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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 PROFESSIONAL FORMATION GUINEUETA. Barcelona.

- Clinical analysis course with an European Foundation 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. γ^+ 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. Daniel). (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 2004 with different contracts linked to different projects.

From 2004 to 2006 as an EMBL Staff (grade 7)

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)

IP and coordinator at the BSC of the PanCancer ICGC-TCGA project (2013).

Vice-president of the Bioinformatics Barcelona (<https://www.bioinformaticsbarcelona.eu/>)

Research trajectory

Summary

My scientific career started when I finished the bachelor in Biology (Biochemistry, Molecular Biology and Genetics) at the University of Barcelona (1994). The first contact with research projects was as Erasmus student at the University of Göttingen in 1994. During that stay, I was in two laboratories working in Molecular Genetics and Biochemistry. Afterwards, I started my PhD in the Manuel Palacin's lab (UB) to study the molecular and biochemical aspects of the amino acid transport under the framework of an FPI fellowship. After five years, I finished the PhD identifying a new amino acid transporter family related to aminoaciduria (see publications between 1998 and 2000). A part of the molecular biology and genetics technics, I also used bioinformatics tools. In 2000, I started my bioinformatics career by joining Dr. Bork group in the EMBL (Heidelberg) by getting an EMBO long-term postdoctoral fellowship. During almost seven years at EMBL I was directly involved in the annotation of genes of the first big eukaryotic genomes sequenced, as well as in the identification and characterization of genetic duplications and pseudogenes, as part of different sequencing and genome analysis consortiums (see publications between 2002 and 2007).

In 2006, I joined the BSC through an "ICREA Research Professor" position. I was awarded with my first National Project from MICINN (BIO2006-15036) as PI, and I started my own group in the BSC doing metagenomics studies as well as annotations of regulating regions in eukaryotic genomes. The participation as PI in another two FP7 projects (MetaHit and MITIN), allowed me to constitute a stable group and to impulse two research lines: (i) studies related to genomics and systems biology; and (ii) genetic variability associated to complex diseases and human gut metagenomics. In parallel, I also pushed other collaborations in the framework of genome annotations, specifically of regulatory regions, where we developed our own tools (ReLA). This led us to participate later in three consortiums funded by the EC, BLUEPRINT and PROCOGEN in the FP7 and T2DSYSTEMS in H2020.

Starting in the context of the ICGC consortium, we contributed over the last years to the genomic study of the CLL (Chronic Lymphocytic Leukemia) genome, by developing and applying SMUFIN, a tool, able to characterize with high detail complex karyotypes associated to aggressive tumors. At the same time, I coordinated the implication of the BSC in the ICGC-PanCancer, as one of the analysis centers, with the objective of studying the complete genome of more nearly 3000 tumors. This research activity has pointed the BSC, as one of the reference centers for the analysis of genomes in biomedicine and, in particular, genomic oncology and its implantation into health care systems, setting up the basis for a personalized medicine. From this activity, we further applied our expertise and methodology to analyze cancer genomes in different contexts. One of these analyses, expands the analysis to other type of DNA, circular DNA, that have a role in the progression of the Neuroblastoma tumor. We are currently applying more sophisticated computational and mathematical models to capture more layers of complexity around disease genomes. These integrative and artificial intelligence-based models can identify more subtle correlations, including with clinical value for prognosis, diagnosis and treatment.

Finally, the group is also coordinating the generation of infrastructures for the management and analysis of genomic data in a Personalized Medicine context. We are coordinators of a project EUCANCAN that aims at implementing a interoperative infrastructure across different personalized medicine nodes (see www.eucancan.com). This activity, together with other regional initiatives for Personalized Medicine, and our implication within the Global Alliance for Genomics and Health, sets our group as one of the reference groups pushing forward genomic biomedical research internationally.

Publications

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

- 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
- Rodríguez-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-Porterías 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
- Nadeu F*, Royo R*, Massoni-Badosa R*, Playa-Albinyana H, 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, Giró A, Verdaguer-Dot N, Romo M, Clot G, Rozman M, Frigola G, Rivas-Delgado A, Baumann T, Alcoceba M, González M, Climent F, Abrisqueta P, Castellví J, Bosch F, Aymerich M, Enjuanes A, Ruiz-Gaspà S, López-Guillermo A, Jares P, Beà S, Capella-Gutierrez S, Gelpí JL, López-Bigas N, **Torrents D**, Campbell PJ, Gut I, Rossi D, Gaidano G, Puente XS, Garcia-Roves PM, Colomer D, Heyn H, Maura F, Martín-Subero JI, Campo E.
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
- Valls-Margarit J*, Galván-Femenía I*, Matías-Sánchez D*, Blay N, Puiggròs M, Carreras A, Salvo C, Cortés B, Amela R, Farre X, Lerga-Jaso J, Puig M, Sánchez-Herrero JF, Moreno V, Perucho M, Sumoy L, Armengol L, Delaneau O, Cáceres M, de Cid R*, **Torrents D***.
GCAT|Panel, a comprehensive structural variant haplotype map of the Iberian population from high-coverage whole-genome sequencing.
Nucleic Acids Res. 2022 Mar 21;50(5):2464-2479. doi: 10.1093/nar/gkac076.
PMID: 35176773
- O'Connor MJ, Schroeder P, Huerta-Chagoya A, Cortés-Sánchez P, Bonàs-Guarch S, Guindo-Martínez M, Cole JB, Kaur V, **Torrents D**, Veerapen K, Grarup N, Kurki M, Rundsten CF, Pedersen O, Brandslund I, Linneberg A, Hansen T, Leong A, Florez JC, Mercader JM.
Recessive Genome-Wide Meta-analysis Illuminates Genetic Architecture of Type 2 Diabetes.
Diabetes. 2022 Mar 1;71(3):554-565. doi: 10.2337/db21-0545.

