

Luis Carvajal-Carmona, PhD

Curriculum Vitae of

Luis G. Carvajal Carmona, PhD

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Education

2003. Ph.D. Human Genetics. Department of Biology. University College London.
1998. B.Sc. Honours (First Class) Animal Science. Universidad Nacional. Colombia.

Employment and appointments

- 2012 - Assistant Professor. Genome Center and Department of Biochemistry and Molecular Medicine, University of California, Davis.
2009-2012 Co-Chair. Colorectal Cancer Group, Latin American Cancer Epidemiology Consortium, NCI.
2008-2012 Senior Research Fellow. Wellcome Trust Centre for Human Genetics, University of Oxford.
2007-2011 Honorary Lecturer. Institute of Cancer, Queen Mary University of London.
2006- Honorary Lecturer. School of Biological Sciences, University of Tolima, Colombia.
2004-2008. Research Fellow. Molecular and Population Genetics Laboratory, London Research Institute.
2000-2003. Research Assistant. Department of Biology, University College London.
1998-2000. Research Assistant. Department of Medical Genetics, Imperial College London.
1996-1998. Undergraduate Research Assistant. School of Medicine, Universidad de Antioquia, Colombia.

Teaching experience

- 2012 Tutor. Cancer Genetics, SSM in Genomic Medicine. DPhil. Clinical Medicine, University of Oxford
2012- Tutor. Population Genetics. Wadham College. BSc. Human Sciences, University of Oxford
2011- Tutor. Human Evolutionary Genetics. B.Sc. Biological Sciences, University of Oxford.
2011- Tutor. Molecular Genetics. B.Sc. in Human Sciences, New College, University of Oxford.
2010- Lecturer. Cancer Genetics. M.Sc. in Experimental Therapeutics, University of Oxford.
2006- Supervisor. Ph.D. and M.Sc. students in Biological Sciences. University of Tolima, Colombia.
2004- Supervisor. Clinical fellows and graduate students. Cancer Research UK London Research Institute and Wellcome Trust Centre for Human Genetics, University of Oxford.
2000-2003. Tutor and Demonstrator. Department of Biology, University College London.
1998-2000. Lecturer. Linkage analysis. MSc in Human Molecular Genetics, Imperial College, London.

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Selected prizes and awards

- 2012. Distinguished Alumni Award. 50th Anniversary, BSc Animal Science. Universidad Nacional, Colombia.
- 2011. Semi-finalist for Trainee Research Award, American Society for Human Genetics for abstract submission to the 12th International Congress of Human Genetics. Montreal, Canada
- 2009. Winner of the Divisional Research Prize. Medical Sciences Division, University of Oxford.
- 2009. Finalist. Neel Award for Young Investigators. International Genetic Epidemiology Society.
- 2008. Winner of the European Association for Cancer Research 40th Anniversary Research Award.
- 2004. Research Fellowship. London Research Institute, United Kingdom
- 2000. Award for best paper published by a member of the University of Antioquia, Colombia
- 1999. Winner of two research awards at the national meetings of Biology and Psychiatry. Colombia.
- 1998-2000. Studentships awarded by The Jackson Laboratory, Rockefeller University, Hoffmann-La Roche and North Carolina State University to attend training courses in genetic analysis.
- 1998. Travel Fellowship to attend the International Conference on Animal Genetics, Auckland, New Zeland
- 1998. Award for best undergraduate research project. Universidad Nacional, Colombia.
- 1992. Studentship for best university entrance exam (among 1000+ entrants). Universidad Nacional, Colombia.

Grants

- 1. The relationship between European admixture and colorectal cancer risk in South American populations.
Role: Principal Investigator. Funded by Cancer Research UK. 2005-2007.
- 2. A genetic study of common hereditary tumours in Hispania and the Americas.
Role: Project Manager. Funded by the European Commission FP7. 2009-2012.
- 3. Rapid HLA haplotyping using second-generation sequencing.
Role: Principal Investigator. Funded by Nuffield Department of Medicine Research Fund. 2010-2012.
- 4. Genetic analyses of breast cancer in admixed populations.
Role: Principal Investigator. Funded by the Glaxo-SmithKline Oncology Ethnic Research Initiative. 2012-2014.

