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Formative and professional positions

High school Period:

SANTA ANA HIGH SCHOOL (California, USA).

- Attendance to the senior course within an international exchange program (from September 1985 to August 1986)

INSTITUT OF PROFESIONAL FORMATION GUINEUETA. Barcelona.

- Clinical analysis course with an European Fundation Fellowship. (1991-1992)

University Studies Period:

UNIVERSITY OF GÖTTINGEN (Germany)

- Stay with an **ERASMUS fellowship** in the laboratories of the Institute of Biochemistry (with Professor R. Zimmermann) and the Institute of Molecular Genetics (with Professor J. Fritz) of the University of Göttingen. (From August of 1993 to August of 1994).

UNIVERSITY OF BARCELONA. Barcelona,

- **Graduated in Biology.** Orientation to Biochemistry and Molecular Biology. (1994)

PhD Period:

UNIVERSITY OF BARCELONA. Barcelona, from Sept. 1994 to Feb. 2000.
Ph.D. in Molecular Biology. Dep.of Biochem. & Mol. Biol.
Supervisor: Professor Manuel Palacín.
Thesis title: "Identification of the light subunit of heteromeric amino acid transporter (LSHAT) family. y^+LAT-1 causes Lysinuric Protein Intolerance (LPI)".
Highest qualifications and **PhD extraordinary prize,** University of Barcelona (2000).

UNIVERSITY OF GIESSEN (Germany).
Pre-doctoral stay in the Laboratory of the Institute of Nutrition (with Professor H. Danniel). (Two months in 1998)

Post-Doc Period:

EUROPEAN MOLECULAR BIOLOGY LABORATORY (EMBL).
(Heidelberg. March 2000 – February 2006)
Post-Doc in the group of Dr. Peer Bork,
From 2000 to 2002 financed through a European Molecular Biology Organization (**EMBO**) **long-term fellowship**.
From 2002 to 2006 with different contracts linked to different projects.
Main Projects: Analysis of gene evolution of Metazoa. Identification and classification of pseudogenes in mammals. Primary Genome sequence annotation.

Principal Investigator Period:

ICREA Research Professor at the Barcelona Supercomputing Center (BSC-CNS),
(February 2006 -)
Principal investigator of the Computational Genomics Group at the Department of Life Sciences, Barcelona Supercomputing Center- Centro Nacional de Supercomputación (BSC-CNS)

Vice-president of the Bioinformatics Barcelona (https://www.bioinformaticsbarcelona.eu/) (2022-present)
Co-coordinator of the GA4GH Cancer Group (2020-2023)

Research trajectory

Summary

My scientific career began in 1994, following the completion of a Bachelor's degree in Biology (Biochemistry, Molecular Biology, and Genetics) at the University of Barcelona. My first research experience was as an Erasmus student at the University of Göttingen, where I worked in two laboratories specializing in Molecular Genetics and Biochemistry. I pursued my PhD in Manuel Palacín's lab (UB) under an FPI fellowship, focusing on the molecular and biochemical mechanisms of amino acid transport. Over five years, I identified a new family of amino acid transporters linked to aminoaciduria (see publications from 1998 to 2000), integrating bioinformatics tools alongside molecular biology and genetics techniques.

In 2000, I transitioned fully into bioinformatics by joining Peer Bork's group at EMBL (Heidelberg) with an EMBO long-term postdoctoral fellowship. During nearly seven years at EMBL, I contributed to the annotation and evolutionary analysis of genes in the first eukaryotic whole-genome sequences, particularly focusing on functional duplications and pseudogenes (see publications from 2002 to 2007).

In 2006, I joined the Barcelona Supercomputing Center (BSC) as an ICREA Research Professor, establishing the Computational Genomics Group. Initially, the group focused on the regulatory aspects of the genome. Through participation in FP7 projects (MetaHit and MITIN), I built a stable team and launched two key research lines: (i) genomics and systems biology, and (ii) genetic variability in complex diseases. Collaborations in genome annotation, particularly for regulatory regions, led to the development of methodology and tools, which facilitated participation in EC-funded consortia (BLUEPRINT, PROCOGEN under FP7, and T2DSystems under H2020).

