

Mario Cáceres Aguilar

1. Personal Information

Current position: ICREA Research Professor, Hospital del Mar Research Institute (IMIM).
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 Webpage: <https://www.imim.es/programesrecerca/informaticabiomedica/genomica-comparada-funcional.html>
 Place of birth: Madrid (Spain) Date of birth: August 13th, 1972
 Researcher ID: A-6379-2009 ORCID: 0000-0002-7736-3251

2. Summary

I obtained my PhD at the Universitat Autònoma de Barcelona working on *Drosophila* chromosomal rearrangements (1995-2000). Then I moved to the USA as a postdoc at the Salk Institute for Biological Studies (2001-2003) and Emory University (2003-2006), where my research shifted to the use of novel genomic techniques to compare gene-expression levels in humans and non-human primates. In 2006 I got a Ramón y Cajal position at the Centre for Genomic Regulation in Barcelona, and focused on the identification of genomic changes with potential functional consequences in the human lineage, such as those associated with expression differences and structural variants. Since 2010 I am an ICREA Research Professor and lead the Comparative and Functional Genomics group, first at the Institut de Biotecnologia i de Biomedicina (IBB) of the UAB and from 2023 at the Research Programme on Biomedical Informatics of the Hospital del Mar Research Institute (IMIM). In particular, one of my main research lines is the evolutionary and functional analysis of polymorphic inversions in the human genome, which aims to investigate the biological significance of these variants at a large scale. To do this my group takes a multidisciplinary approach that combines new genomic methods and bioinformatic analysis of the great wealth of data available, generating results of interest to diverse fields.

As a result of my scientific activity, I have achieved a significant scientific production and co-authored a total of 48 research articles (H-index = 30, I10-index = 41) that stand out for their publication in high-quality journals, including some of the most prestigious ones like *Science*, *Nature*, *PNAS*, *Nature Communications* or *Nature Reviews Genetics*. Several of these articles represented significant advances in the respective fields and have received considerable attention, with an average number of 84 citations per article. In addition, I have succeeded in securing numerous fellowships and grants to fund my research and I have experience in managing big research projects, including an EMBO Postdoctoral Fellowship, the Salk Institute for Biological Studies Presidents' Club Innovation Grant, five 3-year grants from the Spanish Ministry of Science and Education, a 1.5 million euros ERC Starting Grant, and two ERC Proof of Concept grants. I also have experience in supervising students, with 8 doctoral theses (plus 5 more ongoing) and 11 master theses supervised. Finally, I have been distinguished with the 2007 Josep M. Sala-Trepas Award for the scientific trajectory of a young researcher working in the field of gene-expression regulation and the 2021 BioRad Positive Droplet Award on Copy Number Variation.

Quality indicators in Scientific Production:

Total number of publications	48	1st quartile publications (Q1)	42
Publications within 2 first/last authors	37	1st decile publications (D1)	25
Total number of citations	4438*	Average citations/year (2019-2023)	331.2*
H-index	31*	i20-index	44
Doctoral theses supervised	8	Master theses supervised	11

* Citations values obtained from Google Scholar

3. Professional background

- 2/2023-present: ICREA Research Professor and Group leader of the Comparative and Functional Genomics Group, Research Programme on Biomedical Informatics, Hospital del Mar Research Institute (IMIM) (Spain).
- 1/2010-12/2024: ICREA Research Professor and Group leader of the Comparative and Functional Genomics Group, Institut de Biotecnologia i de Biomedicina, Universitat Autònoma de Barcelona (Spain).
- 3/2010-9/2023: Associate Professor, Departament de Genètica i de Microbiologia, Universitat Autònoma de Barcelona (Spain).
- 4/2006-12/2010: Ramón y Cajal Researcher, Genes and Disease Program, Centre for Genomic Regulation, Barcelona (Spain).