PMID: 34862199

- Gomez-Sanchez G, Delgado-Serrano L, Carrera D, **Torrents D**, Berral JL.
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
- Rehm, H., Page A., Smith L, (the GA4GH consortium), ... Birney E. GA4GH:
International policies and standards for data sharing across genomic research and healthcare.
Cell Genomics 2021 Volume: 1, Issue: 2, pp 100029 DOI: 10.1016/J.XGEN.2021.100029
PMID: 35072136
- Alonso L, Piron A, Morán I, Guindo-Martínez M, Bonàs-Guarch S, Atla G, Miguel-Escalada I, Royo R, Puiggròs M, Garcia-Hurtado X, Suleiman M, Marselli L, Esguerra JLS, Turatsinze JV, Torres JM, Nylander V, Chen J, Eliasson L, Defrance M, Amela R; MAGIC, Mulder H, Gloyn AL, Groop L, Marchetti P, Eizirik DL, Ferrer J, Mercader JM, Cnop M, **Torrents D**.
TIGER: The gene expression regulatory variation landscape of human pancreatic islets.
Cell Rep. 2021 Oct 12;37(2):109807. doi: 10.1016/j.celrep.2021.109807.
PMID: 34644572
- Pastor-Ibáñez R, Díez-Fuertes F, Sánchez-Palomino S, Alcamí J, Plana M, Torrents D, Leal L, García F.
Impact of Transcriptome and Gut Microbiome on the Response of HIV-1 Infected Individuals to a Dendritic Cell-Based HIV Therapeutic Vaccine.
Vaccines (Basel). 2021 Jun 24;9(7):694. doi: 10.3390/vaccines9070694.
PMID: 34202658
- Guindo-Martínez M, Amela R, Bonàs-Guarch S, Puiggròs M, Salvoro C, Miguel-Escalada I, Carey CE, Cole JB, Rüeger S, Atkinson E, Leong A, Sanchez F, Ramon-Cortes C, Ejarque J, Palmer DS, Kurki M; FinnGen Consortium, Aragam K, Florez JC, Badia RM, Mercader JM, **Torrents D**.
The impact of non-additive genetic associations on age-related complex diseases.
Nat Commun. 2021 Apr 23;12(1):2436. doi: 10.1038/s41467-021-21952-4.
PMID: 33893285
- Pastor-Ibáñez R, Blanco-Heredia J, Etcheverry F, Sánchez-Palomino S, Díez-Fuertes F, Casas R, Navarrete-Muñoz MÁ, Castro-Barquero S, Lucero C, Fernández I, Leal L, Benito JM, Noguera-Julian M, Paredes R, Rallón N, Estruch R, **Torrents D**, García F.
Adherence to a Supplemented Mediterranean Diet Drives Changes in the Gut Microbiota of HIV-1-Infected Individuals.
Nutrients. 2021 Mar 30;13(4):1141. doi: 10.3390/nu13041141.
PMID: 33808476
- Helmsauer K, Valieva ME, Ali S, Chamorro González R, Schöpflin R, Röefzaad C, Bei Y, Dorado Garcia H, Rodriguez-Fos E, Puiggròs M, Kasack K, Haase K, Keskeny C, Chen CY, Kuschel LP, Euskirchen P, Heinrich V, Robson MI, Rosswog C, Toedling J, Szymansky A, Hertwig F, Fischer M, Torrents D, Eggert A, Schulte JH, Mundlos S, Henssen AG, Koche RP.
Enhancer hijacking determines extrachromosomal circular MYCN amplicon architecture in neuroblastoma.
Nat Commun. 2020 Nov 16;11(1):5823. doi: 10.1038/s41467-020-19452-y.
PMID: 33199677.
- Nadeu F, Martin-Garcia D, Clot G, Díaz-Navarro A, Duran-Ferrer M, Navarro A, Vilarrasa-Blasi R, Kulis M, Royo R, Gutiérrez-Abril J, Valdés-Mas R, López C, Chapaprieta V, Puiggros M, Castellano G, Costa D, Aymerich M, Jares P, Espinet B, Muntañola A, Ribera-Cortada I, Siebert R,

Colomer D, Torrents D, Gine E, López-Guillermo A, Küppers R, Martín-Subero JI, Puente XS, Beà S, Campo E.

Genomic and epigenomic insights into the origin, pathogenesis, and clinical behavior of mantle cell lymphoma subtypes.

Blood. 2020 Sep 17;136(12):1419-1432. doi: 10.1182/blood.2020005289.

PMID: 32584970

- González JR, Ruiz-Arenas C, Cáceres A, Morán I, López-Sánchez M, Alonso L, Tolosana I, Guindo-Martínez M, Mercader JM, Esko T, Torrents D, González J, Pérez-Jurado LA. Polymorphic Inversions Underlie the Shared Genetic Susceptibility of Obesity-Related Diseases.

Am J Hum Genet. 2020 Jun 4;106(6):846-858. doi: 10.1016/j.ajhg.2020.04.017. Epub 2020 May 28.

PMID: 32470372

- ICGC/TCGA Pan-Cancer Analysis of Whole Genomes Consortium.

Pan-cancer analysis of whole genomes.

Nature. 2020 Feb;578(7793):82-93. doi: 10.1038/s41586-020-1969-6. Epub 2020 Feb 5.

PMID: 32025007

- Bailey MH, Meyerson WU, Dursi LJ, Wang LB, Dong G, Liang WW, Weerasinghe A, Li S, Li Y, Kelso S; MC3 Working Group; PCAWG novel somatic mutation calling methods working group (Includes D. Torrents), Saksena G, Ellrott K, Wendl MC, Wheeler DA, Getz G, Simpson JT, Gerstein MB, Ding L; PCAWG Consortium.

Retrospective evaluation of whole exome and genome mutation calls in 746 cancer samples.

Nat Commun. 2020 Sep 21;11(1):4748. doi: 10.1038/s41467-020-18151-y.

PMID: 32958763

- Rodríguez-Martin B, Alvarez EG, Baez-Ortega A, Zamora J, Supek F, Demeulemeester J, Santamarina M, Ju YS, Temes J, Garcia-Souto D, Detering H, Li Y, Rodríguez-Castro J, Dueso-Barroso A, Bruzos AL, Dentro SC, Blanco MG, Contino G, Ardeljan D, Tojo M, Roberts ND, Zumalave S, Edwards PAW, Weischenfeldt J, Puiggròs M, Chong Z, Chen K, Lee EA, Wala JA, Raine K, Butler A, Waszak SM, Navarro FCP, Schumacher SE, Monlong J, Maura F, Bolli N, Bourque G, Gerstein M, Park PJ, Wedge DC, Beroukhim R, Torrents D, Korbel JO, Martincorena I, Fitzgerald RC, Van Loo P, Kazazian HH, Burns KH; PCAWG Structural Variation Working Group, Campbell PJ, Tubio JMC; PCAWG Consortium.

Pan-cancer analysis of whole genomes identifies driver rearrangements promoted by LINE-1 retrotransposition.

Nat Genet. 2020 Mar;52(3):306-319. doi: 10.1038/s41588-019-0562-0. Epub 2020 Feb 5.

