, South Korea
2009 Keynote Address, Annual Meeting for the Korean Society of Medical Biochemistry and Molecular Biology, Korean Society of Medical Biochemistry and Molecular Biology, Seoul, South Korea
2009 Plenary Presentation, Frontiers in Cancer Sciences, Singapore
2009 Seminar, Department of Pathology, Johns Hopkins Hospital, Baltimore, MD
2009 Plenary Presentation, Gordon Conference on Quantitative Genetics and Genomics, Galveston Island, TX
2009 Plenary Presentation, Cambridge Healthtech Institute on Understanding Copy Number Variation, Cambridge Healthtech Institute, San Diego, CA
2009 Platform Presentation (Selected abstract), Annual Meeting of the American College of Medical Genetics, American College of Medical Genetics, Tampa, FL
2009 Plenary Presentation, Genetics and Environmental Mutagenesis Society, Environmental Protection Agency, Research Triangle Park, NC
2009 Seminar, Department of Genetics and Genomics, Mount Sinai Hospital, New York, NY
2009 Plenary Presentation, Molecular Biology of Hearing and Deafness, Harvard Medical School, Boston, MA
2009 Seminar, MGH Reproductive Endocrine Conference Series, Harvard Medical School
2009 Seminar, HMS Pathology Graduate Program Retreat, Harvard Medical School
2009 Seminar, Program in Quantitative Genetics, Harvard School of Public Health
2009 Plenary Presentation, Molecular Biology of Hearing and Deafness, Harvard Medical School
2009 Plenary Presentation, Pathology Retreat, Harvard Medical School
2008 Keynote Address, First Symposium of the Integrated Research Center for Genome Polymorphism, Seoul, South Korea
2008 Keynote Address, 8th Annual Meeting of the Belgium Human Genetics Society, Belgium Human Genetics Society, Leuven, Belgium
2008 Platform Presentation, 3rd Annual DECIPHER meeting, Sanger Center, UK

2008 Plenary Presentation, New Genetics Workshop, Clinical Nutrition Research Center, University of Alabama at Birmingham, Birmingham, AL
2008 Plenary Presentation, Short Course on the Genetics and Epigenetics of Addiction, National Institute on Drug Abuse, National Institutes of Health, Bethesda, MD
2008 Platform Presentation, 1000 Genome Project Meeting, Cold Spring Harbor Laboratories, NY
2008 Platform Presentation, Molecular Cytogenetics Consortium Workshop, Emory University, Atlanta, GA
2008 Plenary Presentation, Genome-wide Association: Genes Environment, and Health Initiative, National Institute of Heart, Lung and Blood, National Institutes of Health, Bethesda, MD
2008 Platform Presentation, Affymetrix Cytogenetics Community Workshop, Affymetrix, Baltimore, MD
2008 Seminar, Department of Human Genetics, Emory University, Atlanta, GA
2008 Seminar, Boston University School of Medicine, Department of Pathology, Boston, MA
2008 Seminar, Distinguished Lecturer Series in Genome-Wide Association Studies, Harvard School of Public Health
2008 Seminar, Center for Integration of Medicine and Innovative Technology (CIMIT), Harvard Medical School
2007 Seminar, University of Alberta, Department of Medical Genetics, Edmonton, Canada
2007 Keynote Address, Wellcome Trust Advanced Course on High Resolution Molecular Cytogenetics, Sanger Center, Hinxton, UK
2007 Keynote Address, INSERM Meeting on Polymorphism and genome rearrangements, Toulon, France
2007 Seminar, University of Michigan, Department of Human Genetics, Ann Arbor, MI
2007 Seminar, Jackson Laboratory, Bar Harbor, ME
2007 Plenary Presentation, World Congress of Psychiatric Genetics, World Congress of Psychiatric Genetics, New York, NY
2007 Platform Presentation, Applied Biosystems Workshop, American Society of Human Genetics Annual Meeting, San Diego, CA
2007 Plenary Presentation, International Society for Genetic Epidemiology Meeting, Harvard School of Public Health, Boston, MA
2007 Plenary Presentation, Microarray CGH Symposium, Agilent Technologies, Boston, MA
2007 Seminar, Framingham Heart Study, Boston University Medical Center, Boston, MA
2007 Seminar, Genetic Grand Rounds, Tufts University, Boston, MA
2007 Department of Experimental Pathology, Beth Israel Deaconess Hospital
2006 Seminar, Centre de Regulacio Genomica, Pompeu Fabra University, Barcelona, Spain
2006 Keynote Address, Wellcome Trust Advanced Course on High Resolution Molecular Cytogenetics, Sanger Center, Hinxton, UK
2006 Seminar, Centro de Ciencias Genomicas – UNAM, Cuernavaca, Mexico
2006 Plenary Presentation, International Conference of Prental Diagnosis, Kyoto, Japan
2006 Plenary Presentation, International Conference of Human Genetics, Brisbane, Australia
2006 Plenary Presentation, Human Genetic Variation Meeting, Hong Kong
2006 Seminar, Prince of Wales Hospital, Department of Obstetrics and Gynecology, Hong Kong
2006 Seminar, Genome Institute of Singapore, Singapore
2006 Plenary Presentation, Congreso Nacional de Medicina Genomica, Mexico City, Mexico
2006 Plenary Presentation, International Symposium on Applied Genomics, Tokyo, Japan
2006 Plenary Presentation, Genome Sequence Variation and Inherited Basis of Common Disease and Complex Traits, Keystone Symposia, Big Sky, MO
2006 Seminar, Arizona State University, Department of Anthropology, Tempe, AZ

