Fecha del CVA	10/01/2019

Parte A. DATOS PERSONALES

Nombre y Apellidos	Marta Eugenia Alarcón Riquelme		
NIE		Edad	
Núm. identificación de investigador	Researcher ID		
	Scopus Author ID		
	Código ORCID		

A.1. Situación profesional actual

Organismo	FUNDACION PUBLICA ANDALUZA PROGRESO Y SALUD		
Dpto. / Centro			
Dirección			
Teléfono		Correo electrónico	luisa.pinel@genyo.es
Categoría profesiona	Cordinadora del Área de Medicina Genomica	Fecha inicio	2015
Espec. cód. UNESCO			
Palabras clave			

A.2. Formación académica (titulo, institución, fecha)

Licenciatura/Grado/Doctorado	Universidad	Año
Professor (catedrático) en Epidemiología Genética de Enfermedades Inflamatorias Crónicas	UPPSALA UNIVERSITY	2009
Docente en Genética Médica	UPPSALA UNIVERSITY	2000
DOCTOR EN FILOSOFÍA / IMMUNOLOGÍA	STOCKHOLM UNIVERSITY. DEPARTAMENTO DE INMUNOLOGÍA DE LA UNIVERSIDAD DE ESTOCOLMO	1994
Licenciada en Medicina	Universidad Nacional Autónoma de México	1985

A.3. Indicadores generales de calidad de la producción científica

Parte B. RESUMEN LIBRE DEL CURRÍCULUM

For over 20 years of my research career I have focused in the identification of the genetic basis of SLE as a first building block towards understanding how such genes lead to cellular abnormalities that eventually lead to clinical disease. A main goal of my research is to understand the mechanisms behind disease pathogenesis and develop therapies.

My early publications dealt with the genetics of lupus, primarily focusing on the identification of susceptibility genes for lupus. The first studies involved multicase families, extended pedigrees obtained from Iceland and Sweden. This led us to identify PDCD1 as involved in lupus susceptibility that we published in Nature Genetics in 2002. The PDCD1 work was one of the first to analyse functional variants and their influence in the function of a gene. After this, other studies followed, including the analysis of IRF5, also published in Nature Genetics in 2006.

With the advent of genome-wide arrays and the creation of the SLEGEN consortium of which I am a founder member, new possibilities opened for the study of the genetics of lupus. As a member of SLEGEN, I participated in the GWAS that identified several new genes, a study identifying ITGAM, and my own independent study where I identified BANK1. These papers were published in the same issue of Nature Genetics in 2008.

I have continued the genetics studies primarily on the identification of genes for lupus in Latin American populations with the understanding of how European admixture contributes to the genetic risk in a unique collection of mestizos with enriched Native American ancestry. Our

GWAS in Latin Americans has been published in Arhtritis & Rheumatology (Alarcón-Riquelme, M.E., et al., 2016).

A second path relates to my discovery of BANK1 and its association with lupus: Our studies have also shown that BANK1 is involved in the TLR7/9 pathway. Our work with BANK1 led us to begin the work on mouse models by setting-up all the systems required. We have found that BANK1 is involved in translation initiation (Wu, et al. J Immunol, 2013). Deficiency of BANK1 abrogates disease phenotypes dependent on TLR7 (Wu, et al., PlosONE, 2016). We are now approaching this work with biochemistry through funding from the MINECO and with continued work on animal models that we are also using to test potential new therapies.

Finally, through our funding from the Innovative Medicines Initiative, we are working on an approach towards diagnosis of systemic autoimmune diseases. The project known as PRECISESADS is providing with the basis for continued study combining mechanisms of disease, reclassification of disease based on pathways and -omics-derived Big Data, integration of such data and clustering analyses. I am certain that our results will reveal clinically useful information that will help in the early diagnosis, not only of SLE, but of the systemic autoimmune diseases, impacting on therapeutic decision-making.

Parte C. MÉRITOS MÁS RELEVANTES (ordenados por tipología)

C.1. Publicaciones

1 Artículo científico. Toro-Dominguez, Daniel; et al. 2018. Longitudinal Stratification of Gene Expression Reveals Three SLE Groups of Disease Activity Progression.Arthritis & rheumatology (Hoboken, N.J.). ISSN 2326-5205.

2 Artículo científico. Delgado-Vega, Angelica M.; et al. 2018. Whole Exome Sequencing of Patients from Multicase Families with Systemic Lupus Erythematosus Identifies Multiple Rare Variants SCIENTIFIC REPORTS. NATURE PUBLISHING GROUP. 8. ISSN 2045-2322.

