



Pablo Mínguez

BIOINFORMATICIAN. Principal Investigator at Genetics Department & Head of Bioinformatics Unit – IIS-Fundación Jiménez Díaz (Madrid)
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Computational biologist interested in gene and protein regulation and their disorders under disease stages. I apply mathematical, statistical and computational methods to study complex systems of regulation from a Systems Biology perspective. Mainly involved in Genomics of Rare Diseases and Personalized Medicine.

WORK EXPERIENCE

- ✓ **January 2017 – present.** Principal Investigator. Genetics Department, IIS-FJD, Madrid, Spain).
- ✓ **September 2015 – present.** Head of the Bioinformatics Unit (IIS-FJD, Madrid, Spain).
- ✓ **September 2009 - January 2015.** Postdoctoral fellow at Bork group (EMBL, Heidelberg).
- ✓ **September 2004 - August 2009.** PhD student & Bridging postdoc (CIPF, Valencia, Spain).
- ✓ **January 2004 - September 2004.** Research associate (Bioinformatician). (European Arabidopsis Stock Centre (University of Nottingham, UK).
- ✓ **June 2003 - October 2003.** Research assistant (Bioinformatician). John Innes Center (Norwich, UK).

EDUCATION

- ✓ **31st October 2008.** PhD in Molecular Biology (Bioinformatics). Universidad Autonoma de Madrid (Madrid, Spain). Thesis Title: Functional profiling of genome-scale experiments. New approaches leading to a systemic analysis.
- ✓ **September 2002 - October 2003.** MSc in Bioinformatics. University of East Anglia (Norwich, UK). Grade Distinction.
- ✓ **September 2000 - September 2001.** Postgraduate Certificate in Education (subject Biology and Geology). Universidad Complutense (Madrid, Spain).
- ✓ **September 1994 - February 2000.** BSc Biological Sciences (Microbiology and environmental biology). Universidad de Alcalá (Madrid, Spain).

COMPLEMENTARY COURSES

- ✓ **12-13 May 2025.** Scientific Communication and Outreach Course. CIBER, ISCIII, Madrid.
- ✓ **March-May 2023.** Entrepreneurship program for Health researchers. UAM, Madrid.
- ✓ **4-8 October 2021.** EMBO Practical course: Research to service: Planning and running a bioinformatics core facility.

MERITS

- ✓ **2023.** R3 Certificate, as independent and established researcher. National Research Agency, Spanish Ministry of Science and Innovation (MICINN).
- ✓ **2022.** Corresponding Academic at the Real Academia Nacional de Medicina (Spanish National Academia of Medicine).

- ✓ **2021.** Miguel Servet II. ISCIII, AES2021.
- ✓ **2017.** Miguel Servet I. ISCIII, AES2017.
- ✓ **2011.** Marie Curie Postdoctoral Fellowship award. Call FP7-PEOPLE-2010-IEF.
- ✓ **2009.** Postdoctoral Fellowship award. Fellowship Programme for Specialization in International Organizations, granted by the MICINN.
- ✓ **2004.** Predoctoral Fellowship award. Genome Canada PhD Fellowship.
- ✓ **2003.** Scholarship Award. MSc dissertation Fellowship (John Innes Centre Norwich, UK).

SCIENTIFIC PUBLICATIONS

124 publications. H-index of 39 and more than > 15,500 citations (source: Google scholar, May-2026). See complete records at: [PUBMED](#), [GOOGLE SCHOLAR](#) & [ORCID](#)