Invited talks and lectures

- 1. Novel approaches to dissect the genetics of cancer. Genome Center, University of California, Davis, 2012.
- 2. Novel approaches to dissect the genetics of cancer: intermediate phenotypes, variation in normal traits and genetic studies in admixed populations. Division of Cancer Epidemiology and Genetics, National Cancer Institute. Bethesda, USA, 2012.
- 3. Novel approaches to dissect the genetics of colorectal cancer. Case Comprehensive Cancer Center, Case Western Reserve University, Cleveland, USA, 2012.
- 4. Advances in colorectal cancer genetics. Division of Surgical Oncology, Brigham and Women Hospital, Boston, USA, 2012
- 5. Strategies to discover cancer genes. Department of Epidemiology, Brown University. Providence, USA, 2012.

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6. Novel approaches to dissect the genetics of cancer: intermediate phenotypes, variation in normal traits and studies in admixed populations. Duncan Cancer Center, Baylor College of Medicine. Houston, USA, 2012
7. Advances in colorectal cancer genetics: fine mapping of known regions, novel variants and associations with cancer initiation. IDIBAPS/Hospital Clinic. Barcelona, Spain, 2011.
8. Colorectal cancer genetics: past, present and future. Einstein College of Medicine, New York, USA. 2011.
9. Current and future approaches aimed at dissecting the genetics of colorectal cancer. Department of Genetics, Stanford University. Palo Alto, USA. 2011.
10. Advances in colorectal cancer genetics: fine mapping of known loci and associations with adenoma risk. Leeds Institute of Molecular Medicine. Leeds, UK. 2011.
11. Colorectal cancer genetics: current and future approaches. Department of Epidemiology and Biostatistics, Memorial Sloan-Kettering Cancer Centre. New York, USA. 2011.
12. UK thyroid cancer genetics study. Peninsula Cancer Network meeting. Lifton, UK. 2010
13. Genetic dissection of complex human diseases using genome-wide association studies. Meeting of the International Society for Forensic Genetic. Buenos Aires, Argentina. 2009.
14. Investigating the genetic basis of colorectal cancer using genome-wide association mapping. UCLA Human Genetics Seminars, Los Angeles, USA. 2009.
15. Genetic history of Latin Americans. Medical Sciences Division, University of Oxford. Oxford, UK. 2009
16. UK-wide thyroid cancer genetics study. NCRI Thyroid Cancer Meeting. London, UK. 2009
17. Genome-wide association studies in cancer. Special Meeting of the Spanish Association for Cancer Research. Santiago de Compostela, Spain. 2009.
18. Genetic history of Antioquia. Department of Human Genetics, UCLA. Los Angeles, USA. 2001.

Selected oral presentations at scientific conferences

1. Genetics of colorectal Adenoma predisposition. 4th meeting of the International Colorectal Cancer Genetics Consortium. Hospital Clinico San Carlos, Madrid, Spain, 2012.
2. Body size and colorectal cancer risk. 3rd Meeting of the CHIBCHA Consortium. Hospital Clinico San Carlos, Madrid, Spain, 2012.
3. Most, but not all, colorectal cancer variants are associated with adenoma predisposition. 12th International Congress of Human Genetics. Montreal, Canada, 2011.
4. Refining the location of four colorectal cancer risk loci. Third meeting of the International Colorectal Cancer Genetics Consortium. MRC Human Genetics Unit, Edinburgh, UK. 2010.
5. Fine mapping of colorectal cancer low penetrance susceptibility loci. Neel Award Finalist session. 18th annual meeting of the International Genetic Epidemiology Society. Honolulu, USA. 2009.
6. Genome-wide association study of tag SNPs identifies five novel colorectal cancer susceptibility loci. Research Award Winners session, 20th meeting of the European Association for Cancer Research. Lyon, France. 2008.
7. Genetic pathways in hyperplastic polyposis syndrome. 2nd Biennial Scientific Meeting of The International Society for Gastrointestinal Hereditary Tumours. Yokohama, Japan. 2007.

Professional affiliations and service

I am member of the American Association for Cancer Research, the American Thyroid Association, the American Society for Human Genetics, the British Association for Cancer Research, the European Association for Cancer Research, the Genetics Society and the International Genetic Epidemiology Society. I am an Associate Editor of BMC Gastroenterology and a Review Editor of Frontiers in Statistical Genetics and Methodology. I have reviewed manuscripts for Archives of Medical Research, Biomedica, BMC Genetics, BMC Medical Genetics, British Journal of Dermatology, Cancer Epidemiology, Cancer Epidemiology Biomarkers and Prevention, Cancer Genetics, Familial Cancer, Gastroenterology, Genes Chromosome & Cancer, Genomics, Gut, Human Molecular Genetics, Molecular Cancer, Molecular Biology Reports, New England Journal of Medicine, Journal of Pathology and PLOS One. I have served as reviewer of grant applications submitted to the Colombian National Science Foundation (Colciencias), The Croatia Science Foundation, the Chilean National Science Foundation (Fondecyt), the UK Medical Research Council and The Wellcome Trust. I have participated in review panels for program grants submitted to Colciencias.