PMID: 32024998

- Koche RP*, Rodríguez-Fos E*, Helmsauer K*, Burkert M, MacArthur IC, Maag J, Chamorro R, Muñoz-Perez N, Puiggròs M, Dorado Garcia H, Bei Y, Röefzaad C, Bardinet V, Szymansky A, Winkler A, Thole T, Timme N, Kasack K, Fuchs S, Klironomos F, Thiessen N, Blanc E, Schmelz K, Künkele A, Hundsdörfer P, Rosswog C, Theissen J, Beule D, Deubzer H, Sauer S, Toedling J, Fischer M, Hertwig F, Schwarz RF, Eggert A, Torrents D*, Schulte JH*, Henssen AG*

Extrachromosomal circular DNA drives oncogenic genome remodeling in neuroblastoma.

Nat Genet. 2019 Dec 16. doi: 10.1038/s41588-019-0547-z.

PMID: 31844324

- Speedy HE, Beekman R, Chapaprieta V, Orlando G, Law PJ, Martín-García D, Gutiérrez-Abril J, Catovsky D, Beà S, Clot G, Puiggròs M, Torrents D, Puente XS, Allan JM, López-Otín C, Campo E, Houlston RS, Martín-Subero JI.

Insight into genetic predisposition to chronic lymphocytic leukemia from integrative epigenomics.

Nat Commun. 2019 Aug 9;10(1):3615. doi: 10.1038/s41467-019-11582-2.

PMID: 31399598

- Miguel-Escalada I, Bonàs-Guarch S, Cebola I, Ponsa-Cobas J, Mendieta-Esteban J, Atla G, Javierre BM, Rolando DMY, Farabella I, Morgan CC, García-Hurtado J, Beucher A, Morán I, Pasquali L, Ramos-Rodríguez M, Appel EVR, Linneberg A, Gjesing AP, Witte DR, Pedersen O, Grarup N,

Ravassard P, Torrents D, Mercader JM, Piemonti L, Berney T, de Koning EJP, Kerr-Conte J, Pattou F, Fedko IO, Groop L, Prokopenko I, Hansen T, Marti-Renom MA, Fraser P, Ferrer J.

Human pancreatic islet three-dimensional chromatin architecture provides insights into the genetics of type 2 diabetes.

Nat Genet. 2019 Jul;51(7):1137-1148. doi: 10.1038/s41588-019-0457-0. Epub 2019 Jun 28. PMID: 31253982

- Warrington NM, Beaumont RN, Stokholm J, Torrents D, Vinding RK, Willems SM, Atalay M, Chawes BL, Kovacs P, Prokopenko I, Tuke MA, Yaghootkar H, Ruth KS, Jones SE, Loh PR, Murray A, Weedon MN, Tönjes A, Stumvoll M, Michaelsen KF, Eloranta AM, Lakka TA, van Duijn CM, Kiess W, Körner A, Niinikoski H, Pakkala K, Raitakari OT, Jacobsson B, Zeggini E, Dedoussis GV, Teo YY, Saw SM, Montgomery GW, Campbell H, Wilson JF, Vrijkotte TGM, Vrijheid M, de Geus EJC, Hayes MG, Kadarmideen HN, Holm JC, Beilin LJ, Pennell CE, Heinrich J, Adair LS, Borja JB, Mohlke KL, Eriksson JG, Widén EE, Hattersley AT, Spector TD, Kähönen M, Viikari JS, Lehtimäki T, Boomsma DI, Sebert S, Vollenweider P, Sørensen TIA, Bisgaard H, Bønnelykke K, Murray JC, Melbye M, Nohr EA, Mook-Kanamori DO, Rivadeneira F, Hofman A, Felix JF, Jaddoe VVW, Hansen T, Pisinger C, Vaag AA, Pedersen O, Uitterlinden AG, Jarvelin MR, Power C, Hyppönen E, Scholtens DM, Lowe WL Jr, Davey Smith G, Timpson NJ, Morris AP, Wareham NJ, Hakonarson H, Grant SFA, Frayling TM, Lawlor DA, Njølstad PR, Johansson S, Ong KK, McCarthy MI, Perry JRB, Evans DM, Freathy RM.

Maternal and fetal genetic effects on birth weight and their relevance to cardio-metabolic risk factors.

Nat Genet. 2019 May;51(5):804-814. doi: 10.1038/s41588-019-0403-1. Epub 2019 May 1. PMID: 31043758

- Aterido A, Cañete JD, Tornero J, Ferrándiz C, Pinto JA, Gratacós J, Queiró R, Montilla C, Torre-Alonso JC, Pérez-Venegas JJ, Fernández Nebro A, Muñoz-Fernández S, González CM, Roig D, Zarco P, Erra A, Rodríguez J, Castañeda S, Rubio E, Salvador G, Díaz-Torné C, Blanco R, Willisch Domínguez A, Mosquera JA, Vela P, Sánchez-Fernández SA, Corominas H, Ramírez J, de la Cueva P, Fonseca E, Fernández E, Puig L, Dauden E, Sánchez-Carazo JL, López-Estebanz JL, Moreno D, Vanaclocha F, Herrera E, Blanco F, Fernández-Gutiérrez B, González A, Pérez-García C, Alperi-López M, Olivé Marques A, Martínez-Taboada V, González-Álvaro I, Sanmartí R, Tomás Roura C, García-Montero AC, Bonàs-Guarch S, Mercader JM, Torrents D, Codó L, Gelpí JL, López-Corbeto M, Pluma A, López-Lasanta M, Tortosa R, Palau N, Absher D, Myers R, Marsal S, Julià A.

Genetic variation at the glycosaminoglycan metabolism pathway contributes to the risk of psoriatic arthritis but not psoriasis.

Ann Rheum Dis. 2018 Dec 14, doi: 10.1136/annrheumdis-2018-214158 PMID: 30552173

- Martín-García D, Navarro A, Valdés-Mas R, Clot G, Gutiérrez-Abril J, Prieto M, Ribera-Cortada I, Woroniecka R, Rymkiewicz G, Bens S, de Leval L, Rosenwald A, Ferry JA, Hsi ED, Fu K, Delabie J, Weisenburger D, de Jong D, Climent F, O'Connor SJ, Swerdlow SH, Torrents D, Beltran S, Espinet B, González-Farré B, Veloza L, Costa D, Matutes E, Siebert R, Ott G, Quintanilla-Martinez L, Jaffe ES, López-Otín C, Salaverria I, Puente XS, Campo E, Beà S.

