

Biomedical Genomics Group, Cancer Program
 Institute for Research in Biomedicine (IRB Barcelona), 08028 - Barcelona. T 934039912.
 nuria.lopez@irbbarcelona.org <http://bbglab.irbbarcelona.org>

SHORT SUMMARY

Nuria Lopez-Bigas is a biologist with a PhD in molecular genetics. She transitioned into bioinformatics during her postdoc at the European Bioinformatics Institute (EBI). Since 2006, she leads a research group in Barcelona focused on the study of cancer from a genomics perspective. Her lab has done important contributions in the identification of cancer genes and driver mutations across cancer types, in the interpretation of tumor genomes for precision cancer medicine, in the discovery of the variation of mutation rate along the genome, in the identification of mutational signatures of cancer treatment and in the identification of genes and mutations involved in clonal hematopoiesis.

ACADEMIC POSITIONS AND EDUCATION

- 2016-present **Group Leader** (ICREA Research Professor) Biomedical Genomics Group
 Institute for Research in Biomedicine (IRB Barcelona) Barcelona, Spain
- 2023-present **Full professor** at the University Pompeu Fabra Barcelona
- 2016 - 2023 **Associate professor** at the University Pompeu Fabra Barcelona
- 2011- 2016 **Group Leader** (ICREA Research Professor) Biomedical Genomics Group
 Universitat Pompeu Fabra Barcelona, Spain
- 2006-2011 **Group Leader** (Ramon y Cajal Researcher) Biomedical Genomics Group
 Universitat Pompeu Fabra Barcelona, Spain
- 2005-2006 **Postdoctoral Researcher** (Human Frontiers Science Program - HFSP)
 Centre for Genomic Regulation (CRG) Barcelona, Spain (Laboratory of
 Roderic Guigó)
- 2002-2005 **Postdoctoral Researcher** (Human Frontiers Science Program - HFSP)
 European Bioinformatics Institute (EBI) Cambridge, UK (Laboratory of
 Christos A. Ouzounis)
- 1998-2002 **PhD student** at Oncologic Research Institute (Laboratory of Xavier Estivill).
 PhD from University of Barcelona Barcelona, Spain
- 1994-1998 **B.Sc. Biology** University of Barcelona Barcelona, Spain

SHORT STAYS IN OTHER SCIENTIFIC INSTITUTIONS

- 2024-25 (12 months) **Visiting Scholar** at University of California San Francisco (UCSF), USA
- 2017 (3 months) **Visiting Professor** at Cancer Research UK Cambridge Institute, UK
- 2013 (3 months) **Visiting Researcher** (EMBO Short Term Fellowship) at MRC –
 Laboratory of Molecular Biology, Cambridge UK Sarah Teichmann Lab
- 2001 (2 months) **Visiting PhD student** at INSERM U254 (Montpellier, France) Prof.
 Remy Pujol and Dr. Lionel Simonneau lab
- 2000 (2 months) **Visiting PhD student** (EMBO Short Term Fellowship) at Sackler School
 of Medicine, Tel Aviv University, Israel Karen Avraham Lab
- 1997 (3 months) **Summer student** at Fred Hutchinson Cancer Research Center
 (FHCRC, Seattle, USA) Ronald Reeder Lab
- 1996 (3 months) **Summer student** at Fred Hutchinson Cancer Research Center
 (FHCRC, Seattle, USA) David Martin Lab

Biomedical Genomics Group, Structural and Computational Biology Programme
 Institute for Research in Biomedicine (IRB Barcelona), 08028 - Barcelona. T 934039912.
 nuria.lopez@irbbarcelona.org <http://bbglab.irbbarcelona.org>

HONORS, AWARDS AND DISTINCTIONS

- 2024 Elected member of the Spanish Royal Academy of Sciences (RAC)
- 2024 Elected member of the Barcelona Royal Academy of Sciences and Art (RACAB - pending entrance speech)
- 2023 Biomedicine Award of Lilly Foundation
- 2022 CGI-clinics EU project granted (coordinator role)
- 2022 Cancer Grand Challenge Award (co-leading project PROMINENT)
- 2022 City of Barcelona Award
- 2022 Innovator Award of the International Society of Computational Biology (ISCB)
- 2022 Aspire Award ASPIRE Award from The Mark Foundation for Cancer Research
- 2021 Elected fellow of the International Society of Computational Biology (ISCB)
- 2019 VIII National "Doctores Diz Pintado" Cancer Research Prize
- 2016 XI Banc de Sabadell Award for Research in Biomedicine
- 2016 Elected Member of the European Molecular Biology Organization (EMBO)
- 2015 European Research Council Consolidator Grant (ERC-consolidator)
- 2011 Selected Senior Research Professor at ICREA (Catalan Institute for Research and Advanced Studies). The mission of ICREA is "to select and hire the most talented and extraordinary scientists and academics to work in Catalonia"
- 2011 Catalan National Award for Young Research Talent (*Premi Nacional de Recerca al Talent Jove*)
- 2006 Career Development Award from the Human Frontiers Science Program Organization (HFSP)
- 2006 EMBO Short Term Fellowship to visit the Laboratory of Molecular Biology (LMB-MRC)
- 2005 Selected for Ramon y Cajal Researcher. Competitive call from the Spanish Ministry of Science to incorporate in the Spanish Research system young researchers with an outstanding track record
- 2003 Long Term Fellowship Award from the Human Frontiers Science Program Organization (HFSP)
- 2000 EMBO Short Term Fellowship to visit Sackler School of Medicine, Tel Aviv University, Israel.

PROFESSIONAL ACTIVITIES AND COMMUNITY SERVICE (selected)

- 2017 - present Chair of the Research Integrity Committee at the Institute for Research in Biomedicine
- 2017 - present Member of the Scientific Board of the Spanish Society Against Cancer (AECC)
- 2021 - present Board member of the European Association of Cancer Research (EACR)

Biomedical Genomics Group, Cancer Program
 Institute for Research in Biomedicine (IRB Barcelona), 08028 - Barcelona. T 934039912.
 nuria.lopez@irbbarcelona.org <http://bbglab.irbbarcelona.org>

2021 - present	Member of the Scientific Council of Gustave Roussy Cancer Center, Paris, France
2024 – present	Leader of National Mirror Group 9. Use case- Cancer (1+Milion Genome Initiative)
2023 - present	Member of the International Scientific Advisory Board at IFOM, Milan (Italy)
2021 - present	Member of the Scientific Advisory Board, CRUK Manchester Institute
2020 - present	Member of the Scientific Advisory Board, VHIO Barcelona
2020 - present	Mentor of the LEAP mentorship program - EMBL
2019 - 2022	Member of the Scientific Advisory Board of Finland Institute of Molecular Medicine
2019	Member of the Scientific Advisory Board of Open Targets (UK) 2016 and
2018	Panel Member for the evaluation of the European Research Council Consolidator Grants
2017	Panel Member for the recruitment of Junior Research Leaders for “la Caixa”
Several years	Evaluator for Wellcome Trust
Several years	Evaluator for Cancer Research UK (CRUK)
Several years	Panel member for the evaluation of Ramon y Cajal Grants and Project Grants from the Spanish Ministry

COMPETITIVE GRANTS AS PRINCIPAL INVESTIGATOR

- **PROMINENT:** Discovering the molecular signatures of cancer PROMotion to INform prevENTION. Cancer GrandChallenges. Co-funded by CRUK, NIH and AECC. Budget for the Group: £ 4,702,838.25. Co-leader. Total project budget: ~25M \$.
- Data-driven cancer genome interpretation for personalised cancer treatment. Horizon Europe HORIZON-HLTH-2021-CARE-05. 2022-2027. Budget for the Group: 2.872.675 €. Coordinator of the Consortium. Ref: 101057509. Total project budget: ~10M €.
- CHEMOHEALTH. Efectos of chemotherapies in healthy tissues. MICINN PID2021-126568OB-I00. 2022-2025. 484.000 Eur. Co-Principal Investigator.
- CIBERONC. Group of the consortium CIBERONC. ISCIII-MICINN. From 2022. Principal Investigator.
- Atheroclonal. Somatic mutations and clonal hematopoiesis as drivers of atherosclerosis: from the laboratory to the clinic. MICINN, Lineas Estratégicas. 2022-2024. 156.601 €. Principal Investigator. Project Coordinator: Jose Javier Fuster, CNIC.
- Myoclonal Somatic mutations and clonal hematopoiesis as predictors and drivers of heart failure progression. La Caixa Foundation HR22-00732. 2022-2025. Project Coordinattor: Jose Fuster.
- IMPACT-Data. Infraestructura de Medicina de Precisión asociada a la Ciencia y Tecnología (IMPACT) de la Acción Estratégica en Salud. 2021-2024. 97.350 €. Project coordinator: BSC.
- EOSC4Cancer. HORIZON-INFRA-2021-EOSC-01. 2022-2025. 65.000 €. Coordinator: Alfonso Valencia.

Biomedical Genomics Group, Structural and Computational Biology Programme
Institute for Research in Biomedicine (IRB Barcelona), 08028 - Barcelona. T 934039912.
nuria.lopez@irbbarcelona.org <http://bbglab.irbbarcelona.org>

- SGR2021. AGAUR. 2022-2025. 60.000 €
- Identifying and functionally characterising colorectal cancer driver mutations. Wellcome Trust. 2019-2024. 277.000 €. Principal investigator. Project coordinator: Professor Richard Houlston.
- Computational Oncology Training Alliance (CONTRA). H2020 Innovative Training Network ITN-766030. European Commission. 01/01/2018-31/12/2021. 495.745 €. Principal investigator.
- Identifying noncoding cancer drivers NONCODRIVERS. ERC Consolidator Grant 682398. European Commission. 01/12/2016-30/11/2021. 1.995.828 €. Principal investigator.
- Discovering the mechanisms of tumorigenesis by driver mutations across tissues. ASPIRE Awards The Mark Foundation for Cancer Research. 2022-2023. 195.308,08 \$. Principal Investigator.
- Damage, repair and mutations caused by alkylating agents along the genome. MINECO RTI2018-094095-B-I00. 01/01/2019-31/08/2022. 387.000 €. Principal Investigator.
- Exploring mechanisms of resistance in adult and pediatric T-Acute Lymphoblastic Leukemia. Asociación Española Contra el Cáncer (AECC). 01/11/2016- 31/10/2022. 216.000 €. Principal investigator.
- BioMedTec, Cancer Genome Interpreter. IRB Barcelona Fundació La Caixa. 2017
- Improving the interpretation of tumor genomes for precision cancer medicine. (SAF-2015-66084-R) MINECO (2016-2018)
- Creating medically-driven integrative bioinformatics applications focused on oncology, CNS disorders and their comorbidities (MedBioinformatics) (H2020 - PHC-32-2014 – 634143). European Commission (2015-2018)
- Conveni programa pilot EDI: IntOGen: integrative oncogenomics cancer browser (EDI-PILOT-2014-9) AGAUR (2014)
- Consolidated group grant for the Computational Genomics group (2014 SGR 1121) AGAUR (2014-2016)
- The Cancer Genome Interpreter (2013110) Fundació La Marató TV3 (2014-2017)
- Plan Nacional: Genómica Integrativa del Cancer en la era de la secuenciación masiva (SAF2012-36199). MICINN (2013-2015)
- Plan Nacional: Integración de datos genómicos para el estudio del Cancer (SAF2009-06954) Spanish Ministry of Science (MCI) (2010-2013)
- INNOCASH: Sistema informático para el manejo, gestión y explotación de datos genómicos (INC-0436 FASE II). Fundación Española para la Ciencia y la Tecnología (FECYT) (2010-2012)
- FPI grant for Carlota Rubio (BES-2013-063354) MINECO (2013-2017)
- Marie Curie grant (Tecniospring) for David Tamborero. (TECSPR13-1-0011) European Commission & ACC10. 2014-2016.
- Causes & Mechanisms of Tumour development: a computational "Human Frontier Science Program". Human Frontiers Science Program Career Development Award. (2006-2010)

Biomedical Genomics Group, Cancer Program
Institute for Research in Biomedicine (IRB Barcelona), 08028 - Barcelona. T 934039912.
nuria.lopez@irbbarcelona.org <http://bbglab.irbbarcelona.org>

PHD THESIS SUPERVISED

Currently supervising four PhD students: Olivia Dove, Stefano Pellegrini, Raquel Blanco and Ferriol Calvet

