Part A. PERSONAL INFORMATION

First Name	Joaquin		
Family Name	Dopazo Blazquez		
Sex	Male	Date of Birth	24/01/1961
ID number Social Security, Passport	24317827I		
URL Web	http://wikis.babelomics.org/jdopazo/		
Email Address	joaquin.dopazo@juntadeandalucia.es		
Open Researcher and Contributor ID (ORCID)		0000-0003-3318-120X	

A.1. Current position

Job Title	Area Director	
Starting date	2016	
Institution	FUNDACION PUBLICA ANDALUZA PROGRESO Y SALUD	
Department / Centre	Area de Bioinformatica Clinica / Fundacion Progreso y Salud.	
Country		Phone Number	
Keywords	Molecular mechanism of disease; Computational biology; Genetics	

A.3. Education

Degree/Master/PhD	University / Country	Year
Doctor en Ciencias Biologicas	Universitat de València	1989
Licenciado en Ciencias Químicas Sección Bioquímica	Universitat de València	1985

Part B. CV SUMMARY

Joaquín Dopazo obtained his PhD in Biology at the University of Valencia in 1989. After several appointments in different research centers and companies he worked for 5 years in Glaxo Wellcome (now Glaxo SmithKline) during the late nineties. There he was in charge of the Bioinformatics unit of the Spanish node, developing methods for bacterial genomic analysis and he participated in several bacterial and fungal genome sequencing projects. In particular, Dr. Dopazo coordinated the assembly and annotation of the bacterial pathogen Streptococcus pneumoniae (Dopazo, 2001, Micr. Drug Res.). In 2000 he moved to the Spanish National Cancer Center (CNIO), where he set up the Bioinformatics group. In the CNIO he coordinated the design of the first Spanish microarray (the Oncochip) in 2000 and he developed the one of the most used resources for transcriptome data analysis and interpretation on the web, the Babelomics. In 2005 Dr. Dopazo moved to the CIPF (Valencia) where he set up the Department of Computational Genomics (formerly Bioinformatics). He served as Scientific director of the Center during 2012. In 2017 He moved to the Clinical Bioinformatics Research Area from the Fundacion Progreso y Salud, In Sevilla, where he coordinates, within the Andalucian Personalized Medicine Program, the introduction of genomic data in the patient electronic health record (eHR). In addition, he heads the Bioinformatics node (BiER) of the CIBERER, where he coordinates a pilot project with seven hospitals across Spain to collect patients genomic data with an advanced software of prioritization (bierapp.babelomics.org). He also heads the node of functional genomics of the Spanish National Institute of Bioinformatics.

The scope of his research has evolved in parallel to the introduction of "Big Data" in the life sciences. He has been working on genomic data integration and, specifically, during the last years, focusing on massive sequence data analysis. Dr. Dopazo's scientific interests revolve around functional genomics, systems biology, artificial intelligence and development of algorithms and software for the analysis of high-throughput data (mainly, but not restricted to, Next Generation Sequencing) and its application to personalized and precision medicine. He is particularly interested in studying disease mechanisms and drug action mechanisms by

modeling complex cellular systems in which signaling, regulation and metabolism knowledge are integrated (see an example with hipathia.babelomics.org/).

He has promoted and coordinated genomic projects such as the Medical Genome Project (www.medicalgenomeproject.com), that sequenced over 1000 patients of inherited diseases to search for new biomarkers and disease genes. He was also involved in international projects such as the MAQC and SEQC (best practices in the use of NGS for finding diagnostic biomarkers).

Part C. RELEVANT ACCOMPLISHMENTS

C.1. Most important publications in national or international peer-reviewed journals, books and conferences

AC: corresponding author. (nº x / nº y): position / total authors. If applicable, indicate the number of citations

1 **Scientific paper**. Pena-Chilet, Maria; Roldan, Gema; Perez-Florido, Javier; et al; Dopazo, Joaquin. 2021. CSVS, a crowdsourcing database of the Spanish population genetic variability.Nucleic acids research. 49-D1, pp.D1130-D1137. ISSN 1362-4962. WOS (0)

2 **Scientific paper**. Lopez-Sanchez, Macarena; Loucera, Carlos; Pena-Chilet, Maria; Dopazo, Joaquin. 2022. Discovering potential interactions between rare diseases and COVID-19 by combining mechanistic models of viral infection with statistical modeling Human Molecular Genetics. Oxford.