3 Artículo científico. Bellocchi, Chiara; et al. 2018. Microbial and metabolic multi-omic correlations in systemic sclerosis patients ANNALS OF THE NEW YORK ACADEMY OF SCIENCES. WILEY. 1421-1, pp.97-109. ISSN 0077-8923, ISSN 1749-6632.

4 Artículo científico. Carnero-Montoro, Elena; Alarcon-Riquelme, Marta E. 2018. Epigenome-wide association studies for systemic autoimmune diseases: The road behind and the road ahead.Clinical immunology (Orlando, Fla.). ISSN 1521-7035.

5 Artículo científico. Barturen, G.; et al. 2018. Moving towards a molecular taxonomy of autoimmune rheumatic diseases.Nature reviews. Rheumatology. 14-3, pp.180. ISSN 1759-4804.

6 Artículo científico. García Pérez, JL.; Alarcón Riquelme, ME.2017. The TREX1 Dinosaur Bites the Brain through the LINE.Cell stem cell. 21-3, pp.287-288. ISSN 1875-9777.

7 Artículo científico. Carmona Sáez, P.; et al. 2017. Metagene projection characterizes GEN2.2 and CAL-1 as relevant human plasmacytoid dendritic cell models.Bioinformatics (Oxford, England). ISSN 1367-4811.

8 Artículo científico. Langefeld, CD.; et al. 2017. Transancestral mapping and genetic load in systemic lupus erythematosus.Nature communications. 8, pp.16021. ISSN 2041-1723.

9 Artículo científico. Steri, M.; et al. 2017. Overexpression of the Cytokine BAFF and Autoimmunity Risk.The New England journal of medicine. 376-17, pp.1615-1626. ISSN 1533-4406.

10 Artículo científico. Teruel, M.; Chamberlain, C.; Alarcón Riquelme, ME.2017. Omics studies: their use in diagnosis and reclassification of SLE and other systemic autoimmune diseases.Rheumatology (Oxford, England). 56-suppl_1, pp.i78. ISSN 1462-0332.

11 Artículo científico. Toro Domínguez, D.; Carmona Sáez, P.; Alarcón Riquelme, ME.2017. Support for phosphoinositol 3 kinase and mTOR inhibitors as treatment for lupus using in-silico drug-repurposing analysis.Arthritis research & therapy. 19-1, pp.54. ISSN 1478-6362.

12 Artículo científico. Muchmore, B.; Alarcón Riquelme, ME.2017. CymeR: cytometry analysis using KNIME, docker and R.Bioinformatics (Oxford, England). 33-5, pp.776-778. ISSN 1367-4811.

13 Artículo científico. Hoffman, RW.; et al. 2017. Gene Expression and Pharmacodynamic Changes in 1,760 Systemic Lupus Erythematosus Patients From Two Phase III Trials of BAFF Blockade With Tabalumab.Arthritis& rheumatology(Hoboken, N.J.). 69-3, pp.643-654. ISSN 2326-5205.

14 Artículo científico. Barturen, G.; Alarcón Riquelme, ME.2017. Systemic Lupus Erythematosus in 2016: Gene expression profiling comes closer to the clinic.Nature reviews. Rheumatology. 13-2, pp.69-70. ISSN 1759-4804.

15 Artículo científico. Márquez, A.; et al. 2017. A combined large-scale meta-analysis identifies COG6 as a novel shared risk locus for rheumatoid arthritis and systemic lupus erythematosus.Annals of the rheumatic diseases. 76-1, pp.286-294. ISSN 1468-2060.

16 Artículo científico. Teruel, M.; Alarcón Riquelme, ME.2016. Genetics of systemic lupus erythematosus and Sjögren's syndrome: an update.Current opinion in rheumatology. 28-5, pp.506-514. ISSN 1531-6963.

17 Artículo científico. Wu, Ying-Yu; et al. 2016. BANK1 Regulates IgG Production in a Lupus Model by Controlling TLR7-Dependent STAT1 Activation PLOS ONE. PUBLIC LIBRARY SCIENCE. 11-5. ISSN 1932-6203.

18 Artículo científico. Alarcon-Riquelme, Marta E; et al. 2016. Genome-Wide Association Study in an Amerindian Ancestry Population Reveals Novel Systemic Lupus Erythematosus Risk Loci and the Role of European Admixture.Arthriti& rheumatology(Hoboken, N.J.). 68-4, pp.932-43. ISSN 2326-5205.

19 Artículo científico. Bentham, James; et al. 2015. Genetic association analyses implicate aberrant regulation of innate and adaptive immunity genes in the pathogenesis of systemic lupus erythematosus NATURE GENETICS. NATURE PUBLISHING GROUP. 47-12, pp.1457-+. ISSN 1061-4036, ISSN 1546-1718.