13 PhD Thesis completed

- | | |
|-------------|--|
| 2019-2023 | Co-Director of the thesis of Hanna Kranas. Title: Computational studies of DNA damage and repair of alkylating agents and UV light University Pompeu Fabra |
| 2019-2022 | Co-Director of the thesis of Claudia Arnedo. Title: Clustering of somatic mutations across cancer genomes University Pompeu Fabra |
| 2019-2022 | Co-Director of the thesis of Jose Bonet. Title: Computational studies of DNA modifications and mutations in bulk and single-cell data University Pompeu Fabra |
| 2017-2021 | Co-Director of the thesis of Inés Sentís. Title: The evolution of T-cell acute lymphoblastic leukemia in adult patients under treatment University Pompeu Fabra |
| 2017-2021 | Co-Director of the thesis of Oriol Pich. Title: The role of mutational processes in the evolution of somatic tissues and malignancy University Pompeu Fabra |
| 2016-2020 | Co-Director of the thesis of Joan Frigola. Title: Coding and noncoding drivers: identification and downstream effects University Pompeu Fabra |
| 2014 - 2017 | Director of the thesis of Carlota Rubio-Perez. Title: “In silico drug prescription to cancer patients”. University Pompeu Fabra. |
| 2012 - 2015 | Director of the thesis of Christian Pérez Llamas. Title “Computational approaches for integrative cancer genomics”. Defended successfully at the UPF on 18th December 2015 |
| 2011 - 2014 | Director of the thesis of Michael P Schroeder. Title: “Analysis and visualization of multidimensional cancer genomics data”. Co-supervised with Dr. Abel Gonzalez-Perez, UPF. Defended successfully at the UPF on 21st November 2014 |
| 2009 - 2014 | Director of the thesis of Alba Jené-Sanz. Title: “Integrative study of the regulatory and epigenomic programs involved in cancer development”. Defended successfully at the UPF on 29th April 2013 |
| 2008 - 2013 | Director of the thesis of Khademul Islam. Title: “Delineating epigenetic regulatory mechanisms of cell proliferation and differentiation”. Co-supervised with Dr. Elizaveta Benevolenskaya, University of Illinois Chicago. Defended successfully at the UPF on 25th June 2012 |
| 2007 - 2011 | Director of the thesis of Gunes Gundem. Title: “Integration of genomic data for the study of cancer”. University Pompeu Fabra. Defended successfully at the UPF on 29th September 2011 |
| 2003 - 2007 | Codirector of Dr. Simon J. Furney thesis. Title: “In silico analysis of human disease and cancer genes”. Defended successfully in the Faculty of Medicine of the University College Dublin on 8th February 2007 |

TRAINING FUTURE SCIENTIFIC LEADERS

Biomedical Genomics Group, Structural and Computational Biology Programme
 Institute for Research in Biomedicine (IRB Barcelona), 08028 - Barcelona. T 934039912.
 nuria.lopez@irbbarcelona.org <http://bbglab.irbbarcelona.org>

Past trainees	Position after leaving my lab
Francisco Martínez-Jiménez	Postdoc at UMC Utrecht – Currently Group Leader at VHIO Barcelona
Sabarinathan Radhakrishnan	Principal Investigator (National Centre for Biological Sciences, Tata Institute of Fundamental Research, Bangalore, India)
Hanna Kranas	Postdoc at Josep Carreras Leukaemia Research Institute (Porta lab)
Oriol Pich	Postdoc at Crick Institute London (Charles Swanton Lab)
Claudia Arnedo-Pac	Postdoc at Cambridge University (Sarah Aitken Lab)
José Bonet Giner	Data architect at Tetrascience, a scientific AI company
Santiago Gonzalez	Computational Biologist at C2i Genomics
Joe Usset	Computational Biologist at Hartwig Medical Foundation
Inés Sentís	Postdoc at CNAG Barcelona (Holger Heyn Lab)
David Tamborero	Scientist, Karolinska Institute, Sweden
Loris Mularoni	Bioinformatics Specialist at Center for Regenerative Medicine Barcelona
Joan Frigola	Postdoc at VHIO, Barcelona (Enriqueta Felip Lab) currently Senior Scientist at AstraZeneca
Carlota Rubio-Perez	Postdoc at VHIO, Barcelona (Joan Seoane Lab) – Currently Oncologist at Clinic Hospital in Barcelona
Michael P. Schroeder	Postdoc (Charité University Hospital, Berlin) – Currently Analytics, ML & Data Science Expert at ERNI
Gunes Gundem	Postdoc at Wellcome Trust Sanger Institute (UK) – Currently Senior Research Fellow (MSKCC, New York)
Christian Perez-Llamas	Senior Software/Data Engineer – Currently at Schibsted Media Group
Alba Jené Sanz	Bioinformatics Unit Coordinator at Barcelona Supercomputing Center
Khademul Islam	Associate Professor (University of Dhaka) – Currently Full Professor
Simon J. Furney	Postdoc at CRUK Manchester Institute - Currently Principal Investigator (Royal College of Surgeons in Ireland)

ORGANIZER OF INTERNATIONAL CONFERENCES (selected)

2024	Co-organizer Barcelona Biomed Conference on CANCER PROMOTION: understanding cancer promotion to inform prevention
2024	Co-organizer Cancer Genomics, Multiomics and Computational Biology Bergamo, Italy : 30 April - 2 May 2024
2022	Co-organizer EACR Congress Translating Biology to Medicine. June 2022 Sevilla, Spain.
2021	Organizer Bioinformatics in Cancer EACR Conference, online
2017 - 2023	Co-organizer. Biannual Cancer Genomics EMBL Conference, Heidelberg (Germany)
2019	Conference Chair, Advances in Computational Biology Conference, ISCB. Barcelona, Spain
2018	Conference Chair, Next Generation Sequencing Conference, ISCB. Barcelona, Spain
2017	Co-organizer. ICREA conference: Across Tumor Heterogeneity and Evolution in Cancer: From In Silico Studies to Clinical Impact, School of Medicine. Barcelona, Spain

Biomedical Genomics Group, Cancer Program
 Institute for Research in Biomedicine (IRB Barcelona), 08028 - Barcelona. T 934039912.
 nuria.lopez@irbbarcelona.org <http://bbglab.irbbarcelona.org>

- 2016 Co-organizer. IV Bioinformatics and Genomics Symposium, Barcelona (Spain)
- 2014 Co-organizer. Beyond the Genome: Cancer Genomics. Harvard Medical School, Boston, USA

TECHNOLOGY TRANSFER ACTIVITIES

- CancerGenomeInterpreter (CGI): Platform to facilitate the interpretation of alterations in a patient's tumor (<https://www.cancergenomeinterpreter.org>). CGI is extensively used in research and clinical settings, with more than 10,000 users per year during the last three years (measured by Google Analytics). We have issued agreements with more than 10 companies and foundations for the use of CGI, including Genomics England (the UK National Initiative for Whole-genome sequencing in the National Health Service (NHS)) and Hartwig Medical Foundation, which provides tumor whole-genome sequencing in clinical practice in partnership with 43 hospitals in the Netherlands. In 2022 an EU project, CGI-Clinics was awarded 10M Euros to further develop and implement CGI in the clinic. CGI-Clinics joins together cancer hospitals in France, Germany, England, Greece and Spain, and cancer patient associations, to develop a carefully designed 5 years project to implement the comprehensive and systematic data-driven interpretation of cancer genomes for precision cancer medicine into clinical settings.
- IntOGen: A framework that uses somatic mutations to identify cancer driver genes and their cancer mutational patterns. (<https://www.intogen.org>). IntOGen methods and the pipeline are provided to the community with a free license that allows academic and commercial use. IntOGen had more than 100,000 unique visitors (according to Google Analytics) in the last years from all around the world.
- Contract of service with INIVATA, clinical cancer genomics company on non-invasive tumour analysis from circulating DNA in Cambridge, UK. October 2014.
- Innovation Teams Program 2014. Catalan Research Foundation. Program to support knowledge transfer. INNOCASH 2010-2011.
- Computational system for managing and exploiting genomic data for personalized medicine. Research Collaboration with Sistemas Genómicos, a biotech company in Valencia, Spain.

INVITED SPEAKER AT MAJOR INTERNATIONAL CONFERENCES (Selected)

- 2024 Keynote Speaker at Cancer Evolution: From Genome to Ecology 2024 conference, Hinxton (UK)
- 2024 Invited Speaker at EACR Cancer Genomics, Multiomics and Computational Biology, Bergamo (Italy)
- 2023 Invited Speaker at VIB Tumor Heterogeneity, Leuven (Belgium)
- 2023 Invited Speaker at 127th International Titisee Conference "Somatic Mosaicism", Titisee (Germany)
- 2023 Invited Speaker at the 10th InterAcademy Workshop, The Korean Academy of Science and Technology (KAST), Frankfurt (Germany)
- 2023 Invited Speaker at 4th Crick International Cancer Conference, London, (UK)
- 2023 Invited Speaker at EACR annual meeting Torino (Italy)
- 2023 Invited Speaker at Gordon Research Conference on Epithelial Differentiation and Keratinization, Barcelona (Spain)

Biomedical Genomics Group, Structural and Computational Biology Programme
Institute for Research in Biomedicine (IRB Barcelona), 08028 - Barcelona. T 934039912.
nuria.lopez@irbbarcelona.org <http://bbglab.irbbarcelona.org>

- 2023 Invited Speaker at Cancer Dependency Map Symposium, Milan (Italy)
- 2023 Invited Speaker and discussant. Forbeck Foundation meeting "Mutational signatures in pediatric cancer-from etiology to therapeutic implications" Asilomar Conference Grounds (USA)
- 2022 Invited Speaker, 34th EORTC-NCI-AACR symposium on Molecular targets and Cancer Therapeutics (ENA 2022), Barcelona (Spain)
- 2022 Keynote Speaker Congress Translating Biology to Medicine. June 2022 Sevilla (Spain)
- 2022 Keynote speaker. Mechanisms of Mutagenesis and Their Clinical Implications. Mutagenesis Gordon Research Seminar (USA)
- 2022 Invited Speaker. EMBO Workshop: The many faces of cancer evolution, Rimini (Italy)
- 2021 Invited Speaker. Symposium on Mutation Signatures and Cancer. National Cancer Institute, NIH. 2021 Bioinformatics in Cancer EACR Conference
- 2021 EACR 2020 Virtual Congress. 18-19 June 2020
- 2020 Discussion Leader. CSHL Biology of Genomes Conference (USA)
- 2019 EMBL Cancer Genomics Conference 2019 (Germany)
- 2019 EACR conference. Defence is the Best Attack: Immuno-Oncology Breakthroughs. Barcelona (Spain)
- 2019 Heterogeneity and Evolution in Cancer Conference at CNIO, Madrid (Spain)
- 2019 III Optimizing Immunotherapy New Approaches, Biomarkers, Sequences and Combinations. IMIM, PRBB Barcelona (Spain)
- 2019 V Simposio Nacional de Genómica Aplicada en Oncología (CNIO), Madrid (Spain)
- 2019 Medical Genetics Session, New Horizons in Genomics, QMUL London (UK)
- 2019 EACR conference. Tracking Cancer: Detection and Monitoring, from Diagnosis to Therapy, EACR, Barcelona (Spain)
- 2019 Keynote Speaker. First Zürich Precision Oncology Symposium, Comprehensive Cancer Center Zürich (Switzerland)
- 2018 Keynote Speaker. Molecular Analysis for Personalised Therapy (MAP) Congress by ESMO. Paris (France)
- 2018 49th International Symposium of the Princess Takamatsu Cancer Research Fund, Tokyo (Japan)
- 2018 Cell Symposia: TCGA Legacy. Multi-omic studies in Cancer, Washington (USA)
- 2018 EMBO Workshop, Cellular signaling and Cancer Therapy, Dubrovnik (Croatia)
- 2018 European Association of Cancer Research (EACR) Biennial Congress, Amsterdam (Netherlands)
- 2018 Keynote Speaker. RECOMB-CCB Cancer Computational Biology, Paris (France)
- 2018 The Impact of Genomics Data on Health: The Role of the European Genome-Phenome Archive (EGA). Barcelona (Spain)
- 2017 Cancer Genomics Conference EMBL, Heidelberg (Germany)
- 2017 Louis Jeantet Symposium, University Medical Center, Geneva (Switzerland)

Biomedical Genomics Group, Cancer Program
 Institute for Research in Biomedicine (IRB Barcelona), 08028 - Barcelona. T 934039912.
 nuria.lopez@irbbarcelona.org <http://bbglab.irbbarcelona.org>

- 2017 St. Gilles Thomas Symposium, Perspectives in cancer genetics and genomics. Paris (France)
- 2017 CRG Symposium: 7th International Workshop on Genomic Epidemiology, Barcelona.
- 2107 Keynote Speaker. BC2 Basel Computational Biology Conference, Basel (Switzerland)
- 2017 Genomics of Common Diseases, Wellcome Genome Campus, Cambridge (UK)
- 2017 EACR: Cancer Genomics 2017, Cambridge (UK) 2017 Invited Speaker
 Major Symposium at AACR Annual meeting 2017, Washington (USA)
- 2017 Gene-Regulatory Systems in Development. Carmona (Spain)
- 2017 EACR, Making it Personal, Cancer Precision Medicine. Meet the expert session. Amsterdam (Netherlands) 2017 ICREA-FIJC Conference, Across tumor heterogeneity and evolution in cancer: from in silico studies to clinical impact, Barcelona (Spain)
- 2017 HUGO-HGM, From Genomics to Therapy, Barcelona (Spain)
- 2016 Genetics Society Autumn Meeting Functional genetic variation in the non-coding genome. London (UK).
- 2016 Canceromatics III: Tumor Heterogeneity, CNIO, Madrid (Spain).
- 2016 Keynote Speaker. European Conference on Computational Biology - ECCB2016, The Hague (Netherlands).
- 2016 Keystone Symposium: Understanding the Function of Human Genome Variation, Uppsala (Sweden).
- 2016 Systematic Functional Annotation of Cancer Variants, "Tumor genomes shed light into mutational processes and cancer vulnerabilities", Cancer Systems Genetics, Heidelberg (Germany).
- 2016 1st European Conference for Translational Bioinformatics, MBI, Copenhagen (Denmark)
- 2016 Applied Bioinformatics in Life Sciences, Leuven, (Belgium).
- 2016 Keystone Symposium: The Cancer Genome, Fairmont Banff Springs, Banff, Alberta, (Canada).
- 2016 Festival of Genomics, London (United Kingdom).
- 2015 EMBL Cancer Genomics conference.
- 2015 EMBO meeting 2015 meeting. Birmingham, UK
- 2015 Advances in Genome Biology and Technology (AGBT) meeting. Marco Island, Florida.