4. Education and Postdoctoral Experience

- 9/2003-4/2006: Post-doctoral fellow, Department of Human Genetics and Yerkes National Primate Research Center, Emory University School of Medicine, Atlanta, GA (USA). Advisors: Todd M. Preuss and James W. Thomas.
- 4/2003-8/2003: Visiting scientist, Departament de Genètica i de Microbiologia, Universitat Autònoma de Barcelona (Spain). Advisors: Alfredo Ruiz and Antonio Barbadilla.
- 1/2001-4/2003: Post-doctoral fellow, Laboratory of Genetics, The Salk Institute for Biological Studies, La Jolla, CA (USA). Advisor: Carrolee Barlow.
- 9/1995-12/2000: Ph.D. in Biology, Departament de Genètica i de Microbiologia, Universitat Autònoma de Barcelona (Spain). Thesis: Chromosomal inversions in *Drosophila*: Molecular origin and evolutionary significance of their size. Advisors: Alfredo Ruiz and Antonio Barbadilla.
- 7/1998-9/1998: Short-research stage, Laboratory of Manyuan Long, Department of Ecology and Evolution, University of Chicago, IL (USA). Duration: 3 months.
- 9/1995-10/1997: Master in Genetics, Departament de Genètica i de Microbiologia, Universitat Autònoma de Barcelona (Spain). Thesis: Inversion length and breakpoint distribution in the *Drosophila buzzatii* species complex. Advisors: Alfredo Ruiz and Antonio Barbadilla.
- 10/1990-7/1995: First degree in Biology (First class honors), Universitat Autònoma de Barcelona (Spain).

5. Honors and Awards

- 2021: BioRad Positive Droplet Award on Copy Number Variation.
- 2010: I3 Program Certification, Ministerio de Ciencia e Innovación (Spain).
- 2008: Research Accreditation of proven research capabilities by the Agency for the Quality of the University System of Catalonia (AQU Catalunya, Spain).
- 2007: Josep M. Sala-Trepapat Award for the scientific trajectory of a young researcher working in the field of gene-expression regulation, Societat Catalana de Biologia (Spain).
- 2001: Doctoral Thesis Special Distinction Award, Universitat Autònoma de Barcelona (Spain).
- 1995: Third Nation-Wide Award for Biology Degree Students, Ministerio de Educación y Ciencia (Spain).
- 1995: Biology Degree Special Distinction Award, Universitat Autònoma de Barcelona (Spain).

6. Funding and Fellowships

- 2023-2026: Research Grant, Agencia Estatal de Investigación (Spain). Title: Complete characterization of human polymorphic inversions with long-read sequence data. Duration: 1/9/2023-31/8/2026. Funding: 237,500 €. PI: Mario Cáceres.
- 2023-2026: Support to Research Groups, Agencia de Gestio d'Ajuts Universitaris i de Recerca, Generalitat de Catalunya (Spain). Title: Genomics, Bioinformatics and Evolutionary Biology Group (GBBE 2021 SGR 00526). Duration: 1/1/2023-31/12/2026. Funding: 40,000 €. PI: Mauro Santos.
- 2021-2024: Industrial Doctorate Grant, Agencia de Gestio d'Ajuts Universitaris i de Recerca, Generalitat de Catalunya (Spain). Title: Exploring genetic risk factors affecting brain function in the context of neurodevelopmental disorders. Duration: 1/9/2021-31/8/2025. Funding: 55,560 €. PI: Laura Pérez Cano and Mario Cáceres.
- 2020-2023: Research Grant, Agencia Estatal de Investigación (Spain). Title: On the role of human recurrent mutations in genome function, evolution and diseases. Duration: 1/6/2020-31/12/2023. Funding: 217,800 €. PI: Mario Cáceres.
- 2017-2021: Support to Research Groups, Agencia de Gestio d'Ajuts Universitaris i de Recerca, Generalitat de Catalunya (Spain). Title: Genomics, Bioinformatics and Evolutionary Biology Group (GBBE 2017 SGR 1379). Duration: 1/4/2017-30/09/2021. Funding: 41,475 €. PI: Alfredo Ruiz.
- 2016-2020: Research Grant, Agencia Estatal de Investigación (Spain). Title: Integrative analysis of the functional impact of inversions in genomes and phenotypic traits. Duration: 30/12/2016-29/12/2020. Funding: 217,800 €. PI: Mario Cáceres.
- 2017-2018: ERC Proof of Concept Grant, European Research Council (EU). Title: Application of genomic inversions as diagnostic markers in precision medicine (IN2DIAG). Duration: 1/7/2017-31/12/2018. Funding: 150,000 €. PI: Mario Cáceres.
- 2014-2016: Research Grant, Agencia Estatal de Investigación (Spain). Title: Integrative analysis of the functional impact of inversions in genomes and phenotypic traits. Duration: 30/12/2016-29/12/2019. Funding: 217,800 €. PI: Mario Cáceres.
- 2015-2016: Reagents research support, Bio-Rad Laboratories (USA). Title: Research study of inversion genotyping by droplet digital PCR (ddPCR). Duration: 12/05/2015-11/11/2016. Funding: 72,727 €. PI: Mario Cáceres.
- 2014-2016: Research Grant, Ministerio de Economía y Competitividad (Spain). Title: Understanding genome variation from nucleotides to phenotypes in *Drosophila* and humans. Duration: 1/1/2014-31/12/2016. Funding: 181,500 €. PI: Mario Cáceres and Antonio Barbadilla.
- 2014-2016: Support to Research Groups, Agencia de Gestio d'Ajuts Universitaris i de Recerca, Generalitat de Catalunya (Spain). Title: Genomics, Bioinformatics and Evolutionary Biology Group (GBBE, 2014-SGR-1346). Duration: 1/1/2014-31/3/2017. Funding: 48,000 €. PI: Alfredo Ruiz.
- 2010-2015: ERC Starting Grant, European Research Council (EU). Title: Evolutionary and functional analysis of polymorphic inversions in the human genome (INVFEST). Duration: 1/2/2010-31/10/2015. Funding: 1,475,377 €. PI: Mario Cáceres.
- 2014-2015: ERC Proof of Concept Grant, European Research Council (EU). Title: A novel high-throughput tool to genotype human genome inversions for personalized medicine (INGENIHUS). Duration: 1/6/2014-31/5/2015. Funding: 149,670 €. PI: Mario Cáceres.
- 2013-2015: Amazon Web Services in Education Team Grant, Amazon (USA). Title: Bioinformatic genotyping of inversions from 1000 Genomes Project data. Duration: 15/04/2013 - 30/04/2015. Funding: 5,000 \$. PI: Mario Cáceres.
- 2010: ICREA Research Professor Position, Institutió Catalana de Recerca i Estudis Avançats (Spain).
- 2007-2009: Research Grant, Ministerio de Educación y Ciencia (Spain). Title: Genomic determinants of gene expression levels in the human brain (BFU2007-60930). Duration: 1/10/2007-31/12/2009. Funding: 105.270 €. PI: Mario Cáceres.