By 2009, the group completely shifted its focus toward biomedicine, exploring the interplay between genome variation and disease. On the cancer front, we contributed to the ICGC consortium, through the genomic study of Chronic Lymphocytic Leukemia (CLL) using newly developed tools to characterize the somatic landscape of tumor genomes. TH egroup also coordinated BSC's role in the ICGC-PanCancer project, by executing and supervising the analysis of nearly 3,000 tumor genomes. Building on this, we later expanded our cancer genomic studies to include different tumor types and analyses.

In parallel, we initiated work on complex diseases, developing comprehensive data analysis approaches to investigate genetic architecture and risk prediction. More recently, the group has begun incorporating lifestyle factors into these studies to create personalized prevention protocols, leveraging AI methods such as machine learning.

In recent years, we've ventured into large-scale infrastructure projects, coordinating the EUCANCan initiative (2019–2024) to create interoperable infrastructures for managing, analyzing, and sharing cancer genomic and clinical data (see www.eucancan.com). These efforts, alongside regional personalized medicine initiatives and involvement with the Global Alliance for Genomics and Health, have positioned our group as a key player in advancing genomic biomedical research on an international scale.

Publications

Some author lists are collapsed for formatting reasons.
Only publications with direct contributions are included

Within the past 5 years

- Martín R, Gaitán N, **Torrents D.**

Protocol for the assessment, improvement, and harmonization of somatic variant calling using ONCOLINER
STAR Protoc. . 2024 Dec 20;6(1):103533. doi: 10.1016/j.xpro.2024.103533. Online ahead of print
PMID: 39708326

- Martín R, Gaitán N, Jarlier F, Feuerbach L, de Soyres H, Arbonés M, Gutman T, Puiggròs M, Ferriz A, Gonzalez A, Estelles L, Gut I, Capella-Gutierrez S, Stein LD, Brors B, Royo R, Hupé P, **Torrents D.**

ONCOLINER: A new solution for monitoring, improving, and harmonizing somatic variant calling across genomic oncology centers.
Cell Genom. 2024 Sep 11;4(9):100639. doi: 10.1016/j.xgen.2024.100639. Epub 2024 Aug 30.
PMID: 39216474

- Terradas M, Schubert SA, Viana-Errasti J, Ruano D, Aiza G, Nielsen M, Marciel P, Tops CM, Parra G, Morreau H, **Torrents D**, van Leerdam ME, Capellá G, de Miranda NC, Valle L, van Wezel T.

Germline NPAT inactivating variants as cause of hereditary colorectal cancer.
Eur J Hum Genet. 2024 Jul;32(7):871-875. doi: 10.1038/s41431-024-01625-8. Epub 2024 May 22.
PMID: 38778081

- Riba M, Sala C, Culhane AC, Flobak Å, Patocs A, Boye K, Plevova K, Pospíšilová Š, 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, Arenas Marquez J, Belien J, De Nicolo A, De Maria R, **Torrents D**, Tonon G.

The 1+Million Genomes Minimal Dataset for Cancer.
Nat Genet. 2024 May;56(5):733-736. doi: 10.1038/s41588-024-01721-x.
PMID: 38702538

- Gómez-Sánchez G, Alonso L, Pérez MÁ, Morán I, **Torrents D**, Berral JL.

Exhaustive Variant Interaction Analysis using Multifactor Dimensionality Reduction.
Res Sq . 2023 Oct 16:rs.3.rs-3401025. doi: 10.21203/rs.3.rs-3401025/v1.
PMID: 37886566

- Rodriguez-Fos E, Planas-Fèlix M, Burkert M, Puiggròs M, Toedling J, Thiessen N, Blanc E, Szymansky A, Hertwig F, Ishaque N, Beule D, **Torrents D**, Eggert A, Koche RP, Schwarz RF, Haase K, Schulte JH, Henssen AG.