20 Artículo científico. Homburger, Julian R.; et al. 2015. Genomic Insights into the Ancestry and Demographic History of South America PLOS GENETICS. PUBLIC LIBRARY SCIENCE. 11-12. ISSN 1553-7404.

21 Artículo científico. Oparina, Nina Y.; et al. 2015. PXK locus in systemic lupus erythematosus: fine mapping and functional analysis reveals novel susceptibility gene ABHD6 ANNALS OF THE RHEUMATIC DISEASES. BMJ PUBLISHING GROUP. 74-3. ISSN 0003-4967, ISSN 1468-2060.

22 Artículo científico. Wu, Ying-Yu; et al. 2015. Concordance of Increased B1 Cell Subset and Lupus Phenotypes in Mice and Humans Is Dependent on BLK Expression Levels Journal of Immunology. 194-12, pp.5692-5702. ISSN 0022-1767.

23 Artículo científico. Vukovic, F; et al. 2015. Systemic lupus erythematosus associates with the decreased immunosuppressivepotential of the IgG glycome Arthritis Rheumatol. 67-11, pp.2978-2989.

24 Artículo científico. Wu, Ying-Yu; et al. 2013. BANK1 Controls CpG-Induced IL-6 Secretion via a p38 and MNK1/2/eIF4E Translation Initiation Pathway JOURNAL OF IMMUNOLOGY. AMER ASSOC IMMUNOLOGISTS. 191-12, pp.6110-6116. ISSN 0022-1767, ISSN 1550-6606.

25 Artículo científico. Martin, Jose-Ezequiel; et al. 2013. A systemic sclerosis and systemic lupus erythematosus pan-meta-GWAS reveals new shared susceptibility loci HUMAN MOLECULAR GENETICS. OXFORD UNIV PRESS. 22-19, pp.4021-4029. ISSN 1460-2083.

26 Artículo científico. Lopez Herraez, David; et al. 2013. Rheumatoid Arthritis in Latin Americans Enriched for Amerindian Ancestry is Associated With Loci in Chromosomes 1, 12, and 13, and the HLA Class II Region ARTHRITIS AND RHEUMATISM. WILEY-BLACKWELL. 65-6, pp.1457-1467. ISSN 0004-3591.

C.2. Proyectos

1 AUTOBANK-El Papel de BANK1 en la Señalización de Células B de TLR y en la Autoinmunidad MINECO. Marta Alarcon Riquelme. (FUNDACION PUBLICA ANDALUZA PROGRESO Y SALUD). 30/12/2016-29/12/2019. 238.000 €.

2 FP 7 Funding Grant Agreement nr°115565, Molecular Reclassification to Find Clinically Useful Biomarkers for Systemic Autoimmune Diseases IMI - Innovative Medicines Initiative. (FUNDACION PUBLICA ANDALUZA PROGRESO Y SALUD). 01/02/2014-31/01/2019. 22.700.000 €.

3 Influence of BANK1 in the In Vivo Development of Lupus (FUNDACION PUBLICA ANDALUZA PROGRESO Y SALUD). 01/03/2014-01/02/2017. 409.076 €.

4 PI12/02558, Lupus Eritematoso Sistémico: Como las variantes genéticas de riesgo dan lugar a alteraciones celulares FONDO DE INVESTIGACION SANITARIA. FONDO DE INVESTIGACION SANITARIA. MARTA EUGENIA ALARCÓN RIQUELME. (GENYO). Desde 01/01/2013. 206.910 €.

5 INVESTIGACIÓN EN CIENCIAS DE LA VIDA Y DE LA MATERIA, Secuenciación Completa del Genoma Exómico del Lupus Eritematoso Sistémico en Familias de Casos Múltiples de Orígen Europeo Funcacion Ramon Areces. MARTA EUGENIA ALARCÓN RIQUELME. Desde 01/06/2012. 101.136 €.

C.3. Contratos

C.4. Patentes

1 Lopez-Escamez, Jose Antonio; Alarcón-Riquelme, Marta Eugenia; Requena-Navarro, Maria Teresa; Cabrera- Martínez, Sonia; Sánchez-Rodriguez, Elena. P-06488. - Diagnóstico genético de la hipoacusia sensorial asociada a enfermedad de Menière o enfermedad autoinmune del oído interno 21/06/2013. Fundación Pública Andaluza Progreso y Salud.

2 Delgado-Vega-,Angelica Maria; Wojcik-,J; Alarcón-Riquelme, Marta Eugenia; Castillejo-López, Casimiro. PCT/EP2010052554. - BANK1 RELATED SNPS AND SLE AND/OR MS SUSCEPTIBILITY10/10/2010 10/10/2010. Merck-Serono.