- 2006-2009: Ramón y Cajal Research Position, Ministerio de Educación y Ciencia (Spain). Title: Gene expression changes in the human brain: Role in cognitive functions and mental disease (RYC-2005-001572). Duration: 10/4/2006-31/12/2009. Funding: 180,400 €. PI: Mario Cáceres.
- 2005: ICREA Junior Research Position, Institució Catalana de Recerca i Estudis Avançats (Spain). (Declined due to incompatibility with Ramón y Cajal Research Position)
- 2001-2002: European Molecular Biology Organization (EMBO) Long Term Post-Doctoral Fellowship.
- 2001-2002: The Salk Institute for Biological Studies Presidents' Club Innovation Grant (USA).
- 1996-1999: Doctoral FI Research Fellowship for Graduate Students, Generalitat de Catalunya (Spain).
- 1998: Short Term Travel Grant, Generalitat de Catalunya (Spain).
- 1995: Research Fellowship for Undergraduate Students, Ministerio de Educación y Ciencia (Spain).

7. Supervising of PhD Students

- 2018-2024: Elena Campoy, Departament de Genètica i de Microbiologia, Universitat Autònoma de Barcelona (Spain). Advisor: Mario Cáceres. Thesis: Bioinformatic analysis of the functional effects of human polymorphic inversions on gene expression, epigenetics and phenotypic traits. Thesis defense: 20/3/2024.
- 2015-2019: Jon Lerga, Departament de Genètica i de Microbiologia, Universitat Autònoma de Barcelona (Spain). Advisor: Mario Cáceres. Thesis: Integrative analysis of the functional consequences of inversions in the human genome. Qualification: Cum laude.
- 2013-2017: Carla Giner, Departament de Genètica i de Microbiologia, Universitat Autònoma de Barcelona (Spain). Advisor: Mario Cáceres. Thesis: Large-scale evolutionary analysis of polymorphic inversions in the human genome. Qualification: Cum laude.
- 2010-2014: Meritxell Oliva, Departament de Genètica i de Microbiologia, Universitat Autònoma de Barcelona (Spain). Advisor: Mario Cáceres. Thesis: Functional impact of polymorphic inversions in the human genome. Qualification: Cum laude.
- 2010-2014: David Vicente, Departament de Genètica i de Microbiologia, Universitat Autònoma de Barcelona (Spain). Advisor: Mario Cáceres. Thesis: Analysis, validation and population study of inversions between two human genomes. Qualification: Excellent.
- 2008-2013: Alexander Martínez (PhD student), Departament de Ciències Experimentals i de la Salut, Universitat Pompeu Fabra, Barcelona (Spain). Advisor: Mario Cáceres. Thesis: Bioinformatic characterization and analysis of polymorphic inversions in the human genome. Qualification: Excellent.
- 2008-2013: Raquel Rubio (PhD student), Departament de Genètica i de Microbiologia, Universitat Autònoma de Barcelona (Spain). Advisor: Mario Cáceres. Thesis: Molecular analysis of the mechanisms involved in THBS4 differential gene expression in the human brain. Qualification: Cum laude.
- 2000-2010: Marta Puig (PhD student), Departament de Genètica i de Microbiologia, Universitat Autònoma de Barcelona (Spain). Advisors: Alfredo Ruiz and Mario Cáceres. Thesis: Functional analysis of position effects of inversion 2j in *Drosophila buzzatii*: Gene *CG13617* silencing and its adaptive significance. Qualification: Cum laude.

