CV summary

Raquel Rabionet, PhD

Genetics, Microbiology and Statistics, University of Barcelona
Tel +34934021494 e-mail: kelly.rabionet@ub.edu
ORCID: 0000-0001-5006-8140 ResearcherID: C-8379-2009

University Education

Biology Degree:
Universitat de Barcelona, 1996
PhD in Biology:
Genetics Department, Universitat de Barcelona, 2002

Research & teaching positions

- Associate Professor (UB) since 2020
- Tenure-track lecturer (UB) 2019-2020
- Postdoc PERIS (IRSJD) 2017-2019
- Assistant Professor (UB) 2015-2019
- Postdoc (CRG) 2011-2017
- Ramon y Cajal Researcher 2005-2010
- Associate in research 2002-2005
- PhD student IRO-CGMM 1997-2002
- Erasmus INSERM u363 1996
- Collaborator student Genetics dep. UB 1994

Grants and Scholarships

- PERIS (IRSJD) 2017-2019
- Ramon y Cajal Contract 2005-2010
- BEFI (ISCIII) 1998-2002
- ERASMUS exchange research grant 1996

Teaching experience

- Undergrad Biomedical Sciences (UB): Molecular Genetics, Human Genetics, Research Project Design, Diagnostic techniques
- coordination Master's thesis program
- Master's: Genetic counselling (UPF), Biotechnology (UB), Genetics and Genomics (UB)
- CRG courses on exome sequencing

Other

Co-Direction of three defended and three ongoing PhD thesis
Reviewer for international journals and AGAUR
AQU accreditation for Lecturer and Researcher

Research lines

Focused on the identification of **genetic factors** involved in heterogeneous and complex disorders (currently, intellectual disability, neuropsychiatric and bone related diseases and stroke) with a focus on structural variation and rare variation.

Scientific Production

H index: **34 (46 considering consortia)**
Articles**: 122**
Of which, consortium articles: **40**
Book chapters: **4**
Citations: **11,745**

Main (first, last, corresponding): Pain, Sci Rep, Plos One, Arthr & Rheum, Genes Brain Behav, Brief Func Genomics, Clin Genet, Hum Genet…

Collaborator: Nature, Nature Genetics, Neurology, Mol Psych, Circ Res, Transl Psych, Lancet, Epigenetics, Hum Mut, J Inv Dermat, Lancet Neurol, Genome Res, Mol Cell Biol…

R&D projects funded

PI: "GENIUS. Genetic influences in ischemic stroke recovery", Fundació la Marató de TV3
Co-PI: "Virus sequence detection in samples from Chronic Fatigue Syndrome" V premio FF2011
Co-PI: "analyzing CNVs in the etiology of lymphoid neoplasms", CIBERESP intramural project

Outreach activities

- Mona de Ciència 2024
- 100ífiques talks promoting STEM for girls
- Science Blogs
- PRBB Open Day volunteer 2013-2015
- "Materia Condensada. Cuinar Ciencia", Arts Santa Mònica exhibit, 2010
- 7th workshop biomedical genomics & proteomics

Raquel Rabionet

Personal information:

Name and surname: Raquel Rabionet Janssen
Date and place of birth: Barcelona, 12/04/1974
Contact information: Department of Genetics, Microbiology and Statistics
Universitat de Barcelona
Diagonal 643
Barcelona 08028
e-mail: kelly.rabionet@ub.edu
ORCID: 0000-0001-5006-8140
ResearcherID: C-8379-2009

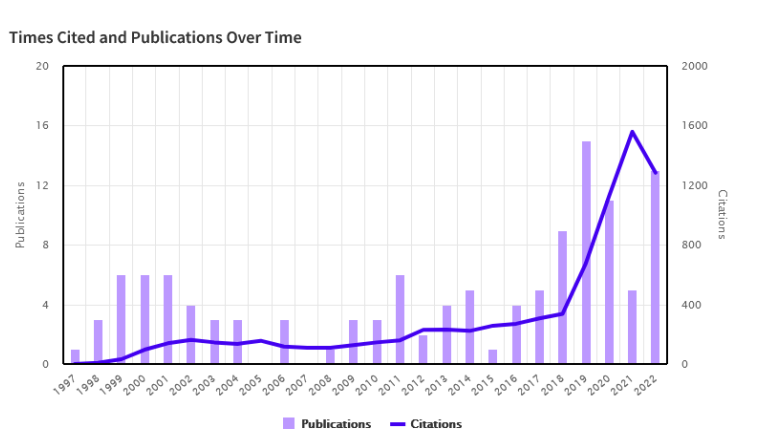
Education:

1996 Bachelor in Biology by the Universidad de Barcelona, Barcelona, Spain.
2002 Doctor in Biology by the Universidad de Barcelona, Barcelona, Spain.

PhD thesis:

Title: Análisis molecular de la sordera debida a mutaciones en el gen GJB2
Directors: Xavier Estivill Pallejà and Maria Lourdes Arbonés de Rafael
University: Universitat de Barcelona **Department**: Department of Genetics
Qualification: excellent cum laude **Defense year**: 2002

Publication metrics:



Total items in list: 122
Sum of times cited: 11745
h-index: 46 (WOS) /42 (Scopus)
updated: 23/05/2023

Professional experience:

2020 – now **Associate Professor,** Department of Genetics, University of Barcelona. I am continuing with the projects on application of NGS to the analysis of complex disorders (OCD, bone-mass phenotypes, stroke recovery, Sézary syndrome) and the functional validation of variants involved in intellectual disability.

May 2019 – Jan 2020 Serra Húnter Lecturer**,** Department of Genetics, University of Barcelona.

2017- 2019 Senior postdoctoral researcher at the department of genetics (university of Barcelona and Fundació Sant Joan de Déu). Leading projects on analysis of structural variation from NGS data, application of NGS to the analysis of genetic factors in complex disorders.

