

Fecha del CVA	28/01/2020
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Parte A. DATOS PERSONALES

Nombre y Apellidos	Elena Goicoechea de Jorge		
DNI	14305533Q	Edad	42
Núm. identificación del investigador	Researcher ID	L-4580-2016	
	Scopus Author ID		
	Código ORCID		

A.1. Situación profesional actual

Organismo	Universidad Complutense de Madrid		
Dpto. / Centro	Department of Microbiology / School of Medicine		
Dirección			
Teléfono	(0034) 665534625	Correo electrónico	egoicoec@ucm.es
Categoría profesional	Contratado Ramón y Cajal	Fecha inicio	2015
Espec. cód. UNESCO	240000 - Ciencias de la Vida		
Palabras clave	Mecanismos moleculares de enfermedad		

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

Licenciatura/Grado/Doctorado	Universidad	Año
Doctorate programme in Molecular Biology	University Autónoma of Madrid	2007
Certificate of advanced study	University Autónoma de Madrid	2002
Bachelor in Biochemistry	University Autónoma de Madrid	2000

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

Artículos totales indexados: 32 (12 de primer autor) 81 % Q1, 17% Q2 y 3% Q3.

Artículos originales: 27

Revisiones: 5

Número citas calculadas (sin citas propias) WOS: 2.801

Índice H (WOS): 23

Parte B. RESUMEN LIBRE DEL CURRÍCULUM

I am a complement biologist whose main research interest is the study of the complement system and its association with disease. My interest in complement began with my doctoral training in Prof. Santiago Rodríguez de Córdoba's group. My thesis focused on the study of the molecular basis of atypical haemolytic uremic syndrome (aHUS), a paradigm of complement-mediated renal disease. I was the first one to demonstrate that, in addition to loss-of-function mutations in complement regulators, gain-of-function mutations in complement activators are associated with aHUS (**Goicoechea de Jorge** et al. PNAS, 2007). I also demonstrated that confluence of multiple inherited risk factors in complement genes is required for aHUS to develop. During my thesis, I also performed work with in collaboration with Prof. Matthew Pickering in London that led to the development of the first animal model of aHUS (Pickering, **Goicoechea de Jorge** et al. J. Exp. Med. 2007).

Once completed my PhD training and motivated by the idea of studying complement dysregulation in vivo, I moved to Imperial College London and joined Prof. Pickering's group as a postdoctoral researcher. During this time, I completed a program of work demonstrating that complement C5 is critical for renal injury to develop in our aHUS model (**Goicoechea de Jorge** et al. JASN, 2011). I also expanded my expertise in complement dysregulation to the study of other complement-mediated renal diseases such as C3 glomerulopathies. I identified the first mutation in complement factor H-related 5 protein (FHR-5) associated with familial C3-glomerulopathy. This work led to a Lancet publication (Gale, **Goicoechea de Jorge**, et al., 2010). This finding opened a new research avenue in the complement field and gave me the opportunity to outline a program of work and secure a Junior Research Fellowship from

Imperial College. Since then, I focused my research in understanding the biology of the FHR1-5 protein family and its association with disease. My major finding was the discovery of the dimerization status for FHR-1, FHR-2 and FHR-5 proteins and the dramatic consequences that the duplication of the dimerization domains has in disease-associated mutant proteins (**Goicoechea de Jorge** et al. PNAS, 2013).

After seven years of a fruitful scientific period in the UK, I was awarded a Ramón y Cajal contract and I settled an independent investigator at the Complutense University Madrid in 2015. Since then, I have continued working on deciphering the biological role of the FHR protein family and its association with renal diseases (i.e. atypical haemolytic uremic syndrome, C3 glomerulopathy and IgA nephropathy) representing one of the main research lines in my lab (**Goicoechea de Jorge** et al. JASN, 2017; Tortajada, Gutiérrez, **Goicoechea de Jorge** et al. Kidney International, 2018). Recently, I also became interested in exploring new scientific avenues. On the one hand, I am investigating the role of complement dysregulation in the pathogenesis of ANCA-associated vasculitis and, on the other hand, I am investigating the potential of microRNAs as biomarkers of the complement-mediated renal diseases. Both lines are being fruitful and manuscripts with the first set of results are on the way.