Full Bibliography (updated January 24th 2013)

Research impact

As of January 24th 2013, my publications have been cited 2,877 times. My H-index is 21 and my i10-index is 33 (for more details see http://scholar.google.co.uk/citations?hl=en&user=-qlpXTIAAAAJ&view_op=list_works&pagesize=100).

2013

51. Fernandez-Rozadilla C, Cazier JB, Tomlinson IP, **Carvajal-Carmona LG**, Palles C, Lamas MJ, Baiget M, López-Fernández LA, Brea-Fernández A, Abulí A, Bujanda L, Clofent J, Gonzalez D, Xicola R, Andreu M, Bessa X, Jover R, Llor X, The EPICOLON Consortium, Moreno V, Castells A, Carracedo A, Castellvi-Bel S, Ruiz-Ponte C. A Colorectal Cancer Genome-Wide Association Study in a Spanish cohort identifies two new candidate colorectal cancer susceptibility variants at 1p33 and 8p12. *BMC Genomics*, In press.
50. Palles, C., Cazier, J., Howarth, K.M., Domingo, E., Jones, A.M., Broderick, P., Kemp, Z., Spain, S., Almeida, E., Salguero, I., Sherborne, A., Chuub, D., **Carvajal-Carmona, L.G.**, Ma, Y., Kaur, K., Dobbins, S., Barclay, E., Gorman, M., Martin, L., Kovac, M., Humphray, S., Lucassen, A., Holmes, C., Bentley, D., Donnelly, P., Taylor, J., Petridis, C., Roylance, R., Sawyer, E., Kerr, D., Clark, S., Grimes, J., Kearsley, S., Thomas, H., McVean, G., Houlston, R., and Tomlinson, I. (2013). Germline mutations in POLE and POLD1 affecting the proofreading domains predispose to colorectal adenomas and carcinomas. *Nat Genet*. In press. doi:10.1038/ng.2503.
49. Fernandez-Rozadilla C, Palles C, **Carvajal-Carmona L**, Peterlongo P, Nici C, Veneroni S, Pinheiro M, Teixeira MR, Moreno V, Lamas M-J, Baiget M, Lopez j-L, Gonzalez D, Brea-Fernandez A, Clofent J, Bujanda L, Bessa X, Andreu M, Xicola R, Llor X, Jover R, Castells A, Castellvi-Bel S, Carracedo A, Tomlinson I, Ruiz-Ponte C.

BMP2/BMP4 colorectal cancer susceptibility loci in Northern and Southern European populations. *Carcinogenesis, in press, 2012*. doi: 10.1093/carcin/bgs357

48. **Carvajal-Carmona LG**, Zauber AG, Jones AM, Howarth K, Wang J, Cheng T, APC Trail Collaborators, APPROVe Trail Collaborators, CORGI Study Collaborators, Colon Cancer Family Registry Collaborators, Riddell R, Lanos A, Morton D, Bertagnolli MM, Tomlinson IPM. Much of the population genetic risk of colorectal cancer is likely to be mediated through susceptibility to adenomas. *Gastroenterology*, 2013, 144:53-55.