2015- 2019 Associate professor, Department of Genetics, University of Barcelona. Average teaching time of 90h/year.

2011- 2017 Senior postdoctoral researcher at the Centre de Regulació Genòmica (CRG-UPF), co-leading several projects, including GWAS analysis of Fibromyalgia, analysis of structural variants from NGS data, application of NGS technologies in the analysis of Mendelian and complex disorders, e.g. intellectual disability or stroke, rare variant association analysis within the PCAWG project, and epigenetic studies of neurodegenerative disorders.

June 2005 – Dec 2010 Ramon y Cajal researcher at the Centre de Regulació Genòmica (CRG-UPF), coordinating several ongoing projects in the group of Xavier Estivill. Analysis of structural variants and their involvement in different complex disorders, such as Fibromyalgia, rheumathoid arthritis or stroke.

June 2002 – May 2005 Postdoctoral stage at the Center for Human Genetics at Duke University, Durham, NC, USA, performing research on the identification and mutational analysis of candidate genes for autism genetic susceptibility.

1997 - 2002 PhD student at the Centro de Genética Médica y Molecular - IRO (CGMM-IRO), analyzing the genetic causes of hereditary hearing loss under the direction of Dr. X. Estivill.

June - December 1996 ERASMUS student (CEE) at INSERM's U-363, Paris, France, performing research on the interaction of the promoter of the Cis (cytokine-inducible SH2-containing protein) gene with STAT5 transcription factor, under the supervision of Dr. S. Chretien.

April - August 1994 Student collaborator at the Genetics Department of Universidad de Barcelona, under the supervision of Dr. J. García-Fernández, working on the homeobox genes in *Dugesia Tigrina*.

Publications:

* indicates corresponding author
º, ⋆ indicates equal contribution

1. Fritz N, Berens S, Dong Y, Martínez C, Schmitteckert S, Houghton LA, … Bustamante M, Estivill X, **Rabionet R**, … Niesler B. *The serotonin receptor 3E variant is a risk factor for female IBS-D.* **J Mol Med (Berl).** 2022;100(11):1617-1627. doi:10.1007/s00109-022-02244-w
   Contribution: Spanish control data. Revision of final manuscript.
   Impact Factor: 5.606 **Quartile and area**: Q1 genetics and heredity

2. Ovejero D, Garcia-Giralt N, Martínez-Gil N, **Rabionet R**, Balcells S, Grinberg D, Pérez-Jurado LA, Nogués, X, Etxebarria-Forondad I. *Clinical description and genetic analysis of a novel familial skeletal dysplasia characterized by high bone mass and lucent bone lesions*. **Bone**. 2022;161:116450. doi:10.1016/j.bone.2022.116450
   Contribution: Analysis of exome sequencing data. Discussion and interpretation of results. Revision of final manuscript.
   Impact Factor: 4.626 **Quartile and area**: Q2 Endocrinology and metabolism

3. Castilla-Vallmanya L, Centeno-Pla M, Serrano M, … **Rabionet Rº**, Balcells Sº, Urreizti Rº. *Advancing in Schaaf-Yang syndrome pathophysiology: from bedside to subcellular analyses of truncated MAGEL2.* **J Med Genet** 2022. Online ahead of print doi: 10.1136/jmg-2022-108690
   Contribution: Main.
   Impact Factor: **Quartile and area**:

4. Martínez-Gil N, OvejeroD, Garcia-Giralt N, Bruque CD, Mellibovsky L, Nogués X, **Rabionet R,** Grinberg D, Balcells S*. Genetic analysis in a familial case with high bone mineral density suggests additive effects at two loci.* **JMBR Plus,** 2022 Feb 18;6(4):e10602. doi: 10.1002/jbm4.10602.
   Contribution: Design and supervision of experimental analysis, discussion and interpretation of results, writing and revision of final manuscript.
   Impact Factor: **Quartile and area**:

5. Martínez-Gil N, Mellibovsky L, Manzano-Lopez González D, Patiño JD, Cozar M, **Rabionet R,** Grinberg D, Balcells S. *On the association between Chiari malformation type 1, bone mineral density and bone related genes*. **Bone Reports***,* 2022 Mar 15;16:101181. doi: 10.1016/j.bonr.2022.101181
   Contribution: Design and supervision of experimental analysis, discussion and interpretation of results, writing and revision of final manuscript..
   Impact Factor: **Quartile and area**:

6. Alvarez-Mora MI, Sánchez A, Rodríguez-Revenga L, Corominas J, **Rabionet R**, Puig S. Madrigal I. *Diagnostic yield of next generation sequencing in 87 families with neurodevelopmental disorders.* **Orphanet J Rare Dis** 2022 17(1):60. doi: 10.1186/s13023-022-02213-z.
   Contribution: Exome analysis of 45 cases. Discussion and interpretation of results. Revision of final manuscript.
   Impact Factor: **Quartile and area**:

7. Domenech L, Willis J, Alemany M, Morell M, Real E, Escaramís G, Bertolín S, Sánchez-Chinchilla D, Balcells S, Segalas C, Estivill X, Menchón JM, Gabaldón T, Alonso Pº, **Rabionet Rº**. *Changes in the stool and oropharyngeal microbiome in obsessive-compulsive disorder* **Scientific Reports,** 2022

Contribution: Main.
Impact Factor: 4.379 **Quartile and area**:1nd quartile, genetics and heredity

8. Garcia-Giralt N, Roca-Ayats N, Abril JF, Martinez-Gil N, Ovejero D, Castañeda S, Nogues X, Grinberg D, Balcells S, **Rabionet R**. *Gene Network of Susceptibility to Atypical Femoral Fractures Related to Bisphosphonate Treatment* **. Genes** (Basel). 2022 Jan 14;13(1):146. doi: 10.3390/genes13010146
Contribution: Main.
Impact Factor: 4.096 **Quartile and area**:2st quartile, multidisciplinary science

9. Munn-Chernoff MA, Johnson EC, Chou YL, […] **Rabionet R** […], Edenberg HJ, Bulik CM, Agrawal A. *Shared genetic risk between eating disorder- and substance-use-related phenotypes: Evidence from genome-wide association studies***. Addict Biol.** 2021;26(1):e12880.