CCND2 and CCND3 hijack immunoglobulin light chain enhancers in cyclin D1-negative mantle cell lymphoma.

Blood. 2018 Dec 11, doi: 10.1182/blood-2018-07-862151 PMID: 30538135

- Galván-Femenía I, Guindo M, Duran X, Calabuig-Fariñas S, Mercader JM, Ramirez JL, Rosell R, Torrents D, Carreras A, Kohno T, Jantus-Lewintre E, Camps C, Peruchó M, Sumoy L, Yokota J, de Cid R.

Genomic profiling in advanced stage non-small-cell lung cancer patients with platinum-based chemotherapy identifies germline variants with prognostic value in SMYD2.

Cancer Treat Res Commun. 2018;15:21-31. doi: 10.1016/j.ctarc.2018.02.003. Epub 2018 Mar 1. PMID: 30207284

- Sadler JBA, Wenzel DM, Williams LK, Guindo-Martínez M, Alam SL, Mercader JM, Torrents D, Ullman KS, Sundquist WI, Martin-Serrano J.

A cancer-associated polymorphism in ESCRT-III disrupts the abscission checkpoint and promotes genome instability.

Proc Natl Acad Sci U S A. 2018 Sep 18;115(38):E8900-E8908. doi: 10.1073/pnas.1805504115. PMID: 30181294

- Galván-Femenía I, Obón-Santacana M, Piñeyro D, Guindo-Martínez M, Duran X, Carreras A, Pluvinet R, Velasco J, Ramos L, Aussó S, Mercader JM, Puig L, Perucho M, Torrents D, Moreno V, Sumoy L, de Cid R.

Multitrait genome association analysis identifies new susceptibility genes for human anthropometric variation in the GCAT cohort.

J Med Genet. 2018 Nov;55(11):765-778. doi: 10.1136/jmedgenet-2018-105437. Epub 2018 Aug 30. PMID: 30166351

- Waage J, Standl M, Curtin JA, Jessen LE, Thorsen J, Tian C, Schoettler N; 23andMe Research Team; AAGC collaborators, Flores C, Abdellaoui A, Ahluwalia TS, Alves AC, Amaral AFS, Antó JM, Arnold A, Barreto-Luis A, Baurecht H, van Beijsterveldt CEM, Bleecker ER, Bonàs-Guarch S, Boomsma DI, Brix S, Bunyavanich S, Burchard EG, Chen Z, Curjuric I, Custovic A, den Dekker HT, Dharmage SC, Dmitrieva J, Duijts L, Ege MJ, Gauderman WJ, Georges M, Gieger C, Gilliland F, Granell R, Gui H, Hansen T, Heinrich J, Henderson J, Hernandez-Pacheco N, Holt P, Imboden M, Jaddoe VWV, Jarvelin MR, Jarvis DL, Jensen KK, Jónsdóttir I, Kabesch M, Kaprio J, Kumar A, Lee YA, Levin AM, Li X, Lorenzo-Diaz F, Melén E, Mercader JM, Meyers DA, Myers R, Nicolae DL, Nohr EA, Palviainen T, Paternoster L, Pennell CE, Pershagen G, Pino-Yanes M, Probst-Hensch NM, Rüschendorf F, Simpson A, Stefansson K, Sunyer J, Sveinbjornsson G, Thiering E, Thompson PJ, Torrent M, Torrents D, Tung JY, Wang CA, Weidinger S, Weiss S, Willemsen G, Williams LK, Ober C, Hinds DA, Ferreira MA, Bisgaard H, Strachan DP, Bønnelykke K.

Genome-wide association and HLA fine-mapping studies identify risk loci and genetic pathways underlying allergic rhinitis.

Nat Genet. 2018 Aug;50(8):1072-1080. doi: 10.1038/s41588-018-0157-1. Epub 2018 Jul 16. PMID: 30013184

- Julià A, López-Longo FJ, Pérez Venegas JJ, Bonàs-Guarch S, Olivé À, Andreu JL, Aguirre-Zamorano MÁ, Vela P, Nolla JM, de la Fuente JLM, Zea A, Pego-Reigosa JM, Freire M, Díez E, Rodríguez-Almaraz E, Carreira P, Blanco R, Taboada VM, López-Lasanta M, Corbeto ML, Mercader JM, Torrents D, Absher D, Marsal S, Fernández-Nebro A.

Genome-wide association study meta-analysis identifies five new loci for systemic lupus erythematosus.

Arthritis Res Ther. 2018 May 30;20(1):100. doi: 10.1186/s13075-018-1604-1. PMID: 29848360

- Bullich G, Domingo-Gallego A, Vargas I, Ruiz P, Lorente-Grandoso L, Furlano M, Fraga G, Madrid Á, Ariceta G, Borregán M, Piñero-Fernández JA, Rodríguez-Peña L, Ballesta-Martínez MJ, Llano-Rivas I, Meñica MA, Ballarín J, Torrents D, Torra R, Ars E.

A kidney-disease gene panel allows a comprehensive genetic diagnosis of cystic and glomerular inherited kidney diseases.

Kidney Int. 2018 Aug;94(2):363-371. doi: 10.1016/j.kint.2018.02.027. Epub 2018 May 22. PMID: 29801666

- Beekman R, Chapaprieta V, Russiñol N, Vilarrasa-Blasi R, Verdager-Dot N, Martens JHA, Duran-Ferrer M, Kulis M, Serra F, Javierre BM, Wingett SW, Clot G, Queirós AC, Castellano G, Blanc J, Gut M, Merkel A, Heath S, Vlasova A, Ullrich S, Palumbo E, Enjuanes A, Martín-García D, Beà S, Pinyol M, Aymerich M, Royo R, Puiggros M, Torrents D, Datta A, Lowy E, Kostadima M, Roller M, Clarke L, Flicek P, Agirre X, Prosper F, Baumann T, Delgado J, López-Guillermo A, Fraser P, Yaspo ML, Guigó R, Siebert R, Martí-Renom MA, Puente XS, López-Otín C, Gut I, Stunnenberg HG, Campo E, Martín-Subero JI.

The reference epigenome and regulatory chromatin landscape of chronic lymphocytic leukemia.

