

## **CURRICULUM VITAE**

**Ferran Casals**

### **EDUCATION / TRAINING**

2012-2014: Incorporation grant, Universitat Pompeu Fabra.  
2008-2011: Postdoc at Université de Montréal.  
2004-2008: Postdoc at Universitat Pompeu Fabra.  
2003: PhD in Biology (Universitat Autònoma de Barcelona).  
1999: Master in Genetics (Universitat Autònoma de Barcelona).  
1997: B. Sc. in Biology (Universitat Autònoma de Barcelona).

### **EMPLOYMENT**

06/2014 – present: Head of the Genomics Core Facility, UPF. Coordinator of UPF the Core Facilities. Researcher and teacher at the Experimental and Health Sciences Department.  
01/2012 – 05/2014: Beatriu de Pinós contract. Institut de Biologia Evolutiva UPF – CSIC, Barcelona, Spain.  
09/2008 – 12/2011: Post-doc at the Centre de Recherche du CHU Sainte-Justine, Université de Montréal, Montréal, Québec, Canada.  
05/2003 – present: Founder and scientific advisory of “Nusos. Activitats Científiques i Culturals, SCCL”, devoted to science pedagogy and divulgation.  
05/2004 – 08/2008: Post-doc at the Evolutionary Biology Unit (Universitat Pompeu Fabra) leaded by Jaume Bertranpetit.  
09/2003–05/2004: Collaborator teacher at the Genetics Unit (Universitat Autònoma de Barcelona). (*Genetics*, and *Molecular Genetics* for Biology students; *Environmental Genetics* and *Experimental Procedures* for Environmental Sciences students).  
01/2002–08/2003. Associate teacher at the Genetics Unit (Universitat Autònoma de Barcelona).(*Genetics*, and *Molecular Genetics* for Biology students; *Environmental Genetics* and *Experimental Procedures* for Environmental Sciences students).

## PUBLICATIONS

- Kuderna L, Solís-Moruno M, Batlle-Masó L, Julià E, Lizano E, Anglada A, Ramírez E, Bote A, Tormo M, Marquès-Bonet T, Fornas O, Casals F. Flow sorting enrichment and nanopore sequencing of chromosome 1 from a Chinese individual. *Frontiers in Genetics*, 2020, 10, 1315.
- Batlle-Masó L, Mensa-Vilaró A, Solís-Moruno M, Marquès-Bonet T, Aróstegui JI, Casals F. Genetic diagnosis of autoinflammatory disease patients using clinical exome sequencing. *European Journal of Medical Genetics*, 2020, 63(5), 103920.
- Costa D, Bonet N, Solé A, González de Aledo-Castillo JM, Sabidó E, Casals F, Rovira C, Nadal A, Marin JL, Cobo T, Castelo R. Genome-wide postnatal changes in immunity following fetal inflammatory response. 2020. *The FEBS journal*, in press.
- Martín-Nalda A, Fortuny C, Rey L, Bunney TD, Alsina L, Esteve-Solé A, Bull D, Anton MC, Basagaña M, Casals F, Deyá A, García-Prat M, Gimeno R, Juan M, Martínez-Banaclocha H, Martínez-García JJ, Mensa-Vilaró A, Rabionet R, Martín-Begue N, Rudilla F, Yagüe J, Estivill X, García-Patos V, Pujol RM, Soler-Palacín P, Katan M, Pelegrín P, Colobran R, Vicente A, Arostegui JI. Severe Autoinflammatory Manifestations and Antibody Deficiency Due to Novel Hypermorphic PLCG2 Mutations. *Journal of Clinical Immunology*, 2020, 40 (7), 987-1000.
- Harding T, Milot E, Moreau C, Lefebvre JF, Bournival JS, Vézina H, Laprise C, Lalueza-Fox C, Anglada R, Loewen B, Casals F, Ribot I, Labuda D. Historical human remains identification through maternal and paternal genetic signatures in a founder population with extensive genealogical record. *American Journal of Physical Anthropology*, 2020, 171(4), 645-658.
- Viñas-Gimenez L, Padilla N, Batlle L, Casals F, Rivière JG, Martínez-Gallo M, de la Cruz X, Colobran R. HLHdb: A comprehensive database on the molecular basis of primary hemophagocytic lymphohistiocytosis. *Frontiers in Immunology*, 2020, 11, 107.
- Barrio PA, García O, Phillips C, Prieto L, Gusmão L, Fernández C, Casals F, Freitas JM, González-Albo MC, Martín P, Mosquera A, Navarro-Vera I, Paredes M, Pérez JA, Pinzón A, Rasal R, Ruiz-Ramírez J, Trindade BR, Alonso A. The first GHEP-ISFG collaborative exercise on forensic applications of massively parallel sequencing. *Forensic Sci Int Genet*. 2020 Sep 18;49:102391.
- Mensa-Vilaró A, Bravo García-Morato M, de la Calle-Martin O, Franco-Jarava C, Martínez-Saavedra MT, González-Granado LI, González-Roca E, Fuster JL, Alsina L, Mutchinick OM, Balderrama-Rodríguez A, Ramos E, Modesto C, Mesa-Del-Castillo P, Ortego-Centeno N, Clemente D, Souto A, Palmou N, Remesal A, Leslie KS, Gómez de la Fuente E, Yadira Bravo Gallego L, Campistol JM, Dhouib NG, Bejaoui M, Dutra LA, Terreri MT, Mosquera C, González T, Cañellas J, García-Ruiz de Morales JM, Wouters CH, Bosque MT, Cham WT, Jiménez-Treviño S, de Inocencio J, Bloomfield M, Pérez de Diego R, Martínez-Pomar N, Rodríguez-Pena R, González-Santesteban C, Soler-Palacín P, Casals F, Yagüe J, Allende LM, Rodríguez-Gallego JC, Colobran R, Martínez-Martínez L, López-Granados E, Aróstegui JI. 2019. Unexpected relevant role of gene mosaicism in patients with primary immunodeficiency diseases. *J Allergy Clin Immunol*. 2019 Jan;143(1):359-368.