10. Ugartondo N, Martínez-Gil N, Esteve M, Garcia-Giralt N, Roca-Ayats N, Ovejero D, Nogués X, Díez-Pérez A, **Rabionet R**, Grinberg D, Balcells S. *Functional Analyses of Four CYP1A1 Missense Mutations Present in Patients with Atypical Femoral Fractures***. Int J Mol Sci.** 2021 Jul 9;22(14):7395. doi: 10.3390/ijms22147395.
Contribution: Supervision of experimental analysis, discussion and interpretation of results, revision of final manuscript.
Impact factor: 5.923 **Quartile and area:** 1st quartile

11. Mohr S, Fritz N, Hammer C, Martínez C, Berens S, Schmitteckert S, Wahl V, Schmidt M, Houghton LA, Goebel-Stengel M, Kabisch M, Götze D, Milovač I, D'Amato M, Zheng T, Röth R, Mönnikes H, Engel F, Gauss A, Tesarz J, Raithel M, Andresen V, Frieling T, Keller J, Pehl C, Stein-Thöringer C, Clarke G, Kennedy PJ, Cryan JF, Dinan TG, Quigley EMM, Spiller R, Beltrán C, Madrid AM, Torres V, Pérez de Arce E, Herzog W, Mayer EA, Sayuk G, Gazouli M, Karamanolis G, Kapur-Pojskič L, Bustamante M, **Rabionet R**, Estivil X, Franke A, Lieb W, Boeckxstaens G, Wouters MM, Simrén M, Rappold GA, Vicario M, Santos J, Schaefert R, Lorenzo-Bermejo J, Niesler B. *The alternative serotonin transporter promoter P2 impacts gene function in females with irritable bowel syndrome***. J Cell Mol Med**. 2021 Aug;25(16):8047-8061. doi: 10.1111/jcmm.16736
Contribution: analysis of Spanish data, revision of final manuscript.
Impact factor: 5.31 **Quartile and area:** 2nd quartile

12. Castilla-Vallmanya L, Gürsoy S, Giray-Bozkaya Ö, Prat-Planas A, Bullich G, Matalonga L, Centeno-Pla M, **Rabionet R**, Grinberg D, Balcells S, Urreizti R. *De Novo PORCN and ZIC2 Mutations in a Highly Consanguineous Family.* **Int J Mol Sci.** 2021 Feb 4;22(4):1549. doi: 10.3390/ijms22041549.
Contribution: exome analysis study supervision, results discussion, revision of final manuscript.
Impact factor: **Quartile and area:** 1st quartile

13. Susak H, Serra-Saurina L, Demidov G**, Rabionet R**, Domènech L, Bosio M, Estivill X, Escaramís G and Ossowski S. *Bayesian Rare Variant Association Test using Integrated Nested Laplace Approximation.* **PLOS Computational Biology,** 2021 Feb 19;17(2):e1007784. doi: 10.1371/journal.pcbi.1007784
Contribution: design and discussion; exome sequencing. Revision of final manuscript.
Impact factor: 4.475 **Quartile and area:** 1st quartile

14. Martín-Nalda A, Fortuny C, Rey CL, Alsina L, Anton MC, Basagaña M, Casals F, Deyá A, García-Prat M, Gimeno R, Juan M, Martinez-Banaclocha H, Martinez-Garcia JJ, Mensa-Vilaró A, **Rabionet R**, Rudilla F, Yagüe J, Estivill X, García-Patos V, Pujol R, Soler-Palacín P, Pelegrín P, Colobran R, Vicente A, Arostegui JI. *Novel and de Novo* Heterozygous *PLCG2 Mutations Causing Severe Cutaneous Inflammation, Profound B Cell Lymphopenia and*

Severe Antibody Deficiency. **Journal of Clinical Immunology**, 2020 Oct;40(7):987-1000. doi: 10.1007/s10875-020-00794-7.
Contribution: Exome analysis of one case family. Discussion of results. Revision of final manuscript.
Impact factor: 6.87 **Quartile and area:** 1st quartile

15. ICGC/TCGA Pan-Cancer Analysis of Whole Genomes Consortium. *Pan-cancer analysis of whole genomes.* **Nature**. 2020;578(7793):82-93
Contribution: high. discussion on experimental design, variant calling methods; analysis of germline data; rare variant association studies on germline data; discussion of all results. Writing. Revision of final draft. Times cited: 779
Impact factor: 44. 7 **Quartile and area:** 1st quartile, D1

16. Alemany-Navarro M, Cruz R, Real E, Segalàs C, Bertolín S, Baenas I, Domènech L, Rabionet R, Carracedo Á, Menchón JM, Alonso P. *Exploring genetic variants in obsessive compulsive disorder severity: A GWAS approach***. J Affect Disord**. 2020 Apr 15;267:23-32.
Contribution: sample preparation for part of the samples. Discussion of results. Writing and revision of final manuscript.
Impact factor: 3.892 **Quartile and area:** 1st quartile, psychiatry

17. Alemany-Navarro M, Cruz R, Real E, Segalàs C, Bertolín S, **Rabionet R**, Carracedo Á, Menchón JM, Alonso P. *Looking into the genetic bases of OCD dimensions: a pilot genome-wide association study***. Transl Psychiatry.** 2020;10(1):151.
Contribution: sample preparation for part of the samples. Discussion of results. Writing and revision of final manuscript.
Impact factor: 4.691 **Quartile and area:** 1st quartile, psychiatry