8. Supervising of Master Students

- 2021-2022: Maria Diaz Ros, Master in Bioinformatics, Universitat Pompeu Fabra (Spain). Advisor: Mario Cáceres. Thesis: Long-term inversion recurrence and segmental duplication conservation during mammalian evolution.
- 2021: Judit Camps Bruch, Master in Advanced Genetics, Universitat Autònoma de Barcelona (Spain). Advisor: Mario Cáceres. Thesis: Evolutionary analysis of inversions in ancient human genomes.

- 2018: Ruth Gómez, Master in Bioinformatics, Universitat Autònoma de Barcelona (Spain). Advisor: Mario Cáceres and Antonio Barbadilla. Thesis: Improving interoperability between InvFEST and PopHuman.
- 2018: Claudia Ramírez, Master in Advanced Genetics, Universitat Autònoma de Barcelona (Spain). Advisor: Mario Cáceres. Thesis: Characterization, validation and genotyping of inversions predicted with new genomic techniques.
- 2018: Miriana Bitonto, Master in Cellular and Molecular Biology, University of Bari Aldo Moro (Italy). Advisor: Francesca Antonacci and Mario Cáceres. Thesis: Genomic inversions in human and non-human primates.
- 2017: Juan Ochoteco, Master in Bioinformatics, Universitat Autònoma de Barcelona (Spain). Advisor: Mario Cáceres. Thesis: Re-annotation of the *Drosophila buzzatii* genome.
- 2017: Jorge de los Santos, Master in Advanced Genetics, Universitat Autònoma de Barcelona (Spain). Advisor: Mario Cáceres. Thesis: Mosaicism detection in human inversions.
- 2017: Teresa Soos, Master in Science, University of Natural Resources and Life Sciences, Vienna (Austria). Advisors: Mario Cáceres and Lukas Mach. Thesis: Validation of polymorphic human inversions and setting up of high-throughput genotyping assays.
- 2016: Noelia Pérez, Master in Advanced Genetics, Universitat Autònoma de Barcelona (Spain). Advisor: Mario Cáceres. Thesis: Detection of mosaicism for human inversions using ddPCR.
- 2015: Jon Lerga, Master in Bioinformatics, Universitat Autònoma de Barcelona (Spain). Advisor: Mario Cáceres. Thesis: Landscape and functional impact of polymorphic inversions on the human genome.
- 2015: Maria Bellet, Master in Advanced Genetics, Universitat Autònoma de Barcelona (Spain). Advisor: Mario Cáceres. Thesis: Validation of differences in gene expression related to human polymorphic inversions.
- 2014: Sergi Hervás, Master in Bioinformatics, Universitat Autònoma de Barcelona (Spain). Advisors: Mario Cáceres and Lorena Pantano. Thesis: Effects of polymorphic inversions on epigenetic regulation marks in humans.
- 2013: Carla Giner, Master in Bioinformatics, Universitat Autònoma de Barcelona (Spain). Advisors: Mario Cáceres and Magda Gayà. Thesis: Evolutionary history of human polymorphic inversions: A bioinformatics approach.