Finally, I am involved in teaching at the Complutense University which allowed me to get the ANECA positive assessment for "Profesor Contratado Doctor". In addition, I obtained the positive assessment for the I3 program.

Parte C. MÉRITOS MÁS RELEVANTES (ordenados por tipología)

C.1. Publicaciones

- 1 **Artículo científico.** Elena Goicoechea de Jorge; et al. 2018. Factor H Competitor Generated by Gene Conversion Events Associates with Atypical Hemolytic Uremic Syndrome. Journal of the American Society of Nephrology. 29-1, pp.240-249.
- 2 **Artículo científico.** Goicoechea de Jorge E; et al. 2013. Dimerization of complement factor H-related proteins modulates complement activation in vivo Proc Natl Acad Sci U S A. 110-12, pp.4685-4690.
- 3 **Artículo científico.** Goicoechea de Jorge E; et al. 2011. The development of atypical hemolytic uremic syndrome depends on complement C5. J Am Soc Nephrol. 22-1, pp.137-145.
- 4 **Artículo científico.** Gale DP; et al. 2010. Identification of a mutation in complement factor H-related protein 5 in patients of Cypriot origin with glomerulonephritis. Lancet. 376-9743, pp.794-801.
- 5 **Artículo científico.** Elena Goicoechea de Jorge; et al. 2007. Gain-of-function mutations in complement factor B are associated with atypical hemolytic uremic syndrome. Proc Natl Acad Sci U S A. 104, pp.240-245.
- 6 **Artículo científico.** Agustín Tortajada; et al. 2017. Elevated factor H-related protein 1 and factor H pathogenic variants decrease complement regulation in IgA nephropathy Kidney International. 92-4, pp.953-963.
- 7 **Artículo científico.** Csicsi, A.I.; et al. 2015. Factor H-related protein 5 (CFHR5) interacts with pentraxin 3 and the extracellular matrix and modulates complement activation Journal of Immunology. 194-10, pp.4963-4973.
- 8 **Artículo científico.** Joseph Caesar; et al. 2014. Competition between antagonistic complement factors for a single protein on N. meningitidis rules disease susceptibility. eLife. 3-e04008.
- 9 **Artículo científico.** MC Pickering; et al. 2013. C3 glomerulopathy: consensus report. Kidney Int. 84-6, pp.1079-1089.
- 10 **Artículo científico.** TH Malik; et al. 2012. A hybrid CFHR3-1 gene causes familial C3 glomerulopathy. J Am Soc Nephrol. 23-7, pp.1155-1160.
- 11 **Artículo científico.** Vernon KA; et al. 2012. Acute presentation and persistent glomerulonephritis following streptococcal infection in a patient with heterozygous complement factor H-related protein 5 deficiency. Am J Kidney Dis. 60-1, pp.121-125.
- 12 **Artículo científico.** S Johnson; et al. 2012. Design and evaluation of meningococcal vaccines through structure-based modification of host and pathogen molecules. PLoS Pathog. 8-10, pp.e1002981.