18. Castilla-Vallmanya L, Selmer KK, Dimartino C, **Rabionet R**, Blanco-Sánchez B, Yang S, Reijnders MRF, van Essen MD AJ, Oufadem M, Vigeland MD, Stadheim B, Houge G, Cox H, Kingston H, Clayton-Smith J, Innis JW, Iascone M, Cereda A, Gabbiadini S, Chung WK, Sanders V, Charrow J, Bryant E, Millichap J, Vitobello A, Thauvin C, Mau-Them FT, Faivre L, Lesca G, Labalme A, Rougeot C, Chatron N, Sanlaville D, Christensen KM, Kirby A, Lewandowski R, Gannaway R, Aly M, Lehman A, Clarke L, Graul-Neumann L, Zweier C, Lessel D, Lozic B, Aukrust I, Peretz R, Stratton R, Smol T, Dieux-Coëslier A, Meira J, Wohler E, Sobreira N, Beaver EM, Heeley J, Briere LC, High FA, Sweetser DA, Walker MA, Keegan CE, Jayakar P, Shinawi M, Kerstjens-Frederikse WS, Earl DL, Siu VM, Reesor E, Yao T, Hegele RA, Vaske OM, Rego S, Undiagnosed Diseases Network, Care4Rare Canada Consortium, Shapiro KA, Wong B, Gambello MJ, McDonald M, Karlowicz D, Colombo R, Serretti A, Pais L, O'Donnell-Luria A, Wray A, Sadedin S, Chong B, Tan TY, Christodoulou J, White SM, Slavotinek A, Barbouth D, Swols DM, Parisot M, Bole-Feysot C, Nitschké P, Pingault V, Munnich A, Cho MT, Cormier-Daire V, Balcells S, Lyonnet S, Grinberg D, Amiel J, Urreizti R & Gordon CT. *Phenotypic spectrum and transcriptomic profile associated with germline variants in TRAF7.* **Genet Med.** 2020 10.1038/s41436-020-0792-7.
Contribution: design and discussion of transcriptomic results, writing, revision of final draft.
Impact Factor: **Quartile and area**: 1st quartile

19. Krzyzewska IM, Maas S, Henneman P, van der Lip K, Venema A, Barañano K, Chassevent A, Aref-Eshghi E, Verheij J, Fukuda T, Ikeda H, Jacquemon ML, Kim HG, Labalme A, Lewis S, Lesca G, Madrigal I, Mahida S, Matsumoto N, **Rabionet R**, Rajcan-Separovic E, Qiao Y, Sadikovic B, Saitsu H, Sweetser D, Alders M, Mannens M. *A genome-wide hypermethylation DNA signature for SETD1B related syndrome.* **Clinical Epigenetics,** 2019 11 (1), 156
Contribution: discussion of necessary implementations, usage testing, betatester and more testing.. Writing, revision of final draft.
Impact Factor: 6.091 **Quartile and area**: 1st quartile, genetics

20. Steyn A, Crowther N, Norris S, **Rabionet R**, Estivill X, Ramsay M. *Epigenetic modification of the pentose phosphate pathway and the IGF-axis in women with gestational diabetes mellitus.* **Epigenomics** 2019 Sep;11(12):1371-1385.
Contribution: Supervision/direction of experiments performed at the CRG. Discussion. Revision of final manuscript.
Impact Factor: 4.404 **Quartile and area**: 1st quartile, genetics

21. Bosio M, Drechsel O, Rahman R, Muyas F, **Rabionet R**, Domenech L, Munell F, Colobran R, Macaya A, Estivill X, Ossowski S. *eDiVA – Classification and Prioritization of Pathogenic Variants for Clinical Diagnostics* **Hum Mut,** 2019; 40(7):865-878.
Contribution: discussion of necessary implementations, usage testing, betatester and more testing.. Writing, revision of final draft.
Impact Factor: 5.359 **Quartile and area**: 1st quartile, Genetics & heredity

22. Muyas F, Bosio M, Puig A, Susak H, Domènech-Salgado L, Escaramís G, Zapata L, Estivill X, **Rabionet R**, Ossowski O *Allele Balance Bias Identifies Systematic Genotyping Errors and False Disease Associations* **Hum Mut,** 2019; 40(1):115-26.
Contribution: discussion. Testing. Genotyping setup and analysis of all validation experiments. Writing, revision of final draft.
Impact Factor: 5.359 **Quartile and area**: 1st quartile, Genetics & heredity

23. Mola M, Soriano-Tarraga C, [...], **Rabionet R**, Vives-Bauzá C, Fernández-Cadenas I, Jiménez-Conde J. *PATJ Low Frequency Variants Are Associated With Worse Ischemic Stroke Functional Outcome*. **Circ Res**, 2019 124(1):114-120.
Contribution: sample preparation for half the samples. Project proposal and experimental design. Discussion of results. Writing and revision of final manuscript.
Impact factor: 15.211 **Quartile and area**: 1st decile, Cardiology

24. Madrigal Iº, **Rabionet Rº,** Sanchez A, AlvaMora MI, Rodríguez-Revenga L, Estivill X, Mila M *Spectrum of clinical hererogeneity of ß-tubulinTUBB5 gene mutations.* **Gene,** 2019 695:12-17.
Contribution: Main. Exome analysis results, prioritization and discussion.
Impact Factor: 2.498 **Quartile and area**: 3rd quartile, Genetics & heredity