9. Teaching Experience

- 2013-2021: One 3-hour session of the Recent Advances in Genetics module of the Master in Advanced Genetics, Universitat Autònoma de Barcelona (Spain).
- 2012-2019: Three 2-hour sessions of the Genomics module of the Master in Bioinformatics for Genomics and Drug Design, Universitat Autònoma de Barcelona (Spain).
- 2010-2012: Three 2-hour sessions of the Genomics and Proteomics module of the Master in Advanced Genetics, Universitat Autònoma de Barcelona (Spain).
- 2006: Session on Brain evolution of the Master in Biomedical Research (BIOMED), Universitat Pompeu Fabra (Spain).
- 2004: Session on Functional Genomics and Microarrays of the First International Postgraduate Course in Genomics, Universitat Autònoma de Barcelona (Spain).
- 2000: Teaching Assistant in Genetics, Universitat Autònoma de Barcelona (Spain).
- 1995: Teaching Assistant in Genetics, Universitat Autònoma de Barcelona (Spain).

10. Other Research Activities

- 2023: Organization of the 11th Bioinformatics and Genomics Symposium of the Societat Catalana de Biologia (15-16/12/2023). Barcelona, Spain.
- 2023: Organization of the 2023 ACGT Computational Genomics Workshop (13-14/4/2023). Sant Feliu de Guixols, Spain.
- 2022: Organization of the 10th Bioinformatics and Genomics Symposium of the Societat Catalana de Biologia (15-16/12/2022). Valencia, Spain.
- 2021: Organization of the 9th Bioinformatics and Genomics Symposium of the Societat Catalana de Biologia (16/12/2021). Online Conference.
- 2020: Organization of the COVID-19 8th Bioinformatics and Genomics Symposium of the Societat Catalana de Biologia (14/12/2020). Online Conference.
- 2019: Organization of the 7th Bioinformatics and Genomics Symposium of the Societat Catalana de Biologia (17/12/2019). Barcelona, Spain.
- 2019-present: Associated Editor for the journal *NAR Genomics and Bioinformatics*.
- 2018: Organization of the 6th Bioinformatics and Genomics Symposium of the Societat Catalana de Biologia (20/12/2018). Barcelona, Spain.
- 2018: Member of the Programme Committee of the Barcelona Next Generation Sequencing Conference 2018 (NGS 2018) (9-11/4/2018). Barcelona, Spain.
- 2017: Organization of the 5th Bioinformatics and Genomics Symposium of the Societat Catalana de Biologia (20/12/2017). Barcelona, Spain.
- 2017-2023: Associated Editor for the journal *Genes*.
- 2017: Member of the Programme Committee of the Barcelona Next Generation Sequencing Conference 2017 (NGS 2017) (3-5/4/2018). Barcelona, Spain.
- 2016: Organization of the 4th Bioinformatics and Genomics Symposium of the Societat Catalana de Biologia (20/12/2016). Barcelona, Spain.
- 2016: Organization of the XXI Seminario de Genética de Poblaciones y Evolución (3-5/10/2016). Sitges, Spain.
- 2015: Guest Editor of 'Special Issue: Functional impact of structural variation in the human genome', Briefings in Functional Genomics, Oxford University Press, Oxford.
- 2014: Organization of the 10th Workshop on Genomics and Proteomics of the Societat Catalana de Biologia (3/10/2014). Barcelona, Spain.
- 2013: Organization of the 9th Workshop on Genomics and Proteomics of the Societat Catalana de Biologia (17/12/2013). Barcelona, Spain.
- 2012: Member of the Scientific Committee of the I Congrés Internacional de Biologia de Catalunya (CIBICAT) of the Societat Catalana de Biologia (9-12/7/2012). Barcelona, Spain.
- 2012: Organization of the Symposium on Structural Evolution of Genomes of the 2012 Annual Meeting of the Society for Molecular Biology and Evolution (23-26/6/2012). Dublin, Ireland.
- 2011-2019: Organization of the bi-monthly Seminar Series on Research in Genomics and Evolution at the Universitat Autònoma de Barcelona. Barcelona, Spain.
- 2011: Organization of the Workshop on Maldi.MS-Imaging for Tissues & Biomedical Proteomics of the Societat Catalana de Biologia (29/11/2011). Barcelona, Spain.
- 2011-2019: Coordinator of the Genomics and Proteomics Section of the Societat Catalana de Biologia.

- 2010: Organization of the 7th Workshop on Biomedical Genomics and Proteomics of the Societat Catalana de Biologia (17/12/2010). Barcelona, Spain.
- 2010-2023: Associated Editor for the journal *BMC Genomics*.
- 2005-present: Scientific reviewer for the journals *Genome Research*, *Nucleic Acids Research*, *Molecular Biology and Evolution*, *Bioinformatics*, *BMC Genomics*, *Human Mutation*, *Genetics*, *Genome Biology and Evolution*, *Genetica*, *Journal of Molecular Evolution*, *Molecular Ecology*, *Evolutionary Bioinformatics*, and *PLoS One*.