- Palomo-Díez S, Gomes C, López-Parra AM, Baeza-Richer C, Cuscó I, Raffone C, García-Arumí E, Vinuesa-Espinosa D, Santos C, Montes N, Rasal R, Escala O, Cuellar J, Subirá E, Casals F, Malgosa A, Tizzano E, Tartera E, Domenech G, Arroyo-Pardo E. Genetic identification of Spanish civil war victims. The state of the art in Catalonia (Northeastern Spain). *Forensic Science International: Genetics Supplement Series*, Volume 7, Issue 1, December 2019, 419-421.
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- De Valles-Ibañez G, Esteve-Solé A, Piquer M, González-Navarro A, Hernández-Rodríguez J, Laayouni H, González-Roca E, Plaza-Martin AM, Deyà-Martínez A, Martín-Nalda A, Martínez-Gallo M, García-Prat M, del Pino L, Cuscó I, Codina-Solà M, Marquès-Bonet T, Bosch E, López-Granados E, Aróstegui JI, Soler-Palacín P, Colobrán R, Yagüe J, Alsina L, Juan M, and Casals F. Evaluating the genetics of common variable immunodeficiency: monogenetic model and beyond. 2018. In press. *Frontiers in Immunology*.
- Mondal M, Casals F, Majumder PP, Bertranpetit J. Reply to 'No evidence for unknown archaic ancestry in South Asia'. *Nat Genet*. 2018 Dec;50(12):1637-1639.
- Schwab et al. 2018. Phenotype, penetrance, and treatment of 133 CTLA-4-insufficient individuals. *J Allergy Clin Immunol*. 2018 Dec;142(6):1932-1946.
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- Mondal M\*, Casals F\* (\* First co-authorship), Xu T, Dall'Olio GM, Pybus M, Netea MG, Comas D, Laayouni H, Li Q, Majumder PP, Bertranpetit J. 2016. Genomic analysis of Andamanese provides insights into ancient human migration into Asia and adaptation. *Nat Genet*. 48(9):1066-70..
- de Valles-Ibáñez G, Hernandez-Rodriguez J, Prado-Martinez J, Luisi P, Marquès-Bonet T, Casals F. 2016. Genetic Load of Loss-of-Function Polymorphic Variants in Great Apes. *Genome Biol Evol*. 26;8(3):871-7.
- Spataro N, Calafell F, Cervera-Carles L, Casals F, Pagonabarraga J, Pascual-Sedano B, Campolongo A, Kulisevsky J, Lleó A, Navarro A, Clarimón J, Bosch E. 2015. Mendelian genes for Parkinson's disease contribute to the sporadic forms of the disease. *Hum Mol Genet*. 1;24(7):2023-34.
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sequencing identifies mutations in the gene TTC7A in French-Canadian cases with hereditary multiple intestinal atresia. *Journal of Medical Genetics* 50(5):324-9.