2006 Keynote Address, American Cytogenetic Conference, Emerald Point, GA
2006 Platform Presentation (Selected abstract), Annual Meeting of the American Society of Human Genetics, American Society of Human Genetics, New Orleans, LA
2006 Seminar, Leukemia and Lymphoma Society Meeting, Leukemia and Lymphoma Society, San Diego, CA
2006 Plenary Presentation, Genome Sequence Variation and Inherited Basis of Common Disease and Complex Traits
2006 Seminar, New England Primate Resource Center, Marlborough, MA
2006 Plenary Presentation, Banbury Center, Cold Spring Harbor Laboratory, NY
2006 Plenary Presentation, Chips to Hits, Boston, MA
2006 Seminar, University of Massachusetts, Department of Laboratory Medicine, Worcester, MA
2006 Plenary Presentation, New England Regional Genetics Group (NERGG) Annual Meeting, New England Regional Genetics Group (NERGG), Durham, NH
2006 Brigham Research Institute Seminar, Brigham and Women's Hospital
2006 Platform Presentation, Broad Retreat 2006, Broad Institute, Boston, MA
2005 Platform Presentation, International Human Genome Meeting, Kyoto, Japan
2005 Platform Presentation, European Congress of Human Genetics, Prague, Czech Republic
2005 Plenary Presentation, Decipher Symposium, Sanger Institute, Hinxton, UK
2005 Keynote Address, Standardization of array-CGH results, Sanger Institute, Hinxton, UK
2005 Plenary Presentation, Genome Structural Variation Symposium, Faculty Club, University of Toronto, Canada
2005 Keynote Address, 7th International Conference on Genetic Variation, Leicester, UK
2005 Seminar, Seoul National University School of Medicine, Stem Cell Research Center, Seoul, South Korea
2005 Seminar, Yonsei University College of Medicine, Cancer Research Center, Seoul, South Korea
2005 Seminar, Yale University, Department of Genetics, New Haven, CT
2005 Seminar, Children's Hospital of Oakland Research Institute, Oakland, CA
2005 Keynote Address, Genomic Variation-Beyond the Genome, San Francisco, CA
2005 Platform Presentation, 4th Structural Birth Defects Meeting, National Institutes of Health, William Bolgers Center, Bethesda, MD
2005 Seminar, University of Utah, Department of Human Genetics, Salt Lake City, UT
2005 Seminar, Genetic Grand Rounds, Tufts University and New England Medical Center, Boston, MA
2005 Plenary Presentation, Microarrays in Medicine, Boston, MA
2005 Platform Presentation, Strategic Conference of Zebrafish Investigators, Mount Desert Island Biological Laboratory, Bar Harbor, ME
2005 Platform Presentation, Gordon Research Conference, Human Genetics and Genomics, Salve Regina University, Newport, RI
2005 Guest Lecturer, Marine Biological Laboratory Course on new genetics technologies, Woods Hole, MA
2005 Seminar, Dana Farber Cancer Institute, Dept Oncology, Boston, MA
2005 Pediatric Grand Rounds, Massachusetts General Hospital, Dept Pediatrics, Boston, MA
2005 Medical and Population Genetics Seminar Series, Broad Institute, Boston, MA
2004 Platform Presentation, Innovative application of CGH microarrays, Annual American Society of Human Genetics Meeting, Los Angeles, CA
2004 Seminar, Genetic Grand Rounds, Erasmus University Medical Center, Department of Surgery and Genetics, Rotterdam, Netherlands
2004 Seminar, University of Toronto Sick Children's Hospital, Department of Genetics, Toronto,