INVITED SEMINARS (Selected)

- 2024 Harvard Medical School, Boston, USA
- 2024 San Francisco, UCSF, Diller Comprehensive Cancer Center, USA
- 2024 University of Colorado, Department of Biochemistry and Molecular Genetics, USA

Biomedical Genomics Group, Structural and Computational Biology Programme
Institute for Research in Biomedicine (IRB Barcelona), 08028 - Barcelona. T 934039912.
nuria.lopez@irbbarcelona.org <http://bbglab.irbbarcelona.org>

- 2024 University of Illinois Chicago, Biochemistry and Molecular Genetics Depart., USA
- 2024 San Francisco, UCSF ImmunoX Program Seminar, USA
- 2024 Cardiff University, virtual Science Seminar Talk
- 2023 Berlin Institute for Medical Systems Biology, Berlin, Germany
- 2023 Izmir University, Izmir, Turkey
- 2023 Koç University, Istanbul University, Istanbul, Turkey
- 2023 CRUK Cambridge Institute, UK - United Kingdom
- 2023 Wellcome Trust Sanger Institute, UK - United Kingdom
- 2023 EMBL Science by the beach, Barcelona - Spain 2021 Broad Institute Seminar Series on Medical and Population Genetics, USA
- 2021 Netherlands Cancer Institute (NKI), NKI Seminars, Netherlands
- 2021 SMB Faculty Seminar Series". Washington University, USA - United States of America (on-line)
- 2020 CNIO Distinguished Seminars, Madrid, Spain
- 2019 Quantitative biology, Bioinformatics, and Genomics Seminar series, DKFZ, Heidelberg, Germany
- 2019 Ludwig Institute Seminar Series at the University of Oxford
- 2019 Memorial Sloan Kettering Cancer Center, New York (USA)
- 2018 Invited lecture at Barts Cancer Institute (BCI), London
- 2018 The University of Texas MD Anderson Cancer Center, Texas (USA)
- 2017 The Gurdon Institute, Cambridge, UK
- 2017 Wellcome Sanger Institute, Hinxton, UK
- 2017 The Francis Crick Institute, London, UK 2016 Institute Gustave Roussy, Paris, France
- 2016 Austrian Academy of Sciences, Research Center for Molecular Medicine, Vienna
- 2016 Cornell University, New York, USA

PUBLIC ENGAGEMENT AND OUTREACH ACTIVITIES (selected)

- Multiple newspaper / radio / documentary appearances showcasing our research (Meet Our Scientists, La Vanguardia, El País, El Punt Avui TV, TVE, TV3, Catalunya Radio...).
- Showing our lab and explaining our work to the general public during PRBB Open Day (2014, 2015, 2016) and IRB Open Day (2017-2023).
- Showing the work of our lab at “Fira de la Recerca en Directe” at Science Museu– Cosmocaixa (2019-2023).
- Multiple seminars at primary schools and high schools about science and cancer genomics.
- Authored a chapter titled «*La misteriosa periodicitat del genoma*» explaining our research for the general public in the book “*Deu aportacions catalanes a la ciència actual*” ed. Galaxia Gutenberg.

Biomedical Genomics Group, Cancer Program
 Institute for Research in Biomedicine (IRB Barcelona), 08028 - Barcelona. T 934039912.
 nuria.lopez@irbbarcelona.org <http://bbglab.irbbarcelona.org>

TEACHING ACTIVITIES

2007-present	Coordinator of Elements of Biocomputing Course - Master in Bioinformatics for Health Sciences at University Pompeu Fabra
2017-present	Co-coordinator of Bioinformatics and High-throughput data analysis - Master in Translational Medicine at University of Barcelona
2018,19	Speaker at Seminar series of the Inter-University Bioinformatics Degree (UPF, UPC, UAB and UB)
2017,18,19	Speaker at Master in Bioinformatics UAB
2014	Teacher at Master in OMICS Data Analysis (UVIC)

PUBLICATIONS

Publications summary

Nuria Lopez-Bigas has authored more than 160 publications. Articles she has co-authored had been cited more than 37,000 times. H-index: 74 (according to google scholar).

For a complete list of publications, visit:

PubMed: <http://www.ncbi.nlm.nih.gov/pubmed/?term=Lopez-Bigas>

Google Scholar: <http://goo.gl/hrCQr7>

ORCID: [0000-0003-4925-8988](https://orcid.org/0000-0003-4925-8988)

10 RECENT SELECTED PUBLICATIONS WITH FEW DESCRIPTIVE SENTENCES

Members of Lopez-Bigas lab are shown in **bold** #join supervisor *Corresponding author

Joseph Usset, Axel Rosendahl Huber, Maria A Andrianova, Eduard Batlle, Joan Carles, Edwin Cuppen, Elena Elez, Enriqueta Felip, Marina Gómez-Rey, Deborah Lo Giacco, Francisco Martinez-Jimenez, Eva Muñoz-Couselo, Lillian L Siu, Josep Taberner, Ana Vivancos, **Ferran Muiños, Gonzalez-Perez* A, López-Bigas* N**. Five latent factors underlie response to immunotherapy. **Nature Genetics, 2024**

Only a subset of the cancer patients treated with immunotherapy response to the treatment. The group of Lopez-Bigas embarked on a comprehensive computational analysis to understand which features of the patient and the tumor define treatment response and survival. They discovered that all biomarkers associated with immunotherapy response and survival collapse into five orthogonal latent factors: tumor mutation burden, T cell effective infiltration, transforming growth factor-beta activity in the microenvironment, prior treatment and tumor proliferative potential. They then built a machine learning classifier that combines the five latent factors to predict response and survival in patients treated with immune checkpoint inhibitors.

Mònica Sánchez-Guixé, Ferran Muiños, Morena Pinheiro-Santin, Víctor González-Huici, Carlos J Rodriguez-Hernandez, Alexandra Avgustinova, Cinzia Lavarino, **Abel González-Pérez#**, Jaume Mora*, **Núria López-Bigas#***. Origins of second malignancies in children and mutational footprint of chemotherapy in normal tissues. **Cancer Discovery 2024**

Cancer in childhood is fortunately relatively rare, suffering two tumors during infancy is extremely surprising. This work describes the study of the cases of four children who have experienced two cancers during childhood. To understand why, Lopez-Bigas team sequenced

Biomedical Genomics Group, Structural and Computational Biology Programme
Institute for Research in Biomedicine (IRB Barcelona), 08028 - Barcelona. T 934039912.
nuria.lopez@irbbarcelona.org <http://bbglab.irbbarcelona.org>

whole-genome sequencing of the two tumors and normal tissue of the four children. Using sophisticated computational techniques they partially reconstructed the evolutionary history of those tumors, finding different cancer evolutionary trajectories in each case. This study also shows that healthy tissues of the patients accumulated mutations due to chemotherapy treatment. Understanding the effect of chemotherapy in healthy tissues is important as a step to uncover the causes of late side effects of the treatment.

Muiños F*, Martínez-Jiménez F, Pich O, Gonzalez-Perez A, Lopez-Bigas N**.** In silico saturation mutagenesis of cancer genes. **Nature 2021**

Not all mutations in cancer genes are cancer driver mutations. Sorting out which are and which aren't is critically important for precision cancer medicine and to understand the mechanisms of tumorigenesis. The group of Nuria Lopez-Bigas have addressed this by discovering (using machine-learning models inspired by evolutionary biology), directly from the somatic mutations in tumors, which are the features that define driver mutations in each gene/cancer type. With models trained per gene/tumor type, they can perform "In silico saturation mutagenesis" to identify all driver mutation in each cancer gene, and study the interplay between mutational processes and driver genes.

Martínez-Jiménez F, Muiños F, Sentís I, Deu-Pons J, Reyes-Salazar I, Arnedo-Pac C, Mularoni L, Pich O, Bonet J, Kranas H, Gonzalez-Perez A, Lopez-Bigas N**.** A compendium of mutational cancer driver genes. **Nature Reviews Cancer 2020**

This manuscript describes the comprehensive approach, named IntOGen, to identify cancer genes across tumor types by detecting signals of positive selection in the pattern of mutations. It reports the results of analyzing more than 28000 tumor genomes with this approach leading to the identification of 568 cancer genes. In addition, it provides an historical review of cancer gene identification.

Pich O, Reyes-Salazar I, Gonzalez-Perez A & Lopez-Bigas N**.** Discovering the drivers of clonal hematopoiesis. **Nature Communications 2022**

Clonal hematopoiesis (CH) has emerged as a prevalent condition across human populations, associated with the development of cardiovascular morbidity, hematopoietic malignancies and aging. Although few genes are known to drive CH upon mutations in Hematopoietic Stem Cells, this list is probably reduced to the tip of the iceberg of mutation frequency. Lopez-Bigas team reasoned that the clonal expansion that drives clonal hematopoiesis is reminiscent of that observed in tumors, and therefore methods to detect positive selection in the mutations of genes across tumors could be used to identify the complete list of CH-related genes. They have repurposed paired tumor/blood genome sequencing data to identify somatic mutations in healthy blood across 12,000 patients. Next, they identified CH driver genes by detecting signals of positive selection in blood somatic mutations. This approach works remarkably well; it recovers all known CH genes, and discovers novel interesting candidates, some of which are supported by prior studies. They also demonstrated that employing all the genes in this novel compendium greatly increases the sensitivity in the detection of CH across donors.

Pich O, Muiños F, Lolkema MP, Steeghs N, Gonzalez-Perez A#, Lopez-Bigas N.** The mutational footprints of cancer therapies. **Nature Genetics 2019**

Cancer therapies produce late side effects which in some cases appear even decades after the patient was treated. These side effects are especially well documented in childhood cancer survivors and include secondary neoplasms, mental disabilities and cardiac problems among others. The mechanisms on how cancer therapies exert these late side effects are not known. Many chemotherapies and also radiotherapy produce mutations in surviving tumor and healthy cells of the patient and it is reasonable to think that these mutations might be involved in causing some of these late side effects. In this work the team measured for the

Biomedical Genomics Group, Cancer Program
Institute for Research in Biomedicine (IRB Barcelona), 08028 - Barcelona. T 934039912.
nuria.lopez@irbbarcelona.org <http://bbglab.irbbarcelona.org>

first time the mutation burden caused by cancer therapies in patient cells. To do that they analyzed more than 3500 whole-genomes of treated metastatic patients to uncover the mutational footprints (or mutational signatures) of the cancer treatments. These signatures allowed them to measure the mutational toxicity of these treatments across patients and organs.

The importance of this work relies not only on the results it reports (namely new mutational signatures of most commonly used chemotherapies and the mutation burden they exert in patient cells) but also in that it paves the way to explore the mechanistic link between cancer treatments and their late side effects.

Gonzalez-Perez A*, Sabarinathan R*, Lopez-Bigas N*. Local Determinants of the Mutational Landscape of the Human Genome. **Cell 2019**

This manuscript is an invited review. It explores recent results, some from Lopez-Bigas lab, on the large variability of mutation rate along the genome. They start by describing the different mutational processes affecting cells from different tissues in our body and the DNA repair pathways involved in fixing DNA damage and mismatches. Next, they detail how chromatin conformation, the presence of nucleosomes and DNA structure interfere both with DNA damage and DNA repair shaping the rate at which mutations are accumulated in our genome.