TOP 10 PAPERS

1. Benítez Y, Uría-Regojo G, **Mínguez P#**. A global survey of systems biology-based predictions of gene-rare disease associations to enhance new diagnoses. Sci Rep. 2026. **IF=3.9**
2. Núñez-Moreno G, Tamayo A, Ruiz-Sanchez C, Cortón M#, **Mínguez P#**. VISOQLR: an interactive tool for the detection, quantification and fine-tuning of isoforms in selected genes using long-read sequencing. Human Genetics. 2023; 142(4):495-506. **IF=5.881**
3. de la Fuente L, Del Pozo-Valero M, Perea-Romero I, Blanco-Kelly F, Fernández-Caballero L, Cortón M, Ayuso C, **Mínguez P#**. Prioritization of New Candidate Genes for Rare Genetic Diseases by a Disease-Aware Evaluation of Heterogeneous Molecular Networks. Int J Mol Sci. 2023 Jan 14;24(2). **IF=6.208**
4. Iancu IF, Perea-Romero I, Núñez-Moreno G, de la Fuente L, Romero R, Ávila-Fernandez A, Trujillo-Tiebas MJ, Riveiro-Álvarez R, Almoguera B, Martín-Mérida I, Del Pozo-Valero M, Damián-Verde A, Cortón M, Ayuso C#, **Mínguez P#**. Aggregated Genomic Data as Cohort-Specific Allelic Frequencies can Boost Variants and Genes Prioritization in Non-Solved Cases of Inherited Retinal Dystrophies. Int J Mol Sci. 2022 Jul 29;23(15). **IF=6.208**
5. Romero R, de la Fuente L, del Pozo-Valero M, Riveiro-Álvarez R, Trujillo-Tiebas MJ, Martín-Mérida I, Ávila-Fernández A, Iancu IF, Perea-Romero I, Núñez-Moreno G, Damián-Verde A, Rodilla C, Almoguera B, Cortón M, Ayuso C, **Mínguez P#**. An evaluation of pipelines for DNA variant detection can guide a reanalysis protocol to increase the diagnostic ratio of genetic diseases. NPJ genomic medicine. 2021. In press. **IF=6.083**
6. Vellosillo P, **Mínguez P#**. A global map of associations between types of protein posttranslational modifications and human genetic diseases. IScience. **2021**;24(8):102917. **IF=6.107**
7. Iancu IF, Avila-Fernandez A, Arteché A, Trujillo-Tiebas MJ, Riveiro-Alvarez R, Almoguera B, Martín-Mérida I, Del Pozo-Valero M, Perea-Romero I, Cortón M, **Mínguez P#**, Ayuso C#. Prioritizing variants of uncertain significance for reclassification using a rule-based algorithm in inherited retinal dystrophies. npj Genomic Medicine. **2021**; 6(1):18. **IF=6.083**
8. Del Puerto-Nevado L*, **Mínguez P*#**, Cortón M, Solanes-Casado S, Prieto I, Mas S, Sanz AB, Gonzalez-Alonso P, Villaverde C, Portal-Núñez S, Aguilera O, Gomez-Guerrero C, Esbrit P, Vivanco F, Gonzalez N, Ayuso C, Ortiz A, Rojo F, Egido J, Alvarez-Llamas G#, Garcia-

Foncillas J. Molecular evidence of field cancerization initiated by diabetes in colon cancer patients. *Molecular Oncology*. **2019**; 13(4):857-872. **IIF=7.449**

9. **Minguez P***, Ivica L*, Parca L, Bork P. "*PTMcode: a database of known and predicted functional associations between post-translational modifications in proteins.*" *Nucl. Acids Res*. **2013**;41(Database issue):D306-11. **IF=16.971**
10. **Minguez P**, Parca L, Diella F, Mende DR, Kumar R, Helmer-Citterich M, Gavin AC, van Noort V, Bork P. "*Deciphering a global network of functionally associated post-translational modifications*" *Molecular Systems Biology*. **2012**; 8:599. **IF=11.429**

BOOK CHAPTERS & REVIEWS

1. Del Puerto-Nevado L*#, **Minguez P*#**, Garcia-Foncillas J. Bioinformática, inteligencia artificial y sistemas cognitivos en medicina. Colección de Bioética y Derecho Sanitario Fundación Merck. 2020; Monografía 25.
2. **Minguez P#**, Bork P. Bioinformatics Analysis of Functional Associations of PTMs. *Methods in molecular biology* (Clifton, N.J.). **2017**; 1558:303-320.
3. **Minguez P**, Dopazo J. "*Functional genomics and networks: new approaches in the extraction of complex gene modules.*" *Expert Rev Proteomics*. **2010**;7(1):55-63.
4. **Minguez P**, Dopazo J. "*Protein Interactions for Functional Genomics.*" . *Biological Data Mining in Protein Interaction Networks*. Hershey, USA: Idea Group Inc (IGI); **2009**.