25. Alemany-Navarro M, Costas J, Real E, Segalas C, Bertolin S, Domenech L, **Rabionet R,** Carracedo A, Menchón JM, Alonso P. *Do polygenic risk and stressful life events predict pharmacological treatment response in obsessive compulsive disorder? A gene-environment interaction approach.* **Transl Psychiatry**. 2019; 9(1):70.
Contribution: sample preparation for part of the samples. Discussion of results. Writing and revision of final manuscript.
Impact factor: 4.691 **Quartile and area:** 1st quartile, psychiatry

26. **Rabionet R***º, Remesal Aº, Mensa-Vilaró A, Murias S, Alcobendas R, Gonzalez-Roca E, Dreschsel O, Ruiz-Ortiz E, Puig A, Comas D, Ossowski S, Yagüe J, MerinoR , Estivill X, Arostegui JI* *LACC1 mutations and juvenile idiopathic arthritis: only restricted to the systemic form?* **Scientific Reports** 2019 9(1):4579.
Contribution: Main. Experimental design. Array Genotyping. Homozygosity mapping. Exome sequencing and analysis. Writing and revising final manuscript.
Impact Factor: 4.122 **Quartile and area**:1st quartile, multidisciplinary science

27. Watson HJ, Yilmaz Z, Thornton LM, […] **Rabionet R** […],Sullivan PF, Breen G, Bulik CM. *Genome-wide Association Study Identifies Eight Loci and Implicates Metabo-Psychiatric Origins for Anorexia Nervosa.* **Nat Genet**. 2019 51(8):1207-1214

Contribution: Spanish patient data. Revising final manuscript

28. Chang L, Patrone CC, Yang W, **Rabionet R**, Espinet B, Gallardo F, Sharma M, Clifton P, Girardi M, Tensen CP, Vermeer M, Geskin L. *An integrated data resource for genomic analysis of cutaneous T-cell lymphoma.* **J invest Dermatol,** 2018 138(12):2681-2683.
Contribution: contribution of Spanis data, CNV reanalysis for12 Spanish samples. Survival data. Revision of final manuscript.
Impact factor: 7.216 **Quartile and area**: 1st quartile, Dermatology

29. Anttila V, Bulik-Sullivan B, […], **Rabionet R**, […], Corvi A, Neale BM, on behalf of the Brainstorm consortium. *Analysis of Shared Heritability in Common Disorders of the Brain.* **Science**. 2018 Jun 22;360(6395):eaap8757

30. Kumar R, Gardner A, Homan C, Douglas E, Mefford H, Wieczorek D, Ludecke HJ, Stark Z, Sadedin S, The Broad CMG, Nowak CB, Douglas J, Parsons G, Mark P, Loidi L, Herman G, Mosher TM, Gillespie MK, Brady L, Tarnopolsky M, Madrigal I, Eiris J, Domenech-Salgado L, **Rabionet R**, Strom TM, Ishihara N, Inagaki H, Kurahashi H, Dudding-Byth T, Palmer E, Field M, Gecz J. *Severe neurocognitive and growth disorders due to variation in THOC2, an essential component of nuclear mRNA export machinery*. **Hum Mut,** 2018 39(8):1126-1138.
Contribution: exome sequencing for Spanish cases (2 mutations). Discussion. Writing and revision of final draft.
Impact Factor: 4.601 **Quartile and area**: Genetics & heredity

31. Urreizti R, Gürsoy S, Castilla-Vallmanya L, Cunill, **R Rabionet** G, Erçal D, Grinberg D, Balcells S. *The ASXL1 mutation p.Gly646Trpfs*12 found in a Turkish boy with Bohring-Opitz Syndrome* **Clin Case Rep** 2018 10;6(8):1452-1456
Contribution: discussion. Bioinformatic analysis of variant frequency in specific populations and disease types in various datasets. Revision of final draft.
Impact factor: **Quartile and area**:

32. Waszak SM*, Tiao G*, Zhu B*, Rausch T*, Muyas Fº, Rodríguez-Martín Bº, **Rabionet Rº**, Yakneen Sº, Escaramis G, [...], Li Y, Ossowski S#, Harismendy O#, Gordenin DA#, Tubio JMC*, De La Vega FM*, Easton DF**, Estivill X**, Korbel JO* . *Germline determinants of the somatic mutation landscape in 2,642 cancer genomes. **BioRxiv** 2017*
Contribution: Main. discussion on experimental design, variant calling methods; analysis of germline data; rare variant association studies; discussion of all results. Writing. Revision of final draft.

33. Carrera C, Jimenez-Conde J, Derdak S, **Rabionet R**, Vives-Bauza C, Soriano-Tárraga C, Giralt-Steihhauer E, Mola M, Diaz-Navarro R, Tur S, Muiño E, Gallego-Fabrega C, Beltran S, Roquer J, Ruiz A, Sotolongo-Grau O, Krupinski J, Lee JM, Cruchaga C, Malik R, Worrall B, Seshadri S, Montaner J, Fernandez-Cadenas I *Whole exome sequence analysis reveals TRPV3 as a possible new risk factor for cardioembolic stroke*. **Thrombosis and Haemostasis**. 2016 Nov 30;116(6):1165-1171
Contribution: exome data for replication dataset. Analysis discussion, writing and revision of final manuscript.
Impact factor: 5.627 **Quartile and area**: 1st quartile, vascular

34. A Prasad, **R Rabionet**, B Espinet, L Zapata, A Puiggros, C Melero, A Puig, Y Sarria-Trujillo, MP Garcia-Muret, T Estrach, O Servitje, I Lopez-Lerma, F Gallardo, RM Pujol, X Estivill *Identification of gene mutations and fusion genes in patients with Sézary Syndrome* **J invest dermatol** 2016. 136 (7):1490-1499

Contribution: *Main*. supervision of whole process, experimental design, CNV analysis, RNA-seq analysis, exome sequencing variant calling, data analysis and discussion. Revision of final manuscript.
Impact factor: 7.216 **Quartile and area**: 1st quartile, Dermatology