- 13 **Artículo científico.** Vernon KA; et al. 2011. Recurrence of complement factor H-related protein 5 nephropathy in a renal transplant. *Am J Transplant.* 11, pp.152-155.
- 14 **Artículo científico.** Goicoechea de Jorge E; Pickering MC. 2010. Atypical hemolytic uremic syndrome: telling the difference between H and Y. *Kidney Int.* 78-8, pp.721-723.
- 15 **Artículo científico.** Fakhouri F; et al. 2010. Treatment with human complement factor H rapidly reverses renal complement deposition in factor H-deficient mice. *Kidney Int.* 78-3, pp.279-286.
- 16 **Artículo científico.** Martínez-Barricarte R; et al. 2009. Lack of association between polymorphisms in C4b-binding protein and atypical haemolytic uraemic syndrome in the Spanish population. *Clin Exp Immunol.* 155-1, pp.59-64.
- 17 **Artículo científico.** Hakobyan S; et al. 2008. Complement factor H binds to denatured rather than to native pentameric C-reactive protein *The Journal of biological chemistry.* 283, pp.30451-30460.
- 18 **Artículo científico.** Montes T; et al. 2008. Genetic deficiency of complement factor H in a patient with age-related macular degeneration and membranoproliferative glomerulonephritis. *Mol Immunol.* 45-10, pp.2897-2904.
- 19 **Artículo científico.** Hakobyan S; et al. 2008. Measurement of factor H variants in plasma using variant-specific monoclonal antibodies: application to assessing risk of age-related macular degeneration. *Invest Ophthalmol Vis Sci.* 49-5, pp.1983-1990.
- 20 **Artículo científico.** Abarategui-Garrido; et al. 2008. Mutations in Proteins of the Alternative Pathway of Complement and the Pathogenesis of Atypical Hemolytic Uremic Syndrome *American Journal of Kidney Diseases.* 52, pp.171-180.
- 21 **Artículo científico.** Saunders R.E.; et al. 2007. The interactive Factor H-atypical hemolytic uremic syndrome mutation database and website: update and integration of membrane cofactor protein and Factor I mutations with structural models. *Hum Mutat.* 28-3, pp.222-234.
- 22 **Artículo científico.** Elena Goicoechea de Jorge; et al. 2007. Spontaneous hemolytic uremic syndrome triggered by complement factor H lacking surface recognition domains. *J Exp Med.* 204-6, pp.1249-1256.
- 23 **Artículo científico.** Esparza-Gordillo J; et al. 2006. Insights into hemolytic uremic syndrome: segregation of three independent predisposition factors in a large, multiple affected pedigree. *Mol. Immunol.* 43, pp.1769-1775.
- 24 **Artículo científico.** Jorge Esparza-Gordillo; et al. 2005. Predisposition to atypical Hemolytic Uremic Syndrome involves the concurrence of different susceptibility alleles in the Regulators of Complement Activation gene cluster in 1q32. *Hum Mol Gen.* 14-5, pp.703-712.
- 25 **Artículo científico.** Uyguner O; et al. 2003. Molecular analyses of the HGO gene mutations in Turkish alkaptonuria patients suggest that the R58fs mutation originated from central Asia and was spread throughout Europe and Anatolia by human migrations *J Inherit Metab Dis.* 26-1, pp.17-23.
- 26 **Artículo científico.** Goicoechea de Jorge E; et al. 2002. Alkaptonuria in the Dominican Republic: identification of the founder AKU mutation and further evidence of mutation hot spots in the HGO gene. *J Med Genet.* 39-7, pp.E40.
- 27 **Artículo científico.** Sánchez-Corral P; et al. 2002. Structural and functional characterization of factor H mutations associated with atypical hemolytic uremic syndrome. *Am J Hum Genet.* 71-6, pp.1285-1295.
- 28 **Capítulo de libro.** S. Rodríguez de Córdoba; E. Goicoechea de Jorge; F. Vivanco. 2007. Complemento *Medicina Interna Farreras-Rozman (Décimoquinta Edición).*
- 29 **Revisión bibliográfica.** Goicoechea de Jorge E; et al. 2018. Common and Rare Genetic Variants of Complement Components In Human Disease *Molecular Immunology.* 102, pp.42-57.
- 30 **Revisión bibliográfica.** Goicoechea de Jorge E; et al. 2018. How novel structures inform understanding of complement function *Seminars in Immunopathology.* 40-1, pp.3-14.
- 31 **Revisión bibliográfica.** Myjali Jozsi; et al. 2015. Factor H-related proteins determine complement-activating surfaces *Trends in Immunology.* 36-6, pp.374-384.
- 32 **Revisión bibliográfica.** de Córdoba SR; de Jorge EG. 2008. Translational mini-review series on complement factor H: genetics and disease associations of human complement factor H. *Clin Exp Immunol.* 151-1, pp.1-13.