Canada

2004 Seminar, Pediatric Grand Rounds, University of Alberta, Faculty of Medicine, Department of Pediatrics, Edmonton, Canada

2004 Amgen Invited Lecturer, Texas Children's Hospital, Baylor College of Medicine, Department of Pediatrics, Houston, TX

2004 Guest Lecturer, MS Degree Program in Genetic Counseling, Brandeis University, Boston, MA

2004 Seminar, Genzyme Genetics, Westborough, MA

2003 Seminar, Vanderbilt University, Department of Medicine, Nashville, TN

2003 Platform Presentation (Selected abstract), Annual Meeting of the American Society of Human Genetics, American Society of Human Genetics, Los Angeles, CA

2003 Seminar, Northeastern University, Department of Pharmaceutical Sciences, Boston, MA

2001 Plenary Presentation, 2001 Annual Clinical Genetics Meeting, Miami, FL

2001 Plenary Presentation, New England Regional Genetics Group Annual Meeting, University of New Hampshire, Durham, NH

2000 Platform Presentation, New England Biological Sciences Association, Boston, MA

2000 Plenary Presentation, Boston Clinical Genetics Group Meeting, Genetics and IVF Institute, Boston, MA

2000 Seminar, Pathology Research Seminar Series, Brigham and Women's Hospital, Division of Molecular Oncology

1998 Plenary Presentation, First International Conference on Mammalian Centromere, Chung Shan Medical & Dental College, Taichung, Taiwan

1998 Seminar, Seoul National University, Department of Obstetrics and Gynecology, Seoul, South Korea

1998 Seminar, Chung Shan Medical and Dental College, Department of Life Sciences, Taichung, Taiwan

1996 Seminar, Baylor College of Medicine, Department of Molecular and Human Genetics, Houston, TX

1995 Platform Presentation, Workshop in Mammalian Chromosome Structure and Function, The Jackson Laboratory, Bar Harbor, ME

1993 Plenary Presentation (Selected abstract), American Cytogenetics Conference, Genetics and IVF Institute, Virginia

MAJOR ADMINISTRATIVE LEADERSHIP POSITIONS

Committee Service

2014-2016 Council Member, Human Genome Organization (HUGO)

2011 Task Force for use of Microarray, International Society for Prenatal Diagnosis

2011 Member, Task Force for use of Microarray Technology in Prenatal Diagnosis, International Society for Prenatal Diagnosis

2010 Consultant, Molecular and Clinical Genetics Panel, Food and Drug Administration (FDA)

2009-present Member, Advisory Committee, International Standardization Cytogenetic Array

2009-present Member, Advisory Committee, International Standardization Cytogenetic Array

2009-2013 GeT-RM CMA Reference Member, Laboratory Science and Standards, Centers for Disease Control and Prevention

2009-2013 Director, Board of Directors, Cancer Cytogenomics Microarray Consortium

2008-2010	Scientific Advisory Board, Center of Excellence for Genome Sciences, Yale University
2007-2013	Working Group, Center for Advanced Molecular Diagnostics, Brigham and Women's Hospital
2007-2010	Member, Program Committee, American Society of Human Genetics
2006-present	Member, Clinical Advisory Committee, NICMS Prenatal Microarray Study
2006-2013	Working Group, Center for Human Genetics, BWH Research Institution
2006-2008	Steering Committee, Structural Variation Consortium, NHGRI/NIH
2005-2006	Steering Committee, Structural Variation & Diseases, Wellcome Trust/Sanger Center
2005	Steering Committee, Standardization of aCGH, Wellcome Trust/Sanger Center

