

Publication list Dr. Oscar Lao

Original Articles in Peer-Reviewed Scientific Journals:

- (1) de Gruijter JM*, **Lao O***, Vermeulen M, Xue Y, Woodwark C, Gillson CJ, Coffey AJ, Ayub Q, Mehdi SQ, Kayser M, Tyler-Smith C. (2009) Contrasting signals of positive selection in genes involved in human skin color variation from tests based on SNP scans and resequencing. Under review in *Mol Biol Evol*. · These authors contributed equally to this work
- (2) Goedbloed M, Vermeulen M, Fang RN, Lembring M, Wollstein A, Ballantyne K, **Lao O**, Brauer S, Krüger C, Roewer L, Lessig R, Ploski R, Dobosz T, Henke L, Henke J, Furtado MR, Kayser M. (2009) Comprehensive mutation analysis of 17 Y-chromosomal short tandem repeat polymorphisms included in the AmpFISTR(R) Yfiler(R) PCR amplification kit. *Int J Legal Med*, Epub April 2009, DOI 10.1007/s00414-009-0342-y.
- (3) Lu TT*, **Lao O***, Nothnagel M, Junge O, Freitag-Wolf S, Caliebe A, Balascakova M, Bertranpetit J, Bindoff LA, Comas D, Holmlund G, Kouvatsi A, Macek M, Mollet I, Parson W, Palo J, Ploski R, Sajantila A, Tagliabracci A, Gether U, Werge T, Rivadeneira F, Hofman A, Uitterlinden AG, Gieger C, Wichmann HE, Rütger A, Schreiber S, Becker C, Nürnberg P, Nelson MR, Kayser M, Krawczak M. (2009) An evaluation of the genetic matched pair study design using genome-wide SNP data from the European population. *Eur J Hum Genet* Epub 21 Jan 2009, doi: 10.1038/ejhg.2008.266 · These authors contributed equally to this work
- (4) Vives S, Gilbert MT, Arenas C, Gigli E, **Lao O**, Lalueza-Fox C. (2008) Statistical analysis of post mortem DNA damage-derived miscoding lesions in Neandertal mitochondrial DNA. *BMC Res Notes*;1:40.
- (5) Kayser M*, **Lao O***, Stoneking M. (2008) East Asian and Melanesian ancestry in Polynesians - Reply to Hedrick. *Am J Hum Genet*;83(1):140-142. · These authors contributed equally to this work
- (6) **Lao O***, Lu TT*, Nothnagel M, Junge O, Freitag-Wolf S, Caliebe A, Balascakova M, Bertranpetit J, Bindoff LA, Comas D, Holmlund G, Kouvatsi A, Macek M, Mollet I, Parson W, Palo J, Ploski R, Sajantila A, Tagliabracci A, Gether U, Werge T, Rivadeneira F, Hofman A, Uitterlinden AG, Gieger C, Wichmann HE, Rütger A, Schreiber S, Becker C, Nürnberg P, Nelson MR, Krawczak M, Kayser M. (2008) Correlation between genetic and geographic structure in Europe. *Curr Biol*; 18(16):1241-8. · These authors contributed equally to this work
- (7) Kayser M, Liu F, Janssens AC, Rivadeneira F, **Lao O**, van Duijn K, Vermeulen M, Arp P, Jhamai MM, van Ijcken WF, den Dunnen JT, Heath S, Zelenika D, Despriet DD, Klaver CC, Vingerling JR, de Jong PT, Hofman A, Aulchenko YS, Uitterlinden AG, Oostra BA, van Duijn CM. (2008) Three genome-wide association studies and a linkage analysis identify HERC2 as a human iris color gene. *Am J Hum Genet*;82(2):411-23.
- (8) Kayser M*, **Lao O***, Saar K, Brauer S, Wang X, Nürnberg P, Trent RJ, Stoneking M. (2008) Genome-wide analysis indicates more Asian than Melanesian ancestry of Polynesians. *Am J Hum Genet*; 82: 1-5. · These authors contributed equally to this work
- (9) **Lao O**, Dupanloup I, Barbujani G, Bertranpetit J, Calafell F. (2008) The Mediterranean Paradox for Susceptibility Factors in Coronary Heart Disease Extends to Genetics. *Ann Hum Genet*; 72(Pt 1):48-56.