- 33 Revisión bibliográfica.** Santiago Rodríguez de Córdoba; Elena Goicoechea de Jorge. 2007. Predisposición genética al síndrome hemolítico urémico atípico. Avances en la elucidación de mecanismos patogénicos y su relevancia en el desarrollo de estrategias terapéuticas Nefrología e hipertensión.
- 34 Revisión bibliográfica.** Rodríguez de Córdoba S; et al. 2004. The human complement factor H: functional roles, genetic variations and disease associations. Mol Immunol.41-4, pp.355-367.

C.2. Proyectos

- 1 Descifrando las bases moleculares de enfermedades renales mediadas por el complemento (RTI2018-095955-B-I00) Ministerio de Ciencia, Innovación y Universidades. Elena Goicoechea de Jorge. (Universidad Complutense de Madrid). 01/01/2019-31/12/2021. 121.000 €.
- 2 Contrato Ramón y Cajal. Complement Physiopathology Elena Goicoechea de Jorge. (Universidad Complutense de Madrid). 25/02/2015-15/06/2020. 40.000 €. Investigador principal.
- 3 El papel de los microRNAs en enfermedades renales mediadas por el sistema del complemento Ministerio de Economía y Competitividad. Elena Goicoechea de Jorge. (Universidad Complutense de Madrid). 2015-2018. 157.300 €. Investigador principal.
- 4 Junior Research Fellow. The biological role of CFHR5 in renal disease Imperial College London. Elena Goicoechea de Jorge. (Imperial College London). 21/09/2011-24/02/2015. 38.000 €. Investigador principal.
- 5 The pathophysiological role of complement regulation in disease injury Matthew Pickering. (Imperial College London). 2008-2011. Miembro de equipo.
- 6 Identificación y caracterización funcional de genes responsables de enfermedades. Bases moleculares del síndrome hemolítico urémico y de la enfermedad de Lafora. Santiago Rodríguez de Córdoba. (Centro de Investigaciones Biológicas). 2005-2008. Miembro de equipo.
- 7 Papel de las proteínas reguladoras del complemento C4BP y Factor H en patología humana Santiago Rodríguez de Córdoba. (Centro de Investigaciones Biológicas). 2003-2005. Miembro de equipo.
- 8 Diagnóstico molecular y aproximaciones a la terapia de deficiencias de proteínas reguladoras del complemento. Factor H y predisposición genética a HUS Santiago Rodríguez de Córdoba. (Centro de Investigaciones Biológicas). 2001-2003. Miembro de equipo.
- 9 El Sistema del Complemento en Salud y Enfermedad. Ayudas para la realización de Programas de Actividades de I+D entre Grupos de Investigación de la Comunidad de Madrid. Comunidad de Madrid. (Universidad Complutense de Madrid). Desde 2018. 828.091,87 €. Miembro de equipo.
- 10 Complemento en Salud y Enfermedad. Redes de Excelencia. MINECO/AEI. (Universidad Complutense de Madrid). Desde 2017. 20.000 €. Investigador principal.
- 11 Desentrañando las bases moleculares del síndrome hemolítico urémico y las glomerulopatías de C3 Fundación Inocente Inocente. Elena Goicoechea de Jorge. (Universidad Complutense de Madrid). Desde 2017. 27.960 €. Investigador principal.

C.3. Contratos

C.4. Patentes

Susan Lea; Matthew Pickering; Elena Goicoechea de Jorge. PCT/GB2014/050258. Patent in Complement System Modulators Reino Unido. 30/01/2014. Imperial College London (Imperial Innovations).