MINISTERIO DE CIENCIA E INNOVACIÓN

Cofinanciado por la Unión Europea

AGENCIA ESTATAL DE INVESTIGACIÓN

CURRICULUM VITAE ABREVIADO (CVA)

***IMPORTANT** – The Curriculum Vitae cannot exceed 4 pages. Instructions to fill this document are available in the website.*

Part A. PERSONAL INFORMATION

First name	Javier		
Family name	Martín Ibáñez		
Gender (*)	Male	Birth date	30/08/1959
Social Security, Passport, ID number	24157941Y		
e-mail	javiermartin@ipb.csic.es	URL: www.ipb.csic.es	
Open Researcher and Contributor ID (ORCID) (*)		0000-0002-2202-0622	
Researcher ID		B-8141-2008	
Scopus		211129010550	

(*) Mandatory

A.1. Current position

Position	Full Profesor, CSIC			
Initial date	2010			
Institution	Consejo Superior de Investigaciones Científicas, CSIC			
Department/Center	Immunology	Instituto de Parasitología y Biomedicina López-Neyra, IPBLN		
Country	Spain		Teleph. number	959181669 636760043
Key words	Autoimmunity, Human Genetics, Genomics, Systemic Sclerosis			

A.2. Previous positions (research activity interuptions, indicate total months)

Period	Position/Institution/Country/Interruption cause
1985-1987	Predoctoral FIS, ISCIII / Dpt Inmunología, Hosp Virg de las Nieves, Spain
1987-1990	Postdoctoral Fellow, Dept of Immunology, Mayo Clinic, USA
1990-2010	Colaborador / Investigador Científico, IPBLN, CSIC, Spain
2010-2012	Scientific Director, Human DNA Bank of Andalusia, Consejeria de Salud, Junta de Andalucía, Spain
2014-2022	Deputy Director, IPBLN, CSIC, Spain

A.3. Education

PhD, Licensed, Graduate	University/Country	Year
MD	University of Granada / Spain	1982
PhD	University of Granada / Spain	1987

Part B. CV SUMMARY *(max. 5000 characters, including spaces)*

My research work as an independent scientist has been focused on the genetic basis of autoimmune diseases, mainly those with a rheumatologic component such as rheumatoid arthritis (RA), systemic lupus erythematosus (SLE), and more recently systemic sclerosis (SSc) and giant cell arteritis (GCA). Our main objective is to identify genetic factors that influence susceptibility and/or severity to these pathologies, which on one hand will help us to know the pathophysiological mechanisms that underlie the development of these diseases, and on the other can allow us to develop new and more specific diagnostic tools and therapeutic targets.

Our group has carried out numerous genetic studies in RA, SLE and GCA, noting the participation in large-scale genetic studies, GWAS o Immunochip, with international consortia

that have yielded a remarkable progress in the identification of risk genes for autoimmune diseases, and which results were published in prestigious journals such as Nature (2), Nat Genet (8), Nat Commun (3) or Am J Hum Genet (8), Plos Genet (4) or Hum Mol Genet (8),

However, our most noteworthy scientific achievements come from the work on SSc genetics that have positioned our group as the international leader in the genetic/genomics of SSc. Remarkably, we carried out the first genome wide association study - GWAS - in SSc (*Radstake et al, Nat Genet, 2010*) that was co-directed by our group. Subsequently we performed follow-up (*Bossini-Castillo et al., Ann Rheum Dis 2011, Hum Mol Genet. 2012*; *Martin JE et al., Hum Mol Genet. 2012*; *Lopez-Isac et al, Arthritis Rheum, 2016; Ann Rheum Dis 2017*) and fine-mapping studies in SSc (*Gorlova et al., PLos Genet, 2011*; *Mayes et al. Am J Hum Genet, 2014*) identifying new genetic risk loci for SSc.