Nat Med. 2018 Jun;24(6):868-880. doi: 10.1038/s41591-018-0028-4. Epub 2018 May 21. PMID:29785028

- Bonàs-Guarch S, Guindo-Martínez M, Miguel-Escalada I, Grarup N, Sebastian D, Rodriguez-Fos E, Sánchez F, Planas-Fèlix M, Cortes-Sánchez P, González S, Timshel P, Pers TH, Morgan CC, Moran I, González JR, Andersson EA, Díaz C, Badia RM, Udler M, Flannick J, Jørgensen T, Linneberg A, Jørgensen ME, Witte DR, Christensen C, Brandslund I, Appel EV, Scott RB, Luan J, Langenberg C, Wareham NJ, InterAct Consortium, The SIGMA T2D Consortium, Pedersen O, Zorzano A, Florez JC, Hansen T, Ferrer J, Mercader JM and Torrents D.
A Comprehensive Reanalysis Of Publicly Available GWAS Datasets Reveals An X Chromosome Rare Regulatory Variant Associated With High Risk For Type 2 Diabetes.
Nat. Comms, 2018; Jan 22;9(1):321. doi: 10.1038/s41467-017-02380-9.
PMID: 29358691
- Chinnaswamy S, Wardzynska A, Pawelczyk M, Makowska J, Skaaby T, Mercader JM, Ahluwalia TS, Grarup N, Guindo-Martinez M, Bisgaard H, Torrents D, Linneberg A, Bønnelykke K, Kowalski ML.
A functional IFN- λ 4-generating DNA polymorphism could protect older asthmatic women from aeroallergen sensitization and associate with clinical features of asthma.
Sci Rep. 2017 Sep 5;7(1):10500. doi: 10.1038/s41598-017-10467-y.
PMID: 28874741
- Mercader JM, Liao RG, Bell AD, Dymek Z, Estrada K, Tukiainen T, Huerta-Chagoya A, Moreno-Macías H, Jablonski KA, ...45 other authors...Blangero J, Duggirala R, Saxena R, MacArthur D, Ferrer J, McCarroll SA, Torrents D, Knowler WC, Baier LJ, Burtt N, González-Villalpando C, Haiman CA, Aguilar-Salinas CA, Tusié-Luna T, Flannick J, Jacobs SBR, Orozco L, Altshuler D, Florez JC.
A Loss-Of-Function Splice Acceptor Variant in IGF2 is Protective for Type 2 Diabetes.
Diabetes. 2017 Aug 24. pii: db170187. doi: 10.2337/db17-0187. [Epub ahead of print]
PMID: 28838971
- Wu H, Esteve E, Tremaroli V, Khan MT, Caesar R, Mannerås-Holm L, Ståhlman M, Olsson LM, Serino M, Planas-Fèlix M, Xifra G, Mercader JM, Torrents D, Burcelin R, Ricart W, Perkins R, Fernández-Real JM, Bäckhed F.
Metformin alters the gut microbiome of individuals with treatment-naive type 2 diabetes, contributing to the therapeutic effects of the drug.
Nat Med. 2017 Jul;23(7):850-858. doi: 10.1038/nm.4345. Epub 2017 May 22.
PMID: 28530702
- Henssen AG, Koche R, Zhuang J, Jiang E, Reed C, Eisenberg A, Still E, MacArthur IC, Rodríguez-Fos E, Gonzalez S, Puiggròs M, Blackford AN, Mason CE, de Stanchina E, Gönen M, Emde AK, Shah M, Arora K, Reeves C, Succi ND, Perlman E, Antonescu CR, Roberts CWM, Steen H, Mullen E, Jackson SP, Torrents D, Weng Z, Armstrong SA, Kentsis A.
PGBD5 promotes site-specific oncogenic mutations in human tumors.
Nat Genet. 2017 Jul;49(7):1005-1014. doi: 10.1038/ng.3866. Epub 2017 May 15.
PMID: 28504702
- Carreras-Badosa G, Bonmatí A, Ortega FJ, Mercader JM, Guindo-Martínez M, Torrents D, Prats-Puig A, Martínez-Calcerrada JM, DE Zegher F, Ibáñez L, Fernández-Real JM, Lopez-Bermejo A, Bassols J.
Dysregulation of placental mirna in maternal obesity is associated with pre-and post-natal growth.
J Clin Endocrinol Metab. 2017 Mar 20. doi: 10.1210/jc.2017-00089. [Epub ahead of print]
PMID: 28368446
- Fernández JM, de la Torre V, Richardson D, Royo R, Puiggròs M, Moncunill V, Fragkogianni S, Clarke L; BLUEPRINT Consortium., Flicek P, Rico D, Torrents D, Carrillo de S. Pau E, Valencia A.
The BLUEPRINT Data Analysis Portal.
Cell Syst. 2016 Nov 23;3(5):491-495.e5. doi: 10.1016/j.cels.2016.10.021.
PMID: 27863955

