Jose A Caparros-Martin

Biologist | Molecular geneticist | Data Science

CONTACT

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SUMMARY

Results-driven molecular geneticist and data scientist with over 15 years of international and national experience in medical research. Proficient in several coding languages and dataflow management tools. Strong background in problem-solving and governance. Skilled in managing large datasets to support strategic initiatives.

SKILLS

Experienced in PC2 laboratory practices, molecular biology and molecular genetics techniques.
Bash, R, Python,
statistical and data analysis of high dimensional datasets,
community involvement,
project management, ethics, governance, grantsmanship.
scientific and lay writing.

EDUCATION

1998 - 2002	Degree in Biology. University of Alicante (Alicante, Spain)
2002 - 2006	PhD in Biotechnology, Molecular and Cell Biology Institute (IBMCP-CSIC). Polytechnic University of Valencia-UPV (Spain). Qualification: Excellent *Cum Laude*.

EXPERIENCE

2007 - 2008	Postdoctoral researcher, Molecular and Cell Biology Institute (IBMCP-CSIC). Polytechnic University of Valencia-UPV (Spain).
2008 - 2015	Postdoctoral training in molecular genetics and Researcher at CIBER of Rare Diseases (CIBERER, Madrid, Spain).
2016-2020	Research Fellow, Curtin University (Perth, Australia).
2020-2025	Research Fellow, Telethon Kids Institute (Perth, Australia).
2017 – Curr.	Organising Committee of the WA symposium on microbiome.
2021 – Curr.	Member of the working group of the Australasian Human Microbiome Research Network.
2021 – Curr.	Pacific Organising Committee of the Microbiome Virtual International Forum working group.
2022 – 2025	Member of the Wal-yan Respiratory Research Centre strategic subcommittee.
2023 – 2025	Reviewer for the Low-Risk Ethic Committee of the Child and Adolescence Health Services at Perth Children Hospital.
2023 – Curr.	Member of the School Board at Nedlands Primary School managing the STEM portfolio.

TEACHING

2021-2022. Master of System Medicine from the Australian National Phenome Centre at Murdoch University. Microbiome bioinformatics analysis class.

CAREER SUMMARY: I hold a degree in Biology from the University of Alicante (Spain) and a PhD in Biotechnology from the Polytechnic University of Valencia (Spain). I have received laboratory training in molecular genetics and bioinformatics. My work has gained international recognition, with publications in high-impact journals. I have established laboratory and computational workflows on microbiome analysis. At the Wal-yan Respiratory Research Centre, I led research on host-microbiota interaction in respiratory and gastrointestinal diseases and was accepted in the 2024 The Kids Institute Emerging Leaders program.

RESEARCH IMPACT: Among my scientific achievements is a novel mechanism with translational potential to explain the gut-lung axis in Cystic Fibrosis respiratory disease. In 2017, I led the first report involving the interaction between host genetics and the murine microbiota in the secondary effects associated with the treatment with lipid lowering medications. I have also led/participated in the identification of mutations in three new genes (*SP7*, *BMP1* and *WNT1*) as the genetic cause of Osteogenesis Imperfecta in humans, the identification of specific variants in the gene *WDR35* as the genetic cause of Ellis-van Creveld syndrome in humans, and a new causative gene of congenital multiminicore myopathy (*FXR1*). I also led the functional characterization of the Ellis-van Creveld proteins Evc and Evc2, and their role as mediators of Hedgehog signalling in mouse models.

SUPERVISION AND MENTORING: I have officially co-supervised 1 PhD student (UWA), 2 senior Thesis (honours equivalent), 2 Master's Thesis (awarded with the highest qualification), one Honours Student (Curtin University, first class) and a postdoctoral researcher. I am currently supervising two PhD students.