In addition, we have performed meta-GWAS in autoimmunity (*Martin JE et al., Hum Mol Genet. 2013*; *Marquez et al., Genome Med, 2017;Acosta-Herrera et al, Ann Rheum Dis, 2018*), that have provide to be useful tools to identify genes that may serve as targets for drug discovery and/or repositioning in these diseases. Remarkably, we recently conducted the largest GWAS in SSc (*Lopez-Isac et al, Nat Commun, 2019*) identifying 27 loci independently associated with SSc and the most relevant molecular pathways implicated in the disease. Furthermore, data mining analysis of SSc GWAS data allowed us a comprehensive analysis of the MHC region in SSc identifying differential HLA associations by clinical and serological subtypes with possible application as biomarkers of disease severity and progression (*Acosta-Herrera et al, Ann Rheum Dis, 2021*). In addition, based on GWAS data, we successfully implemented a genomic risk score (GRS) in SSc with potential application in the clinic (*Bossini-Castillo et al., Ann Rheum Dis, 2021*). Recently we described that copy number (CN) polymorphisms of complement C4 are implicated in the sex-biased vulnerability observed in SSc (*Kerick et al, NPJ Genome Med, 2022*).

On the other hand, we have contributed to the better understanding of the pathogenesis of SSc by investigating the differential whole blood gene expression occurring in SSc patients through a genome-wide transcriptome analysis (*Beretta et al., Ann Rheum Dis, 2020*) and by the identification of the genetic variants that affect gene expression (eQTLs) in SSc (*Kerick et al., Arthritis Rheum, 2021*). In a very recent study, we used promoter capture HiC (pCHi-C) in two of the most relevant cell types in SSc pathogenesis, CD4+ T cells and CD14+ monocytes from SSc patients and healthy controls, identifying new target genes and confirming others for SSc GWAS loci in these two cell types (*Gonzalez-Serna et al, Arthritis Rheum, 2022*, Oct 25, Online ahead of print).

Our medium-term research is aimed at deciphering the functional consequences of genetic variants associated to SSC through the application of new genomic, epigenomic and transcriptomic techniques.

H-index: ISI/ Scopus=73 ; Google Scholar=89 / i10-index= 485
Total citations: Scopus= 24312; Google Scholar= 36333
Last five years Google Scholar: H-index= 53, i10-index=279, citations= 14653
http://www.ipb.csic.es
https://orcid.org/0000-0002-2202-0622
http://scholar.google.com/citations?user=j-lW-IEAAAAJ
https://www.scopus.com/authid/detail.uri?authorId=8157312000

Part C. RELEVANT MERITS

C.1. Publications

C1a- Publications as senior author

Most relevant publications close related to the project

1. Acosta-Herrera M, Kerick M, Lopéz-Isac E, et al., Martin J (23/23). Comprehensive analysis of the major histocompatibility complex in systemic sclerosis identifies differential HLA associations by clinical and serological subtypes. ***Ann Rheum Dis.*** 2021;80:1040–1047. doi: 10.1136/annrheumdis-2018-214127. IF:19.1; Citations = 14
2. Bossini-Castillo L, Villanueva-Martin G, Kerick M et al., and, Martin J (23/23). Genomic Risk Score impact on susceptibility to systemic sclerosis. ***Ann Rheum Dis***. 2021;80:118-127. doi: 10.1136/annrheumdis-2020-218558. IF:19.1; Citations =11
3. Kerick M, González-Serna D, Carnero-Montoro E, et al, and Martin J (15/15). eQTL analysis in systemic sclerosis identifies new candidate genes associated with multiple

aspects of disease pathology. ***Arthritis Rheumatol*** 2021;73:1288–1300. doi: 10.1002/art.41657; IF: 9.586; Citations = 6