2012

47. Dunlop, MG, Dobbins SE, Farrington SM, Jones AM, Palles C, Whiffin N, Tenesa A, Spain S, Broderick P, Ooi LY, Domingo E, Smillie C, Henrion M, Frampton M, Martin L, Grimes G, Gorman M, Semple C, Ma YP, Barclay E, Prendergast J, Cazier J-B, Olver B, Penegar S, Lubbe S, Chander I, **Carvajal-Carmona LG**, Ballereau S, Lloyd A, Vijayakrishnan J, Zgaga L, Rudan I, Theodoratou E, Starr JM, Deary I, Kirac I, Kovacević D, Aaltonen LA, Renkonen-Sinisalo L, Mecklin J-P, Matsuda K, Nakamura Y, Okada Y, Gallinger S, Duggan DJ, Conti D, Newcomb P, Hopper J, Jenkins MA, Schumacher F, Casey G, Easton D, Shah M, Pharoah P, Lindblom A, Liu T, Smith CG, West H, Cheadle JP, Midgley R, Kerr DJ, Campbell H, Tomlinson IP, Houlston RS. Common variation near CDKN1A, POLD3 and SHROOM2 influences colorectal cancer risk. *Nature Genetics*, 2012, 44: 770-776.
46. Dunlop MG, Tenesa A, Farrington SM, Ballereau S, Brewster DH, Kessler T, Pharoah P, Schafmayer C, Hampe J, Volzke H, Chang-Claude J, Hoffmeister M, Brenner H, von Holst S, Picelli S, Lindblom A, Jenkins MA, Hopper JL, Casey G, Duggan D, Newcomb PA, Abul A, Bessa Z, Ruiz-Ponte C, Castellví-Bel S, Niittymäki I, Tuupanen S, Karhu A, Aaltonen L, Zanke B, Hudson T, Gallinger S, Barclay E, Martin L, Gorman M, **Carvajal-Carmona L**, Walther A, Kerr D, Lubbe S, Broderick P, Chandler I, Pittman A, Penegar S, Campbell H, Tomlinson H, Houlston RS. Cumulative impact of common genetic variants and other risk factors on colorectal cancer risk in 42,103 individuals. *Gut*, 2012, doi:10.1136/gutjnl-2011-300537.
45. Jones AM, Howarth KM, Martin L, Gorman M, Mihai R, Moss L, Auton A, Lemon C, Mehanna H, Mohan H, Clarke SEM, Wadsley J, Macias E, Coatesworth A, Beasley M, Roques T, Martin C, Ryan P, Gerrard G, Power D, Bremner C, The TCUKIN Consortium, Tomlinson I, **Carvajal-Carmona LG**. Thyroid cancer susceptibility polymorphisms: confirmation of loci on chromosomes 9q22 and 14q13, validation of a recessive 8q24 locus and failure to replicate a locus on 5q24. *Journal of Medical Genetics*. 2012, 49:158-163.
44. Houlston RS, Members of COGENT (COGENT: Aaltonen LA, Brenner H, Buch S, Campbell H, Carracedo A, **Carvajal-Carmona L**, Castells A, Castellví-Bel S, Cheadle JP, Devilee P, Dunlop M, Echeverry MM, Gallinger S, Galvan A, Hampe J, Hemminki K, Ho JW, Hofstra RMW, Hudson TJ, Kirac I, Lerch MM, Li L, Lindblom A, Lipton L, Matsuda K, Maughan TS, Moreno V, Morreau H, Naccardi A, Nakamura Y, Peterlongo P, Pharoah PD, Sieber O, Radice P, Ruiz-Ponte C, Schafmayer C, Schmidt CA, von Schönfels W, Schreiber S, Scott R, Sham P,

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43. Spain SL, **Carvajal-Carmona LG**, Howarth KM, Jones AM, Su Z, Cazier JB, Aaltonen LA, Pharoah P, Kerr DJ, Cheadle J, Li L, Vodicka P, Sieber O, Lipton L, Gibbs P, Martin NG, Montgomery GW, Young J, Baird PN, Ruiz-Ponte C, Fernandez-Rozadilla C, Carracedo A, Castells A, Castellvi-Bel S, Dunlop M, Houlston RS, Tomlinson IPM. Refinement of the associations between colorectal cancer and polymorphisms on chromosomes 1q41 and 12q13.13. *Human Molecular Genetics*, 2012, 21:934-946.
42. Jones AM, Beggs AD, **Carvajal-Carmona L**, Walker M, Howarth K, Farrington S, Tenesa A, Ballereau S, Campbell H, Zuber A, Bertagnolli M, Kerr D, Hodgson SV, Dunlop M, Tomlinson I: TERC polymorphisms are associated both with susceptibility to colorectal cancer and with longer telomeres. *Gut*, 2012, 61:248-254.