Pich O, Muiños F, Radhakrisnan S, Reyes-Salazar I, Gonzalez-Perez A#, Lopez-Bigas N#*. Somatic and germline mutation periodicity follow the orientation of the DNA minor groove around nucleosomes. **Cell 2018**

This work started when Nuria Lopez-Bigas and her team realized that there is a stunning 10bp periodicity in the rate of somatic mutations in nucleosome covered regions. They observed this initially in tumor genomes. They thought it may be related to the long-studied WW 10bp periodicity in the eukaryotic genomes. Thus, they wanted to determine the causes of this periodicity and its link with the evolution of eukaryotic genomes. The analysis of somatic mutations in thousands of tumors, variation and divergence in Human and Arabidopsis, and maps of DNA damage in human and yeast cells, allowed them to conclude that the structure of DNA and the way it wraps around the nucleosomes is a major influence to DNA damage (UV-light damage) and to the efficiency of DNA repair (Nucleotide Excision Repair and Base Excision Repair), thus leading to a periodic mutation pattern.

The importance of this work is twofold: from one side it is a major advance in our understanding of DNA damage and DNA repair (and on particular on how the structure of DNA and its wrapping around histones affects them) and from another side it provides an explanation on how these factors shape the evolution of eukaryotic genomes leading to patterns like the mysterious 10bp WW periodicity.

Frigola J, Sabarinathan R, Mularoni L, Muiños F, Gonzalez-Perez A, López-Bigas* N. Reduced mutation rate in exons due to differential mismatch repair. **Nature Genetics, 2017**

This manuscript describes that exons and introns have a different rate of somatic mutations. This difference is most prominent in tumors with mutations in the polymerase (e.g. POLE). POLE mutant tumors accumulate a large number of mutations due to mismatches created during replication. The team finds that the mutation rate difference between exons and introns is caused by differential activity of mismatch repair in those regions. The importance of this manuscript is framed in the increase in our understanding on DNA repair along the genome and how it shapes the rate of mutations in different regions.

Sabarinathan R, Mularoni L, Deu-Pons J, Gonzalez-Perez A, Lopez-Bigas N*. Nucleotide excision repair is impaired by binding of transcription factors to DNA. **Nature, 2016**

Biomedical Genomics Group, Structural and Computational Biology Programme
Institute for Research in Biomedicine (IRB Barcelona), 08028 - Barcelona. T 934039912.
nuria.lopez@irbbarcelona.org <http://bbglab.irbbarcelona.org>

This work was initially motivated by the interest of the group in finding driver mutations in non-coding regions. When analyzing mutations in promoters in melanoma samples they found a high number of mutations in transcription factor binding sites (TFBS). They suspected that this accumulation was not related to their role as cancer driver mutations. Thus, they turned our attention to Nucleotide Excision Repair, the pathway involved in repairing UV-light DNA adducts. They showed that when there is a protein binding to the DNA (i.e. a transcription factor) NER activity is impaired, thus leading to a more than fivefold increase in the mutation rate in TFBS.

Biomedical Genomics Group, Cancer Program
 Institute for Research in Biomedicine (IRB Barcelona), 08028 - Barcelona. T 934039912.
 nuria.lopez@irbbarcelona.org <http://bbglab.irbbarcelona.org>

FULL LIST OF PUBLICATIONS

Refereed publications since independent position

- Riba M, Sala C, Culhane AC, Flobak A, Patocs A, Boye K, Plevova K, Pospíšilová S, Gandolfi G, Morelli MJ, Bucci G, Edsjö A, Lassen U, Al-Shahrour F, Lopez-Bigas N, Hovland R, Cuppen E, Valencia A, Poirel HA, Rosenquist R, Scollen S, Juan Arenas Marquez J, Belien J, De Nicolo A, De Maria R, Torrents D & Tonon G. The 1+Million Genomes Minimal Dataset for Cancer, *Nature Genetics*, 2024, doi.org/10.1038/s41588-024-01721-x
- Martinez-Millan D, Brando F, Grau ML, Sanchez-Guixe M, Lopez-Elorduy C, Reyes-Salazar I, Deu-Pons J, Lopez-Bigas N, Gonzalez-Perez A. OpenVariant: a toolkit to parse and operate multiple input file formats. *Bioinformatics*, doi: 10.1093/bioinformatics/btae714
- Aya F, Lanuza-Gracia P, González-Pérez A, Bonnal S, Mancini E, López-Bigas N, Arance A, Valcárcel J. Genomic deletions explain the generation of alternative BRAF isoforms conferring resistance to MAPK inhibitors in melanoma, *Cell reports*, 2024 doi.org/10.1016/j.celrep.2024.114048
- Sánchez-Guixé M, Muiños F, Pinheiro-Santin M, González-Huici V, Rodríguez-Hernandez CJ, Avgustinova A, Lavarino C, González-Pérez A, Mora J, López-Bigas N. Origins of Second Malignancies in Children and Mutational Footprint of Chemotherapy in Normal Tissues. *Cancer Discovery*, 2024, doi.org/10.1158/2159-8290.CD-23-1186
- Ramos-Rodríguez M, Subirana-Granés M, Norris R, Sordi V, Fernández A, Fuentes-Páez G, Pérez-González B, Berenguer C, Raurell-Vila H, Chowdhury M, Corripio R, Partelli S, López-Bigas N, Pellegrini S, Montanya E, Nacher M, Falconi M, Layer R, Rovira M, González-Pérez A, Piemonti L, Pasquali L. Implications of noncoding regulatory functions in the development of insulinomas, *Cell Genomics*, 2024, DOI: 10.1016/j.xgen.2024.100604
- Anderson CJ, Talmane L, Luft J, Connelly J, Nicholson MD, Verburg JC, Pich O, Campbell S, Giaisi M, Wei P, Sundaram V, Connor F, Ginno PA, Sasaki T, Gilbert DM, Liver Cancer Evolution Consortium, López-Bigas N, Semple CA, Odom DT, Aitken SJ & Taylor MS. Strand-resolved mutagenicity of DNA damage and repair, 2024, *Nature*, DOI: 10.1038/s41586-024-07490-1
- Demajo S, Ramis-Zaldivar JE, Grau ML, Andrianova M, Muiños F, López-Bigas N, Gonzalez-Perez A. Identification of Clonal Hematopoiesis Driver Mutations through In Silico Saturation Mutagenesis, *Cancer Discovery*, 2024, doi.org/10.1158/2159-8290.CD-23-1416
- Gonzalez S, Lopez-Bigas N & Gonzalez-Perez A 2023, 'Copy number footprints of platinum-based anticancer therapies', *Plos Genetics*, 19, 2, e1010634.
- Mangiante L, Alcalá N, Sexton-Oates A, et al. 2023, 'Multiomic analysis of malignant pleural mesothelioma identifies molecular axes and specialized tumor profiles driving intertumor heterogeneity', *Nature Genetics*, 55, 4, 607-618.
- Pich O, Reyes-Salazar I, Gonzalez-Perez A & Lopez-Bigas N 2022, 'Discovering the drivers of clonal hematopoiesis', *Nature Communications*, 13, 1, 4267.
- Bonet J, Chen M, Dabad M, Heath S, Gonzalez-Perez A, Lopez-Bigas N & Lagergren J, 2022 'DeepMP: a deep learning tool to detect DNA base modifications on Nanopore sequencing data', *Bioinformatics*, 38, Issue 5, pp 1235–1243.
- Palafox, Marta; Monserrat, Laia; Bellet, Meritxell; Villacampa, Guillermo; Gonzalez-Perez, Abel; Oliveira, Mafalda; Braso-Maristany, Fara; Ibrahimi, Nusaibah; Kannan, Srinivasaraghavan; Mina, Leonardo; Herrera-Abreu, Maria Teresa; Odena, Andreu; Sanchez-Guixe, Monica; Capelan, Marta; Azaro, Analia; Bruna, Alejandra; Rodriguez, Olga; Guzman, Marta; Grueso, Judit; Viaplana, Cristina; Hernandez, Javier; Su, Faye; Lin,

Biomedical Genomics Group, Structural and Computational Biology Programme
Institute for Research in Biomedicine (IRB Barcelona), 08028 - Barcelona. T 934039912.
nuria.lopez@irbbarcelona.org <http://bbglab.irbbarcelona.org>

- Kui; Clarke, Robert B.; Caldas, Carlos; Arribas, Joaquin; Michiels, Stefan; Garcia-Sanz, Alicia; Turner, Nicholas C.; Prat, Aleix; Nuciforo, Paolo; Dienstmann, Rodrigo; Verma, Chandra S.; Lopez-Bigas, Nuria; Scaltriti, Maurizio; Arnedos, Monica; Saura, Cristina; Serra, Violeta 2022, 'High p16 expression and heterozygous RB1 loss are biomarkers for CDK4/6 inhibitor resistance in ER+ breast cancer', *Nature Communications*, 13, 1, 5258.
- Arnedo-Pac, Claudia; Lopez-Bigas, Nuria; Muinos, Ferran 2022, 'Predicting disease variants using biodiversity and machine learning', *Nature Biotechnology*, 40, pages 27–28.
- Nadeu, F; Royo, R; Massoni-Badosa, R; Garcia-Torre, B; Duran-Ferrer, M; Dawson, KJ; Kulis, M; Diaz-Navarro, A; Villamor, N; Melero, JL; Chapaprieta, V; Dueso-Barroso, A; Delgado, J; Moia, R; Ruiz-Gil, S; Marchese, D; Verdaguer-Dot, N; Romo, M; Rozman, M; Frigola, G; Rivas-Delgado, A; Baumann, T; Alcoceba, M; Gonzalez, M; Climent, F; Abrisqueta, P; Castellvi, J; Bosch, F; Aymerich, M; Enjuanes, A; Ruiz-Gaspa, S; Lopez-Guillermo, A; Jares, P; Bea, S; Colomer, D; Lopez-Bigas, N; LIGelpi, J; Torrents, D; Campbell, PJ; Gut, I; Garcia-Roves, PM; Rossi, 2022, 'Detection of early seeding of Richter transformation in chronic lymphocytic leukemia', *Nature Medicine* 28, pages1662–1671
- Connelly, J.; Luft, J.; Anderson, C. J.; Bankhead, P.; Connor, F.; Flicek, P.; Lopez-Bigas, N.; Semple, C. A.; Odom, D. T.; Aitken, S. J.; Taylor, M. S. 2022, 'Using Machine Learning to Infer Whole Genome Duplication from Tumour Nuclear Morphology', *Journal Of Pathology*, 256, S5 - S5.
- Stobbe MD, Gonzalez-Perez A, Lopez-Bigas N & Glynne Gut I 2022, 'Ten simple rules for a successful international consortium in big data omics', *Plos Computational Biology*, 18, 10, e1010546.
- Muinos F, Martinez-Jimenez F, Pich O, Gonzalez-Perez A, Lopez-Bigas N 2021, 'In silico saturation mutagenesis of cancer genes', *Nature*, 596, pages428–432 .
- Frigola J, Sabarinathan R, Gonzalez-Perez A, Lopez-Bigas N 2021, 'Variable interplay of UV-induced DNA damage and repair at transcription factor binding sites', *Nucleic Acids Research*, 49, 2, 891-901.
- Pich O, Cortes-Bullich A, Muinos F, Pratcorona M, Gonzalez-Perez A, Lopez-Bigas N 2021, 'The evolution of hematopoietic cells under cancer therapy', *Nature Communications*, 12, 1, 4803.
- Woolston, Andrew; Barber, Louise J.; Griffiths, Beatrice; Pich, Oriol; Lopez-Bigas, Nuria; Matthews, Nik; Rao, Sheela; Watkins, David; Chau, Ian; Starling, Naureen; Cunningham, David; Gerlinger, Marco 2021, 'Mutational signatures impact the evolution of anti-EGFR antibody resistance in colorectal cancer', *Nature Ecology & Evolution*, .
- Peri, Aviyah; Greenstein, Erez; Alon, Michal; Pai, Joy A.; Dingjan, Tamir; Reich-Zeliger, Shlomit; Barnea, Eilon; Barbolin, Chaya; Levy, Ronen; Arnedo-Pac, Claudia; Kalaora, Shelly; Dassa, Bareket; Feldmesser, Ester; Shang, Ping; Greenberg, Polina; Levin, Yishai; Benedek, Gil; Levesque, Mitchell P.; Adams, David J.; Lotem, Michal; Wilmott, James S.; Scolyer, Richard A.; Joensson, Goeran B.; Admon, Arie; Rosenberg, Steven A.; Cohen, Cyrille J.; Niv, Masha Y.; Lopez-Bigas, Nuria; Satpathy, Ansuman T.; Friedman, Nir; Samuels, Yardena 2021, 'Combined presentation and immunogenicity analysis reveals a recurrent RAS.Q61K neoantigen in melanoma', *Journal Of Clinical Investigation*, 131, 20, e129466.
- Zhang T;; Gonzalez-Perez A; Martinez-Jimenez F;... Lopez-Bigas N; .; Landi MT 2021, 'Genomic and evolutionary classification of lung cancer in never smokers', *Nature Genetics*, 53, 9, 1348.

