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3.3.5 Complement Defects in Human Phatology

Publications: 9 | **Q1:**7

COMPOSITION

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STRATEGIC OBJETIVES

- 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.
- Biochemical and molecular diagnosis of Hereditary and Acquired forms of bradykinin-mediated Angioedema.
- Pathogenic mechanisms of adquired lipodistrhphies: Barraquer-Simons syndrome, and Lawrence syndrome.
- Complement defects in Age Related Macular Degeneration

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RESEARCH ACTIVITY

Master Theses

- **García Sánchez MR.** Impact of FH desialylation on the regulation of the Alternative Pathway and contribution to pathology[dissertation]. *Madrid: UCM: 2023(16/07/2023). Director: Sánchez-Corral Gómez MP, Corvillo Rodríguez F.*

Publications

- **Bakkaloglu SA, Vidal E, Bonthuis M, Neto G, Paripovic D, Asberg A, Hijosa MM, Vondrak K, Jankauskiene A, Roussinov D, Awan A, Jager KJ.** European chronic kidney disease registries for children not on kidney replacement therapy: tools for improving health systems and patient-centred outcomes. Clin Kidney J. 2023; 16(11): 1980-5. Article. IF: 3.9; Q1
- **Caravaca-Fontán F, Cavero T, Díaz-Encarnación 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, Avila A, Bravo L, Espinosa N, Allende N, de la Nieta MDS, Rodríguez E, Rivas B, 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.** Clinical profiles and patterns of kidney disease progression in c3 glomerulopathy. Kidney360. 2023; 4(5): 659-72. Article. IF: 3.2; Q1
- **Corvillo F, Abel BS, López-Lera A, Ceccarini G, Magno S, Santini F, Araújo-Vilar D, Brown RJ, Nozal P, López-Trascasa M.** Characterization and clinical association of autoantibodies against perilipin 1 in patients with acquired generalized lipodystrophy. Diabetes. 2023; 72(1): 71-84. Article. IF: 6.2; Q1
- **García-Solís B, van den Rym AD, Martínez-Martínez L, Franco T, Pérez-Caraballo JJ, Markle J, Cubillos-Zapata C, Marín A, Recio MJ, Regueiro JR, Navarro-Zapata A, Mestre-Durán C, Ferreras C, Cotázar CM, Mena R, de la Calle-fabregat C, López-Lera A, Arquero MF, Pérez-Martínez A, López-Collazo E, Sánchez-Ramón S, Casanova JL, Martínez-Barricarte R, de la Calle-Martín O, de Diego RP.** Inherited human ezrin deficiency impairs adaptive immunity. J Allergy Clin Immun. 2023; 152(4): 997-1009. Article. IF: 11.4; D1
- **Iglesias GT, Fernández-Fournier M, López-Molina M, Piniella D, Laso-García F, Gómez-de Frutos MC, Alonso-López E, Botella L, Chamorro B, Sánchez-Velasco S, Puertas I, Barranco AT, Nozal P, Díez-Tejedor E, Gutiérrez-Fernández M, Otero-Ortega L.** Dual role of peripheral B cells in multiple sclerosis: emerging remote players in demyelination and novel diagnostic biomarkers. Front Immunol. 2023; 14: 1224217. Article. IF: 5.7; Q1
- **Molina LD, Gallego LYB, Nozal P, Soto-Serrano Y, Martínez-Feito A, Reche-Yebra K, González-Torbay A, de la Cámara RCM, Gianelli C, Cámara C, González-García J, González-Muñoz M, Rodríguez-Pena R, Granados EL.** Detection of specific RBD+ IgG+ memory B cells by flow cytometry in healthcare workers and patients with inborn errors of immunity after BNT162b2 m RNA COVID-19 vaccination. Front Immunol. 2023; 14: 1136308. Article. IF: 5.7; Q1
- **Plasencia-Rodríguez C, Martínez-Feito A, Hernández M, del Pino-Molina L, Novella-Navarro M, Serrano Y, González-Muñoz M, Peiteado D, Bonilla G, Monjo I, Nuño L, Tornero C, López-Granados E, Balsa A, Nozal P.** Immune response after SARS-CoV-2 vaccination in patients with inflammatory immune-mediated diseases receiving immunosuppressive treatment. Allergy Asthma Clin Immunol. 2023; 19(1): 71. Article. IF: 2.6; Q2
- **Zirngibl M, Buder K, Luithle T, Tönshoff B, Weitz M, (Espinosa Román L).** Diagnostic and therapeutic management of vesico-ureteral reflux in pediatric kidney transplantation-Results of an online survey on behalf of the European Society for Paediatric Nephrology. Pediatr Transplant. 2023; 27(2): e14449. Article. IF: 1.2; Q3
- **Zurowska A, Drozynska-Duklas M, Topaloglu R, Bouts A, Boyer O, Shenoy M, Vivarelli M, (Melgosa Hijosa, M).** Rituximab-associated hypogammaglobulinemia in children with idiopathic nephrotic syndrome: results of an ESPN survey. Pediatr Nephrol. 2023; 38(9): 3035-42. Article. IF: 2.6; Q1

Research projects

- **Sánchez-Corral Gómez P.** Contrato Miguel Servet Categoria A (CES07/030). CM. 2008-2025. 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 (PI19/00970). ISCIII. 2020-2024. Managment centre: FIBHULP
- **López Trascasa M.** Estudio de las bases moleculares 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 proteolisis del FH del Complemento y relevancia en la patologia del Síndrome Hemolítico-Urémico Atípico. Fundación SENEFRO. 2022-Ongoing. Managment centre: FIBHULP
- **Sánchez-Corral Gómez P.** Caracterización de patrones de expresión del FH y proteínas FHRS del complemento mediante western-blot e inteligencia artificial. correlación genotipo-fenotipo en patología renal y ocular (PI22/00211). ISCIII. 2023-2025. Managment centre: FIBHULP
- **Sánchez-Corral Gómez P.** El sistema del complemento en salud y enfermedad (Complemento III-CM) (P2022/BMD-7278). CM. 2023-Ongoing. Managment centre: FIBHULP

Cibers and Retics

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

Clinical trials

- **Espinosa Román, Laura.** Estudio con enmascaramiento doble, controlado con placebo, de aumento progresivo de la dosis para evaluar la eficacia, la seguridad y la farmacocinética de la voclosporina en adolescentes con nefritis lúpica.
  Type/Phase: III
  Sponsored by: Aurinia Pharmaceuticals Inc
  Signed date: 06/01/2023
- **López Lera, Alberto.** Identificación y caracterización funcional de variantes genéticas asociadas a fenotipos clínicos específicos en angioedema hereditario por déficit de C1 inhibidor.
  Type/Phase: No EPA
  Sponsored by: Fundacio Hospital Vall D'Hebron
  Signed date: 28/07/2023

Patents and trademarks

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

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