LEADERSHIP, COMMUNITY ENGAGEMENT AND INVOLVEMENT: I engage with the Child and Adolescent CF Consumer Reference Group and supports events like the Telethon Family Festival and the 65 Roses Day for Cystic Fibrosis. I have collaborated in different working groups with peers at local, national, and international levels. I am also co-leading the establishment of a consumer reference group on paediatric inflammatory bowel disease. Beyond my primary research work, I also participate in community-oriented initiatives, such as the School Program at Telethon Kids Institute. I am also in the School Board of Nedlands Primary School managing the STEM portfolio and have volunteered several years as coach and team manager in my son's rugby team (Nedlands RUFC).

PEER REVIEW AND GRANT ASSESSMENT: I am on the editorial board of BioTech (ISSN 2673-6284) managing 1-2 manuscripts a year. I am frequently asked to review manuscripts for a range of journals (*Publons profile* (https://publons.com/author/1318551/jose-caparros-martin#profile), including top tier journals such as *Gut, European Respiratory Journal*, *Communication Biology* or *BMC Genetics*. I have reviewed research projects for the Australian Research Council (ARC Discovery, 2020), the Australian National Health Medical Research Council (NHMRC, 2017-2019, 2022, 2023, Investigator grants (2024)), the Italian Ministry of Health (2022,2023), and Diabetes Australia (2017-2018).

CONTRIBUTION TO SCIENTIFIC AND PUBLIC OUTREACH EVENTS: I have been invited to speak to >20 different research forums, including local and national scientific forums, as well as seminars to a non-science audience. In 2016 I was invited by the Australian Academy of Sciences during the last Theo Murphy Frontiers of Sciences Microbiome Symposium in Adelaide, which highlights the relevance of my work on the study of microbial communities. In 2017, I was also selected by the Australian Academy of Sciences to participate as

Microbiome expert in the Think Tank "Rethinking food and nutrition science". The discussion paper resulting of this forum contributed to the consultation process during the development of a decadal plan for the discipline of nutrition in Australia. Since 2017 I am part of the organizing committee of the WA symposium on microbiome. Since 2021 I am part of the Australasian Human Microbiome Research Network, as well as the Microbiome Virtual International Forum working groups. Since 2022, I am part of the Wal-yan Respiratory Research Centre strategic subcommittee. Since 2023 I am appointed as reviewer for the Low-Risk Ethic Committee of the Child and Adolescence Health Services at Perth Children's Hospital.

CONTRIBUTIONS TO SCIENCE: I have published 45 original research papers (19 of them as first and/or senior author) in highly ranked international peer-reviewed journals (29 of them in Q1 Journals, including 3 papers highlighted by Faculty of 1000 as excellent reports), 5 reviews, 4 preprints and 2 book chapters. I have been cited over 2220 times (average citation per publication > 50) and have an *h*-index of 21 (source Google Scholar and Web of Science as on January 2025). Additionally, my research has been cited across 8 policy documents (https://policyprofiles.sagepub.com/dashboard) demonstrating that my work is considered authoritative and influential within the policy-making community.

TOP 5 PUBLICATIONS IN THE LAST 5 YEARS (Listed in descending order of publication date – full list of publications can be found at the end of the CV):

1. **Caparrós-Martín JA**, Saladié M, Agudelo-Romero P, Nichol KS, Reen FJ, Moodley Y, Stick SM, Wark PA, O'Gara F (2024). Bile acids in lower airways is associated with airway microbiota changes in chronic obstructive pulmonary disease: an observational study. *BMJ Open Respiratory Research.* 11(1):e002552 http://dx.doi.org/10.1136/bmjresp-2024-002552.
   Q1 Journal. Ranked 21st in Pulmonary and Respiratory Medicine as per SJR, Jan 2025. Citations: 0 – Published in December 2024.

2. Frayman KB, Macowan M, **Caparrós-Martín JA**, Ranganathan SC, Marsland B on behalf of SYNERGY CF (2024). The microbial and metabolic landscape of infant Cystic Fibrosis: the gut-lung axis. *European Respiratory Journal.* 63(5):2302290.
   Q1 Journal. Ranked 4th in Pulmonary and Respiratory Medicine, and 50th in Medicine (miscellaneous) as per SJR, Jan 2025. Citations: 4.