Professional Societies

2002-present	Member, American Board of Medical Genetics
1994-present	Member, Canadian Society of Biochemistry and Molecular Biology
1993-present	Member, American College of Medical Genetics
1992-present	Member, American Society of Human Genetics

Grant Review Activities

2009-present	Genes Health and Development Study Section, Standing Study Section Member, *Ad hoc* Reviewer
2009-2010	Pilot Grants, Autism Speaks
2009	ARRA RC1 Grants, NIH, *Ad hoc* Reviewer
2009	Harvard Catalyst Pilot Grants, Harvard Catalyst Program, *Ad hoc* Reviewer
2008	Genes Health & Development Study Section, NIH, *Ad hoc* Reviewer
2008	Clinical / Biomedical R&D, US Dept of Veterans Affairs, *Ad hoc* Reviewer
2008	Wellcome Trust Individual Grants, Wellcome Trust, *Ad hoc* Reviewer
2007	Program Project Grants in Molecular Oncology, NIH/NCI, *Ad hoc* Reviewer
2006	Child Health Research Foundation, Cure Kids New Zealand, *Ad hoc* Reviewer
2005	National Science Foundation, *Ad hoc* Reviewer
2004	Genome Canada Grant Competition, NIH/NCRR, *Ad hoc* Reviewer
2010	GWAS Sequencing Grants, NIH, *Ad hoc* Reviewer

Editorial Activities

***Ad hoc* Reviewer:**

American Journal of Human Genetics
Genetics in Medicine
Genome Research
Human Genetics
Human Molecular Genetics
Nature
Nature Genetics
Nature Methods
Nature Reviews Genetics
Nucleic Acids Research
Proceedings of the National Academy of Sciences USA
Science
Trends in Genetics

Other Editorial Roles:

2012-present	Editorial Board, Experimental and Molecular Medicine
2010-present	Editorial Advisor, Genes and Genomics
2009-2011	Associate Editor, American Journal of Human Genetics

HONORS AND PRIZES

2014	Citation Laureate, Thompson Reuters
2012	Fellow, AAAS, American Association for the Advancement of Science
2012	Chen Global Investigator Award, Human Genome Organization (HUGO)
2012	Vandenberghe Chair Award, Katholic University of Leuven, Belgium
2010	George W. Brumley, Jr., MD Memorial Award, Duke University, Durham, North Carolina, USA
2008	Ho-Am Prize in Medicine, Ho Am Foundation, Seoul, South Korea
2008	C. Thomas Caskey Lectureship, University of South Carolina, Columbia, South Carolina, USA
2007	American Association for Cancer Research Inaugural Team Award, American Association for Cancer Research
2002	Stanley L. Robbins Research Award, Department of Pathology, Brigham and Women's Hospital
1996-1998	NSERC Postdoctoral Fellowship, Natural Sciences and Engineering Research Council of Canada
1996	MRC Postdoctoral Fellowship, Medical Research Council of Canada
1994-1996	AHFMR PhD Studentship, Alberta Heritage Foundation for Medical Research
1994	75th Anniversary Faculty of Medicine Scholarship, University of Alberta
1993	American Cytogenetics Conference Student Award, Genetics and IVF Institute

CLINICAL ACTIVITIES AND INNOVATIONS

Current Certification

2002-present American Board of Medical Genetics (Clinical Cytogenetics)

Practice Activities

2002-2013 Clinical Cytogenetic signout, Brigham and Women's Hospital, 20%

Technological and Other Scientific Innovations

US patent assigned: #7,718,369 – Recurrent gene fusions in prostate cancer
Chinnaiyan A, Tomlins S, Rhodes D, Mehra R, Rubin MA, Sun X-W, Demichelis F, Perner S, Lee C, inventors; Regents of the University of Michigan and The Brigham and Women's Hospital, Inc. assignees.

US patent pending – FISH assay for EML4 and ALK fusion in lung cancer
Lee C, Murphy C, Janne P, inventors; Brigham and Women's Hospital and Dana Farber Cancer Institute, Inc. assignees.