11. Patents

- 2013 **M. Cáceres**, S. Villatoro, and C. Aguado. An in vitro method of genotyping multiple inversions. EU Patent Application EP13382296.5. Priority date: 23/7/2013.

12. Publications

- 2024 S. Azidane Chenlo, X. Gallego, L. Durham, M. Cáceres, E. Guney, L. Pérez-Cano. Identification of novel driver risk genes in CNV loci associated with neurodevelopmental disorders. *HGG Advances* (Submitted).
- 2023 COVID-19 Host Genetics Initiative (including J. Lerga-Jaso and **M. Cáceres**). A second update on mapping the human genetic architecture of COVID-19. *Nature* 621: E7–E26.
- 2022 J. Valls-Margarit, I. Galván-Femenía, D. Matias-Sánchez, N. Blay, M. Puiggròs, A. Carreras, C. Salvoró, B. Cortés, R. Amela, X. Farre, J. Lerga-Jaso, M. Puig, J. F. Sánchez-Herrero, V. Moreno, M. Perucho, L. Sumoy, Ll. Armengol, O. Delaneau, **M. Cáceres**, R. de Cid, D. Torrents. GCAT|Panel, a comprehensive structural variant haplotype map of the Iberian population from high-coverage whole-genome sequencing. *Nucleic Acids Research* 50: 2464-2479
- 2022 F. Degenhardt*, D. Ellinghaus*, S. Juzenas*, J. Lerga-Jaso*, M. Wendorff*, D. Maya-Miles*,... (+ 323 authors)... T. L. Lenz[§], R. Asselta[§], R. de Cid[§], L. Valenti[§], T. H. Karlsen[§], **M. Cáceres**[§], A. Franke[§]. Detailed stratified GWAS analysis for severe COVID-19 in four European populations. *Human Molecular Genetics*, 31: 3945-3966.
- 2022 E. Campoy*, M. Puig*, I. Yakymenko, J. Lerga-Jaso, **M. Cáceres**[§]. Genomic architecture and functional effects of human inversion supergenes. *Philosophical Transactions of the Royal Society B* 377: 20210209.
- 2021 COVID-19 Host Genetics Initiative (including J. Lerga-Jaso and **M. Cáceres**). Mapping the human genetic architecture of COVID-19 by worldwide meta-analysis. *Nature* 600: 472–477.
- 2020 F. A. M. Maggiolini, A. D. Sanders, C. J. Shew, A. Sulovari, Y. Mao, M. Puig, C. R. Catacchio, M. Dellino, D. Palmisano, L. Mercuri, M. Bitonto, D. Porubský, **M. Cáceres**, E. E. Eichler, M. Ventura, M. Y. Dennis, J. O. Korbé, F. Antonacci. Single-cell strand sequencing of a macaque genome reveals multiple nested inversions and breakpoint reuse during primate evolution. *Genome Research* 30: 1680-1693 (2020).
- 2020 M. Puig, J. Lerga-Jaso, C. Giner-Delgado, S. Pacheco, D. Izquierdo, A. Delprat, J. F. Regan, G. Karlin-Neumann, **M. Cáceres**[§]. Determining the impact of uncharacterized inversions in the human genome by droplet digital PCR. *Genome Research* 30: 724-735.
- 2019 C. Giner-Delgado*, S. Villatoro*, J. Lerga-Jaso*, M. Gayà-Vidal, M. Oliva, D. Castellano, L. Pantano, B. Bitarello, D. Izquierdo, I. Noguera, I. Olalde, A. Delprat, A. Blancher, C. Lalueza-Fox, T. Esko, P. O'Reilly, A. Andrés, L. Ferretti, M. Puig, **M. Cáceres**[§]. Evolutionary and functional impact of common polymorphic inversions in the human genome. *Nature Communications* 10: 4222.
- 2017 D. Vicente-Salvador*, M. Puig*, M. Gayà-Vidal, S. Pacheco, C. Giner-Delgado, I. Noguera, D. Izquierdo, A. Martínez-Fundichely, A. Ruiz-Herrera, X. Estivill, C. Aguado, J. I. Lucas-Lledó, and **M. Cáceres**[§]. Detailed analysis of inversions predicted between two human genomes: errors, real polymorphisms, and their origin and population distribution. *Human Molecular Genetics* 26: 567-581.