3. McGuiness AJ, Stinson LF, Snelson M, Loughman A, Stringer A, Hannan AJ, Cowan CSM, Jama HA, **Caparrós-Martín JA**, West M, Wardill HR on behalf of the AHMRN (2024). From hype to hope in microbiome science. Considerations in conducting robust microbiome science. *Brain Behav Immun*. 115:120-130. doi: 10.1016/j.bbi.2023.09.022.
   Q1 Journal. Ranked 1st in Endocrine and Autonomic Systems, 2nd in Behavioral Neuroscience, and 20th in Immunology as per SJR, Jan 2025. Citations: 7.

4. **Caparrós-Martín JA**, Saladié M, Agudelo-Romero P, Reen FJ, Ware RS, Sly, PD, Stick SM, O'Gara F on behalf of COMBAT Study Group. (2023). Detection of Bile Acids in Bronchoalveolar Lavage Fluid Defines Early Pathobiological Events in Infants with Cystic Fibrosis. *Microbiome*. 11,132. https://doi.org/10.1186/s40168-023-01543-9.
   Q1 Journal. Ranked 6th in Microbiology as per SJR, Jan 2025. Citations: 8.

5. Blake SJ, James J, Ryan FJ, **Caparrós-Martín JA,** Eden GL, Tee YC, Salamon JR, Benson SC, Tumes DJ, Sribnaia A, Stevens NE, Finnie JW, Kobayashi H, White DL, Wesselingh SL, O'Gara F, Lynn MA, Lynn DJ (2021). The immunotoxicity, but not anti-tumor efficacy, of anti-CD40 and anti-CD137 immunotherapies is dependent of the gut microbiota. *Cell Reports Medicine* 2(12):100464.
   Q1 Journal. Ranked 15th in Biochemistry, Genetics and Molecular Biology (miscellaneous) as per SJR, Jan 2025. Citations 23.

RESEARCH SUPPORT IN THE LAST 5 YEARS (Listed in descending order of grant date):

I have a record of grant success through competitive funding schemes (> 6 million AUD) from major funding bodies including the NHMRC and the WA Department of Health, including projects in which I am chief and co-chief investigator.

2025-2028. Western Australia Child Research fund. 599,135 AUD$ for measuring respiratory function at 5 years of age in the participants of the AERIAL study, a longitudinal birth cohort evaluating epithelial factors influencing susceptibility to viral respiratory infections and respiratory morbidity in the first years of life.
Role: Associate Investigator.

2023-2024. Telethon Kids Institute Theme Collaboration Award. 125,000 AUD$ for establishing the biobank from the CIRCA-DIEM study.
Role: Co-Chief Investigator.

2023-2024. Telethon Kids Institute Theme Collaboration Award. 122,795 AUD$ for establishing bioinformatics capacity at Telethon Kids Institute.
Role: Co-Chief Investigator.

2023-2024. Wal-yan Respiratory Research Centre Inspiration Award. 98,483 AUD$ for developing a new laboratory test at-point-of-care.
Role: Co-Chief Investigator.

2020-2025. NHMRC Synergy Grant (APP1183640). 5,000,000 AUD$.
Role: Associate Investigator.

2020-2023. Health Research Board (Ireland). Reference ILP-POR-2019-004. 367,992€.
Role: Co-chief investigator.

2019-2021. Endeavour Project grant. Funded by Endeavour College of Natural Health 21,000 AUD$.
Role: Chief Investigator.

2019-2021. Project grant. Funded by Irish Thoracic Society/Health Research Board. Reference MRCG-2018-16. 115,000 €.
Role: Associate Investigator.