3 **Scientific paper**. Gundogdu, Pelin; Loucera, Carlos; Alamo-Alvarez, Inmaculada; Dopazo, Joaquin; Nepomuceno, Isabel. 2022. Integrating pathway knowledge with deep neural networks to reduce the dimensionality in single-cell RNA-seq data BioData Mining. BioMed Central. 15-1, pp.1-21.

4 **Scientific paper**. Casimiro-Soriguer, Carlos S; Loucera, Carlos; Pena-Chilet, Maria; Dopazo, Joaquin. 2022. Towards a metagenomics machine learning interpretable model for understanding the transition from adenoma to colorectal cancer Sci Rep. Nature Publishing Group. 12-1, pp.450-450.

5 **Scientific paper**. Ostaszewski, Marek; Niarakis, Anna; Mazein, Alexander; et al; others. 2021. COVID19 Disease Map, a computational knowledge repository of virus--host interaction mechanisms Molecular systems biology. 17-10, pp.e10387-e10387.

6 **Scientific paper**. Rian, Kinza; Hidalgo, Marta R.; Cubuk, Cankut; et al; Dopazo, Joaquin. 2021. Genome-scale mechanistic modeling of signaling pathways made easy: A bioconductor/cytoscape/web server framework for the analysis of omic data COMPUTATIONAL AND STRUCTURAL BIOTECHNOLOGY JOURNAL. 19, pp.2968-2978. ISSN 2001-0370. WOS (0)

7 **Scientific paper**. Carlos Loucera; Marina Esteban-Medina; Kinza Rian; Matias Falco; Joaquin Dopazo; Maria Peña-Chilet. 2020. Drug repurposing for COVID-19 using machine learning and mechanistic models of signal transduction circuits related to SARS-CoV-2 infection Signal Transduction and Targeted Therapy. Nature. 5-1, pp.290.

8 **Scientific paper**. Martin-Broto, Javier; Hindi, Nadia; Grignani, Giovanni; et al; Lopez-Martin, Jose A. 2020. Nivolumab and sunitinib combination in advanced soft tissue sarcomas: a multicenter, single-arm, phase Ib/II trial.Journal for immunotherapy of cancer. 8-2. ISSN 2051-1426. WOS (0)

9 **Scientific paper**. Haibe-Kains, Benjamin; Adam, George Alexandru; Hosny, Ahmed; et al; Aerts, Hugo J. W. L.2020. Transparency and reproducibility in artificial intelligence NATURE. 586-7829, pp.E14-+. ISSN 1476-4687. WOS (1)

10 **Scientific paper**. Lopez-Lopez, Daniel; Loucera, Carlos; Carmona, Rosario; et al; Dopazo, Joaquin. 2020. SMN1copy-number and sequence variant analysis from next-generation sequencing data HUMAN MUTATION. WILEY. 41-12, pp.2073-2077. WOS (0)

11 **Scientific paper**. Cubuk, Cankut; Can, Fatma E; Pena-Chilet, Maria; Dopazo, Joaquin. 2020. Mechanistic Models of Signaling Pathways Reveal the Drug Action Mechanisms behind Gender-Specific Gene Expression for Cancer Treatments.Cells. 9-7. WOS (0)

12 <u>**Scientific paper**</u>. Ostaszewski, Marek; Mazein, Alexander; Gillespie, Marc E.; et al; Schneider, Reinhard. 2020. COVID-19 Disease Map, building a computational repository of SARS-CoV-2 virus-host interaction mechanisms SCIENTIFIC DATA. 7-1. WOS (0)

13 <u>**Scientific paper**</u>. Martin-Broto, Javier; Cruz, Josefina; Penel, Nicolas; et al; Stacchiotti, Silvia. 2020. Pazopanib for treatment of typical solitary fibrous tumours: a multicentre, single-arm, phase 2 trial LANCET ONCOLOGY. 21-3, pp.456-466. ISSN 1470-2045. WOS (1)

14 <u>**Scientific paper**</u>. Pena-Chilet, Maria; Esteban-Medina, Marina; Falco, Matias M.; Rian, Kinza; Hidalgo, Marta R.; Loucera, Carlos; Dopazo, Joaquin. 2019. Using mechanistic models for the clinical interpretation of complex genomic variation SCIENTIFIC REPORTS. 9. ISSN 2045-2322. WOS (0)

15 <u>**Scientific paper**</u>. Esteban Medina, M.; Peña Chilet, M.; Loucera, C.; Dopazo, J.2019. Exploring the druggable space around the Fanconi anemia pathway using machine learning and mechanistic models.BMC bioinformatics. 20-1, pp.370. ISSN 1471-2105.