Mutational topography reflects clinical neuroblastoma heterogeneity.
Cell Genom. 2023 Sep 7;3(10):100402. doi: 10.1016/j.xgen.2023.100402.
PMID: 37868040

- Quintana I, Terradas M, Mur P, Te Paske IBAW, Peters S, Spier I, Steinke-Lange V, Maestro C, **Torrents D**, Puiggròs M, Royo R, Tonda R, Parra G, Piscia D, Beltrán S, Navarro M, Piñol V, Brunet J, Gonzalez-Abuin N, Aiza G, Sommer A, van Herwaarden Y, Astuti G, Holinski-Feder E, Hoogerbrugge N, de Voer RM, Aretz S, Capellá G, Valle L.

Wnt genes in colonic polyposis predisposition.
Genes Dis. 2022 Dec 29;10(3):753-757. doi: 10.1016/j.gendis.2022.12.002.
PMID: 37396538

- Font-Porterias N, García-Fernández C, Aizpurua-Iraola J, Comas D, **Torrents D**, de Cid R, Calafell F.
Sequence diversity of the uniparentally transmitted portions of the genome in the resident population of Catalonia.
Forensic Sci Int Genet. 2022 Nov;61:102783. doi: 10.1016/j.fsigen.2022.102783. Epub 2022 Sep 28
PMID: 36240588

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Detection of early seeding of Richter transformation in chronic lymphocytic leukemia.
Nat Med. 2022 Aug;28(8):1662-1671. doi: 10.1038/s41591-022-01927-8. Epub 2022 Aug 11.
PMID: 35953718

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Recessive Genome-Wide Meta-analysis Illuminates Genetic Architecture of Type 2 Diabetes.
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Clustering and graph mining techniques for classification of complex structural variations in cancer genomes.
Sci Rep. 2022 Feb 28;12(1):3244. doi: 10.1038/s41598-022-07211-6.
PMID: 35228601

- COVID-19 Host Genetics Initiative (Includes **D. Torrents**).
Mapping the human genetic architecture of COVID-19.
Nature. 2021 Dec;600(7889):472-477. doi: 10.1038/s41586-021-03767-x. Epub 2021 Jul 8.
PMID: 35922517

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International policies and standards for data sharing across genomic research and healthcare.
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TIGER: The gene expression regulatory variation landscape of human pancreatic islets.
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The impact of non-additive genetic associations on age-related complex diseases.
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Genomic and epigenomic insights into the origin, pathogenesis, and clinical behavior of mantle cell lymphoma subtypes.
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Retrospective evaluation of whole exome and genome mutation calls in 746 cancer samples.
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Research Grants

Active during the last 5 years

- **Project title:** Integrative analysis of genetic, clinical and lifestyle factors involved in the risk and prevention of complex diseases (PREVDIS)

Agency: Plan Nacional I+D+i, Ministerio de Educación y Ciencia.
Coordinator: David Torrents (BSC)
Duration: 04/12/2024-31/12/2027
Amount for the group: 194K Euros

- **Project title:** Extreme Near-Data Processing Platform (NEARDATA)

Agency: Horizon Europe_Cluster 4_HORIZON-CL4-2022-DATA-01-05.
Coordinator: University of Rovira i Virgili (Tarragona)
Duration: 01/01/2023-31/12/2025
Amount for the group: 170K Euros

- **Project title:** Grup Consolidat AGAUR

Agency: Catalan Administration AGAUR
Coordinator: David Torrents (BSC)
Duration: 01/01/2021-31/12/2025
Amount for the group: 60K Euros

- **Project title:** Clinical impact of genomic anlysis in Blood Noeplasms (PREGENLINF)

Agency: PMP21-ISCIII. Instituto de Salud Carlos III
Coordinator: IDIBAPS (Barcelona)
Duration: 07/11/2022-31/12/2025
Amount for the group: 278K Euros

- **Project title:** Development and integration of integrated aritficial intellingence models for the prediction of T2D risk (AI4T2D)

Agency: Spanish Plan Complementario de las CC.AA.
Coordinator: David Torrents (BSC)
Duration: 07/03/2023-31/12/2024
Amount for the group: 100K Euros

- **Project title:** Benchmarking platform for the analysis of genomic structural variation and its application to cancer and healthy neural tissues

Agency: Plan Nacional I+D+i, Ministerio de Educación y Ciencia.
Coordinator: David Torrents (BSC)
Duration: 01/09/2021-31/08/2024
Amount for the group: 120K Euros

- **Project title:** EUCANCan; A federated network of aligned and interoperable infrastructures for the homogeneous analysis and sharing of cancer genomics data.