4. Beretta L, Barturen G, Vigone B, et al., and Martin J (19/19). Genome-wide whole blood transcriptome profiling in a large European cohort of systemic sclerosis patients. ***Ann Rheum Dis*** 2020;79:1218-1226. doi:10.1136/annrheumdis-2020-217116 IF= 19.1; Citations = 16
5. López-Isac E, Acosta-Herrera M, Kerick M, et al., and Martin J (44/44). GWAS for systemic sclerosis identifies multiple risk loci and highlights fibrotic and vasculopathy pathways. ***Nat Commun*** 2019 Oct 31;10(1):4955. doi: 10.1038/s41467-019-12760-y. IF= 12.12; Citations = 58. *Comments in: Clarke, J. New genetic risk loci found for SSc. Nat Rev Rheumatol 16, 4 (2020) doi:10.1038/s41584-019-0342-3.*
6. Acosta-Herrera M, Kerick M, González-Serna D et al., and Martin J (14/14). Genome-wide meta-analysis reveals shared new loci in systemic seropositive rheumatic diseases. ***Ann Rheum Dis*** 2019 Mar;78(3):311-319. doi: 10.1136/annrheumdis-2018-214127. IF=16.1; Citations = 56. *Comments in: Yamamoto K, Okada Y. Shared genetic factors and their causality in autoimmune diseases. Ann Rheum Dis. 2019 Nov;78(11):1449-1451*
7. Márquez A, Kerick M, Zhernakova A et al., and Martín J (19/19). Meta-analysis of Immunochip data of four autoimmune diseases reveals novel single-disease and cross-phenotype associations. ***Genome Med*** *2018; 20;10(1):97.* doi: 10.1186/s13073-018-0604-8 IF: 10.95; Citations = 52
8. Mayes MD, Bossini-Castillo L, Gorlova O, et al., Martin J (76/76). Immunochip analysis identifies multiple susceptibility loci for systemic sclerosis. ***Am J Hum Genet***. 2014;94:47-61. doi: 10.1016/j.ajhg.2013.12.002. IF= 10.93; Citations= 152
9. Gorlova O, Martin JE, Rueda B, et al. Martin J (70/70). Identification of Novel Genetic Markers Associated with Clinical Phenotypes of Systemic Sclerosis through a Genome-Wide Association Strategy. ***PLoS Genet.*** *2011 Jul;7(7):e1002178.* doi: 10.1371/journal.pgen.1002178. IF= 8.69 ; Citations = 183
10. Radstake TRDJ*, Gorlova O*, Rueda B*, et al., Koeleman BPC*, Martín J* , Mayes MD* , (62/63), Genome-wide association study of systemic sclerosis identifies CD247 as a new susceptibility locus. ***Nat Genet*** 2010, 42:426-429. **these authors contributed equally to this work.* doi: 10.1038/ng.565. IF= 36.37 ; Citations= 311

C1b- Collaborative Publications (5 most relevant)

1. Ishigaki K, Sakaue S, Terao C, et al., Raychaudhuri S. (88/91) Multi-ancestry genome-wide association analyses identify novel genetic mechanisms in rheumatoid arthritis. ***Nat Genet.*** 2022 Nov;54(11):1640-1651.
2. Westra HJ, Martínez-Bonet M, Onengut-Gumuscu S, et al., Raychaudhuri S (8/19). Fine-mapping and functional studies highlight potential causal variants for rheumatoid arthritis and type 1 diabetes. ***Nat Genet*** *2018 Oct;50(10):1366-1374.*
3. Bentham J, Morris DL, Cunninghame Graham DS, et al., Vyse TJ. (7/17) Genetic association analyses implicate aberrant regulation of innate and adaptive immunity genes in the pathogenesis of systemic lupus erythematosus. ***Nat Genet.*** 2015; 47: 1457-1464.
4. Lenz TL, Deutsch AJ, Han B, et al., Raychaudhuri S (18/37) Widespread non-additive and interaction effects within HLA loci modulate the risk of autoimmune diseases. ***Nat Genet****.* 2015; 47:1085-1090.
5. Okada Y, Wu D, Trynka G, et al., Plenge RM (55/95). Genetics of rheumatoid arthritis contributes to biology and drug discovery. ***Nature*****.** 2014 506:376-81

C.2. Congress.

5 most relevant invited lectures related to the project

1.- *Studying the genetic basis of autoimmune diseases*. 16th International Workshop on Scleroderma Research, 27th-31th July, 2019, St John's College, Cambridge, UK

2.- *Immunogenetics of systemic sclerosis: Defining heridability, functional variants and shared-autoimmunity pathways*. 15th International Workshop on Scleroderma Research, August, 5-9, 2017, Pittsburgh, PN. USA.