SOFTWARE AND DATABASES:

Agudelo-Romero P, Sharma A, Kicic A, Conradie T, **Caparros-Martin J**, Stick SM (2025). A Nextflow-Based Automated Pipeline for Viral Assembly and Characterisation (EVEREST). Zenodo. https://doi.org/10.5281/zenodo.14963685

Agudelo-Romero P, Sharma A, Kicic A, Conradie T, **Caparros-Martin J**, Stick SM (2024). Database for EVEREST (pipEline for Viral assEmbly and chaRactEriSaTion). (0.0.4b) [Data set]. Zenodo. https://doi.org/10.5281/zenodo.14279485

Agudelo-Romero P, Conradie T, **Caparros-Martin JA**, Stick SM, Hakkaart C, and Sharma A. (2024). Nextflow biowiki. Zenodo. https://doi.org/10.5281/zenodo.13989753

Agudelo-Romero P, Conradie T, Kicic A, **Caparros-Martin JA** and Stick SM. (2024). EVEREST_meta (v0.1.0). Zenodo. https://doi.org/10.5281/zenodo.10487446

Agudelo-Romero, P., Sharma, A., Conradie, T., Kicic, A., Caparros-Martin, J., & Stick, S. (2023). EVEREST-nf (Version 1.0.0) [Computer software]. https://github.com/agudeloromero/everest_nf

REFERENCES:

Former managers:

Professor Stephen Stick
Perth Children Hospital (Perth, Australia)
Stephen.stick@health.wa.gov.au

Professor Fergal O'Gara
University College Cork (Cork, Ireland)
f.ogara@ucc.ie

FULL PUBLICATION LIST:

Peer-reviewed original publications.

45. Montgomery ST, Carr PG, **Caparrós-Martín JA** (2025). Optimisation of DNA extraction from nasal lining fluid to assess the nasal microbiome using third-generation sequencing. *Accepted in American Journal of Respiratory Cell and Molecular Biology.*

44. Agudelo-Romero P*, **Caparrós-Martín JA***, Sharma A, Saladie M, Stick SM, O'Gara F (2024). Virome Assembly Reveals Draft Genomes of Native Pseudomonas Phages isolated from a Paediatric Bronchoalveolar Lavage Sample. *Microbiology Resource Announcement* e0103024. doi: 10.1128/mra.01030-24.
*** Equal contribution**

43. **Caparrós-Martín JA**, Saladié M, Agudelo-Romero P, Nichol KS, Reen FJ, Moodley Y, Stick SM, Wark PA, O'Gara F (2024). Bile acids in lower airways is associated with airway

microbiota changes in chronic obstructive pulmonary disease: an observational study. *BMJ Open Respiratory Research.* 11(1):e002552 *http://dx.doi.org/10.1136/bmjresp-2024-002552*

42. Agudelo-Romero P*, **Caparrós-Martín JA***, Sharma A, Saladie M, Sly PD, Stick SM, O'Gara F (2024). A near-complete genome of the uncultured *Staphylococcus aureus* Phage COMBAT-CF_PAR01, isolated from the lungs of an infant with Cystic Fibrosis. *Microbiology Resource Announcement* 13(12):e0104724. doi: 10.1128/mra.01047-24
*** Equal contribution**

41. Hancock DG, Kicic-Starcevich E, Sondag T, Rivers R, McGee K, Karpievitch YV, D'Vaz N, Agudelo-Romero P, **Caparrós-Martín JA**, Iosifidis T, Kicic A, Stick SM (2024). Real time monitoring of respiratory viral infections in cohort studies using a smartphone app. *iScience* 27(10): 110912.

40. Conradie T, **Caparrós-Martín JA**, Egan S, Kicic A, Kõks S, Stick SM, Agudelo-Romero P (2024). Exploring the complexity of the human respiratory virome through an *in-silico* analysis of shotgun metagenomic data retrieved from public repositories. *Viruses* 16(6):953.