- 2015 M. Puig, D. Castellano, L. Pantano, C. Giner-Delgado, D. Izquierdo, M. Gayà-Vidal, J. I. Lucas-Lledó, T. Esko, C. Terao, F. Matsuda, and **M. Cáceres**[§]. Functional impact and evolution of a novel human polymorphic inversion that disrupts a gene and creates a fusion transcript. *PLoS Genetics* 11(10): e1005495. doi:10.1371/journal.pgen.1005495.
- 2015 M. Puig, S. Casillas, S. Villatoro, and **M. Cáceres**[§]. Human inversions and their functional consequences. *Briefings in Functional Genomics* 14: 369-379.
- 2015 **M. Cáceres**[§]. Structural variants, much ado about nothing? *Briefings in Functional Genomics* 14: 303-304.
- 2014 J. I. Lucas-Lledó, D. Vicente-Salvador, C. Aguado, and **M. Cáceres**. Population genetic analysis of bi-allelic structural variants from low-coverage sequence data with an expectation-maximization algorithm. *BMC Bioinformatics* 15:163.
- 2014 C. Aguado, M. Gayà-Vidal, S. Villatoro, M. Oliva, D. Izquierdo, C. Giner, V. Montalvo, J. García-González, A. Martínez-Fundichely, L. Capilla, A. Ruiz-Herrera, X. Estivill, M. Puig, and **M. Cáceres**[§]. Validation and genotyping of multiple human polymorphic inversions mediated by inverted repeats reveals a high degree of recurrence. *PLoS Genetics* 10(3): e1004208. doi:10.1371/journal.pgen.1004208.
- 2014 J. R. González, A. Cáceres, T. Esko, I. Cusco, M. Puig, M. Esnaola, V. Siroux, E. Bouzigon, R. Nadif, E. Reinmaa, L. Milani, M. Bustamante, D. Jarvis, J. M. Antó, J. Sunyer, F. Demenais, M. Kogevinas, A. Metspalu, **M. Cáceres**, and L. A. Pérez-Jurado. A common 16p11.2 inversion underlies the joint susceptibility to asthma and obesity. *American Journal of Human Genetics* 94: 361-372.
- 2013 A. Martínez-Fundichely, S. Casillas, R. Egea, M. Ràmia, A. Barbadilla, L. Pantano, M. Puig, and **M. Cáceres**[§]. InvFEST, a database integrating information of polymorphic inversions in the human genome. *Nucleic Acids Research* 42 (D1): D1027-D1032.
- 2013 J. Prado-Martínez, I. Hernando-Herraez, B. Lorente-Galdos, M. Dabad, O. Ramírez, C. Baeza-Delgado, C. Morcillo-Suarez, C. Alkan, F. Hormozdiari, E. Rainieri, J. Estelle, M. Fernández-Callejo, M. Valles, L. Ritscher, T. Schöneberg, E. de la Calle-Mustienes, S. Casillas, R. Rubio-Acero, M. Melé, J. Engelken, **M. Cáceres**, J. L. Gómez Skarmeta, M. Gut, J. Bertranpetit, I. G. Gut, T. Abello, E. E. Eichler, I. Mingarro, C. Lalueza-Fox, A. Navarro, and T. Marques-Bonet. The genome sequencing of an albino western gorilla reveals inbreeding in the wild. *BMC Genomics* 14: 363.
- 2013 R. Cagliani, F. Guerini, R. Rubio-Acero, F. Baglio, D. Forni, C. Agliardi, L. Griffanti, M. Fumagalli, U. Pozzoli, S. Riva, E. Calabrese, M. Sikora, F. Casals, G. P. Comi, N. Bresolin, **M. Cáceres**, M. Clerici, and M. Sironi. Long-standing balancing selection in the *THBS4* gene: influence on sex-specific brain expression and grey matter volumes in Alzheimer's disease. *Human Mutation* 34: 743-753.
- 2013 L. Bassaganyas, E. Riveira-Muñoz, M. García-Aragonés, J. R. González, **M. Cáceres**, Ll. Armengol, and X. Estivill. Worldwide population distribution of the common *LCE3C-LCE3B* deletion associated with psoriasis and other autoimmune disorders. *BMC Genomics* 14: 261.
- 2013 G. Escaramís, C. Tornador, L. Bassaganyas, R. Rabionet, J. M. C. Tubio, A. Martínez-Fundichely, **M. Cáceres**, M. Gut, S. Ossowski, and X. Estivill. PeSV-Fisher: Identification of somatic and non-somatic structural variants using next generation sequencing data. *PLoS One* 8: e63377.
- 2013 J. I. Lucas-Lledó and **M. Cáceres**[§]. On the power and the systematic biases of the detection of chromosomal inversions by paired-end genome sequencing. *PLoS One* 8: e61292.
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13. Oral Presentations at International Meetings and Invited Seminars