Biomedical Genomics Group, Cancer Program

Institute for Research in Biomedicine (IRB Barcelona), 08028 - Barcelona. T 934039912.

nuria.lopez@irbbarcelona.org <http://bbglab.irbbarcelona.org>

Borgsmueller N, Bonet J, Marass F, Gonzalez-Perez A, Lopez-Bigas N & Beerenwinkel N 2020. Bayesian non-parametric clustering of single-cell mutation profiles. *Bioinformatics*, 36, 19, 4854 - 4859.

Sentis I, Gonzalez S, Genesca E, Garcia-Hernandez V, Muinos F, Gonzalez C, Lopez-Arribillaga E, Gonzalez J, Fernandez-Ibarrondo L, Mularoni L, Espinosa L, Bellosillo B, Ribera JM, Bigas A, Gonzalez-Perez A, Lopez-Bigas N 2020, 'The evolution of relapse of adult T cell acute lymphoblastic leukemia', *Genome Biology*, 21, 1, 284.

Rheinbay E et al. 2020, 'Analyses of non-coding somatic drivers in 2,658cancer whole genomes', *Nature*, 578, 7793, 102 - 111.

Aitken SJ, Anderson CJ, Connor F, Pich O, Sundaram V, Feig C, Rayner TF, Lukk M, Aitken S, Luft J, Kentepozidou E, Arnedo-Pac C, Beentjes S, Davies SE, Drews RM, Ewing A, Kaiser VB, Khamseh A, López-Arribillaga E, Redmond AM, Santoyo-Lopez J, Sentís I, Talmane L, Yates AD, Semple CA, López-Bigas N, Flicek P, Odom DT, Taylor MS 2020. "Pervasive lesion segregation shapes cancer genome evolution", *Nature*, 583, pages265–270

Popa, Alexandra; Genger, Jakob-Wendelin; Nicholson, Michael D.; Penz, Thomas; Schmid, Daniela; Aberle, Stephan W.; Agerer, Benedikt; Lercher, Alexander; Endler, Lukas; Colaco, Henrique; Smyth, Mark; Schuster, Michael; Grau, Miguel L.; Martinez-Jimenez, Francisco; Pich, Oriol; Borena, Wegene; Pawelka, Erich; Keszei, Zsofia; Senekowitsch, Martin; Laine, Jan; Aberle, Judith H.; Redlberger-Fritz, Monika; Karolyi, Mario; Zoufaly, Alexander; Maritschnik, Sabine; Borkovec, Martin; Hufnagl, Peter; Nairz, Manfred; Weiss, Gunter; Wolfinger, Michael T.; von Laer, Dorothee; Superti-Furga, Giulio; Lopez-Bigas, Nuria; Puchhammer-Stockl, Elisabeth; Allerberger, Franz; Michor, Franziska; Bock, Christoph; Bergthaler, Andreas 2020, 'Genomic epidemiology of superspreading events in Austria reveals mutational dynamics and transmission properties of SARS-CoV-2 Alexandra', *Science Translational Medicine*, 12, 573, eabe2555.

Alexandrov, Ludmil B; Kim, Jaegil; Haradhvala, Nicholas J; Huang, Mi Ni; Tian Ng, Alvin Wei; Wu, Yang; Boot, Arnoud; Covington, Kyle R; Gordenin, Dmitry A; Bergstrom, Erik N; Islam, S M Ashiqul; Lopez-Bigas, Nuria; Klimczak, Leszek J; McPherson, John R; Morganello, Sandro; Sabarinathan, Radhakrishnan; Wheeler, David A; Mustonen, Ville; Getz, Gad; Rozen, Steven G; Stratton, Michael R 2020, 'The repertoire of mutational signatures in human cancer.', *Nature*, 578, 7793, 94 - 101.

Martinez-Jimenez F, Muinos F, Sentis I, Deu-Pons J, Reyes-Salazar I, Arnedo-Pac C, Mularoni L, Pich O, Bonet J, Kranas H, Gonzalez-Perez A, Lopez-Bigas N 2020, 'A compendium of mutational cancer driver genes', *Nature Reviews Cancer*, 20, pages555–572.

Popa A, Genger JW, Nicholson MD, Penz T, Schmid D, Aberle SW, Agerer B, Lercher A, Endler L, Colaço H, Smyth M, Schuster M, Grau ML, Martínez-Jiménez F, Pich O, Borena W, Pawelka E, Keszei Z, Senekowitsch M, Laine J, Aberle JH, Redlberger-Fritz M, Karolyi M, Zoufaly A, Maritschnik S, Borkovec M, Hufnagl P, Nairz M, Weiss G, Wolfinger MT, Laer D, Superti-Furga G, Lopez-Bigas N, Puchhammer-Stöckl E, Allerberger F, Michor F, Bock C and Andreas Bergthaler A, Genomic epidemiology of superspreading events in Austria reveals mutational dynamics and transmission properties of SARS-CoV-2, *Science Translational Medicine* (2020) Vol. 12, Issue 573, eabe2555

Wagner, Alex H.; Walsh, Brian; Mayfield, Georgia; Tamborero, David; Sonkin, Dmitriy; Krysiak, Kilannin; Deu-Pons, Jordi; Duren, Ryan P.; Gao, Jianjiong; McMurphy, Julie; Patterson, Sara; Fitz, Catherine; Pitel, Beth A.; Sezerman, Ozman U.; Ellrott, Kyle; Warner, Jeremy L.; Rieke, Damian T.; Aittokallio, Tero; Cerami, Ethan; Ritter, Deborah I.; Schriml, Lynn M.; Freimuth, Robert R.; Haendel, Melissa; Raca, Gordana; Madhavan, Subha; Baudis, Michael; Beckmann, Jacques S.; Dienstmann, Rodrigo; Chakravarty, Debyani; Li,

Biomedical Genomics Group, Structural and Computational Biology Programme
 Institute for Research in Biomedicine (IRB Barcelona), 08028 - Barcelona. T 934039912.
 nuria.lopez@irbbarcelona.org <http://bbglab.irbbarcelona.org>

- Xuan Shirley; Mockus, Susan; Elemento, Olivier; Schultz, Nikolaus; Lopez-Bigas, Nuria; Lawler, Mark; Goecks, Jeremy; Griffith, Malachi; Griffith, Obi L.; Margolin, Adam A. 2020, 'A harmonized meta-knowledgebase of clinical interpretations of somatic genomic variants in cancer', *Nature Genetics*, 52, 4, 448 - +.
- McGuire AL, Gabriel S, Tishkoff SA, Wonkam A, Chakravarti A, Furlong EMM, Treutlein B, Meissner A, Chang HY, Lopez-Bigas N, Segal E, Kim JS, 2020, 'The road ahead in genetics and genomics', *Nature Reviews Genetics*, 21, pages581–596 .
- Pich O, Muiños F, Lolkema MP, Steeghs N, Gonzalez-Perez A & Lopez-Bigas N 2019, 'The mutational footprints of cancer therapies', *Nature Genetics*, 51, 12, 1732-1740
- Hernández-Sánchez M, Kotaskova J, Rodríguez AE, Radova L, Tamborero D, Abáigar M, Plevova K, Benito R, Tom N, Quijada-Álamo M, Bikos V, Martín AA, Pal K, García de Coca A, Doubek M, López-Bigas N, Hernández-Rivas JM & Pospisilova S 2019, 'CLL cells cumulate genetic aberrations prior to the first therapy even in outwardly inactive disease phase Comprehensive Characterization of Cancer Driver Genes and Mutations', *Leukemia*, 33, 2, 518 - 522.
- Martínez-Jiménez F, Muiños F, López-Arribillaga E, López-Bigas N & González-Pérez A 2020, 'Systematic analysis of alterations in the ubiquitin proteolysis system reveals its contribution to driver mutations in cancer', *Nature Cancer*, 1, 1, 122 - +.
- Gonzalez-Perez A, Sabarinathan R & Lopez-Bigas N 2019, 'Local Determinants of the Mutational Landscape of the Human Genome', *Cell*, 177 (1):101-114.
- Marquès I Bonet, Tomàs. 2019, DEU APORTACIONS CATALANES A LA CIÈNCIA ACTUAL, Galaxia Gutenberg.
- Arnedo-Pac C, Mularoni L, Muinos F, Gonzalez-Perez A & Lopez-Bigas N 2019, 'OncodriveCLUSTL: a sequence-based clustering method to identify cancer divers', *Bioinformatics*, 35, 22, 4788-4790
- Pich O, Muiños F, Sabarinathan R, Reyes-Salazar I, Gonzalez-Perez A, Nuria Lopez-Bigas N. Somatic and germline mutation periodicity follow the orientation of the DNA minor groove around nucleosomes. *Cell* 2018. 175 (4) pp 902-904
- Martínez-Ricarte F, Mayor R, Martínez-Sáez E, Rubio-Pérez C, Pineda E, Cordero E, Cicuéndez M, Poca MA, López-Bigas N, Ramon Y Cajal S, Vieito M, Carles J, Tabernero J, Vivancos A, Gallego S, Graus F, Sahuquillo J, Seoane J. Molecular Diagnosis of Diffuse Gliomas through Sequencing of Cell-Free Circulating Tumor DNA from Cerebrospinal Fluid. *Clinical Cancer Research*. 2018
- Bailey MH, Tokheim C, Porta-Pardo E, Sengupta S, Bertrand D, Weerasinghe A, Colaprico A, Wendl MC, Kim J, Reardon B, Ng PK, Jeong KJ, Cao S, Wang Z, Gao J, Gao Q, Wang F, Liu EM, Mularoni L, Rubio-Perez C, Nagarajan N, Cortés-Ciriano I, Zhou DC, Liang WW, Hess JM, Yellapantula VD, Tamborero D, Gonzalez-Perez A, Suphavitai C, Ko JY, Khurana E, Park PJ, Van Allen EM, Liang H; MC3 Working Group; Cancer Genome Atlas Research Network, Lawrence MS, Godzik A, Lopez-Bigas N, Stuart J, Wheeler D, Getz G, Chen K, Lazar AJ, Mills GB, Karchin R, Ding L. Comprehensive Characterization of Cancer Driver Genes and Mutations. *Cell*. 2018.
- Sabarinathan R, Pich O, Martincorena I, Rubio-Perez C, Juul M, Wala J, Schumacher S, Shapira O, Sidiropoulos N, Waszak S, Tamborero D, Mularoni L, Rheinbay E, Hornshoj H, Deu-Pons J, Muinos F, Bertl J, Guo Q, Weischenfeldt J, Korbel JO, Getz G, Campbell PJ, Pedersen JS, Beroukhim R, Perez-Gonzalez A, Lopez-Bigas N, PCAWG Drivers and Functional Interpretation Group, ICGC/TCGA Pan-Cancer Analysis of Whole Genomes Net. The whole-genome panorama of cancer drivers. 2017, Pre-print. doi: 10.1101/190330

Biomedical Genomics Group, Cancer Program

Institute for Research in Biomedicine (IRB Barcelona), 08028 - Barcelona. T 934039912.