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- F Casals, J Bertranpetit. 2012. Human genetic variation, shared and private. *Science* 337(6090), 39 - 40. 2012.
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Bertranpetit. 2011. Similarity in recombination rate estimates highly correlates with genetic differentiation in humans. *PLoS One* Mar 28;6(3):e17913.

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### **Book chapters**

- Elena Bosch and Ferran Casals. 2016. Next Generation Sequencing for Rare Diseases. *Genome-Wide Association Studies: From Polymorphism to Personalized Medicine*, Cambridge University Press.
- Ferran Casals and Elena Bosch. 2016. Next Generation Sequencing for Complex Disorders. *Genome-Wide Association Studies: From Polymorphism to Personalized Medicine*, Cambridge University Press.



**Principal Investigator/Coordinator in FUNDED PROJECTS**

PROJECT TITLE: New Experimental and Methodological Approaches to the Study of Autoinflammatory Diseases.

REFERENCE: RTI2018-096824-B-C22.

FINANCING ENTITY: Ministerio de Ciencia, Innovación y Universidades, Plan Estatal, Spain.

DURATION: 2019 – 2021.

PRINCIPAL RESEARCHER: Ferran Casals, Universitat Pompeu Fabra.

ROLE (Ferran Casals): Principal investigator.

TOTAL AMOUNT: 193,600 €.

PROJECT TITLE: Genetic mechanisms of primary immunodeficiencies: identification and contribution of germline and somatic mutations.

REFERENCE: SAF2015-68472-C2-2-R.

FINANCING ENTITY: Ministerio de Economía y Competitividad, Plan Nacional, Spain.

DURATION: 2016 – 2018.

PRINCIPAL RESEARCHER: Ferran Casals, Universitat Pompeu Fabra.

ROLE (Ferran Casals): Principal investigator.

TOTAL AMOUNT: 108,900 €.

PROJECT TITLE: Development of a population genetics database from El Salvador.

FINANCING ENTITY: UPF Solidaria.

DURATION: 2018.

PRINCIPAL RESEARCHER: Ferran Casals, Universitat Pompeu Fabra.

ROLE (Ferran Casals): Principal investigator.

TOTAL AMOUNT: 3,000 €.

PROJECT TITLE: Medical Genomics of Common Variable Immunodeficiency (CVID).

REFERENCE: SAF2012-35025.

FINANCING ENTITY: Ministerio de Economía y Competitividad, Plan Nacional, Spain

DURATION: 2013 – 2015.

PRINCIPAL RESEARCHER: Ferran Casals, Universitat Pompeu Fabra.

ROLE (Ferran Casals): Principal Investigator.

TOTAL AMOUNT: 106,000 €.

PROJECT TITLE: Application of next-generation sequencing technologies to the study of archaeological sites in Catalonia.

FINANCING ENTITY: Biological Sciences Section, Institut d'Estudis Catalans.

DURATION: 2017.

PRINCIPAL RESEARCHER: Jaume Bertranpetit, Universitat Pompeu Fabra.

ROLE (Ferran Casals): Coordinator.

TOTAL AMOUNT: 6,000 €.

PROJECT TITLE: Population genetic and functional analyses of maintenance of DNA sequence variability in response to infectious agents (human innate immune system and other responses). REFERENCE: PRI-PIBIN-2011-0942.

DURATION: 2011 – 2014.

FINANCING ENTITY: Ministerio de Economía y Competitividad. Subprograma de fomento de la cooperación científica internacional

PRINCIPAL RESEARCHER: Jaume Bertranpetit, Universitat Pompeu Fabra.

ROLE (Ferran Casals): Co-Principal Investigator.

TOTAL AMOUNT: 96,000 €.

PROJECT TITLE: Innovation in Teaching: Implementation of Behavioural Genetics practical sessions for Zoology.

REFERENCE: PlaQUID 2012.

DURATION: 2013.

FINANCING ENTITY: Universitat Pompeu Fabra.

PRINCIPAL RESEARCHER: Ferran Casals.

ROLE (Ferran Casals): Principal investigator.

TOTAL AMOUNT: 1,500 €.

PROJECT TITLE: Next Generation sequencing approaches for functional characterization of undefined primary immunodeficiency. REFERENCE: CIHR 120299. DURATION: 2012 – 2013.

FINANCING ENTITY: Canadian Institute of Health and Research.

PRINCIPAL RESEARCHER: Philip Awadalla, Université de Montréal.

ROLE (Ferran Casals): Co-Applicant.

TOTAL AMOUNT: 77,509.13 €.

