



3.3.5 Diagnosis and Treatment of Diseases Associated with Abnormalities of the Complement System Group

Publications: 8

Q1: 6

COMPOSITION

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STRATEGIC OBJECTIVE

This is a national reference group for the study of primary or acquired defects in the complement system.

The main aim is to gain an understanding of the molecular mechanisms underlying in complement deficiency or dysregulation in several human diseases, the majority of which are classified as rare diseases.

In this context, the group implements and applies various methodological strategies (immunological, biochemical, proteomic and genetic) to detect clinically relevant deficiencies or functional defects in complement components. By following a clearly translational approach,

the group also develops new diagnostic tests which are implemented in the clinical practice, especially for treatment adjustment and patient follow-up.

The research lines could be classified as:

screening and characterization of genetic or acquired complement defects that cause renal pathology (I), functional and molecular diagnosis of deficiencies affecting individual components of the complement system (II), diagnosis and molecular studies in Hereditary Angioedema and screening for disease modifying genes (III), study of pathogenic mechanisms in acquired lipodystrophies (IV).



RESEARCH LINES

- Screening and characterization of genetic and acquired Complement defects in renal disease.
- Biochemical and molecular diagnosis of isolated Complement deficiencies.
- Characterization of autoantibodies in acquired generalized lipodystrophy.

- Screening and molecular characterization of disease modifying genes in Hereditary Angioedema.
- Genetic and immunological mechanisms associated with Complement dysregulation in partial lipodystrophy.

RESEARCH ACTIVITY

● Doctoral theses

Gómez Delgado I. Contribución del FH y las proteínas FHRs del Complemento en la predisposición y evolución del Síndrome Hemolítico-Urémico Atípico y la Nefropatía por IgA [dissertation]. Madrid: Universidad Autónoma de Madrid: 2022 (14/07/2022).

Director: Sánchez-Corral Gómez P.

European Mention



● Publications

- Caravaca-Fontán F, Díaz-Encarnación M, Cabello V, Ariceta G, Quintana LF, Marco H, Barros X, Ramos N, Rodríguez-Mendiola N, Cruz S, Fernández-Juarez G, Rodríguez A, de José AP, Rabasco C, Rodado R, Fernández L, Gómez VP, Ávila A, Bravo L, Espinosa N, Allende N, de la Nieta MDS, Rodríguez E, Olea T, Melgosa M, Huerta A, Miquel R, Mon C, Fraga G, de Lorenzo A, Draibe J, Cano-Megías M, González F, Shabaka A, López-Rubio ME, Fenollosa MA, Martín-Penagos L, Da Silva I, Titos JA, de Córdoba SR, de Jorge EG, Praga M. Longitudinal change in proteinuria and kidney outcomes in C3 glomerulopathy. *Nephrol Dial Transpl.* 2022; 37(7): 1270-80. Article. IF: 6.1; Q1
- Caravaca-Fontan F, Rivero M, Caverio T, Díaz-Encarnacion M, Cabello V, Ariceta G, Quintana LF, Marco H, Barros X, Ramos N, Rodríguez-Mendiola N, Cruz S, Fernández-Juárez G, Rodríguez A, de José AP, Rabasco C, Rodado R, Fernández L, Pérez-Gómez V, Ávila A, Bravo L, Espinosa N, Allende N, de la Nieta MDS, Rodríguez E, Olea T, Melgosa M, Huerta A, Miquel R, Mon C, Fraga G, de Lorenzo A, Draibe J, González F, Shabaka A,

López-Rubio ME, Fenollosa MA, Martín-Penagos L, Da Silva I, Titos JA, de Córdoba SR, de Jorge EG, Praga M. Development and validation of a nomogram to predict kidney survival at baseline in patients with C3 glomerulopathy. *Clin Kidney J.* 2022; 15(9): 1737-46. Article. IF: 4.6; Q1