39. Frayman KB, Macowan M, **Caparrós-Martín JA**, Ranganathan SC, Marsland B on behalf of SYNERGY CF (2024). The microbial and metabolic landscape of infant Cystic Fibrosis: the gut-lung axis. *European Respiratory Journal.* 63(5):2302290.

38. Kicic-Starcevich E, Hancock D, Iosifidis T, Agudelo-Romero P, **Caparrós-Martín JA**, Silva D, Turkovic L, Le Souef PN, Bosco A, Martino DJ, Kicic A, Prescott S, Stick SS (2024). Airway epithelium respiratory illnesses and allergy (AERIAL) birth cohort: study protocol. *Frontiers in Allergy* 5:1349741.

37. Caparrós-Martín JA, Maher P, Ward NC, Saladie M, Agudelo-Romero SP, Chan DC, Watts GF, O'Gara F (2024). Analysis of the gut microbiota and related metabolites following PCSK9 inhibition in patients with elevated lipoprotein (a). *Microorganisms.* 12(1), 170.

36. Ahmad AF, **Caparrós-Martín JA**, Gray N, Lodge S, Wist J, Lee S, O'Gara F, Ward NC, Dwivedi G (2024). Gut microbiota and metabolomics profiles in patients with chronic stable angina and acute coronary syndrome. *Physiological Genomics.* 56(1):48-64. doi: 10.1152/physiolgenomics.00072.2023.

35. McGuiness AJ, Stinson LF, Snelson M, Loughman A, Stringer A, Hannan AJ, Cowan CSM, Jama HA, **Caparrós-Martín JA**, West M, Wardill HR on behalf of the AHMRN (2024). From hype to hope in microbiome science. Considerations in conducting robust microbiome science. *Brain Behav Immun*. 115:120-130. doi: 10.1016/j.bbi.2023.09.022.

34. Ahmad AF, **Caparrós-Martín JA**, Gray N, Lodge S, Wist J, Lee S, O'Gara F, Ward NC, Dwivedi G (2023). Insights into the associations between the gut microbiome, its metabolites and heart failure. *Am J Physiol Heart Circ Physiol.* doi: 10.1152/ajpheart.00436.2023

33. Barbeito P, Martin-Morales R, Palencia-Campos A, Cerrolaza J, Rivas-Santos C, Gallego-Colastra L, **Caparrós-Martín JA**, Martin-Bravo C, Martin-Hurtado A, Sanchez-Bellver L, Marfany G, Ruiz-Perez VL, Garcia-Gonzalo FR (2023). EVC-EVC2 complex stability and ciliary targeting are regulated by modification with ubiquitin and SUMO. *Front Cell Dev Biol.* 11:1190258.

32. Malhotra P, Palanisamy R, **Caparrós-Martín JA**, Falasca M (2023). Bile acids and microbiota interplay in pancreatic cancer. *Cancers.* 15(14), 3573.

31. Caparrós-Martín JA, Saladié M, Agudelo-Romero P, Reen FJ, Ware RS, Sly, PD, Stick SM, O'Gara F on behalf of COMBAT Study Group. (2023). Detection of Bile Acids in Bronchoalveolar Lavage Fluid Defines Early Pathobiological Events in Infants with Cystic Fibrosis. *Microbiome.* 11,132.
https://doi.org/10.1186/s40168-023-01543-9

30. Ahmad AF, **Caparrós-Martín JA**, Lee S, O'Gara F, Ward NC, Yeap B, Ballal M, Dwivedi G (2023). Altered gut microbiome and metabolites following bariatric surgery. *Microorganisms* 11(5):1126.

29. Luque J, Mendes I, Gómez B, Morte B, López de Heredia M, Herreras E, Corrochano V, Bueren J, Gallano P, Artuch R, Fillat C, Pérez-Jurado LA, Montoliu L, Carracedo Á, Millán JM, Webb SM, Palau F; **CIBERER Network**, Lapunzina P. (2022). CIBERER: Spanish national network for research on rare diseases: A highly productive collaborative initiative. *Clin Genet* 101(5-6):481-493.