nuria.lopez@irbbarcelona.org <http://bbglab.irbbarcelona.org>

- Tamborero D, Rubio-Perez C, Muinos F, Sabarinathan R, Piulats JMM, Muntasell A, Dientsmann R, Lopez-Bigas N, Gonzalez-Perez A. A pan-cancer landscape of interactions between solid tumors and infiltrating immune cell populations. 2017, *Clinical Cancer Research* DOI: 10.1158/1078-0432.CCR-17-3509
- Tamborero D, Rubio-Perez C, Deu-Pons J, Schroeder MP, Vivancos A, Rovira A, Tusquets I, Albanell J, Rodon J, Tabernero J, de Torres C, Dienstmann R, Gonzalez-Perez A, Lopez-Bigas N. Cancer Genome Interpreter Annotates The Biological And Clinical Relevance Of Tumor Alterations. 2018, *Genome Medicine*. doi.org/10.1186/s13073-018-0531-8
- Frigola J, Sabarinathan R, Mularoni L, Muiños F, Gonzalez-Perez A, López-Bigas N. Reduced mutation rate in exons due to differential mismatch repair. *Nature Genetics*, 2017 49(12), pp. 1684-1692, doi:10.1038/ng.3991
- Karube K, Enjuanes A, Dlouhy I, Jares P, Martin-Garcia D, Nadeu F, Ordóñez GR, Rovira J, Clot G, Royo C, Navarro A, Gonzalez-Farre B, Vaghefi A, Castellano G, Rubio-Perez C, Tamborero D, Briones J, Salar A, Sancho JM, Mercadal S, Gonzalez-Barca E, Escoda L0, Miyoshi H, Ohshima K, Miyawaki K, Kato K, Akashi K, Mozos A, Colomo L, Alcoceba M, Valera A, Carrió A, Costa D, Lopez-Bigas N, Schmitz R, Staudt LM, Salaverria I, López-Guillermo A, Campo E. Integrating genomic alterations in diffuse large B-cell lymphoma identifies new relevant pathways and potential therapeutic targets. *Leukemia*, 2017 DOI: 10.1038/leu.2017.251
- Porta-Pardo E, Kamburov A, Tamborero D, Pons T, Grases D, Valencia A, Lopez-Bigas N, Getz G, Godzik A, Comparison of algorithms for the detection of cancer drivers at subgene resolution. *Nat Methods*. 2017 Jul 17. doi: 10.1038/nmeth.4364.
- Hayward NK, Wilmott JS, Waddell N, Johansson PA, Field MA, Nones K, Patch AM, Kakavand H, Alexandrov LB, Burke H, Jakrot V, Kazakoff S, Holmes O, Leonard C, Sabarinathan R, Mularoni L, Wood S, Xu Q, Waddell N, Tembe V, Pupo GM, De Paoli-Iseppi R, Vilain RE, Shang P, Lau LMS, Dagg RA, Schramm SJ, Pritchard A, Dutton-Regester K, Newell F, Fitzgerald A, Shang CA, Grimmond SM, Pickett HA, Yang JY, Stretch JR, Behren A, Kefford RF, Hersey P, Long GV, Cebon J, Shackleton M, Spillane AJ, Saw RPM, López-Bigas N, Pearson JV, Thompson JF, Scolyer RA, Mann GJ. Whole genome landscapes of major melanoma subtypes. *Nature*, 2017
- Griffith M, Spies NC, Krysiak K, McMichael JF, Coffman AC, Danos AM, Ainscough BJ, Ramirez CA, Rieke DT, Kujan L, Barnell EK, Wagner AH, Skidmore ZL, Wollam A, Liu CJ, Jones MR, Bilski RL, Lesurf R, Feng Y, Shah NM, Bonakdar M, Trani L, Matlock M, Ramu A, Campbell KM, Spies GC, Graubert AP, Gangavarapu K, Eldred JM, Larson DE, Walker JR, Good BM, Wu C, Su AI, Dienstmann R, Margolin A, Tamborero D, Lopez-Bigas N, Jones SJM, Bose R, Spencer DH, Wartman LD, Wilson RK, Mardis ER, Griffith OL. 2016. CIViC: A resource for crowdsourcing the clinical interpretation of variants in cancer. *Nature Genetics*, 2017 49, 170–174
- Mateo F, Arenas EJ, Aguilar H, Serra-Musach J, de Garibay GR, Boni J, Maicas M, Du S, Iorio F, Herranz-Ors C, Islam A, Prado X, Llorente A, Petit A, Vidal A, Català I, Soler T, Venturas G, Rojo-Sebastian A, Serra H, Cuadras D, Blanco I, Lozano J, Canals F, Sieuwerts AM, de Weerd V, Look MP, Puertas S, García N, Perkins AS, Bonifaci N, Skowron M, Gómez-Baldó L, Hernández V, Martínez-Aranda A, Martínez-Iniesta M, Serrat X, Cerón J, Brunet J, Barretina MP, Gil M, Falo C, Fernández A, Morilla I, Pernas S, Plà MJ, Andreu X, Seguí MA, Ballester R, Castellà E, Nellist M, Morales S, Valls J, Velasco A, Matias-Guiu X, Figueras A, Sánchez-Mut JV, Sánchez-Céspedes M, Cordero A, Gómez-Miragaya J, Palomero L, Gómez A, Gajewski TF, Cohen EE, Jesiotr M, Bodnar L, Quintela-Fandino M, López-Bigas N, Valdés-Mas R, Puente XS, Viñals F, Casanovas O, Graupera M, Hernández-Losa J, Ramón Y Cajal S, García-Alonso L, Saez-Rodriguez J, Esteller M, Sierra A, Martín-Martín N, Matheu A, Carracedo A, González-Suárez E, Nanjundan M, Cortés J, Lázaro C, Odero MD, Martens JW, Moreno-Bueno G, Barcellos-

Biomedical Genomics Group, Structural and Computational Biology Programme
 Institute for Research in Biomedicine (IRB Barcelona), 08028 - Barcelona. T 934039912.
 nuria.lopez@irbbarcelona.org <http://bbglab.irbbarcelona.org>

- Hoff MH, Villanueva A, Gomis RR, Pujana MA. Stem cell-like transcriptional reprogramming mediates metastatic resistance to mTOR inhibition. *Oncogene*. 2016.
- Rubio-Perez C, Deu-Pons J, Tamborero D, Lopez-Bigas N, Gonzalez-Perez A. Rational design of cancer gene panels with OncoPaD. *Genome Medicine*. 2016. 8 (1): 98
- Sabarinathan R, Mularoni L, Deu-Pons J, Gonzalez-Perez A, Lopez-Bigas N. Nucleotide excision repair is impaired by binding of transcription factors to DNA. *Nature*, 2016; 532 (7598): 264-7.
- lorio F, Knijnenburg TA, Vis DJ, Bignell GR, Menden MP, Schubert M, Aben N, Gonçalves E, Barthorpe S, Lightfoot H, Cokelaer T, Greninger P, van Dyk E, Chang H, de Silva H, Heyn H, Deng X, Egan RK, Liu Q, Mironenko T, Mitropoulos X, Richardson L, Wang J, Zhang T, Moran S, Sayols S, Soleimani M, Tamborero D, Lopez-Bigas N, Ross-Macdonald P, Esteller M, Gray NS, Haber DA, Stratton MR, Benes CH, Wessels LFA, Saez-Rodriguez J, McDermott U, Garnett MJ. A Landscape of Pharmacogenomic Interactions in Cancer. *Cell*, 2016; 166 (3):740-754.
- Loris Mularoni, Radhakrishnan Sabarinathan, Jordi Deu-Pons, Abel Gonzalez-Perez, Núria López-Bigas. OncodriveFML: A general framework to identify coding and non-coding regions with cancer driver mutations. *Genome Biology*, 2016 17:128
- Frigola J, Iturbide A, Lopez-Bigas N, Peiro S, Gonzalez-Perez A. Altered oncomodules underlie chromatin regulatory factors driver mutations. *Oncotarget*. 2016 Apr 15. 7(21):30748-59
- Puente XS, Beà S, Valdés-Mas R, Villamor N, Gutiérrez-Abril J, Martín-Subero JI, Munar M, Rubio-Pérez C, Jares P, Aymerich M, Baumann T, Beekman R, Belver L, Carrio A, Castellano G, Clot G, Colado E, Colomer D, Costa D, Delgado J, Enjuanes A, Estivill X, Ferrando AA, Gelpí JL, González B, González S, González M, Gut M, Hernández-Rivas JM, López-Guerra M, Martín-García D, Navarro A, Nicolás P, Orozco M, Payer ÁR, Pinyol M, Pisano DG, Puente DA, Queirós AC, Quesada V, Romeo-Casabona CM, Royo C, Royo R, Rozman M, Russiñol N, Salaverría I, Stamatopoulos K, Stunnenberg HG, Tamborero D, Terol MJ, Valencia A, López-Bigas N, Torrents D, Gut I, López-Guillermo A, López-Otín C, Campo E. Non-coding recurrent mutations in chronic lymphocytic leukaemia. *Nature*. 2015 Jul 22.
- Mutation Consequences and Pathway Analysis working group of the International Cancer Genome Consortium. Pathway and network analysis of cancer genomes. *Nat Methods*. 2015 Jun 30;12(7):615-21.
- B Barneda-Zahonero, O Collazo, A Azagra, I Fernández-Duran, J Serra-Musach, Abul BMMK Islam, N Vega-García, R Malatesta, M Camós, A Gómez, L Román-González, AVidal, N López-Bigas , A Villanueva, M Esteller, M Parra. The transcriptional repressor HDAC7 promotes apoptosis and c-Myc downregulation in particular types of leukemia and lymphoma. *Cell death & disease*. 2015, 6 (2), e1635
- Carlota Rubio-Perez, David Tamborero, Michael P. Schroeder, Albert A. Antolín, Jordi Deu-Pons, Christian Perez-Llamas, Jordi Mestres, Abel Gonzalez-Perez, and Nuria Lopez-Bigas. In silico prescription of anticancer drugs to cohorts of 28 tumor types reveals unexploited targeting opportunities. 2015. *Cancer Cell*.
- Di Vona C, Bezdán D, Islam AB, Salichs E, López-Bigas N, Ossowski S, de la Luna S. Chromatin-wide Profiling of DYRK1A Reveals a Role as a Gene-Specific RNA Polymerase II CTD Kinase. *Mol Cell*, 2015
- Leiserson MD, Vandin F, Wu HT, Dobson JR, Eldridge JV, Thomas JL, Papoutsaki A, Kim Y, Niu B, McLellan M, Lawrence MS, Gonzalez-Perez A, Tamborero D, Cheng Y, Ryslik GA, Lopez-Bigas N, Getz G, Ding L, Raphael BJ. Pan-cancer network analysis identifies

Biomedical Genomics Group, Cancer Program

Institute for Research in Biomedicine (IRB Barcelona), 08028 - Barcelona. T 934039912.

nuria.lopez@irbbarcelona.org <http://bbglab.irbbarcelona.org>

- combinations of rare somatic mutations across pathways and protein complexes. *Nat Genet*, 2015
- Ferreira PG, Jares P, Rico D, Gómez-López G, Martínez-Trillos A, Villamor N, Ecker S, González-Pérez A, Knowles DG, Monlong J, Johnson R, Quesada V, Gouin A, Djebali S, López-Guerra M, Colomer D, Royo C, Cazorla M, Pinyol M, Clot G, Aymerich M, Rozman M, Kulis M, Tamborero D, Papasaikas P, Blanc J, Gut M, Gut I, Puente XS, Pisano DG, Martin-Subero JI, López-Bigas N, López-Guillermo A, Valencia A, López-Otín C, Campo E, Guigo R. Transcriptome characterization by RNA sequencing identifies a major molecular and clinical subdivision in chronic lymphocytic leukemia. *Genome Res*, 2014; 24 (2): 212-226
- Millán-Ariño L, Islam AB, Izquierdo-Bouldstridge A, Mayor R, Terme JM, Luque N, Sancho M, López-Bigas N, Jordan A. Mapping of six somatic linker histone H1 variants in human breast cancer cells uncovers specific features of H1.2. *Nucleic Acids Res*, 2014; 42 (7): 4474-4493
- Deu-Pons J, Schroeder MP, Lopez-Bigas N. jHeatmap: an interactive heatmap viewer for the web. *Bioinformatics*, 2014; 15; 30(12): 1757-8
- Vilarrasa-Blasi J, Gonzalez-Garcia MP, Frigola D, Fabregas N, Alexiou KG, Lopez-Bigas N, Rivas S, Jauneau A, Lohmann JU, Benfey PN, Ibanes M, Cano-Delgado AI. Regulation of Plant Stem Cell Quiescence by a Brassinosteroid Signaling Module. *Dev Cell*, 30 (1):36-47
10.1016/j.devcel.2014.05.020 JUL 2014
- Truscott M, Islam ABMMK, Lightfoot J, Lopez-Bigas N, Frolov MV. An Intronic microRNA Links Rb/E2F and EGFR Signaling. *Plos Genet*, 2014; 10(7) :e1004493
- Hoadley KA, Yau C, Wolf DM, Cherniack AD, Tamborero D, Ng S, Leiserson MD, Niu B, McLellan MD, Uzunangelov V, Zhang J, Kandoth C, Akbani R, Shen H, Omberg L, Chu A, Margolin AA, Van't Veer LJ, Lopez-Bigas N, Laird PW, Raphael BJ, Ding L, Robertson AG, Byers LA, Mills GB, Weinstein JN, Van Waes C, Chen Z, Collisson EA; Cancer Genome Atlas Research Network, Benz CC, Perou CM, Stuart JM. Multiplatform Analysis of 12 Cancer Types Reveals Molecular Classification within and across Tissues of Origin. *Cell*, 2014; 14; 158(4):929-44
- Schroeder MP, Rubio-Perez C, Tamborero D, Gonzalez-Perez A, Lopez-Bigas N. OncodriveROLE classifies cancer driver genes in loss of function and activating mode of action. *Bioinformatics*, 2014; 30(17):i549-i555
- Gobbi A, Iorio F, Dawson KJ, Wedge DC, Tamborero D, Alexandrov LB, Lopez-Bigas N, Garnett MJ, Jurman G, Saez-Rodriguez J. Fast randomization of large genomic datasets while preserving alteration counts. *Bioinformatics*, 2014; 30(17):i617-i623
- Vento-Tormo R, Rodríguez-Ubreva J, Di Lisio L, Islam AB, Urquiza JM, Hernando H, López-Bigas N, Shannon-Lowe C, Martínez N, Montes-Moreno S, Piris MA, Ballestar E. NF-KappaB directly mediates epigenetic deregulation of common microRNAs in Epstein-Barr virus-mediated transformation of B-cells and in lymphomas. *Nucleic Acids Res*, 2014; 42 (17): 11025-11039
- Guiu J, Bergen D, De Pater E, Islam A, Ayllón V, Gama-Norton L, Ruiz-Herguido C, González J, López-Bigas N, Menéndez P, Dzierzak E, Espinosa L, Bigas A*. Identification of Cdca7 as a novel Notch transcriptional target involved in hematopoietic stem cell emergence. *J Exp Med*, 2014; 211 (12): 2411-2423
- Biton A, Bernard-Pierrot I, Lou Y, Krucker C, Chapeaublanc E, Rubio-Pérez C, López-Bigas N, Kamoun A, Neuzillet Y, Gestraud P, Grieco L, Rebouissou S, de Reyniès A, Benhamou S, Le Bret T, Southgate J, Barillot E, Allory Y, Zinovyev A, Radvanyi F. Independent component analysis uncovers the landscape of the bladder tumor transcriptome and reveals insights into luminal and Basal subtypes. *Cell Rep*, 2014;9(4):1235-45