35. Pulit SL, McArdle PF, Wong Q, Malik R, Gwinn K, Achterberg S, Algra A, Amouyel P, Anderson C, [...], **Rabionet R,** [...], Woo D, Woodfield R, Wu O, Xu H, Zonderman AB, Worrall BB, de Bakker PIW, Kittner SJ, Mitchell BD, Rosand J *Loci associated with ischaemic stroke and its subtypes (SiGN): a genome-wide association study.* **Lancet Neurol** 2016. 15 (2):174-184
Contribution: DNA extraction for Spanish cases, genotyping data for Spanish controls, revision of final manuscript.
Impact factor: 21.9 **Quartile and area**: 1st quartile, Clinical Neurology

36. Escaramís G, Docampo E, **Rabionet R*.** *A decade of structural variants: description, history and methods to detect structural variation*. **Brief Funct Genomics.** 2015 pii: elv014
Contribution: main.
Impact factor: 3.427 **Quartile and area**: 1st quartile, Biotechnology and applied microbiology

37. Petersen BS, Spehlmann ME, Raedler A, Stade B, Thomsen I, **Rabionet R**, Rosenstiel P, Schreiber S, Franke A. *Whole genome and exome sequencing of monozygotic twins discordant for Crohn's disease*. **BMC Genomics** 2014. 15:564
Contribution: CNV analysis of whole genome and whole exome data.
Impact factor: 4.04 **Quartile and area**: 1st quartile, Genetics and Heredity

38. Docampo E, Escaramís G, Gratacòs M, Villatoro S, Puig A, Kogevinas M, Collado A, Carbonell J, Rivera J, Vidal J, Alegre J, Estivill X*º, **Rabionet R*º**. *Genome-wide analysis of single nucleotide polymorphisms and copy number variants in fibromyalgia suggest a role for the central nervous system*. **Pain**. 2014 Jun;155(6):1102-9.
Contribution: Main. Experimental design, direction of all experiments and analysis. Discussion. Writing.
Impact factor: 5.836 **Quartile and area**: 1st quartile, Clinical Neurology

39. Madrigal I, Alvarez-Mora MI, Karlberg O, Rodríguez-Revenga L, Elurbe DM, **Rabionet R**, Mur A, Ramos FJ, Ballesta F, Sauer S, Syvänen AC, Milà M. *Efficient application of next-generation sequencing for the diagnosis of genetic syndromes with rare variants.* **J clin pathol** 2014 Dec; 67(12):1099-103
Contribution: analysis of whole exome data. Writing.
Impact factor: 2.551 **Quartile and area**: 2nd quartile, Pathology

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Contribution: Genotypes for Spanish controls. Revision of final manuscript

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Contribution: Genotypes for Spanish controls. Revision of final manuscript.
Impact factor: 15.147 **Quartile and area**: 1st quartile, Psychiatry

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Contribution: main. Experimental design, direction of all experiments and analysis. Discussion. writing
Impact factor: 3.534 **Quartile and area:** 1st[d] quartile, Multidisciplinary sciences

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Contribution: discussion, linkage disequilibrium, genotyping.
Impact factor: 2.36 **Quartile and area**: 3rd quartile, Genetics and Heredity

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Contribution: CNV analysis in part of samples.
Impact factor: 13.852 **Quartile and area**: 1st quartile, Genetics and Heredity

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Contribution: discussion, input in definition of parameters, testing. Writing. Revision of final manuscript.
Impact factor: 3.534 **Quartile and area**: 1st quartile, Multidisciplinary sciences

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Contribution: Main.
Impact factor: 3.597 **Quartile and area:** 1st quartile, Behavioral sciences

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Contribution: analysis of Spanish samples. Revision of final manuscript.
Impact factor: 5.398 **Quartile and area:** 1st quartile, Neurosciences

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Contribution: genotyping of Spanish samples. Revising manuscript.
Impact factor: 23.462 **Quartile and area**: 1st quartile, Clinical Neurology

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Contribution: Methylation analysis. Revising manuscript.
Impact factor: 7.636 **Quartile and area**: 1st quartile, Genetics and heredity

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Contribution: Main
Impact factor: 3.128 **Quartile and area**: 2nd quartile, Genetics and heredity

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Contribution: Main. Experimental design, direction of all experiments and analysis. Discussion. writing
Impact factor: 7.866 **Quartile and area**: 1st quartile, Rheumatology

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Contribution: genotyping of Spanish samples. Revising manuscript.
Impact factor: 10.746 **Quartile and area**: 1st quartile, Neurosciences

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Contribution: main. Experimental design, direction of all experiments and analysis. Discussion. writing
Impact factor: 8.435 **Quartile and area:** 1st quartile, Rheumatology

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Contribution: started the project. Experimental design. analysis of part of the samples. Writing.
Impact factor: 3.488 **Quartile and area:** 1st quartile, Clinical Neurology

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Contribution: Main. Genotyping all samples, experimental design, discussion, writing.
Impact factor: 4.156 **Quartile and area**: 1st quartile, Genetics and heredity

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Contribution: discussion, writing.
Impact factor: 4.351 **Quartile and area:** 1st quartile, Biology

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Contribution: partial analysis of samples. Experiment design, discussion
Impact factor: 4.351 **Quartile and area**: 1st quartile, Rheumatology

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Contribution: analysis Spanish samples.
Impact factor: 4.139 **Quartile and area:** 1st quartile, Biochemistry & Molecular biology

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Contribution: writing.
Impact factor: 1.965 **Quartile and area:** 3rd quartile, Genetics and heredity

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Contribution: meeting report
Impact factor: 6.153 **Quartile and area:** 1st quartile, Genetics and heredity

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Contribution: main
Impact factor: 0.574 **Quartile and area**: 4th quartile, Genetics and heredity

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Contribution: Main
Impact factor: 8.25 **Quartile and area**: 1st quartile, Psychiatry

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Contribution: discussion, writing
Impact factor: 2.855 **Quartile and area**: 2nd quartile, Biochemistry & Molecular Biology

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Contribution: Main.