16 <u>**Scientific paper**</u>. Chacón Solano, E.; León, C.; Díaz, F.; et al; Del Río, M.2019. Fibroblasts activation and abnormal extracellular matrix remodelling as common hallmarks in three cancer-prone genodermatoses.The British journal of dermatology. 181-3, pp.512-522. ISSN 1365-2133.

17 <u>**Scientific paper**</u>. Cubuk, Cankut; Hidalgo, Marta R.; Amadoz, Alicia; Pujana, Miguel A.; Mateo, Francesca; Herranz, Carmen; Carbonell-Caballero, Jose; Dopazo, Joaquin. 2018. Gene Expression Integration into Pathway Modules Reveals a Pan-Cancer Metabolic Landscape CANCER RESEARCH. 78-21, pp.6059-6072. ISSN 0008-5472. WOS (0)

18 <u>**Scientific paper**</u>. Ferreira, PG.; Muñoz Aguirre, M.; Reverter, F.; et al; Guigó, R.2018. The effects of death and post-mortem cold ischemia on human tissue transcriptomes.Nature communications. 9-1, pp.490. ISSN 2041-1723. https://doi.org/10.1038/s41467-017-02772-x

19 <u>**Scientific paper**</u>. Roca Ayats, N.; Balcells, S.; Garcia Giralt, N.; et al; Díez Pérez, A.2017. GGPS1 Mutation and Atypical Femoral Fractures with Bisphosphonates.The New England journal of medicine. 376-18, pp.1794-1795. ISSN 1533-4406.

20 <u>**Scientific paper**</u>. Hidalgo, MR.; Cubuk, C.; Amadoz, A.; Salavert, F.; Carbonell Caballero, J.; Dopazo, J.2017. High throughput estimation of functional cell activities reveals disease mechanisms and predicts relevant clinical outcomes.Oncotarget. 8-3, pp.5160-5178. ISSN 1949-2553.

21 <u>**Scientific paper**</u>. Falco, MM.; Bleda, M.; Carbonell Caballero, J.; Dopazo, J.2016. The pan-cancer pathological regulatory landscape.Scientific reports. 6, pp.39709. ISSN 2045-2322.

22 <u>**Scientific paper**</u>. Salavert, F.; Hidago, MR.; Amadoz, A.; Çubuk, C.; Medina, I.; Crespo, D.; Carbonell Caballero, J.; Dopazo, J.2016. Actionable pathways: interactive discovery of therapeutic targets using signaling pathway models.Nucleic acids research. 44-W1, pp.W212. ISSN 1362-4962.

23 <u>**Scientific paper**</u>. Dopazo, J.; Amadoz, A.; Bleda, M.; et al; Antiñolo, G.2016. 267 Spanish Exomes Reveal Population-Specific Differences in Disease-Related Genetic Variation.Molecular biology and evolution. ISSN 1537-1719.

24 <u>**Scientific paper**</u>. Porta Pardo, E.; Garcia Alonso, L.; Hrabe, T.; Dopazo, J.; Godzik, A.2015. A Pan-Cancer Catalogue of Cancer Driver Protein Interaction Interfaces.PLoS computational biology. 11-10, pp.e1004518. ISSN 1553-7358.

25 <u>**Scientific paper**</u>. Hernansaiz Ballesteros, RD.; Salavert, F.; Sebastián León, P.; Alemán, A.; Medina, I.; Dopazo, J.2015. Assessing the impact of mutations found in next generation sequencing data over human signaling pathways.Nucleic acids research. 43-W1, pp.W270. ISSN 1362-4962.

C.3. Research projects and contracts

1 <u>**Project**</u>. Medicina interceptiva, descubrimiento de dianas y reutilización de fármacos mediante modelos mecanísticos y aprendizaje supervisado. (FUNDACION PUBLICA ANDALUZA PROGRESO Y SALUD). 01/09/2021-31/03/2024. 242.000 €.