SIGNIFICANT SCIENTIFIC PROJECTS

- ✓ **PMP24/00024 (IMPACT-Data 2)** - "Plataforma Digital de IMPACT para la captura, integración y modelado de datos biomédicos asociados a la Cohorte y proyectos IMPACT-PMP". ISCIII. 2025-2027. **Principal Investigator IIS-FJD team.**
- ✓ **RED2024-153760-E** - EXPANSION DEL INSTITUTO NACIONAL DE BIOINFORMATICA (INB) PARA INCREMENTAR SU IMPACTO INTERNACIONAL. Ministerio de Ciencia, Innovación y Universidades. REDES 2024. **Principal Investigator IIS-FJD team.**
- ✓ **PI22/00579** - DB4DISCOVERY. Desarrollo de métodos de priorización y descubrimientos de variantes mediante el reuso de información genómica agregada en una base de datos específica de una cohorte. ISCIII. 2023-2025. **Principal Investigator.**
- ✓ **RRI22/00005** - Caminando hacia la Ciencia Abierta: implementación del acceso abierto a los datos ómicos en el IIS-FJD. ISCIII, Convocatoria de Investigación e Innovación. **Principal Investigator.**
- ✓ **ACCI2022** - Desarrollo de un repositorio de flujos de trabajo de análisis genómicos. CIBERER, ISCIII. **Principal Investigator IIS-FJD team.**
- ✓ **IMP/00019** - Infraestructura de Medicina de Precisión asociada a la Ciencia y Tecnología (IMPACT). **Data Science**. ISCIII. 2021-2023. **Principal Investigator IIS-FJD team.**
- ✓ **IMP/00009** - Infraestructura de Medicina de Precisión asociada a la Ciencia y Tecnología (IMPACT). **Genomic Medicine**. ISCIII. 2021-2023. **Coordinator of the data sharing/data discovery working group.**
- ✓ **UnCOVer (Grant agreement ID: 101016216)** - Unravelling data for rapid evidence-based response to COVID-19. European Commission, H2020. 15/11/2020-14/11/2022. Project team member.

- ✓ **COV20_00181.** STOP-CORONAVIRUS: Factores clínicos, inmunológicos, genómicos, virológicos y bioéticos de COVID-19. ISCIII. 01/05/2020 – 30/04/2021. Project team member.
- ✓ **PID10291.** Deciphering the molecular basis of ophtalmogenetic diseases: sequencing the whole genome using a long reads approach (WGS4EyeRare). H2020 (EasiGenomics program). 2020-2021. Project team member.
- ✓ **ACCI 2019** - Bases metodológicas y organizativas para Mejorar la Eficiencia en el Diagnóstico de pacientes con Enfermedades raras No Diagnosticadas mediante medicina genómica (MEDENoD). CIBERER, ISCIII. 2020-2021. Project team member.
- ✓ **RED2018-102404-T** - Afianzando la red bioinformatica traslacional TRANSBIONET. Ministerio de Ciencia, Innovación y Universidades. 2020-2021. Project team member.
- ✓ **PI18/00579** - Desarrollo de algoritmos bioinformáticos basados en redes y Biología de Sistemas para la búsqueda de nuevos genes responsables de enfermedades raras de origen genético. ISCIII, AES2018. **Principal Investigator.**
- ✓ **CP16/00116** - Dissecting the role of protein post-translational modifications in human diseases. ISCIII, Miguel Servet project. AES2016. **Principal Investigator.**
- ✓ **S2017/BMD-3721** - Red de recursos genomicos, funcionales, clínicos y terapéuticos para el estudio de las enfermedades raras neurológicas. C.A. de Madrid. Project team member.
- ✓ **CIVP18A3862** - A knowledge-based pipeline to increase the diagnosis rate of Rare Diseases using deep sequencing. Fundación Ramón Areces 2017. Project team member.
- ✓ **PIE13/00051.** Diabetes Cancer Connect: Targeting common inflammatory markers and signaling pathways in diabetes mellitus and cancer. ISCIII. Proyecto Integrado IIS Projects Call del ISCIII (2014-2016). Project team member.
- ✓ **IntoAct – FP7-PEOPLE.** Assessment of the regulatory mechanisms of different protein post-translational modifications by studying their role within a realistic in silico model of the cell. **Principal Investigator, Marie Curie Fellow.** FP7- PEOPLE-2010-IEF.