Biomedical Genomics Group, Structural and Computational Biology Programme
 Institute for Research in Biomedicine (IRB Barcelona), 08028 - Barcelona. T 934039912.
 nuria.lopez@irbbarcelona.org <http://bbglab.irbbarcelona.org>

- Hernando H, Shannon-Lowe C, Islam AB, Al-Shahrour F, Rodriguez-Ubreva J, Rodriguez-Cortez VC, Javierre BM, Mangas C, Fernandez AF, Parra M, Delecluse HJ, Esteller M, Lopez-Granados E, Fraga MF, Lopez-Bigas N, Ballestar E. The B cell transcription program mediates hypomethylation and overexpression of key genes in Epstein-Barr virus-associated proliferative conversion. *Genome Biol*, 2013; 14 (1): R3
- Tamborero D, Lopez-Bigas N, Gonzalez-Perez A. Oncodrive-CIS: A Method to Reveal Likely Driver Genes Based on the Impact of Their Copy Number Changes on Expression. *PLoS One*, 2013; 8(2)
- de la Rica L, Urquiza JM, Gómez-Cabrero D, Islam AB, Lopez-Bigas N, Tegnér J, Toes RE, Ballestar E. Identification of novel markers in rheumatoid arthritis through integrated analysis of DNA methylation and microRNA expression. *J Autoimmun*, 2013; 41 6-16
- Barneda-Zahonero B, Roman-Gonzalez L, Collazo O, Rafati H, Islam AMMK, Bussmann LH, di Tullio A, De Andres L, Graf T, Lopez-Bigas N, Mahmoudi T, Parra M. HDAC7 Is a Repressor of Myeloid Genes Whose Downregulation Is Required for Transdifferentiation of Pre-B Cells into Macrophages. *Plos Genetics*, 9 (5):e1003503
- Cases M, Furlong LI, Albanell J, Altman RB, Bellazzi R, Boyer S, Brand A, Brookes AJ, Brunak S, Clark TW, Gea J, Ghazal P, Graf N, Guigo R, Klein TE, Lopez-Bigas N, Maojo V, Mons B, Musen M, Oliveira JL, Rowe A, Ruch P, Shabo A, Shortliffe EH, Valencia A, van der Lei J, Mayer MA, Sanz F. Improving data and knowledge management to better integrate health care and research. *J Intern Med* 2013; 274 (4): 321-8
- International Cancer Genome Consortium Mutation Pathways and Consequences Subgroup of the Bioinformatics Analyses Working Group, Gonzalez-Perez A, Mustonen V, Reva B, Ritchie GR, Creixell P, Karchin R, Vazquez M, Fink JL, Kassahn KS, Pearson JV, Bader GD, Boutros PC, Muthuswamy L, Ouellette BF, Reimand J, Linding R, Shibata T, Valencia A, Butler A, Dronov S, Flicek P, Shannon NB, Carter H, Ding L, Sander C, Stuart JM, Stein LD, Lopez-Bigas N. Computational approaches to identify functional genetic variants in cancer genomes. *Nat Methods*, 2013; 10 (8): 723-9
- Mulero MC, Ferres-Marco D, Islam A, Margalef P, Pecoraro M, Toll A, Drechsel N, Charneco C, Davis S, Bellora N, Gallardo F, López-Arribillaga E, Asensio-Juan E, Rodilla V, González J, Iglesias M, Shih V, Alba M, Di Croce L, Hoffmann A, Miyamoto S, Villà-Freixa J, Lopez-Bigas N, Keyes WM, Domínguez M, Bigas A, Espinosa L. Chromatin-Bound IκBα Regulates a Subset of Polycomb Target Genes in Differentiation and Cancer. *Cancer Cell*, 2013; 24 (2): 151-166
- Jene-Sanz A, Renta V, Vilcova AV, Khramtsova GF, Khramtsov AI, Olopade OI, Lopez-Bigas N, Benevolenskaya EV. Expression of Polycomb targets predicts breast cancer prognosis. *Mol Cell Biol*, 2013; 33 (19): 3951-3961
- Tamborero D, Gonzalez-Perez A, Lopez-Bigas N. OncodriveCLUST: exploiting the positional clustering of somatic mutations to identify cancer genes. *Bioinformatics*, 2013; 29 (18): 2238-44
- Cancer Genome Atlas Research Network (including Lopez-Bigas N, González-Pérez A, Tamborero D), Weinstein JN, Collison EA, Mills GB, Shaw KM, Ozenberger BA, Ellrot K, Shmulevich I, Sander C, Stuart JM. The Cancer Genome Atlas Pan-Cancer Analysis Project. *Nat Genet*, 2013; 45: 1113–1120
- Gonzalez-Perez A, Jene-Sanz A, Lopez-Bigas N. The mutational landscape of chromatin regulatory factors across 4623 tumor samples. *Genome Biol*, 2013; 14(9):r106
- Tamborero D, Gonzalez-Perez A, Perez-Llamas C, Deu-Pons J, Kandoth C, Reimand J, Lawrence MS, Getz G, Bader GD, Ding L, Lopez-Bigas N. Comprehensive identification of mutational cancer driver genes across 12 tumor types. *Sci Rep-UK*, 2013; 3

Biomedical Genomics Group, Cancer Program

Institute for Research in Biomedicine (IRB Barcelona), 08028 - Barcelona. T 934039912.

nuria.lopez@irbbarcelona.org <http://bbglab.irbbarcelona.org>

- Gonzalez-Perez A, Perez-Llamas C, Deu-Pons J, Tamborero D, Schroeder MP, Jene-Sanz A, Santos A, Lopez-Bigas N. IntOGen-mutations identifies cancer drivers across tumor types. *Nat Methods*, 2013; 10 (11): 1081-1082
- Ambrus AM, Islam AB, Holmes KB, Moon NS, Lopez-Bigas N, Benevolenskaya EV, Frolov MV. Loss of dE2F Compromises Mitochondrial Function. *Dev Cell*, 2013; 25; 27(4): 438-51
- Milne R, La Vecchia C, Van Steen K, Hahn S, Buchholz M, Costello E, Esposito I, Hoheisel JD, Lange B, Lopez-Bigas N, Michalski CW, Real FX, Brand A, Malats N. EU Pancreas: An Integrated European Platform for Pancreas Cancer Research - from Basic Science to Clinical and Public Health Interventions for a Rare Disease. *Public Health Genomics*. 2013; 16 (6): 305-312
- Rafael-Palou X, Schroeder MP, Lopez-Bigas N. SVGMap: configurable image browser for experimental data. *Bioinformatics*, 2012; 28 (1):119-120
- Ingles-Esteve J, Morales M, Dalmases A, Garcia-Carbonell R, Jene-Sanz A, Lopez-Bigas N, Iglesias M, Ruiz-Herguido C, Rovira A, Rojo F, Albanell J, Gomis RR, Bigas A, Espinosa L. Inhibition of Specific NF-kappa B Activity Contributes to the Tumor Suppressor Function of 14-3-3 sigma in Breast Cancer. *PLOS ONE*, 2012; 7 (5) e38347
- Ng S, Collisson EA, Sokolov A, Goldstein T, Gonzalez-Perez A, Lopez-Bigas N, Benz C, Haussler D, Stuart JM. PARADIGM-SHIFT predicts the function of mutations in multiple cancers using pathway impact analysis. *Bioinformatics*, 2012; 15;28(18): i640-i646
- Gonzalez-Perez A, Lopez-Bigas N. Functional impact bias reveals cancer drivers. *Nucleic Acids Res*, 2012; 40; 21: e169
- Beshiri ML, Holmes KB, Richter WF, Hess S, Islam AB, Yan Q, Plante L, Litovchick L, Gévry N, Lopez-Bigas N, Kaelin WG Jr, Benevolenskaya EV. Coordinated repression of cell cycle genes by KDM5A and E2F4 during differentiation. *P Natl Acad Sci USA*, 2012; 109 (45): 18499-18504
- Pippa R, Espinosa L, Gundem G, Garcia-Escudero R, Dominguez A, Orlando S, Gallastegui E, Saiz C, Besson A, Pujol MJ, Lopez-Bigas N, Paramio JM, Bigas A, Bachs O. p27(Kip1) represses transcription by direct interaction with p130/E2F4 at the promoters of target genes. *Oncogene*, 2012; 31 (38):4207-4220
- Tamborero D, Lopez-Bigas N and Gonzalez-Perez A. Oncodrive-CIS: a method to reveal likely driver genes based on the impact of their copy number changes on expression. *PLoS ONE*. 2013
- Gonzalez-Perez A, Deu-Pons J and Lopez-Bigas N. Improving the prediction of the functional impact of cancer mutations by baseline tolerance transformation. *Genome Medicine* 2012. 4:89 doi:10.1186/gm390s
- Bayarmagnai Nicolay BN, Islam A, Lopez-Bigas N and Maxim V. Frolov. Drosophila GAGA factor is required for full activation of the dE2F1-Yki/Sd transcriptional program. *Cell Cycle* 2012. 11, 22. Pages 4191 - 4202
- Gundem G and Lopez-Bigas N. Sample level enrichment analysis unravels shared stress phenotypes among multiple cancer types. *Genome Medicine* 2012. 10.1186/gm327
- Rafael-Palou X, Schroeder MP and Lopez-Bigas N. SVGMap: configurable image browser for experimental data. *Bioinformatics* 2012. 28(1):119-20.
- Guberman JM et al (including Gundem G, Perez-Llamas C, Lopez-Bigas N). BioMart Central Portal: an open database network for the biological community. *Database* 2011. 2011 Sep 18;2011:bar041.
- Truscott M, Islam AB, Lopez-Bigas A, and Frolov MV. mir-11 limits the pro-apoptotic function of its host gene, dE2f1. *Genes and Development* 2011. 25(17):1820-34.