2. **Project**. IMPACT Data. Alfonso Valencia. (Barcelona Supercomputing Center). 01/01/2022-31/12/2023. 4.549.380 €.
3. **Project**. PE-0002-2018, Desarrollo de un sistema bioinformático de apoyo a la decisión en medicina genómica de precisión para el tratamiento personalizado del cáncer en el sistema de salud. Consejería de Salud de la Junta de Andalucía. Joaquin Dopazo Blázquez. (FUNDACION PUBLICA ANDALUZA PROGRESO Y SALUD). 01/01/2019-31/12/2022. 196.586,03 €.
4. **Project**. 813533, MLFPM: Machine Learning Frontiers for Personalized Medicine. H2020. (FUNDACION PUBLICA ANDALUZA PROGRESO Y SALUD). 01/01/2019-31/12/2022.
5. **Project**. Modelo mecanístico basado en inteligencia artificial para la reutilización de fármacos contra la infección por SARS-CoV-. Instituto de Salud Carlos III. (FUNDACION PUBLICA ANDALUZA PROGRESO Y SALUD). 08/05/2020-07/02/2021. 57.000 €.
6. **Project**. COVID-0012-2020, Secuenciación del genoma del virus SARS-CoV-2 para el seguimiento y manejo de la epidemia del Covid-19 en Andalucía y la generación rápida de biomarcadores pronósticos y de respuesta a tratamiento. Consejería de Salud de la Junta de Andalucía. (FUNDACION PUBLICA ANDALUZA PROGRESO Y SALUD). 17/04/2020-16/09/2020. 103.000 €.
7. **Project**. 676559, ELIXIR-EXCELERATE implementation and drive early user exploitation across the life-sciences. EU H2020-INFRADEV-1-2015-1. Niklas Bloomberg. (Fundacion Publica Andaluza Progreso y Salud). 01/07/2015-31/07/2019.
8. **Project**. DTS16/00139, Desarrollo de un sistema integrado para la medicina personalizada orientado al diagnóstico y descubrimiento de genes de enfermedad usando secuenciación de nueva generación.. MINECO. (FUNDACION PUBLICA ANDALUZA PROGRESO Y SALUD). 01/01/2017-31/12/2017. 50.050 €.
9. **Project**. 316861, MLPM2012: Machine Learning for Personalized Medicine. EU FP7-PEOPLE Project. (Centro de Investigación Príncipe Felipe). 01/01/2013-31/12/2016. 3.758.057 €.
10. **Project**. Large-scale drug repurposing in rare diseases by genomic Big Data analysis with machine learning methods. FUNDACION BANCO BILBAO-VIZCAYA. Joaqui Dopazo. (FUNDACION PUBLICA ANDALUZA PROGRESO Y SALUD). From 01/06/2019. 94.300 €.
11. **Contract**. VALIDACIÓN DE LA SECUENCIACIÓN MASIVA COMO HERRAMIENTA PARA LA DECISIÓN DE TRATAMIENTO EN CÁNCER DE PULMÓN Y DESARROLLO DE UNA HERRAMIENTA BIOINFORMÁTICA PARA EL MANEJO DE LOS DATOS GENÓMICOS Consejería de Salud de la Junta de Andalucía. Joaquin Dopazo. (Fundacion Progreso y Salud). 01/11/2018-01/11/2020. 115.000 €.
12. **Contract**. NAGEN Proyecto Genoma Navarra. Proyecto Piloto sobre Uso Clínico de Medicina Genómica en Servicios Públicos de Salud de Navarra Fundacion Miguel Servet. Joaquin Dopazo Blázquez. 01/01/2018-01/01/2020. 133.000 €.
13. **Contract**. NAGEN Proyecto Genoma Navarra. Proyecto Piloto sobre Uso Clínico de Medicina Genómica en Servicios Públicos de Salud de Navarra Fundacion Miguel Servet. Joaquin Dopazo Blázquez. 01/01/2017-01/01/2018. 31.214 €.
14. **Contract**. Medical Genome Project Roche Diagnostics; Consejería de Salud, Junta de Andalucia. Joaquin Dopazo Blazquez. 16/04/2012-16/04/2014.

C.4. Activities of technology / knowledge transfer and results exploitation

1. Joaquin Dopazo Blazquez. 2107208395155. Mamapred Spain. 20/07/2021. Fundación Instituto valenciano de oncología. Durviz, Diagnóstico e Investigación, S.L.U.
2. J. Dopazo. ES200100045. GENE EXPRESSION DATA SELF-ORGANIZING ALGORITHM Spain. Alma bioinformática SA.