Impact factor: 2.019 **Quartile and area:** 3rd quartile, Neurosciences

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Contribution: main, experimental design, cloning, part of the experiments.
Impact factor: 3.429 **Quartile and area**: 2nd quartile, Genetics & heredity

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Contribution: Main
Impact factor: 7.162 **Quartile and area:** 1st quartile, Medicine research experimental

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Contribution: analysis of Spanish samples.
Impact factor: 3.303 **Quartile and area**: 1st quartile, Medicine research experimental

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Contribution: Analysis of Spanish samples.
Impact factor: 10.542 **Quartile and area:** 1st quartile, Genetics & Heredity

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Contribution: discussion, writing.
Impact factor: 9.318 **Quartile and area**: 1st quartile, Genetics & Heredity

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Contribution: experimental design, writing.
Impact factor: 3.173 **Quartile and area:** 2nd quartile, Genetics & Heredity

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Contribution: Main. Characterization of all Spanish samples.
Impact factor: 3.422 **Quartile and area**: 2nd quartile, Genetics & Heredity

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Contribution: Main.
Impact factor: 3.666 **Quartile and area**: 1st quartile, Genetics & Heredity

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Contribution: experimental design, writing.
Impact factor: 3.666 **Quartile and area**: 1st quartile, Genetics & Heredity

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Contribution: experimental design, genotyping, writing.
Impact factor: 10.351 **Quartile and area**: 1st quartile, Genetics & Heredity

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Contribution: Main. Design, Analysis of >2000 samples, writing. Times cited: 316
Impact factor: 3.175 **Quartile and area**: 2nd quartile, Genetics & Heredity

76. Lopez-Bigas N, **Rabionet R**, de Cid R, Govea N, Gasparini P, Zelante L, Arbones ML, Estivill X. *Splice-site mutation in the PDS gene may result in intrafamilial variability for deafness in Pendred syndrome*. **Hum Mutat**. 1999; 14(6):520-6.
Contribution: experimental design, analysis of some patients, writing.
Impact factor: 2.642 **Quartile and area:** 2nd quartile, Genetics & Heredity

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Contribution: analysis of Spanish patients
Impact factor: 10.426 **Quartile and area**: 1st quartile, Genetics & Heredity

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Contribution: analysis of Spanish cases
Impact factor: 30.693 **Quartile and area**: 1st quartile, Genetics & Heredity

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Contribution: Main. Design, analysis of 50% greek samples, writing.
Impact factor: 1.391 **Quartile and area**: 3rd quartile, Genetics & Heredity

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Contribution: main
Impact factor: 2.986 **Quartile and area**: 2nd quartile, Genetics & Heredity

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preferentially with two associated STAT5A dimers: a distinct biochemical difference between STAT5A and STAT5B. **Mol Cell Biol**. 1998 Oct; 18(10):5852-60
Contribution: EMSA and luciferase assays
Impact factor: 9.866 **Quartile and area**: 1st quartile, Biochemistry & Molecular Biology

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Contribution: analysis of Spanish families, writing. Times cited: 504
Impact factor: 11.793 **Quartile and area**: 1st quartile, Medicine, general & internal

Publications as part of a consortium:

1. Mishra A, Malik R, Hachiya T, ... COMPASS Consortium; INVENT Consortium; Dutch Parelsnoer Initiative (PSI) Cerebrovascular Disease Study Group; Estonian Biobank; PRECISE4Q Consortium; FinnGen Consortium; **NINDS Stroke Genetics Network (SiGN)**; MEGASTROKE Consortium; SIREN Consortium; China Kadoorie Biobank Collaborative Group; VA Million Veteran Program; International Stroke Genetics Consortium (ISGC); Biobank Japan; CHARGE Consortium; GIGASTROKE Consortium,.... Kamatani Y, Dichgans M, Debette S. *Stroke genetics informs drug discovery and risk prediction across ancestries.* **Nature**. 2022;611(7934):115-123.

2. Bullich G, Matalonga L, Pujadas M, ...Pérez-Jurado LA, Beltran S, **Undiagnosed Rare Disease Program of Catalonia (URD-Cat) Consortium**. Systematic Collaborative *Reanalysis of Genomic Data Improves Diagnostic Yield in Neurologic Rare Diseases.* **J Mol Diagn.** 2022;24(5):529-542. doi:10.1016/j.jmoldx.2022.02.003

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4. PCAWG Transcriptome Core Group; Calabrese C, Davidson NR, Demircioğlu D, Fonseca NA, He Y, Kahles A, Lehmann KV, Liu F, Shiraishi Y, Soulette CM, Urban L, Greger L, Li S, Liu D, Perry MD, Xiang Q, Zhang F, Zhang J, Bailey P, Erkek S, Hoadley KA, Hou Y, Huska MR, Kilpinen H, Korbel JO, Marin MG, Markowski J, Nandi T, Pan-Hammarström Q, Pedamallu CS, Siebert R, Stark SG, Su H, Tan P, Waszak SM, Yung C, Zhu S, Awadalla P, Creighton CJ, Meyerson M, Ouellette BFF, Wu K, Yang H; PCAWG Transcriptome Working Group; Brazma A, Brooks AN, Göke J, Rätsch G, Schwarz RF, Stegle O, Zhang Z; PCAWG Consortium. *Genomic basis for RNA alterations in cancer.* **Nature**. 2020;578(7793):129-136.