3.- *Genomics in Scleroderma.* 14th International Workshop on Scleroderma Research. August 1-5, 2015. St John's College, Cambridge, UK,

4.- *Genetic of Systemic Sclerosis: a roadmap for a complex disease*. 4EME Seminarie de la Fedération de Médicine Translationnelle de Strasbourg –FMTS-, 3 Diciembre, 2014 Strasbourg, France.
5.- *Genetic of Complex Diseases.* 12th International Workshop on Scleroderma Research - 23-27 July 2011 - Trinity College, Cambridge UK.

C.3. Research projects.
1.- Project reference: RTI2018-101332-B-I00. Funding entity: Ministry of Science, Innovation and Universities. Title: *Deciphering the molecular basis of systemic sclerosis*. PI: Javier Martin Ibáñez. Duration 1/1/2019-31/12/2022. Financing received: 350,000 €.
2.- Project reference RD21/0002/0039. Funding entity: Red de Investigación Cooperativa Orientadas a Resultados en Salud (RICORS). Carlos III Health Institute. Title: *Research Network on Inflammatory Diseases (REI)*. PI: Javier Martín Ibáñez. Duration: 2022-2024. Financing received: 109.403,80 €
3.- Project reference 339/C/2020. Fundació la Marató de TV3. Title: *Design of an integrative patients stratification approach for the systemic sclerosis management*. Coordinator: Javier Narvaez. PI subproject: Javier Martín Ibáñez. Duration: 2021-2024. Financing: 379.020,52 €
4.- Project reference P18-RT-4442. Consejería de Transformación Económica, Industria, Conocimiento y Universidades, Junta de Andalucía, Título: *Estudio de las bases moleculares de la esclerosis sistémica mediante secuenciación del transcriptoma de linfocitos T y fibroblastos a nivel de una única célula.* PI: Javier Martín Ibáñez. Duration: 2020-2022. Financing received: 140.352,00 €
5.- Project reference: No 831,434. Project funded by Innovative Medicines Initiative (IMI) Program of the European Commission and the European Federation of Pharmaceutical Industries and Associations (EFPIA). Title: *Taxonomy, Treatment, Targets and Remission :Identification of the Molecular Mechanisms of non-response to Treatments, Relapses and Remission in Autoimmune, Inflammatory, and Allergic Conditions (3TR)*. Coordinator: Marta Alarcón-Riquelme. PI: Javier Martín. Duration: 2019-2025. Financing received to the group: 285,000 €.
6.- Project reference: H2020- Marie Skłodowska-Curie Actions-Innovative Training Networks-European training networks (MSCA- ITN-ETN) Title: *Health Data Linkage for Clinical Benefit* (HELICAL). Funding entity: European Union. Coordiantor: Mark Little. PI: Javier Martín. Duration: 2019-2022. Financing received to the group: 265,000 €
7.- Funding entity: CYTED: Programa Iberoamericano de Ciencia y Tecnología para el Desarrollo . Title: *Red Iberoamericana de Medicina Genómica en la Enfermedad de Chagas - RIMGECH-*.Coordinador: Javier Martín Ibáñez. Duration: 2017-2020. Financing received: 150.000 €.
8.- Project reference: SAF2015-66761-P. Funding entity: Ministry of Science, Innovation and Universities. Título: *Molecular Basis of Systemic Sclerosis: integrating genomics and trnascriptomics.* PI: Javier Martín. Duration: 2016-2019. Financing received: 296.450 €.
9.- Project reference: RD16/0012/0013. Funding entity: RETIC Program (Thematic Networks of Cooperative Research). Carlos III Health Institute Title: *Research Network on Inflammation and Rheumatic Diseases (RIER)*. Principal investigator: Javier Martín Ibáñez.; Duration 1/1/2017 -31/12/2021; Financing received: 260,000 €.
10.- Project reference: nº 115,565. Funding entity: Innovative Medicines Initiative (IMI) Program of the European Commission and the European Federation of Pharmaceutical Industries and Associations (EFPIA) Título: *Molecular Reclassification to Find Clinically Useful Biomarkers for Systemic Autoimmune Diseases (PRECISESAD*). Coordinatorr: Marta Alarcón-Riquelme. WP leader: Javier Martín Ibáñez. Duration: 2014-2019. Financing received to the group: 825.000 euros.