

1

Introduction

2

Executive
summary

3

Research
areas
and groups

4

Associated
clinicians



3.5.9 Human Genetic Variability

Publications: 13 | Q1: 5

COMPOSITION

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Catedrática de Genética del Departamento de Medicina. Universidad Europea de Madrid

- **Beatriz Antón Pascual.** *Médico Adjunto en Oncología Médica. Hospital Universitario Doce de Octubre*
- **Alberto Carretero González.** *Facultativo Especialista de Área en Oncología Médica. Hospital Universitario de Getafe*
- **María Gaibar Alonso.** *Profesora Titular. Universidad Camilo José Cela*
- **Diego Malón Jiménez.** *Médico Adjunto en Oncología Médica. Hospital Universitario de Fuenlabrada*
- **Apolonia Novillo Villajos.** *Profesora Permanente Laboral. Universidad Complutense de Madrid*
- **José María Rodríguez Piñas.** *Profesor Adjunto. Universidad Europea de Madrid*
- **Alicia Romero Lorca.** *Catedrática de Bioquímica y Biología Molecular. Universidad Europea de Madrid*
- **David Sanz Rosa.** *Director de Investigación. Universidad Europea de Madrid*

STRATEGIC OBJECTIVES

- The ultimate goal of our group is to contribute to pharmacogenetics and precision medicine. We aim to identify genetic biomarkers that can be used to predict which patients will experience the best response, benefit, and outcome, and the least toxicity, to specific drugs.

Planned research projects:

- Genetic biomarkers of toxicity in cancer immunotherapy
- Genetic biomarkers of efficacy in cancer immunotherapy
- Global genetic variability of polymorphisms associated with COVID severity

Transfer of results:

- Contributions to national and international conferences
- Publications in JCR journals
- Training and education program:
- Students of medicine, nursing, etc., will be able to join the group as doctoral students

Diffusion:

- Publications in JCR journals and at national and international conferences
- Guides for doctors

Internationalization:

- The team will apply for European programs when specific programs related to our research become available. Quality program for gender equality: Gender equality is achieved in our group because women and men have the same conditions and opportunities to belong to and work with us.

RESEARCH LINES

- Pharmacogenetics and cancer:
- Pharmacogenetics in breast cancer
- Pharmacogenetics in colorectal cancer
- Pharmacogenetics and immunotherapy
- Genetic variability of human populations





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RESEARCH ACTIVITY

Final Degree Theses

- **Simón Zuquin PS.** Del gen al tratamiento: explorando el rol de las mutaciones en PBRM1 y su impacto en la inmunoterapia del carcinoma renal. [dissertation]. MADRID: UCM: 2025(23/01/2025). Director: Novillo Villajos A.
- **Castro Sánchez-Clemente S.** Análisis de mutaciones en KMT2D en pacientes con cáncer de vejiga y su repercusión en inmunoterapia.[dissertation]. MADRID: UCM: 2025(30/05/2025). Director: Novillo Villajos A.

Publications

- **Farinea G, Antón-Pascual B, Villa JLC, Riesco-Martínez MD.** Change in MMR status as a mechanism of acquired resistance to immunotherapy in a dMMR colorectal cancer. *BMJ Case Rep.* 2025; 18(12): e264115. Article. IF: 0.4; Q4
- **Neil-Sztramko SE, Bland KA, Alibhai SMH, Capozzi LC, Chalmers A, Copp J, Dinardo R, Gimenez DM, Dufresne SS, Lambert LK, Langelier DM, Lane KN, Mackenzie K, McDonald K, Mcneely ML, Nadler MB, Papadakis J, Prestley N, Simmons C, Ashbury FD, Hart NH, Campbell KL.** The Bone Metastases and Exercise (BME) Hub: a MASCC-endorsed knowledge mobilization initiative to advance exercise recommendations for people with bone metastases. *Support Care Cancer.* 2025; 33(12): 1073. Editorial Material. IF: 3.4; D1
- **Capdevila J, Hernando J, Molina-Cerrillo J, Viñuales MB, García-Carbonero R, Teulé A, Custodio A, Jiménez-Fonseca P, López C, Hierro C, Carmona-Bayonas A, Alonso V, Llanos M, Sevilla I, García-Álvarez A, Alonso-Gordoa T, Jiménez IG, Antón-Pascual B, Sánchez AM, Grande E.** Cabozantinib plus atezolizumab in advanced, progressive endocrine malignancies: a multicohort, basket,

phase II trial (CABATEN/GETNE-T1914). *Clin Cancer Res.* 2025; 31(22): 4655-63. Article. IF: 10.9; D1