Biomedical Genomics Group, Structural and Computational Biology Programme
Institute for Research in Biomedicine (IRB Barcelona), 08028 - Barcelona. T 934039912.
nuria.lopez@irbbarcelona.org <http://bbglab.irbbarcelona.org>

- Islam AB, Richter WF, Jakobs L, Lopez-Bigas N, Benevolenskaya EV. Coregulation of Histone Modifying Enzymes in Cancer. *PLoS ONE* 2011. 6(8): e24023.
- Perez-Llamas C#, Gundem G#, Lopez-Bigas N. Integrative cancer genomics (IntOGen) in BioMart. Database 2011. Sep 7;2011:bar039.
- Perez-Llamas C and Lopez-Bigas N. Gitools: analysis and visualisation of genomic data using interactive heat-maps. *PLoS One* 2011. 6(5): e19541.
- Gonzalez-Perez A and Lopez-Bigas N. Improving the assessment of the outcome of non-synonymous SNVs with a Consensus deleteriousness score (Condel). *Am J Hum Genet* 2011. 88(4):440-449.
- Nicolay BN, Bayarmagnai B, Islam AB, Lopez-Bigas N, Frolov MV. Cooperation between dE2F1 and Yki/Sd defines a distinct transcriptional program necessary to bypass cell cycle exit. *Genes Dev* 2011. Feb 15;25(4):323-35.
- Islam AB, Richter WF, Lopez-Bigas N, Benevolenskaya EV. Selective targeting of histone methylation. *Cell Cycle* 2011. 10(3):413-24
- The International Cancer Genome Consortium (including Lopez-Bigas N). International network of cancer genome projects. *Nature* 2010. 464, 993-998
- Gunes Gunes, Christian Perez-Llamas, Alba Jené, Khademul Islam, Anna Kedzierska, Jordi Deu-Pons, Simon J. Furney and Nuria Lopez-Bigas. IntOGen: A novel framework for integration and data-mining of multidimensional oncogenomic data. *Nature Methods* 2010. 7, 92-93
- Holger Richly, Luciana Rocha-Viegas, Joana Domingues Ribeiro, Santiago Demajo, Gunes Gundem, Nuria Lopez-Bigas, Tekeya Nakagawa, Sabine Rospert, Takashi Ito and Luciano Di Croce. Transcriptional Activation of Polycomb-repressed genes by the H2A-ubiquitin binding protein ZRF1. *Nature* 2010. 468, 1124-1128.
- Aguilar H, Sole X, Bonifaci N, Serra-Musach J, Islam A, Lopez-Bigas N, Méndez-Pertuz M, Beijersbergen RL, Lázaro C, Urruticoechea A, Pujana MA. Biological reprogramming in acquired resistance to endocrine therapy of breast cancer. *Oncogene* 2010. Aug 16.
- Ferreiro I#, Joaquin M#, Islam A, Gomez G, Barragan M, Lombardía L, Domínguez O, Pisano DG, Lopez-Bigas N, Nebreda AR, Posas F. Whole genome analysis of p38 SAPK-mediated gene expression upon stress. *Genome Biology* 2010.1;11(1):144
- Beshiri ML, Islam A, DeWaal DC, Richter WF, Love J, Lopez-Bigas N, Benevolenskaya EV. Genome-wide Analysis using ChIP to Identify Isoform-specific Gene Targets. *JoVE* doi: 10.3791/2101.
- Mascarell-Creus A, Cañizares J, Vilarrasa J, Mora-García S, Blanca J, Gonzalez-Ibeas D, Saladié M, Roig C, Deleu W, Picó B, López-Bigas N, Aranda MA, Garcia-Mas J, Nuez F, Puigdomènech P, Caño-Delgado A. An oligo-based microarray offers novel transcriptomic approaches for the analysis of pathogen resistance and fruit quality traits in melon (*Cucumis melo* L.) *BMC Genomics* 2009. 10(1):467.
- Rodilla, V., Villanueva, A., Obrador-Hevia, A., Robert-Moreno, A., Fernández-Majada, V., Grilli, A., López-Bigas, N., Bellora, N., Albà, M., Duñach, M, Sanjuan, X., Gonzalez, S., Gridley, T., Capella, G., Bigas, A., Espinosa, LI. Notch is an Essential Component Downstream of β -catenin in Colorectal Cancer. *PNAS* 2009. 106(15):6315-20.
- Solé, X., Bonifaci, N., López-Bigas, N., Berenguer, A., Hernández, P., Reina, O., Maxwell, C., Aguilar, H., Urruticoechea, A., de Sanjosé, S., Comellas, F., Capellá, G., Moreno, V., Pujana, MA. Biological convergence of cancer signatures. *PLoS ONE* 2009. 4(2): e4544

Biomedical Genomics Group, Cancer Program

Institute for Research in Biomedicine (IRB Barcelona), 08028 - Barcelona. T 934039912.

nuria.lopez@irbbarcelona.org <http://bbglab.irbbarcelona.org>

Furney SJ, Calvo B, Larranaga P, Lozano JA, Lopez-Bigas N. Prioritization of candidate cancer genes - an aid to oncogenomic studies. *Nucleic Acids Research* 2008. e115.

Lopez-Bigas N, Kisiel TA, Volkert T, Gupta S, Love J, Murria H.L., Young R.A., and Benevolenskaya E.V. Genome-wide analysis of the H3K4 histone demethylase RBP2 reveals a transcriptional program controlling differentiation. *Molecular Cell* 2008. 31:520-530.

The STAR consortium (including Shikhagaie M and Lopez-Bigas N). SNP and haplotype mapping for genetic analysis in the Rat. *Nature Genetics* 2008. 40:560-6.

Subhajyoti De, Lopez-Bigas N, Sarah A Teichmann. Patterns of evolutionary constraints on genes in humans. *BMC Evolutionary Biology* 2008. 8:275.

Lopez-Bigas N, De S. & Teichmann SA. Evolvability and protein divergence in the evolution of *Homo sapiens*. *Genome Biology* 2008. 9:R33.

Furney SJ#, Madden SF#, Kisiel TA, Higgins DG and Lopez-Bigas N. (2007). Distinct patterns in the regulation and evolution of human cancer genes. *In Silico Biology* 2008. 8:33-46.

The ENCODE Project Consortium (including Lopez-Bigas N). Identification and analysis of functional elements in 1% of the human genome by the ENCODE pilot project. *Nature* 2007. 447(7146): 799-816.

Calvo, B., Lopez-Bigas N, Furney SJ, Larranaga P, Lozano JA. A partially supervised classification approach to dominant and recessive human disease gene prediction. *Comput Methods Programs Biomed* 2007. 85(3): 229-37.

Marques-Bonet T , Sanchez-Ruiz J, Armengol L, Khaja R , Bertranpetit J, Rocchi M, Gazave E, Lopez-Bigas N, Navarro A. On the association between chromosomal rearrangements and genic evolution in humans and chimpanzees. *Genome Biology* 2007. 8(10).

Furney SJ, Albà MM, Lopez-Bigas N. Differences in the evolutionary history of disease genes affected by dominant or recessive mutations. *BMC Genomics* 2006. 7(0):165.

Refereed publications during postdoc position

Furney SJ, Higgins DG, Ouzounis CA, & Lopez-Bigas N. Structural and functional properties of genes involved in human cancer. *BMC Genomics*. 2006, 22:269-77.

Tiffin, N., Adie, E., Turner, F., Brunner, H.G., van Driel, M.A., Oti, M., Lopez-Bigas, N., Ouzonis, C. A., Perez-Iratxeta, C., Andrade, M., Adeyemo, A., Patti, M.E., Semple, C. & Hide, W. In silico candidate disease gene identification: a concert of methods identifies type 2 diabetes and obesity candidate genes. *Nucleic Acids Res*. 2006. 34(10):3067-81.

Lopez-Bigas N, Blencowe BJ & Ouzounis CA. Highly consistent patterns for inherited human diseases at the molecular level. *Bioinformatics*. 2006, 11;7:3

Karp PD, Ouzounis CA, Moore-Kochlacs C, Goldovsky L, Kaipa P, Ahren D, Tsoka S, Darzentas N, Kunin V & Lopez-Bigas N. Expansion of the BioCyc collection of pathway/genome databases to 160 genomes. *Nucleic Acids Res*. 2005, 33:6083-9.

Goldovsky L, Janssen P, Ahren D, Audit B, Cases I, Darzentas N, Enright AJ, Lopez-Bigas N, Peregrin-Alvarez JM, Smith M, Tsoka S, Kunin V, Ouzounis CA. CoGenT++: an extensive and extensible data environment for computational genomics. *Bioinformatics*. 2005, 21:3806-10.

Lopez-Bigas N, Audit B, Parra G, Ouzounis CA, Guigo R. Are splicing mutations the most frequent cause of hereditary disease? *FEBS letters* 2005, 28:1900-1903.

Lopez-Bigas N, Ouzounis CA. Genome-wide identification of genes likely to be involved in human genetic disease. *Nucleic Acids Research* 2004, 32:3108-3114.

Biomedical Genomics Group, Structural and Computational Biology Programme
Institute for Research in Biomedicine (IRB Barcelona), 08028 - Barcelona. T 934039912.
nuria.lopez@irbbarcelona.org <http://bbglab.irbbarcelona.org>

Janssen PJ, Audit B, Cases I, Darzentas N, Goldovsky L, Kunin V, Lopez-Bigas N, Peregrin-Alvarez JM, Pereira-Leal JB, Tsoka S, Ouzounis CA Beyond 100 genomes. *Genome Biology* 2003, 4:402

Refereed publications arising from PhD work

Rabionet, R, Morales-Peralta, E, Lopez-Bigas, N, Arbonés, ML and Estivill, X. A novel G21R mutation of the GJB2 gene causes autosomal dominant non-syndromic congenital deafness in a Cuban family. *Genetics and Molecular Biology*. 2006. 29(3):443-445.

Lopez-Bigas N, Rabones ML, Estivill X, Simonneau L, Expression profiles of the connexin genes, Gjb1 and Gjb3, in the developing cochlea. *Mechanisms of Development* 2002. 119 Suppl 1:S111-5

Rabionet R, López-Bigas N, Rabones ML, Estivill X. Connexin mutations in hearing loss, dermatological and neurological disorders. *TRENDS in Mol Med* 2002 8:205-212

Lopez-Bigas N, Melchionda S, Gasparini P, Borragan A, Arbones ML, Estivill X. A common frameshift mutation and other variants in GJB4 (connexin 30.3): Analysis of hearing impairment families. *Hum Mutat* 2002 19:458

Lopez-Bigas N, Olive M, Rabionet R, Ben-David O, Martinez-Matos JA, Bravo O, Banchs I, Volpini V, Gasparini P, Avraham KB, Ferrer I, Arbones ML, Estivill X. Connexin 31 (GJB3) is expressed in the cochlea and peripheral and auditory nerves and causes neuropathy and hearing impairment. *Hum Mol Genet* 2001 10:947-952

Lopez-Bigas N, Rabionet R, Arbonès ML, Estivill X. R32W variant in connexin 31: mutation or polymorphism for deafness and skin disease?. *Eur J Hum Genet* 2001 9:70

Lopez-Bigas N, Melchionda S, de Cid R, Grifa A, Zelante L, Govea N, Arbones ML, Gasparini P, Estivill X. Identification of five new mutations of PDS/SLC26A4 in Mediterranean families with hearing impairment. *Hum Mutat* 2001 18:548

Lopez-Bigas N, Rabionet R, Martinez E, Bravo O, Giron J, Borragan A, Pellicer M, Arbonés ML, Estivill X. Mutations in the mitochondrial tRNA Ser(UCN) and in the GJB2 (connexin26) gene are not modifiers of the age at onset and severity of hearing loss in Spanish patients with the 12S rRNA A1555G mutation. *Am J Hum Genet* 2000 66:1465-1467

Lopez-Bigas N, Rabionet R, Martinez E, Banchs I, Volpini V, Vance JM, Arbonès ML, Estivill X. Identification of seven new variants (five nucleotide and two amino acid substitutions) in the Connexin31 (GJB3) gene. *Hum Mutat* 2000 15:481-482

Rabionet R, Zelante L, López-Bigas N, D'Agruma L, Melchionda S, Restagno G, Arbonès ML, Gasparini P, Estivill X. Molecular basis of childhood deafness due to mutations in the GJB2 (Connexin26) gene. *Hum Genet* 2000 106:40-44

Grifa A, Wagner CA, D'Ambrosio L, Melchionda S, Bernardi F, López-Bigas N, Rabionet R, Arbonès R, Della Monica M, Estivill X, Zelante L, Lang P, Gasparini P. Mutations in GJB6 cause a form of nonsyndromic autosomal dominant deafness at DFNA3 locus. *Nature Genet* 1999 23:16-18

Lopez-Bigas N, Rabionet R, de Cid R, Govea N, Gasparini P, Arbonès ML, Estivill X. A splice site mutation in the PDS gene may result in intrafamilial variability for deafness in Pendred syndrome. *Hum Mutat* 1999. 14:520-526

Torrioni A, Cruciani F, Rengo C, Sellitto D, López-Bigas N, Rabionet R, Govea N, Lopez De Munain A, Sarduy M, Romero L, Villamar M, del Castillo I, Moreno F, Estivill X, Scozzari R. The A1555G mutation in the 12S rRNA gene of human mtDNA: Recurrent origins and

Biomedical Genomics Group, Cancer Program
Institute for Research in Biomedicine (IRB Barcelona), 08028 - Barcelona. T 934039912.
nuria.lopez@irbbarcelona.org <http://bbglab.irbbarcelona.org>

founder event in families affected by sensorineural deafness. Am J Hum Genet 1999
65:1349-1358

Non-refereed publications and book chapters

David Tamborero, Abel Gonzalez-Perez and Lopez-Bigas N. Identification of oncogenic driver mutations Experimental Medicine. Vol 32 No 12 2014.

Simon J. Furney, Gunes Gunes and Nuria Lopez-Bigas. Oncogenomics. Book Chapter in “Genetics of Complex Human Diseases: A Laboratory Manual” CSHL Press 2009.

Lopez-Bigas N, Alba MM, Eduardo E. Genomics and Bioinformatics lecture book. Catalanian Open University (UOC) 2005.

Lopez-Bigas N, Rabionet R, Estivill X. Genética de la sordera: Identificando las moléculas de la audición (Genetics of Deafness: Finding the molecules for hearing). Investigación & Ciencia 2000 Diciembre.

Lopez-Bigas N, Govea N, Rabionet R, Arbonés ML, Estivill X. Genética y Genómica de la sordera no sindrómica (Genetics and Genomics of non-syndromic deafness). In: Tratado de Otorrinolaringología Pediátrica 2000. 227-235.