TEACHING EXPERIENCE

- ✓ Teaching subject “Omics data analysis”, Master in Bioinformatics, UAX.
- ✓ Teaching in Master in Bioinformatics, Universidad de Valencia. Subject: Estudios *in silico* en Biomedicina.
- ✓ Teaching in Master in Bioinformatics applied to Personalized Medicine and Health, Escuela Nacional de Sanidad and ISCIII. Subject: Analysis of omics data using networks.
- ✓ Coordination and teaching in the training activities of the Bioinformatics Unit at the IIS-FJD: course “Introducción al análisis de datos ómicos”, 10-11 october 2022.
- ✓ Universidad Internacional Menéndez Pelayo Summer course 2023 – Rare diseases.
- ✓ Regular one day Seminars at Bioinformatics Masters. Universidad Politécnica de Madrid, Universidad Autónoma de Madrid.
- ✓ Several workshops at the IIS-FJD: VII Curso en Avances en Nefrología Clínica, II Jornadas UAM y VIH.
- ✓ **Other Bioinformatics courses.** Introducción a análisis ómicos (IIS-FJD). DiGtoP- Bioinformatics Course on Protein Interactions and Networks (EMBL, Heidelberg, Germany), International Course of Massive Data Analysis (CIPF, Valencia, Spain).

TUTOR EXPERIENCE

- ✓ **Supervised dissertations: 2 PhDs:** 1) *Estudio del papel de las modificaciones de proteínas en enfermedades genéticas*. Student: Perceval Velloso Gonzalez. Director: Pablo Mínguez. UAM, 2022. 2) *Caracterización molecular, epidemiológica y funcional de variantes de significado incierto (VUS) en distrofias de retina, mediante el empleo de métodos bioinformáticos y de Biología de Sistemas*. Student: Ionut-Florin Iancu. Directors: Pablo Mínguez/Carmen Ayuso. UAM, 2022. 3 **ongoing PhD thesis (UAM)**: Graciela Uría Regojo (PFIs), Lucía López López (Conchita Rábago), Yolanda Benítez (Bioinfo-ISCI).
- ✓ **9 Master Thesis** (2018-2024) Master in Bioinformatics, ISCI/ENS, UAM, UOC, Université Rennes 1
- ✓ **6 Final Degree Projects** (2018-2025) Grado de Biotecnología, UPM.

BIOINFORMATICS SOFTWARE & DATABASES

- ✓ Collaborative DNaseSeq pipeline (<https://github.com/CIBERER/CBRA>).
- ✓ DNaseSeq pipeline (<https://github.com/TBLabFJD/PARROT-FJD>).
- ✓ Long Reads DNaseSeq pipeline (<https://github.com/TBLabFJD/WHALE>).
- ✓ VISOQLR (<https://github.com/TBLabFJD/VISOQLR>).
- ✓ PriorR (<https://github.com/TBLabFJD/PriorR>).
- ✓ GLOWgenes (www.glowgenes.org). Prediction of gene-disease associations.
- ✓ Machine Learning (<https://github.com/pminguez/MachineLearning4UnbalancedData>).
- ✓ PTMcode (<http://ptmcode.embl.de>): Database of known and predicted functional associations between protein posttranslational modifications.
- ✓ BABELOMICS (<http://babelomics.org>): Several tools: SNOW, Tissues Mining Tool (former versions), MarmiteScan & Marmite (former versions).

OTHER ACTIVITIES

- ✓ Member of the Real Academia Nacional de Medicina (https://bit.ly/PM_RANM).
- ✓ Member of the National Mirror Group 5 of the B1MG project.
- ✓ Member of collaboration networks: CIBERER, TransBioNet, IMPaCT-Data, IMPaCT-Genómica, RAREGenomics.
- ✓ Congresses/Seminars/Courses organizer: SEBiBC2026 (<https://www.congresosebirc.com/index.php?go=comite>), JBI2025 (<https://jbi2025.cnbcsc.es/committee>), BioinfoCAM (<https://bit.ly/BioinfoCAM2022>), Omics Course (<https://bit.ly/OmicsCourse>), Science week activities (www.semanacienciamadrid.org) and more.
- ✓ Reviewer in several Journals (Nature Communications, Briefings in Bioinformatics, NAR, Bioinformatics, PLoS Comp Bio, and others).
- ✓ Projects' reviewer (ISCI, CAM, Junta Andalucía, ISMB).
- ✓ Member of the Bioinformatics Spanish Society (SEBiBC).
- ✓ Associate editor in journals: International Journal of Molecular Sciences (section Molecular Informatics); Frontiers in Cell and Developmental Biology (section Molecular and Cellular Pathology); Frontiers in Bioinformatics (section Protein Bioinformatics).