Agency: EU Horizon 2020- SC1-BHC-2018-2020
Coordinator: David Torrents (BSC)
Duration: 01/01/2019-31/07/2023
Amount for the group: 800K Euros

- **Project title:** CLLEvolution: Genomic and epigenomic drivers of disease evolution in chronic lymphocytic leukemia targets for clinical intervention.

Agency: La CAIXA-Recercaixa, Projectes de Salut
Coordinator: IDIBAPS (Barcelona)
Duration: 01/12/2019-31/12/2021
Amount for the group: 90K Euros

- **Project title:** TransTumVar: beyond the identification and classification of structural variation across different tumors: new approaches and translation into the clinics

Agency: Ministerio de Economía y Competitividad
Coordinator: David Torrents
Duration: from 1/1/2018 to 31/12/2021
Amount for the group: 160K Euros

> ***5 years ago.***

- **Project title:** Development of a systems biomedicine approach for risk identification, prevention and treatment of type 2 diabetes (667191-2); T2DSystem

Agency: Horizon 2020-PHC-2015
Coordinator: ULB (Brussels)
Duration: from 1/1/2016 to 1/1/2019

- **Project title:** Functional and clinical impact of genomic analysis in cll (PMP15/00007). PerMed.

Agency: Instituto de Salud Carlos III (ISCIII)
Coordinator: Elias Campo (IDIBAPS, Barcelona)
Duration: from Jan 2016 to Dec 2019

- **Project title:** SMUFIN

Agency: Caixaimpulse. La Caixa
Coordinator: David Torrents
Duration: from 1/1/2015 to 1/1/2018

- **Project title:** Identificacion y caracterizacion de reordenamientos cromosomicos complejos en multiples genomas de diferentes tipos de tumor. (SAF2014-60293-R)

Agency: Plan Nacional, Ministerio de Economía y Competitividad.
Duration: from Jan 2014 to Dec 2017

- **Project title:** Understanding of the conifer genome (ProCoGen) (FP7-KBBE-2011-5: 289841)

Agency: European 7th Framework Programme
Coordinator: Carmen Diaz-Sala
Duration: from 1/1/2011 to 1/1/2015

- **Project title:** Systems biology analysis of immune tolerance in organ transplantation

Agency: Fundació Marató TV3
Coordinator: Juanjo Lozano
Duration: from 1/1/2013 to 31/12/2015

- **Project title:** A *BLUEPRINT* of haematopoietic epigenomes (BLUEPRINT) (HEALTH.2011.2.1.1-1 – FP7 FP7-KBBE-2011-5)
Agency: European 7th Framework Programme
Coordinator: Willcocks S.
Duration: from 1/1/2011 to 1/1/2016

- **Project title:** Metagenomics of the human intestinal tract (METAHIT, HEALTH-F4- 2007-201052).
Agency: European 7th Framework Programme
Coordinator: Dr. S. Dusko Ehrlich (INRA)
Duration: from 1/1/2008 to 1/1/2012

- **Project title**: Integration of the systems models of mitochondrial functiona and insulin signalling, and its application in the study of complex diseases (MITIN, HEALTH-F4-2008-223450).
Agency: European 7th Framework Programme
Partner: WP1 leader, David Torrents Arenales (coordinator: Antonio Zorzano, IRB)
Duration: from 11/11/2008 to 11/11/2011

- **Project title:** Estudios a gran escala y en detalle de las estrategias y mecanismos responsables de la evolución génica en vertebrados y su implicación en la generación de diversidad biológica (BIO2006-15036).
Agency: Plan Nacional I+D+i, Ministerio de Educación y Ciencia.
Principal Investigator: David Torrents Arenales
Duration: from 01/10/2006 to 30/09/2009