- **Pathak N, Di Iorio M, Giménez DM, Berner-Wygoda Y, Savill J, Aljuhani A, Mittal A, Kumar V, Amir E.** Adverse effect of trastuzumab deruxtecan in solid tumours: A systematic review and meta-analysis. *Crit Rev Oncol Hemat.* 2025; 213: 104787. Review. IF: 6.2; Q1
- **Rodríguez-Piñas JM, Romero-Lorca A, Gaibar M, Novillo A, Rubio M, Malón D, Antón-Pascual B, Camacho FI, Cavanagh M, Luján DR, Martín AM, Khedaoui R, Moreno D, Pinedo F, Boiza M, García-Adrián S, Gómez P, Marrupe D, Fernández-Santander A.** Tumour SNPs associated with immune-related hepatitis in patients with melanoma receiving immune checkpoint inhibitors. *Biomedicine.* 2025; 13(10): 2351. Article. IF: 4.5; Q2
- **Chang K, Delavan HM, Yip E, Kasap C, Zhu J, Lodha R, Liao SY, Berman SC, Carretero-González A, Basar M, Ozcan GG, Teo MY, Solit DB, Rosenberg JE, Al-Ahmadie H, Ding CCK, Chan EMY, Steri V, Porten SP, Koshkin VS, Friedlander TW, Feng FY, Lee JK, Wiita AP, Chu CE, Chou J.** Modulating the PPAR γ pathway upregulates NECTIN4 and enhances chimeric antigen receptor (CAR) T cell therapy in bladder cancer. *Nat Commun.* 2025; 16(1): 8215. Article. IF: 18.1; D1
- **Berner-Wygoda Y, Amir E, Blumenfeld P, Li M, Malon D, Pathak N, Savil J, Zarbiv Y, Kumar V.** Efficacy and safety of metastatic directed treatment of oligometastatic disease: results of a meta-analysis. *Adv Radiat Oncol.* 2025; 10(7): 101797. Review. IF: 2.9; Q2
- **Berner-Wygoda Y, Li M, Perlou C, Malon D, Pathak N, Di Iorio M, Savill J, Aljuhani A, Nadler MB, Amir E.** Safety and effectiveness of adriamycin and cyclophosphamide followed by dose dense (every 2 weeks) paclitaxel given with

trastuzumab as neoadjuvant treatment in her2-positive early-stage breast cancer. *Clin Breast Cancer.* 2025; 25(6): e750-5. Article. IF: 2.7; Q3

- **Rubio-Cuesta B, Carretero-Puche C, Llamas P, Sarmiento J, Gil-Calderón B, Lens-Pardo A, Antón-Pascual B, Rubio-González E, Cámara-Jurado M, Salamanca J, Rueda-Fernández D, Delcuratolo MD, Soldevilla B, García-Carbonero R.** Co-targeting SRC overcomes resistance to BRAF inhibitors in colorectal cancer. *Brit J Cancer.* 2025; 133(3): 404-19. Article. IF: 7.8; Q1
- **Pathak N, Kumar S, Giménez DM, Di Iorio M, Savill J, Berner-Wygoda Y, Li M, Valiente CM, Cuthbert D, Gupta A, Arteaga DP, Batra A, Amir E, Mittal A.** Continuing cyclin-dependent kinase 4/6 inhibitors beyond progression in advanced breast cancer: a meta-analysis. *Cancers (Basel).* 2025; 17(10): 1609. Review. IF: 4.8; Q2
- **del Olmo-García M, Hernández-Rienda L, García-Carbonero R, Hernando J, Custodio A, Antón-Pascual B, Gomez M, Milla SP, Suárez L, Bellver M, Alonso V, Serrano R, Valdés N, Melián M, Febrero B, Sampedro-Nuñez MA, Biarnes J, Díaz-Pérez JA, Molina-Cerrillo J, López C, Olmos MAM, Merino-Torres JF, Capdevila J, Pla MA, NUTRIGETNE Grp.** Nutritional status and quality of life of patients with advanced gastroenteropancreatic neuroendocrine neoplasms in Spain: the NUTRIGETNE (GETNE-S2109) study. *Oncologist.* 2025; 30(2): oyae343. Article. IF: 4.7; Q2
- **Lee CL, Martínez E, Gimenez DM, Muniz TP, Butler MO, Saibil SD.** Female oncofertility and immune checkpoint blockade in melanoma: where are we today? *Cancers (Basel).* 2025; 17(2): 238. Review. IF: 4.8; Q2
- **Malón D, Molto C, Prasla S, Cuthbert D, Pathak N, Berner-Wygoda Y, Di Iorio M, Li M, Savill J, Mittal A, Amir E, Jhaveri K, Nadler**

MB. Steatotic liver disease in metastatic breast cancer treated with endocrine therapy and CDK4/6 inhibitor. *Breast Cancer Res Tr.* 2025; 210(2): 405-16. Article. IF: 3.3; Q2

Research projects

- **Antón Pascual B.** Evaluación del impacto de la práctica ejercicio físico en salud osteomuscular, emocional y tumoral en pacientes con tumores neuroendocrinos gastroenteropancreáticos u origen desconocido avanzados: FIT-NETs. NET España. 2023-2025. **Managment centre:** Hospital 12 de Octubre
- **Gaibar Alonso M.** Análisis de la expresión de microRNAs como posibles biomarcadores de toxicidad en inmunoterapia en cáncer genitourinario. Universidad Camilo José Cela. 2024-2025. **Managment centre:** Universidad Camilo Jose Cela
- **Fernández-Santander A.** Estudio de polimorfismos de tipo SNP y CNV como posibles marcadores de toxicidad en inmunoterapia aplicada a tumores genitourinarios. FIBHULP. 2024-2025. **Managment centre:** IdiPAZ
- **Fernández-Santander A.** NALUNG – Multi-Omics Analysis for Predicting Response and Adverse Events to Neoadjuvant Treatment in Resectable NSCLC (EPPERMED2025-267-NALUNG). FIBHULP. 2025-2028. **Managment centre:** FIBHULP
- **Antón Pascual B.** Evaluación del impacto de la práctica ejercicio físico en salud osteomuscular, emocional y tumoral en pacientes con tumores neuroendocrinos gastroenteropancreáticos u origen desconocido avanzados: FIT-NETs. NET España. 2023-2025. **Managment centre:** Hospital 12 de Octubre