- Horikoshi M, Beaumont RN, Day FR, Warrington NM, Kooijman MN, et al... & Freathy RM. Genome-wide associations for birth weight and correlations with adult disease. **Nature**. 2016 Oct 13;538(7624):248-252. doi: 10.1038/nature19806. PMID: 27680694
- Heyn H, Vidal E, Ferreira HJ, Vizoso M, Sayols S, Gomez A, Moran S, Boque-Sastre R, Guil S, Martinez-Cardus A, Lin CY, Royo R, Sanchez-Mut JV, Martinez R, Gut M, Torrents D, Orozco M, Gut I, Young RA, Esteller M
Epigenomic analysis detects aberrant super-enhancer DNA methylation in human cancer. **Genome Biol**. 2016 Jan 26;17(1):11. doi: 10.1186/s13059-016-0879-2. PMID: 26813288
- Eiler A, Mondav R, Sinclair L, Fernandez-Vidal L, Scofield DG, Schwientek P, Martinez-Garcia M, Torrents D, McMahon KD, Andersson SG, Stepanauskas R, Woyke T, Bertilsson S.
Tuning fresh: radiation through rewiring of central metabolism in streamlined bacteria. **ISME J**. 2016 Jan 19. doi: 10.1038/ismej.2015.260. [Epub ahead of print] PMID: 26784354
- Carreras-Badosa G, Bonmatí A, Ortega FJ, Mercader JM, Guindo-Martínez M, Torrents D, Prats-Puig A, Martínez-Calcerrada JM, Platero-Gutierrez E, De Zegher F, Ibáñez L, Fernandez-Real JM, Lopez-Bermejo A, Bassols J.
Altered Circulating miRNA Expression Profile in Pregestational and Gestational Obesity. **J Clin Endocrinol Metab**. 2015 Nov;100(11), doi: 10.1210/jc.2015-2872. Epub 2015 Sep 25. PMID: 26406295
- Alioto TS, Buchhalter I, Derdak S, Hutter B, Eldridge MD, Hovig E, Heisler LE, Beck TA, Simpson JT, Tonon L, Sertier AS, Patch AM, Jäger N, Ginsbach P, Drews R, Paramasivam N, Kabbe R, Chotewutmontri S, Diessl N, Previti C, Schmidt S, Brors B, Feuerbach L, Heinold M, Gröbner S, Korshunov A, Tarpey PS, Butler AP, Hinton J, Jones D, Menzies A, Raine K, Shepherd R, Stebbings L, Teague JW, Ribeca P, Giner FC, Beltran S, Raineri E, Dabad M, Heath SC, Gut M, Denroche RE, Valdés-Mas R, Nakken S, Vodák D, Bower L, Lynch AG, Anderson CL, Waddell N, Pearson JV, Grimmond SM, Peto M, Spellman P, He M, Kandoth C, Lee S, Zhang J, Létourneau L, Ma S, Seth S, Torrents D, Xi L, Wheeler DA, López-Otín C, Campo E, Campbell PJ, Boutros PC, Puente XS, Gerhard DS, Pfister SM, McPherson JD, Hudson TJ, Schlesner M, Lichter P, Eils R, Jones DT, Gut IG. A comprehensive assessment of somatic mutation detection in cancer using whole-genome sequencing. **Nat Commun**. 2015 Dec 9;6:10001. doi: 10.1038/ncomms10001. PMID: 26647970
- Ortega FJ, Mercader JM, Moreno-Navarrete JM, Nonell L, Puigdecenet E, Rodriguez-Hermosa JI, Rovira O, Xifra G, Guerra E, Moreno M, Mayas D, Moreno-Castellanos N, Fernández-Formoso JA, Ricart W, Tinahones FJ, Torrents D, Malagón MM, Fernández-Real JM.
Surgery-induced weight loss is associated with the downregulation of genes targeted by microRNAs in adipose tissue. **J Clin Endocrinol Metab**. 2015 Aug 7;jc20152357 PMID: 26252355
- 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, Berver L, Carrio A, Castellano G, Clot G, Enjuanes A, Estivill X, Ferrando , Gelpí JL, , López-Guerra M, Martín-García D, Navarro A, Nicolás P, Orozco M, , 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. doi: 10.1038/nature14666 PMID: 26200345
- Guillén Y, Rius N, Delprat A, Williford A, Muyas F, Puig M, Casillas S, Ràmia M, Egea R, Negre B, Mir G, Camps J, Moncunill V, Ruiz-Ruano FJ, Cabrero J, de Lima LG, Dias GB, Ruiz JC,

Kapusta A, Garcia-Mas J, Gut M, Gut IG, Torrents D, Camacho JP, Kuhn GC, Feschotte C, Clark AG, Betrán E, Barbadilla A, Ruiz A

Genomics of ecological adaptation in cactophilic *Drosophila*

Genome Biol Evol. 2014 Dec 31;7(1):349-66. doi: 10.1093/gbe/evu291

PMID: 25552534

- Moncunill, V*, Gonzalez S*, Bea, Andrieux L., Salaverria I., Royo C., Martinez L., Puiggròs M., Segura-Wang M., Stütz A., Navarro A., Royo R., Gelpí J.L.L., Gut I.G., López-Otín C., Orozco M., Korbel J.O., Campo E., Puente X. & Torrents D.

Comprehensive characterization of complex structural variation in cancer by directly comparing genome sequence reads.

Nature Biotechnology, Oct. 2014; doi:10.1038/nbt.3027

PMID: 25344728

- Fernandez L, Mercader JM, Planas-Fèlix M, Torrents D. Adaptation to environmental factors shapes the organization of regulatory regions in microbial communities.

BMC Genomics. 2014 Oct 8;15(1):877. doi: 10.1186/1471-2164-15-877

PMID: 25294412

- Parcerisas A, Rubio SE, Muhaisen A, Gómez-Ramos A, Pujadas L, Puiggros M, Rossi D, Ureña J, Burgaya F, Pascual M, Torrents D, Rábano A, Avila J, Soriano E.

Somatic Signature of Brain-Specific Single Nucleotide Variations in Sporadic Alzheimer's Disease.

J Alzheimers Dis. 2014 Jul 11. [Epub ahead of print]

PMID: 25024348

- Bønnelykke K, Sleiman P, Nielsen K, Kreiner-Møller E, Mercader JM, Belgrave D, den Dekker HT, Husby A, Sevelsted A, Faura-Tellez G, Mortensen L, Paternoster L, Flaaten R, Mølgaard A, Smart D, Thomsen PF, Rasmussen M, Bonàs-Guarch S, Holst C, Nohr EA, Yadav R, March ME, Blicher T, Lackie P, Jaddoe V, Simpson A, Holloway JW, Duijts L, Custovic A, Davies D, Torrents D, Gupta R, Hollegaard MV, Hougaard D, Hakonarson H, Bisgaard H.

A genome-wide association study identifies CDHR3 as a susceptibility locus for early childhood asthma with severe exacerbations.

Nature Genetics. 2014 Jan;46(1):51-5. doi: 10.1038/ng.2830.

PMID: 24241537

- Durán E, Djebali S, González S, Flores O, Mercader JM, Guigó R, Torrents D, Soler-López M, Orozco M.

Unravelling the hidden DNA structural/physical code provides novel insights on promoter location

Nucleic Acids Res. 2013, Aug;41(15):7220-30. doi: 10.1093/nar/gkt511

PMID: 23761436

- Pueyo N, Ortega FJ, Mercader JM, Moreno-Navarrete JM, Sabater M, Bonàs S, Botas P, Delgado E, Ricart W, Martinez-Larrad MT, Serrano-Ríos M, Torrents D, Fernández-Real JM

Common genetic variants of surfactant protein-D (SP-D) are associated with type 2 diabetes.

PLoS One. 2013;8(4):e60468. doi: 10.1371/journal.pone.0060468.

PMID: 23577114

- Mercader J, Puiggros M, Segrè AV... et al... DIAGRAM cons, MITIN cons. ...Torrents D.

Identification of novel type 2 diabetes candidate genes involved in the crosstalk between the mitochondrial and the insulin signaling systems.

PLoS Genetics, 2012 Dec 6; doi:10.1371/journal.pgen.1003046

PMID: 23236286

- Pérez A, Castellazzi CL, Battistini F, Collinet K, Flores O, et al... Orozco M.