- **Project title:** Genome re-modelling in evolution: functional annotation of tandem gene duplications in drosophila and other invertebrates
Agency: Fundación BBVA.
Principal Investigator: Miguel Manzanares (CSIC, Madrid)
Duration: from 2004 to 2007

Contracts and technology transfer

Contract. Analysis of potential pleiotropic effects of specific target genes, through a comprehensive PheWAS analysis across a large population. Almirall S.A.. David Torrents. 13/03/2023-12/03/2024. 104.680,00 €.
Contract. Identification of new gene–disease associations for atopic dermatitis as a dermatology indication of interest for Almirall using advanced genomic and computational technologies. Almirall-Prodesfarma, S.A.. David Torrents. 17/12/2020-17/06/2021. 50.000 €.
Contract. Analysis of genetic data for dermatological indications. Almirall-Prodesfarma, S.A.. David Torrents. 01/10/2018-31/01/2020. 80.000 €.

Patent: A computer-implemented and reference-free method for identifying variants in nucleic acid sequences (EP16178577.9); Registered: May 2016; Inventors: David Torrents, David Carrera, Mercè Planes, Jordà Polo.

Teaching

Master in Biomedicine and health. Faculty of Medicine (University of Barcelona (2014- to present)
Master in Biomedicine, Bioinformatics and Genomics (University of Barcelona, 2006-to present)
Molecular Evolution Course (University of Barcelona)
Phylogeny and genealogy course (University of Barcelona)
Advanced bioinformatics, Biotechnology studies (University of Vic)
Comparative Genomics (University of Barcelona)
Bioinf. approaches in Biomedical Res. (Oncological Institute, IDIBELL)

Directed Thesis

During the last 5 years:

PhD Student: Ana Dueso
Title: Somatic processed pseudogenes and micropeptides in cancer: insights from large-scale genomic studies
Organism: UNIVERSITY OF BARCELONA, BIOMEDICINE
Year: 2024

PhD Student: Romina Royo
Title: Development and application of methodologies and infrastructures for cancer genome analysis within personalized medicine.
Organism: UNIVERSITY OF BARCELONA, BIOMEDICINE
Year: 2023

PhD Student: Lorena Alonso
Title: From the discovery of epistatic events in type 2 diabetes mellitus to the study of related gene expression regulatory variation.
Organism: UNIVERSITY OF BARCELONA, BIOMEDICINE
Year: 2023

PhD Student: Luisa Delgado
Title: Identification and characterization of new complex patterns of structural dna and rna alterations in cancer.
Organism: UNIVERSITY OF BARCELONA, BIOMEDICINE
Year: 2021

PhD Student: Jordi Valls
Title: Comprehensive identification and characterization of germline structural variation within the iberian population.
Organism: UNIVERSITY OF BARCELONA, BIOMEDICINE
Year: 2021

PhD Student: Mercè Planas
Title: Detection and classification of somatic structural variants, and its application in the study of neural development
Organism: UNIVERSITY OF BARCELONA, BIOMEDICINE
Year: 2020

PhD Student: Elias Rodriguez
Title: Study of complex chromosomal rearrangements in cancer. the role of extrachromosomal circular dna as a genome remodeler in neuroblastoma
Organism UNIVERSITY OF BARCELONA, BIOMEDICINE
Year: 2020

> 5 years

PhD Student: Marta Guindo
Title: A systematic and comprehensive approach for large genome-wide association studies
Organism: University of Barcelona, Biomedicine
Year: 2019

PhD Student: Sílvia Bonàs
Title: Implementation of a novel analytical framework for large-scale genetic data. extending the genetic architecture of type 2 diabetes beyond common variants
Organism: University of Barcelona, Biomedicine
Year: 2017

PhD Student: Santiago Gonzalez
Title: Identification and characterization of non-coding genomic variations associated to cancer disease
Organism: University of Barcelona, Biomedicine.
Year: 2016

PhD Student: Leyden Fernandez
Title: The role of genomic regulatory regions in the adaptation of prokaryotes to environmental factors
Organism: Universitat de Barcelona (UB)
Year: 2015