28. Blake SJ, James J, Ryan FJ, **Caparrós-Martín JA,** Eden GL, Tee YC, Salamon JR, Benson SC, Tumes DJ, Sribnaia A, Stevens NE, Finnie JW, Kobayashi H, White DL, Wesselingh SL, O'Gara F, Lynn MA, Lynn DJ (2021). The immunotoxicity, but not anti-tumor efficacy, of anti-CD40 and anti-CD137 immunotherapies is dependent of the gut microbiota. *Cell Reports Medicine* 2(12):100464.

27. Liu Y, Croft KD, **Caparrós-Martín JA,** O'Gara F, Mori TA, Ward NC (2021). Beneficial effects of inorganic nitrate in non-alcoholic fatty liver disease. *Archives of Biochemistry and Biophysics.* 711:109032.

26. Woods DF, Flynn S, **Caparrós-Martín JA,** Stick SM, Reen FJ, O'Gara F (2021). Systems biology and bile acid signaling in microbiome-host interactions in the Cystic Fibrosis lung. *Antibiotics.* 10(77):766.

25. Flynn S*, Reen FJ*, **Caparrós-Martín JA*,** Woods DF, Peplies J, Ranganathan SC, Stick SM, O'Gara F (2020). Bile acid signal molecules associate temporally with respiratory inflammation and microbiome signatures in clinically stable Cystic Fibrosis patients. *Microorganisms.* 8(11):1741.
*** Equal contribution**

24. Saladie M, **Caparrós-Martín JA,** Agudelo-Romero P, Wark PAB, Stick SM, O'Gara F (2020). Microbiomic analysis on low abundant respiratory biomass samples; improved recovery of microbial DNA from bronchoalveolar lavage fluid. *Frontiers in Microbiology.*11:572504.

23. Piceci-Sparascio F, Palencia-Campos A, Soto-Bielicka P, Danzi A, Guida V, Rosati J, **Caparrós-Martín JA**, Torrente I, D'Asdia MC, Versacci P, Briuglia S, Lapunzina P, Tartaglia M, Marino B, Digilio MC, Ruiz-Perez VL, De Luca A. (2020). Common atrium/atrioventricular canal defect and postaxial polydactyly: a mild clinical subtype of Ellis-

van Creveld syndrome caused by hypomorphic mutations in the *EVC* gene. *Human Mutation* 41(12):2087-2093.

22. Caparrós-Martín JA*, Reen FJ*, Flynn S*, Woods DF, Peplies J, Agudelo-Romero SP, Ranganathan SC, Stick SM, O'Gara F (2020). The detection of bile acids in the lungs of paediatric Cystic Fibrosis patients is associated with altered inflammatory patterns. *Diagnostics.* 10(5):282.
*** Equal contribution**

21. Caparrós-Martín JA*, McCarthy-Suarez, Culiañez-Macia FA (2019). Structural features of substrate ambiguity in a HAD phosphosugar phosphatase of Arabidopsis thaliana. *Biology (Basel)*. 8(4). pii: E77.
* Corresponding author

20. Ahmad AF, Dwivedi G, O'Gara F, **Caparrós-Martín JA**, Ward NC (2019). The gut microbiome and atherosclerosis: current knowledge and clinical potential. *AJP-Heart and Circulatory Physiology*. 317(5):H923-H938.

19. Estañ MC, Fernandez-Nuñez E, Zaki MS, Esteban MI, Donkervoort S, Hawkins C, **Caparrós-Martín JA**, Saade DD, Nevado J, Lamuedra A, Largo R, Herrero-Beaumont G, Regadera J, Hernandez-Chico C, Tizzano EF, Martinez-Glez V, Carvajal JJ, Zong R, Nelson D, Otaify GA, Temtamy S, Aglan M, Issa M, Bönnemann CG, Lapunzina P, Yoon G, Ruiz-Perez VL. (2019). Recessive mutations in muscle-specific isoforms of FXR1 cause congenital multi-minicore myopathy. *Nature Communications*. 10(1):797.