Impact of methylation on the physical properties of DNA.

Biophys J. 2012 May 2;102(9):2140-8.

PMID: 22824278

- Tomato Genome Consortium
The tomato genome sequence provides insights into fleshy fruit evolution.
Nature. 2012 May 30;485(7400):635-41. doi: 10.1038/nature11119.
PMID: 22660326
- Adams D, Altucci L, Antonarakis SE, Ballesteros J, Beck S, et al... Willcocks S.
BLUEPRINT to decode the epigenetic signature written in blood.
Nature Biotechnology. 2012 Mar 7;30(3):224-6. doi: 10.1038/nbt.2153.
PMID: 22398613
- González S, Montserrat-Sentís B, Sánchez F, Puiggròs M, Blanco E, Ramirez A, Torrents D.
ReLA, a local alignment search tool for the identification of distal and proximal gene regulatory regions and their conserved transcription factor binding sites.
Bioinformatics. 2012 Mar 15;28(6):763-70. Epub 2012 Jan 16.
PMID: 22253291
- Puente XS, Pinyol M, Quesada V, Conde L, Ordóñez GR, et al... López-Otín C, Campo E.
Whole-genome sequencing identifies recurrent mutations in chronic lymphocytic leukaemia.
Nature. 2011 Jun 5;475(7354):101-5. doi: 10.1038/nature10113.
PMID: 21642962
- Arumugam M, Raes J, Pelletier E, Le Paslier D, Yamada T, et al... Bork P.
Enterotypes of the human gut microbiome.
Nature. 2011 May 12;473(7346):174-80. Epub 2011 Apr 20. Erratum in: *Nature*. 2011 Jun 30;474(7353):666.
PMID: 21508958
- Carlos Quijano, Pavel Tomancak, Jesus Lopez-Marti, et al... Torrents D* and Manzanares M.*
Selective maintenance of Drosophila tandemly-arranged duplicated genes during evolution
Genome Biology, 2008 Dec 16; 9(12):R176
PMID: 19087263
- Casagrande F, Ratera M, Valencia E, Lopez JM, Torrents D, Engel A, Palacin M, Fotiadis D.
Projection structure of a member of the amino acid/polyamine/organocation transporter superfamily.
J Biol Chem. 2008, Nov 28;283(48):33240-8. Epub 2008 Sep 25.
PMID: 18819925
- Casals F, Ferrer-Admetlla A, Chillarón J, Torrents D, Palacín M, Bertranpetit J.^{[L]_{SEP}}
Is there selection for the pace of successive inactivation of the arpAT gene in primates?^{[L]_{SEP}}
J Mol Evol. 2008, Jul;67(1):23-8. Epub 2008 Jun 20.^{[L]_{SEP}}
PMID:18566733
- Goñi JR, Fenollosa C, Pérez A, Torrents D, Orozco M.
DNAlive: a tool for the physical analysis of DNA at the genomic scale.
Bioinformatics. 2008, Aug 1;24(15):1731-2. Epub 2008 Jun 9.
PMID: 18544548
- Goñi JR, Pérez A, Torrents D & Orozco M.
Determining promoter location based on DNA structure first principle calculations
Genome Biology 2007, Dec. 11; 8:R263
PMID: 18072969
- Reig N, Del Rio C, Casagrande F, Ratera M, Gelpi JL, Torrents D, at al ... Palacin M.
Functional and Structural Characterization of the First Prokaryotic Member of the L-Amino Acid Transporter (LAT) Family: A MODEL FOR APC TRANSPORTERS.
J Biol Chem. 2007 May 4;282(18):13270-81.
PMID: 17344220

- Suyama M, Harrington E, Bork P*, Torrents D*.
Identification and analysis of genes and pseudogenes within duplicated regions in the human and mouse genomes.
PLoS Comput Biol. **2006** Jun 30;2(6):e76. Epub 2006 May 16.
PMID: 16846249
- Suyama M*, Torrents D*, Bork P.
PAL2NAL: robust conversion of protein sequence alignments into the corresponding codon alignments.
Nucleic Acids Res. **2006** Jul 1;34(Web Server issue):W609-12.
PMID: 16845082
- Chimpanzee Sequencing and Analysis Consortium (includes **D. Torrents**)
Initial sequence of the chimpanzee genome and comparison with the human genome.
Nature **2005** Sep 1;437(7055):69-87.
PMID: 16136131
- Hillier LW & Chromosomes 2 and 4 sequencing and analysis group (includes **D. Torrents**)
Generation and annotation of the DNA sequences of human chromosomes 2 and 4.
Nature **2005** Apr 7;434(7034):724-31.
PMID: 15815621
- Fernandez E, Torrents D, Zorzano A, Palacin M, Chillaron J.
Identification and functional characterization of a novel low affinity aromatic-preferring amino acid transporter (arpAT). One of the few proteins silenced during primate evolution.
J Biol Chem. **2005** May 13;280(19):19364-72. Epub 2005 Mar 9.
PMID: 15757906
- Chapter: Pseudogenes
David Torrents
Encyclopedia of Genetics, Genomics, Proteomics and Bioinformatics
Ed: John Wiley & Sons, Ltd, 200
- Zdobnov EM, Campillos M, Harrington ED, Torrents D, Bork P.
Protein coding potential of retroviruses and other transposable elements in vertebrate genomes.
Nucleic Acids Res. **2005** Feb 16;33(3):946-54. Print 2005.
PMID: 15716312
- International Chicken Genome Sequencing Consortium
Sequence and comparative analysis of the chicken genome provide unique perspectives on vertebrate evolution.
Nature **2004** Dec 9;432(7018):695-716. Erratum in: *Nature.* **2005** Feb 17;433(7027):777.
PMID: 15592404
- Waterston RH, Hillier LW, Fulton LA, Fulton RS, Graves TA, Pepin KH, Bork P, Suyama M, **Torrents D**, Chinwalla AT, Mardis ER, McPherson JD, Wilson RK.
The human genome: genes, pseudogenes, and variation on chromosome 7.
Cold Spring Harb Symp Quant Biol. **2003**;68:13-22. Review.
PMID: 15338598
- Rat Genome Sequencing Project Consortium.
Genome sequence of the Brown Norway rat yields insights into mammalian evolution.
Nature **2004** Apr 1;428(6982):493-521.
PMID: 15057822