- Delgado IG, Sánchez-Corral P. Contribution of functional and quantitative genetic variants of Complement Factor H and Factor H-Related (FHR) proteins on renal pathology. *Nefrología.* 2022; 42(3): 280-9. Review. IF: 2.6; Q3
- Drouet C, López-Lera A, Ghannam A, López-Trascasa M, Cichon S, Ponard D, Parsopoulou F, Grombrikova H, Freiburger T, Rijavec M, Veronez CL, Pesquero JB, Germeis AE. SERPING1 variants and C1-INH biological function: A close relationship with C1-INH-HAE. *Front Allergy.* 2022; 3: 835503. Review. Not Indexed
- Espinosa LG, Santovena AZ, Blanco JN, Alvario MG, Feito JB, Hijosa MM. Spontaneous remission in a child with an NPHS1-based congenital nephrotic syndrome. *Clin Kidney J.* 2022; 15(10): 1969-70. Letter. IF: 4.6; Q1
- Fernández-Fournier M, Lacruz L, Nozal P, Chico JL, Barranco AT, Otero-Ortega L, Corral I, Carrasco A. The study of neural antibodies in neurology: A practical summary. *Front Immunol.* 2022; 13: 1043723. Review. IF: 7.3; Q1
- Márquez-Tirado B, Gutiérrez-Tenorio J, Tortajada A, Continente LL, Caravaca-Fontán F, Malik TH, Montero RR, Elias S, González AS, Fernández-Juárez G, Sánchez-Corral P, Pickering MC, Praga M, de Córdoba SR, de Jorge EG. Factor H-related protein 1 drives disease susceptibility and prognosis in C3 glomerulopathy. *J Am Soc Nephrol.* 2022; 33(6): 1137-53. Article. IF: 13.6; D1
- Sánchez-Zapardiel E, Alos M, Nozal P, González-Muñoz M, Frauca-Remacha E, Gavilán LB, Quiles MJ, Hierro L, López-Granados E. Humoral and

cellular immune responses to Pfizer-BioNTech BNT162b2 SARS-CoV-2 vaccine in adolescents with liver transplantation: Single center experience. *Front Immunol.* 2022; 13: 1049188. Article. IF: 7.3; Q1

● Research projects

Nozal Aranda P, Corvillo Rodríguez F. Detección y caracterización de autoanticuerpos en pacientes con lipodistrofia generalizada adquirida (síndrome de Lawrence). Asociación de Familiares y Afectados de Lipodistrofia (AELIP). 2019-2022.

Managment centre: CIBERER

Sánchez-Corral Gómez P. Contrato Miguel Servet Categoría A (CES07/030). CM. 2008-2025.

Managment centre: FIBHULP

Sánchez-Corral Gómez P. El sistema del complemento en salud y enfermedad (Complemento II-CM) (S2017/BMD-3673). CM. 2018-2021.

Managment centre: FIBHULP

Sánchez-Corral Gómez P. Perfiles cuantitativos de las proteínas FH/FHRS del complemento como biomarcadores de predisposición y pronóstico en el síndrome hemolítico-urémico primario y secundario (P119/00970). ISCIII. 2020-2022.

Managment centre: FIBHULP

López Trascasa M. Estudio de las bases mo-

leculares del Síndrome Hemolítico Urémico Atípico y creación de un registro español de SHU-Atípico. (PI-1106). Secugen S.L. 2011-Ongoing.

Managment centre: FIBHULP

Sánchez-Corral Gómez P. Caracterización funcional de la deglicosilación y proteólisis del FH del Complemento y relevancia en la patología del Síndrome Hemolítico-Urémico Atípico. Fundación SENEPRO. 2022-Ongoing.

Managment centre: FIBHULP

● Cibers and Retics

Sánchez-Corral Gómez P. Rare Diseases Networking Biomedical Research Centre. (CIBERer). ISCIII. (31/12/2022). FIBHULP

López Trascasa M. The European Consortium of Lipodystrophies. (ECLIP). EU. (31/12/2023). FIBHULP

● Patents and trademarks

López Trascasa M, authors; FIBHULP, assignee. Brand name: COMPLEMENTest; CM 009.658.791; 2011, January 14.