- (10) Sampietro ML, **Lao O**, Caramelli D, Lari M, Pou R, Martí M, Bertranpetit J, Lalueza-Fox C. (2007) Palaeogenetic evidence supports a dual model of Neolithic spreading into Europe. *Proc Biol Sci*; 274(1622):2161-7.
- (11) Myles S, Hradetzky E, Engelken J, **Lao O**, Nürnberg P, Trent RJ, Wang X, Kayser M, Stoneking M. (2007) Identification of a candidate genetic variant for the high prevalence of type II diabetes in Polynesians. *Eur J Hum Genet*; 15(5):584-9.
- (12) Sistonen J, Sajantila A, **Lao O**, Corander J, Barbujani G, Fuselli S. (2007) CYP2D6 worldwide genetic variation shows high frequency of altered activity variants and no continental structure. *Pharmacogenet Genomics*; 17(2):93-101.
- (13) Kayser M, Brauer S, Cordaux R, Casto A, **Lao O**, Zhivotovsky LA, Moyse-Faurie C, Rutledge RB, Schiefenhoewel W, Gil D, Lin AA, Underhill PA, Oefner PJ, Trent RJ, Stoneking M. (2006) Melanesian and Asian Origins of Polynesians: mtDNA and Y-chromosome Gradients Across the Pacific. *Mol Biol Evol*; 23(11):2234-44.
- (14) Sampietro ML, Gilbert MT, **Lao O**, Caramelli D, Lari M, Bertranpetit J, Lalueza-Fox C. (2006) Tracking down Human Contamination in Ancient Human Teeth. *Mol Biol Evol*; 23(9):1801-1807.
- (15) **Lao O**, van Duijn K, Kersbergen P, de Knijff P, Kayser M. (2006) Proportioning whole-genome single-nucleotide-polymorphism diversity for the identification of geographic population structure and genetic ancestry. *Am J Hum Genet*; 78(4):680-90.
- (16) Gonzalez-Neira A, Ke X, **Lao O**, Calafell F, Navarro A, Comas D, Cann H, Bumpstead S, Ghori J, Hunt S, Deloukas P, Dunham I, Cardon LR, Bertranpetit J. (2006) The portability of tagSNPs across populations: a worldwide survey. *Genome Res*; 16(3):323-30.
- (17) Gardner M, Gonzalez-Neira A, **Lao O**, Calafell F, Bertranpetit J, Comas D. (2006) Extreme population differences across Neuregulin 1 gene, with implications for association studies. *Mol Psychiatry*; 11(1):66-75.
- (18) Marques-Bonet T*, **Lao O***, Goertsches R, Comabella M, Montalban X, Navarro A. (2005) Association Cluster Detector: a tool for heuristic detection of significance clusters in whole-genome scans. *Bioinformatics*; 21 Suppl 2:ii180-ii181. * Shared authorship.
- (19) Marques-Bonet T*, **Lao O***, Goertsches R, Montalban X, Navarro A, Comabella M. (2005) A tool for detection of association clusters in whole genome scans. *Multiple Sclerosis*; 11(Suppl 1): S112-S113. · These authors contributed equally to this work
- (20) Sampietro ML, Caramelli D, **Lao O**, Calafell F, Comas D, Lari M, Agusti B, Bertranpetit J, Lalueza-Fox C. (2005) The genetics of the pre-Roman Iberian Peninsula: a mtDNA study of ancient Iberians. *Ann Hum Genet*; 69(Pt 5):535-48.
- (21) Ramirez-Soriano A, **Lao O**, Soldevila M, Calafell F, Bertranpetit J, Comas D. (2005) Haplotype tagging efficiency in worldwide populations in CTLA4 gene. *Genes Immun*; 6(8):646-57.
- (22) Kayser M, **Lao O**, Anslinger K, Augustin C, Bargel G, Edelmann J, Elias S, Heinrich M, Henke J, Henke L, Hohoff C, Illing A, Jonkisz A, Kuzniar P, Lebioda A, Lessig R, Lewicki S, Maciejewska A, Monies DM, Pawlowski R, Poetsch M, Schmid D, Schmidt U, Schneider PM, Stradmann-Bellinghausen B, Szibor R, Wegener R, Wozniak M, Zoledziwska M, Roewer L, Dobosz T, Ploski R. (2005) Significant genetic differentiation between Poland and Germany follows present-day political borders, as revealed by Y-chromosome analysis. *Hum Genet*; 117(5):428-43.