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41. Abulí A, Fernandez-Rozadilla C, Alonso-Espinaco V, Muñoz J, Gonzalo V, Bessa X, González D, Clofent J, Cubiella J, Morillas JD, Rigau J, Latorre M, Fernández-Bañares F, Peña E, Riestra S, Payá A, Jover R, Xicola RM, Llor X, **Carvajal-Carmona L**, Villanueva CM, Moreno V, Piqué JM, Carracedo A, Castells A, Andreu M, Ruiz-Ponte C, Castellví-Bel S, for the Gastrointestinal Oncology Group of the Spanish Gastroenterological Association. Case-control study for colorectal cancer genetic susceptibility in EPICOLON: previously identified variants and mucins. *BMC Cancer*, 2011, 11:339.
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39. **Carvajal-Carmona LG**, Cazier J-B, Jones A, Howarth K, Broderick P, Pittman A, Dobbins S, Tenesa A, Farrington S, Prendergast J, Theodoratou E, Barnetson R, Conti D, Newcomb P, Hopper JL, Jenkins MA, Gallinger S, Zanke B, Duggan DJ, Kerr D, Campbell H, Casey G, Houlston R, Dunlop M, Tomlinson I. Fine-mapping of colorectal cancer susceptibility loci at 8q23.3, 16q22.1 and 19q13.11 refines regional association signals and in silico analyses suggests functional variation and identifies unexpected candidate target genes. *Human Molecular Genetics*, 2011, 20 (14): 2879-2888.

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38. Tomlinson IPM, **Carvajal-Carmona LG***, Dobbins SE, Tenesa A, Jones A, Howarth K, Palles C, Broderick P, Jaeger EM, Farrington S, Lewis A, Prendergast JG, Pittman AM, Theodoratou E, Olver B, Barnetson RA, Penegar S, Barclay E, Whiffin N, Martin L, Lloyd A, Gorman M, The COGENT Consortium, The CORGI collaborators, Ruiz-Ponte C, Fernandez-Rozadilla C, Castells A, Carracedo A, Castellvi-Bel S, Duggan D, Conti D, Cazier J-B, Kerr DK, Campbell H, Sieber O, Lipton L, Gibbs P, Montgomery G, Young J, Baird P, Zanke B, Gallinger S, Newcomb P, Hopper J, Jenkins MA, Aaltonen LA, Cheadle J, Pharoah P, Casey G, Dunlop MG, Houlston RS. The *GREM1*, *BMP4* and *BMP2* loci harbor multiple common susceptibility variants for colorectal cancer. *PLOS Genetics*, 2011, 7: e1002105.
37. Alonso-Espinaco V, Giráldez M, van der Klift H, Muñoz J, Balaguer F, Ocaña T, Madrigal I, Jones A, Echeverry M, Velez A, Tomlinson I, Milà M, Wijnen J, Trujillo C, **Carvajal-Carmona L**, Castells A, Castellvi-Bel S: Novel *MLH1* duplication identified in Colombian families with Lynch Syndrome. *Genetics in Medicine*, 2011, 13 (2): 155-160.
36. Ho J, Choi AS, Lee YF, Hui TC, Cherny SC, Garcia-Barcelo MM, **Carvajal-Carmona L**, Liu R, To SH, Yau T-K, Chung CC, Yau CC, Hui SM, Lau PY, Yuen CH, Wong YW, Ho S, Fung SS, Tomlinson IP, Houlston RS, Cheung KK, Sham PC: Replication Study of SNP Associations for Colorectal Cancer in Hong Kong Chinese. *British Journal of Cancer*, 2011: 104(2): 369-375.

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35. Venkatachalam R, Ligtenberg M, Hoogerbrugge N, Schackert H, Goossens M, Hahn M, Kamping E, Vreede L, Hoenselaar E, van der Looij E, Görgens H, Churchman M, **Carvajal-Carmona L**, Tomlinson I, de Bruijn D, van Kessel A, Kuiper R: Germline epimutation of the tumor suppressor gene *PTPRJ* in early onset familial colorectal cancer. *Gastroenterology*, 2010, 139: 2221-2224.
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33. **Carvajal-Carmona, LG:** Genetic dissection of intermediate phenotypes as a way to discover novel cancer susceptibility alleles. *Current Opinions in Genetics and Development*, 2010, 20:308–314.
32. **Carvajal-Carmona LG:** Challenges in the identification and use of rare disease-associated predisposition variants. *Current Opinions in Genetics and Development*, 2010, 20:227–281.
31. **Carvajal-Carmona LG, Silver A, Tomlinson I:** Molecular Genetics of Familial Adenomatous Polyposis. *Hereditary Colorectal Cancer*. Pages 45-66. Edited by Rodriguez-Bigas M, Cutait R, Lynch P, Tomlinson I, Vasen H. Springer, New York.
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25. **Carvajal-Carmona LG[‡]**, Spain S, Kerr D, Houlston R, Cazier JB, Tomlinson I: Common variation at the adiponectin locus is not associated with colorectal cancer risk in the UK. *Human Molecular Genetics*, 2009, 18(10):1889-1892.
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