18. Tan S, **Caparrós-Martín JA**, Matthews VB, Koch H, O'Gara F, Croft KD, Ward NC (2018). Isoquercetin and inulin synergistically modulate the gut microbiome to prevent development of the metabolic syndrome in mice fed a high fat diet. *Scientific Reports*. 8(1), 10100.

17. Phelan JP, Reen FJ **Caparrós-Martín JA**, O'Connor R, O'Gara F (2017). Rethinking the bile acid/gut microbiome axis in cancer. *Oncotarget.* 8(70):115736-115747.

16. Caparrós-Martín JA*, Lareu RR*, Ramsay JP, Peplies J, Reen J, Koch H, Ward NC, Croft KD, Newsholme P, Hughes JD, O'Gara F (2017). Statin therapy causes gut dysbiosis in mice through a PXR-dependent mechanism. *Microbiome.* 5(1):95.

15. Caparrós-Martín JA, Aglan M, Temtamy S, Otaify GA, Valencia M, Nevado J, Vallespin E, Del Pozo A, Prior de Castro C, Calatrava-Ferreras L, Gutierrez P, Bueno AM, Sagastizabal B, Guillen-Navarro E, Ballesta-Martinez M, Gonzalez V, Basaran SY, Buyukoglan R, Sarikepe B, Espinoza-Vadez C, Cammarata-Scalisi F, Martinez-Glez V, Heath KE, Lapunzina P, Ruiz-Perez VL. (2017). Molecular spectrum and differential diagonosis in patients referred with sporadic or autosomal recessive osteogenesis imperfecta. *Molecular Genetics and Genomic Medicine.* 5(1):28-39.

14. Caparrós-Martín JA, De Luca A, Cartault F, Aglan M, Temtamy S, Otaify GA, Mehrez M, Valencia M, Vázquez L, Alessandri JL, Nevado J, Rueda-Arenas I, Heath KE, Digilio MC, Dallapiccola B, Goodship JA, Mill P, Lapunzina P, Ruiz-Perez VL. (2015). Specific variants in WDR35 cause a distinctive form of Ellis-van Creveld syndrome by disrupting the

recruitment of the EvC complex and SMO into the cilium. *Hum Mol Genet.* 15;24(14):4126-37.

13. Caparrós-Martín JA, McCarthy-Suárez I, Culiáñez-Macià FA. (2014). The kinetic analysis of the substrate specificity of motif 5 in a HAD hydrolase-type phosphosugar phosphatase of Arabidopsis thaliana. *Planta*. 240(3):479-87.

12. Valencia M, **Caparrós-Martin JA,** Sirerol-Piquer MS, García-Verdugo JM, Martínez-Glez V, Lapunzina P, Temtamy S, Aglan M, Lund AM, Nikkels PG, Ruiz-Perez VL, Ostergaard E. (2014). Report of a newly indentified patient with mutations in BMP1 and underlying pathogenetic aspects. *Am J Med Genet A*. 164A(5):1143-50.

11. Caparrós-Martin JA, Valencia M, Pulido V, Martínez-Glez V, Rueda-Arenas I, Amr K, Farra C, Lapunzina P, Ruiz-Perez VL, Temtamy S, Aglan M. (2013). Clinical and molecular analysis in families with autosomal recessive osteogenesis imperfecta identifies mutations in five genes and suggests genotype-phenotype correlations. *Am J Med Genet A*. 161A(6):1354-69.