- (23)González-Neira G, Calafell F, Navarro A, **Lao O**, Cann H, Comas D, Bertranpetit J. (2004) Geographic stratification of linkage disequilibrium: a worldwide population study in a region of chromosome 22. *Hum Genomics*; 1(6):399-409.
- (24)Aldea A, Calafell F, Aróstegui JI, **Lao O**, Rius F, Plaza S, Masó M, Vives J, Yagüe J. (2004) The west side story: *MEFV* haplotype in Spanish FMF patients and in controls. Evidences of high LD and a recombination “hot-spot” at the *MEFV* locus. *Hum Mutat*; 23(4):399.
- (25)Comas D, Plaza S, Wells RS, Yuldasheva N, **Lao O**, Calafell F, Bertranpetit J. (2004) Admixture, migrations, and dispersals in Central Asia: evidence from maternal DNA lineages. *Eur J Hum Genet*, 12(6):495-504.
- (26) Andres AM, Soldevila M, **Lao O**, Volpini V, Jacobs HT, Saitou N, Hayasaka I, Calafell F, Bertranpetit J. (2004) Comparative genetics of functional trinucleotide tandem repeats in humans and apes. *J Mol Evol*, 59(3):329-339.
- (27)Andres AM, **Lao O**, Soldevila M, Calafell F, Bertranpetit J. (2003) Dynamics of CAG repeat loci revealed by the analysis of their variability. *Hum Mutat*, 21(1):61-70.
- (28)**Lao O**, Andres AM, Mateu E, Bertranpetit J, Calafell F. (2003) Spatial patterns of cystic fibrosis mutation spectra in European populations. *Eur J Hum Genet*, 11(5):385-94.
- (29)Mateu E, Calafell F, **Lao O**, Bonne-Tamir B, Kidd JR, Pakstis A, Kidd KK, Bertranpetit J. (2001) Worldwide genetic analysis of the CFTR region. *Am J Hum Genet*, 68(1):103-17. 119.

Invited Review Articles in Peer-Reviewed Scientific Journals:

- (1) **Lao O** and Kayser M. (2008) Positive selection of the *OCA2/HERC2* region and human pigmentation traits. *Cell Science Reviews*. Vol 5 No 2, http://www.cellscience.com/Reviews18/Positive_selection_OCA2.html.
- (2) **Lao O** and Kayser M (2008) Mechanismen und Auswirkungen natürlicher Selektion (Mechanisms and effects of natural selection). (Overview paper) Special issue on population genetics of the *Zeitschrift für Medizinische Genetik*, 20:308–314, online published 1. September 2008, DOI 10.1007/s11825-008-0122-y (in German)

Book Chapters:

- (1) **Lao O** and Kayser M (2009) Op het eerste gezicht: Hoe het klimaat onze huidskleur dicteerde (At the first glance: how climate has influenced our skin color) In: *Evolutie zit in je genen: over Darwin en genomics (Evolution is in your genes: on Darwin and genomics)* Geraedts JPM, de Knijff P, Everdingen JJE, van den Broek J, Smits R (Eds), Stichting Bio-Wetenschappen en Maatschappij / Netherland Research Council (NWO), Den Haag, The Netherlands, 2009, 47-54
- (2) **Lao O** and Kayser M (2009) Human relationships inferred from genetic variation. In: *Encyclopaedia of Life Sciences*, Hildegard Kehrler-Sawatzki (Ed), John Wiley & Sons, Inc, US, in press

Invited Lectures at Scientific Workshops and Courses:

- (1) “Ascertaining AIMs for detecting population substructure in the European continent”. **O Lao**. First Workshop on Whole-Genome Association Studies. University Pompeu Fabra. Barcelona, November 2006.