- 2023 Invited speaker at the Nanopore Day, Madrid 2023, Madrid, Spain.
- 2023 Department of Biological Sciences, University at Buffalo (online seminar).
- 2023 PRBB Computational Genomics Seminars, Parc de Recerca Biomèdica de Barcelona, Barcelona, Spain.
- 2023 Port d'Informació Científica, Universitat Autònoma de Barcelona, Barcelona, Spain.
- 2022 School of Biosciences; University of Kent (online seminar).
- 2021 Máster Universitario en Bioinformática, Universidad de Murcia (online seminar).
- 2020 Master in Genetics and Genomics, Universitat de Barcelona (online seminar).
- 2019 Icahn School of Medicine at Mount Sinai, New York, USA.
- 2018 Invited speaker at the 4th Annual Danish Bioinformatics Conference 2018, Odense, Denmark.
- 2017 IBB PI Seminar Series, Universitat Autònoma de Barcelona, Barcelona, Spain.
- 2017 Barcelona Next Generation Sequencing Conference 2017 (NGS 2017): Structural variation and population genomics, Barcelona, Spain.
- 2016 XXI Seminario de Genética de Poblaciones y Evolución, Sitges, Spain.
- 2016 Seminar Series on Research in Genomics and Evolution, Universitat Autònoma de Barcelona, Barcelona, Spain.
- 2015 Institut de Medicina Predictiva i Personalitzada del Càncer (IMPPC), Barcelona, Spain.
- 2015 Society for Molecular Biology and Evolution (SMBE) 2015 Annual Meeting, Vienna, Austria.
- 2014 The 1000 Genomes Project and Beyond Conference, Wellcome Trust, Cambridge, United Kingdom.
- 2014 Invited speaker at the 14th International Gene Forum 2014: Omics Meets Genomics, Estonian Genome Foundation, Tartu, Estonia.
- 2014 Institut de Biologia Evolutiva (CSIC-UPF), Barcelona, Spain.
- 2013 Invited speaker at the 12th Swedish Bioinformatics Workshop, University of Skövde, Skövde, Sweden.
- 2013 Universidad de Barcelona, Barcelona, Spain.
- 2013 XXIX Congreso de la Sociedad Española de Genética, Girona, Spain.

- 2011 Seminar Series on Research in Genomics and Evolution, Universitat Autònoma de Barcelona, Barcelona, Spain.
- 2010 Barcelona Biomedical Research Park (PRBB), Barcelona, Spain.
- 2010 The IX CRG Annual Symposium on Medical Genome Sequencing: Understanding the Genomes of Disease, Centre for Genomic Regulation, Barcelona, Spain.
- 2010 1st International Medical Research Association (IMRA) Conference, Universitat Autònoma de Barcelona, Barcelona, Spain.
- 2008 Society for Molecular Biology and Evolution (SMBE) 2008 Annual Meeting, Barcelona, Spain.
- 2007 Current Trends in Biomedicine Workshop on Deciphering the Regulatory Genome: Development, Evolution and disease, Universidad Internacional de Andalucía, Baeza, Spain.
- 2007 Departament de Genètica i de Microbiologia, Universitat Autònoma de Barcelona, Spain.
- 2004 Graduate Program in Population Biology, Ecology and Evolution, Emory University, Atlanta, Georgia, USA.
- 2003 Department of Human Genetics, Emory University School of Medicine, Atlanta, Georgia, USA.
- 2003 Departament de Ciències Experimentals i de la Salut, Universitat Pompeu Fabra, Barcelona, Spain.
- 2002 Department of Evolutionary Genetics, Max Planck Institute for Evolutionary Anthropology, Leipzig, Germany.
- 2002 Cold Spring Harbor Laboratory Meeting on Human Origins & Disease, Cold Spring Harbor Laboratory, New York, USA.
- 2002 Departament de Genètica i de Microbiologia, Universitat Autònoma de Barcelona, Barcelona, Spain.
- 2000 The Salk Institute for Biological Studies, San Diego, California, USA.
- 1999 40th Annual Drosophila Research Conference, Genetics Society of America, Bellevue, Washington, USA.