- Suyama M*, **Torrents D***, Bork P.
BLAST2GENE: a comprehensive conversion of BLAST output into independent genes and gene fragments.
Bioinformatics. **2004** Aug 12;20(12):1968-70. Epub 2004 Mar 22.
PMID: 15037510
- **Torrents D***, Suyama M*, Zdobnov E, Bork P.
A genome-wide survey of human pseudogenes.
Genome Res. 2003 Dec;13(12):2559-67.
PMID: 14656963
- Hillier LW, ... Chromosome 7 sequencing and analysis group
The DNA sequence of human chromosome 7.
Nature **2003** Jul 10;424(6945):157-64.
PMID: 12853948
- **Torrents, D.**, Suyama, M. & Bork, P
Chapter: Pseudogenes and Genomes
Bioinformatics and genomes, Editor: Miguel Andrade, Horizon Sci. Press, 2003
- Fernandez E, **Torrents D**, Chillaron J, Martin Del Rio R, Zorzano A, Palacin M.
Basolateral LAT-2 has a major role in the transepithelial flux of L-cystine in the renal proximal tubule cell line OK.
J Am Soc Nephrol. **2003** Apr;14(4):837-47.
PMID: 12660317
- Mouse Genome Sequencing Consortium
Initial sequencing and comparative analysis of the mouse genome.
Nature **2002** Dec 5;420(6915):520-62.
PMID: 12466850
- Zdobnov EM, von Mering C, Letunic I, **Torrents D**, Suyama M, et al ... Kafatos FC, Bork P.
Comparative genome and proteome analysis of *Anopheles gambiae* and *Drosophila melanogaster*.
Science **2002** Oct 4;298(5591):149-59.
PMID: 12364792
- Torras-Llort M, **Torrents D**, Soriano-Garcia JF, Gelpi JL, Estevez R, Ferrer R, Palacin M, Moreto M.
Sequential amino acid exchange across b(0,+)-like system in chicken brush border jejunum.
J Membr Biol. **2001** Apr 1;180(3):213-20.
PMID: 11337893
- Mykkanen J, **Torrents D**, Pineda M, Camps M, Yoldi ME, ... Palacin M, Aula P.
Functional analysis of novel mutations in y⁺LAT-1 amino acid transporter gene causing lysinuric protein intolerance (LPI).
Hum Mol Genet. **2000** Feb 12;9(3):431-8.
PMID: 10655553
- Feliubadalo L, Font M, Purroy J, Rousaud F, Estivill X, Nunes V, et al... Palacin M; International Cystinuria Consortium.
Non-type I cystinuria caused by mutations in SLC7A9, encoding a subunit (b⁰⁺AT) of rBAT.
Nat Genet. **1999** Sep;23(1):52-7.
PMID: 10471498

- Pineda M, Fernandez E, **Torrents D**, Estevez R, Lopez C, Camps M, Lloberas J, Zorzano A, Palacin M.
Identification of a membrane protein, LAT-2, that Co-expresses with 4F2 heavy chain, an L-type amino acid transport activity with broad specificity for small and large zwitterionic amino acids.
J Biol Chem. **1999** Jul 9;274(28):19738-44.
PMID: 10391915
- **Torrents D**, Mykkanen J, Pineda M, Feliubadalo L, Estevez R, de Cid R, Sanjurjo P, Zorzano A, Nunes V, Huoponen K, Reinikainen A, Simell O, Savontaus ML, Aula P, Palacin M.
Identification of SLC7A7, encoding y⁺LAT-1, as the lysinuric protein intolerance gene.
Nat Genet. **1999** Mar;21(3):293-6.
PMID: 10080182
- **Torrents D**, Estevez R, Pineda M, Fernandez E, Lloberas J, Shi YB, Zorzano A, Palacin M.
Identification and characterization of a membrane protein (y⁺L amino acid transporter-1) that associates with 4F2hc to encode the amino acid transport activity y⁺L. A candidate gene for lysinuric protein intolerance.
J Biol Chem. **1998** Dec 4;273(49):32437-45.
PMID: 9829974

Patents

- 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.

Selected Research Grants

- **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
- **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:** INTEGRATION OF THE SYSTEMS MODELS OF MITOCHONDRIAL FUNCTION 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:** 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:** 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:** 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:** 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:** SMUFIN
Agency: Caixaimpulse. La Caixa
Coordinator: David Torrents
Duration: from 1/1/2015 to 1/1/2018
- **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:** 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: Miriam Cnop
Duration: from 1/1/2016 to 1/1/2019
- **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
- **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: Elias Campo
Duration: from 1/1/2019 to 31/12/2021
- **Project title:** EUCANCan; A FEDERATED NETWORK OF ALIGNED AND INTEROPERABLE INFRASTRUCTURES FOR THE HOMOGENEOUS ANALYSIS AND SHARING OF CANCER GENOMICS DATA.
Agency: Horizon 2020- SC1-BHC-2018-2020
Coordinator: David Torrents
Duration: from 1/1/2019 to 31/12/2022
- **Project title:** PLATAFORMA PARA LA EVALUACION DEL ANALISIS GENOMICO DE LA VARIACION ESTRUCTURAL Y SU APLICACION A CANCER Y A TEJIDOS NEURONALES SANOS.
Agency: Agencia Estatal. Plan nacional.
Coordinator: David Torrents
Duration: from 1/9/2021 to 01/09/2024

Teaching (examples, not up to date)

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

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

PhD Student: Santiago Gonzalez Rosado

Title: "IDENTIFICATION AND CHARACTERIZATION OF NON-CODING GENOMIC VARIATIONS ASSOCIATED TO CANCER DISEASE"

Organism: University of Barcelona, Biomedicine.

Year: 2016

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: Marta Guindo

Title: "A SYSTEMATIC AND COMPREHENSIVE APPROACH FOR LARGE GENOME-WIDE ASSOCIATION STUDIES"

Organism: University of Barcelona, Biomedicine

Year: 2019

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

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: Jordi Valls

Title: "COMPREHENSIVE IDENTIFICATION AND CHARACTERIZATION OF GERMLINE STRUCTURAL VARIATION WITHIN THE IBERIAN POPULATION"

Organism: UNIVERSITY OF BARCELONA, BIOMEDICINE

Year: 2021

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: 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: Romina Royo

Title: DEVELOPMENT AND APPLICATION OF METHODOLOGIES AND INFRASTRUCTURES FOR CANCER GENOME ANALYSIS WITHIN PERSONALIZED MEDICINE.

Organism: UNIVERSITY OF BARCELONA, BIOMEDICINE

Year: 2023

David Torrents Arenales, PhD.