- (2) Jornadas Bioinformatica VIII. Invited speaker. **O Lao**. "Optimal algorithms for ascertaining ancestry sensitive markers (ASMs) in human populations" Ciudad de las Ciencias. Valencia, February 2008.
- (3) Programme Workshop on Phylogeny & Genetics in microbiology and virology. Invited speaker. **O Lao**. "Detecting signatures of selection in higher organisms". May 2008
- (4) Annual AGD Meeting. **O Lao**. "Genetic ancestry testing in human populations". Koln University. Postdam (Germany), September 2008
- (5) DIMACS Workshop on Algorithmics in Human Population-Genomics. **O Lao**. "Efficient algorithms for ascertaining markers for controlling for population substructure". DIMACS. New Jersey (US), May 2008

Oral Presentations at Scientific Conferences (non-invited):

- (1) II Jornadas españolas de Bioinformática. **Lao O**, Andrés A, Bertranpetit J, Calafell F. "Mantenimiento de la diversidad genómica en enfermedades mendelianas: factores poblacionales vs. factores genómicos". Málaga, June 2001.
- (2) II Jornada de Biología Evolutiva. **Lao O**, Bertranpetit J, Calafell F. "Els patrons espacials de l'espectre de mutacions de la β -talassèmia reflecteixen l'aparició recent de la malària". Barcelona, July 2002.
- (3) 2nd International Symposium of DNA polymorphisms in Human populations. **Lao O**, Dupanloup I, Barbujani G, Bertranpetit J, Calafell F. "Geographic structure of variation at SNPs conferring risk for Coronary Heart Disease". Paris, November 2003.
- (4) IV Jornada de Biología Evolutiva. **Lao O**, Malats N, Dupanloup I, Barbujani G, Bertranpetit J, Calafell F. "Anàlisi del patró de distribució de polimorfismes associats a malalties complexes en poblacions humanes". Barcelona, 2003.
- (5) Human Genome Meeting 2006. **Lao O**, van Duijn K, Kersbergen P, de Kniff P, Kayser M. "Proportioning whole-genome single-nucleotide-polymorphism diversity for identification of geographic population structure and genetic ancestry". Helsinki, May 2006.
- (6) *European Mathematical Genetics Meeting 2008 (EMGM-2008)*. **Lao O**, Aulchenko Y, Kayser M. "In silico genome-wide association mapping of complex traits". Rotterdam, March 2008.

Poster Presentations at Scientific Conferences:

- (1) Cold Spring Harbor Laboratory Meeting on Human Evolution. Poster: Lao O, Andrés A, Bertranpetit J, Calafell F: "Geographic patterns of mutation diversity point to population factors in shaping mutation spectra in cystic fibrosis and phenylketonuria". Cold Spring Harbor (NY, USA), November 2002.
- (2) Human Genome Meeting 2004. Lao O, Dupanloup I, Barbujani G, Bertranpetit J, Calafell F. "Geographic structure of variation at SNPs conferring risk for Coronary Heart Disease". Berlin, March 2004.
- (3) SMBE meeting 2008. Lao O, Aulchenko Y, Kayser M. "In silico genome-wide association mapping of complex traits". Barcelona, June 2008.
- (4) SMBE meeting 2008. Martinez-Marigorta U, Lao O, Casals F, Calafell F, Bosch E, Morcillo C, Navarro A. "The effect of population genetic differentiation on the replicability of association studies". Barcelona, June 2008.
- (5) XX International congress of genetics 2008. Lao O, Lu TT, Nothnagel M, Junge O, Freitag-Wolf S, Caliebe A, Kayser M, Krawczak M. "Ascertaining Ancestry Sensitive Markers for Investigating Geographic Differentiation in the European Continent". Berlin, July 2008