10. Keupp K, Beleggia F, Kayserili H, Barnes AM, Steiner M, Semler O, Fischer B, Yigit G, Janda CY, Becker J, Breer S, Altunoglu U, Grünhagen J, Krawitz P, Hecht J, Schinke T, Makareeva E, Lausch E, Cankaya T, **Caparrós-Martín JA**, Lapunzina P, Temtamy S, Aglan M, Zabel B, Eysel P, Koerber F, Leikin S, Garcia KC, Netzer C, Schönau E, Ruiz-Perez VL, Mundlos S, Amling M, Kornak U, Marini J, Wollnik B. (2013). Mutations in WNT1 cause different forms of bone fragility. *Am J Hum Genet.* 4;92(4):565-74.
Highlighted by Faculty of 1000 as a very good report. (http://f1000.com/prime/717989964).

9. Caparrós-Martín JA, McCarthy-Suárez I, Culiáñez-Macià FA. (2013). HAD hydrolase function unveiled by substrate screening: enzymatic characterization of Arabidopsis thaliana subclass I phosphosugar phosphatase AtSgpp. *Planta*. 237(4):943-54.

8. Caparrós-Martín JA*, Valencia M*, Reytor E*, Pacheco M, Fernandez M, Perez-Aytes A, Gean E, Lapunzina P, Peters H, Goodship JA, Ruiz-Perez VL. (2013). The ciliary Evc/Evc2 complex interacts with Smo and controls Hedgehog pathway activity in chondrocytes by regulating Sufu/Gli3 dissociation and Gli3 trafficking in primary cilia. *Hum Mol Genet.* 1;22(1):124-39.
*** Equal contribution**

7. Puig-Hervás MT, Temtamy S, Aglan M, Valencia M, Martínez-Glez V, Ballesta-Martínez MJ, López-González V, Ashour AM, Amr K, Pulido V, Guillén-Navarro E, Lapunzina P, **Caparrós-Martín JA**, Ruiz-Perez VL. (2012). Mutations in PLOD2 cause autosomal-recessive connective tissue disorders within the Bruck syndrome-osteogenesis imperfecta phenotypic spectrum. *Hum Mutat.* 33(10):1444-9.

6. Martínez-Glez V*, Valencia M*, **Caparrós-Martín JA***, Aglan M*, Temtamy S*, Tenorio J, Pulido V, Lindert U, Rohrbach M, Eyre D, Giunta C, Lapunzina P, Ruiz-Perez VL. (2012). Identification of a mutation causing deficient BMP1/mTLD proteolytic activity in autosomal recessive osteogenesis imperfecta. *Hum Mutat.* 33(2):343-50.
Highlighted by Faculty of 1000 as a good report. (http://f1000.com/prime/13396964).
*** Equal contribution**

5. Pacheco M, Valencia M, **Caparrós-Martín JA**, Mulero F, Goodship JA, Ruiz-Perez VL. (2012). Evc works in chondrocytes and osteoblasts to regulate multiple aspects of growth plate development in the appendicular skeleton and cranial base. *Bone*. 50(1):28-41.

4. Romá-Mateo C, Sacristán-Reviriego A, Beresford NJ, **Caparrós-Martín JA**, Culiáñez-Macià FA, Martín H, Molina M, Tabernero L, Pulido R. (2011). Phylogenetic and genetic linkage between novel atypical dual-specificity phosphatases from non-metazoan organisms. *Mol Genet Genomics.* 285(4):341-54.

3. Lapunzina P*, Aglan M*, Temtamy S*, **Caparrós-Martín JA**, Valencia M, Letón R, Martínez-Glez V, Elhossini R, Amr K, Vilaboa N, Ruiz-Perez VL. (2010). Identification of a frameshift mutation in Osterix in a patient with recessive osteogenesis imperfecta. *Am J Hum Genet.* 9;87(1):110-4.
Highlighted by Faculty of 1000 as a very good report (http://f1000.com/prime/4015963).
*** Equal contribution**

2. Kupke T, **Caparrós-Martín JA,** Malquichagua Salazar KJ, Culiáñez-Macià FA. (2009). Biochemical and physiological characterization of Arabidopsis thaliana AtCoAse: a Nudix CoA hydrolyzing protein that improves plant development. *Physiol Plant.* 135(4):365-78.

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Publications under review.

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