5. Alexandrov LB, Kim J, Haradhvala NJ, Huang MN, Tian Ng AW, Wu Y, Boot A, Covington KR, Gordenin DA, Bergstrom EN, Islam SMA, Lopez-Bigas N, Klimczak LJ, McPherson JR, Morganella S, Sabarinathan R, Wheeler DA, Mustonen V; PCAWG Mutational Signatures Working Group; Getz G, Rozen SG, Stratton MR; **PCAWG Consortium**. *The repertoire of mutational signatures in human cancer*. **Nature**. 2020;578(7793):94-101

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LncRNA census. reveals evidence for deep functional conservation of long noncoding RNAs in tumorigenesis. **Commun Biol.** 2020;3(1):56.

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19. Cmero M, Yuan K, Ong CS, Schröder J; PCAWG Evolution and Heterogeneity Working Group; Corcoran NM, Papenfuss T, Hovens CM, Markowetz F, Macintyre G; **PCAWG Consortium.** *Inferring structural variant cancer cell fraction*. **Nat Commun.** 2020;11(1):730.

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22. Zhang Y, Chen F, Fonseca NA, He Y, Fujita M, Nakagawa H, Zhang Z, Brazma A; PCAWG Transcriptome Working Group; PCAWG Structural Variation Working Group; Creighton CJ; **PCAWG Consortium.** *High-coverage whole-genome analysis of 1220 cancers reveals hundreds of genes deregulated by rearrangement-mediated cis-regulatory alterations.* **Nat Commun.** 2020;11(1):736.

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International Stroke Genetics Consortium (ISGC). *Contribution of Common Genetic Variants to Risk of Early Onset Ischemic Stroke.* **Neurology**. 2022 Aug 31;99(16):e1738-54. doi: 10.1212/WNL.0000000000201006.

26. Li CH, Prokopec SD, Sun RX, Yousif F, Schmitz N; PCAWG Tumour Subtypes and Clinical Translation, Boutros PC; **PCAWG Consortium**. *Sex differences in oncogenic mutational processes*. **Nat Commun**. 2020;11(1):4330

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34. Huckins LM, Hatzikotoulas K, Southam L, […]; **Eating Disorder Working Group of the Psychiatric Genomics Consortium,** […], Zeggini E. *Investigation of common, low-frequency and rare genome-wide variation in anorexia nervosa.* **Mol Psychiatry**. 2018 May;23(5):1169-1180.

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Hakonarson H; Price Foundation Collaborative Group.; Children's Hospital of Philadelphia/Price Foundation., Herpertz-Dahlmann B, de Zwaan M, Herzog W, Ehrlich S, Zipfel S, Egberts KM, Adan R, Brandys M, van Elburg A, Boraska Perica V, Franklin CS, Tschöp MH, Zeggini E, Bulik CM, Collier D, Scherag A, Müller TD, Hebebrand J. *Evidence for three genetic loci involved in both anorexia nervosa risk and variation of body mass index* **Mol Psychiatry**. 2017 Feb;22(2):192-201.

36. Li D, Chang X, Connolly JJ, Tian L, Liu Y,[..], **Eating Disorders Working Group of the Psychiatric Genomics Consortium,** Price Foundation Collaborative Group, Hakonarson H. *A genome-wide association study of anorexia nervosa suggests a risk locus implicated in dysregulated leptin signaling*. **Sci Rep**. 2017;7(1):3847.

37. Duncan, Yilmaz Z, Gaspar H, […], **Eating Disorders Working Group of the Psychiatric Genomics Consortium**, […], Bulik CM. *Significant Locus and Metabolic Genetic Correlations Revealed in Genome-Wide Association Study of Anorexia Nervosa.* **Am J Psychiatry.** 2017; 174(9):850-858

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39. Soriano-Tárraga C, Jiménez-Conde J, Giralt-Steinhauer E, Mola M, Vivanco-Hidalgo RM, Ois A, Rodríguez-Campello A, Cuadrado-Godia E, Sayols-Baixeras S, Elosua R, Roquer J; **GeneStroke Consortium**.; "The Spanish Stroke Genetics Consortium" Epigenome-wide association study identifies TXNIP gene associated with type 2 diabete*s mellitus and sustained hyperglycemia.* **Human Molecular Genetics**. 2016 Apr 30;9(4):e96543.

40. Soriano-Tárraga C, Jiménez-Conde J, Giralt-Steinhauer E, Mola M, Ois A, Rodríguez-Campello A, Cuadrado-Godia E, Fernández-Cadenas I, Carrera C, Montaner J, Elosua R, Roquer J; **GeneStroke**.; "The Spanish Stroke Genetics Consortium" *Global DNA methylation of ischemic stroke subtypes.* **PLoS One**. 2014 Apr 30;9(4):e96543.

Non-indexed publications:

Rabionet R, Balcells S *Las enfermedades humanas complejas y el mendelismo* En: Ruiz-Rejón C, Navajas-Pérez R, de la Herrán R, Robles F (Eds) **La herencia del Mendelismo** (2022) Pp 307-317

Rabionet R *Genética de las enfermedades complejas: patología cardiovascular, patologías mediadas por mecanismos autoinmunes y patologías psiquiátricas*. En: Farreras, Rozman (Eds) **Tratado de medicina interna** (2020) (pp)

Rabionet R *Genética de las enfermedades complejas: enfermedades cardiovasculares*. En: Farreras, Rozman (Eds) **Tratado de medicina interna** (2016) Pp 1187-1189

Rabionet R *Genética de las enfermedades complejas: enfermedades inflamatorias*. En: Farreras, Rozman (Eds) **Tratado de medicina interna** (2016) Pp 1185-1186

Estivill X, **Rabionet R**, Gratacos M. *Genética de las enfermedades complejas*. En: Farreras, Rozman (Eds) **Tratado de medicina interna** (2008). Pp 1263-73 2008

Raquel Rabionet (2002) *Análisis Molecular de la Sordera Debida a Mutaciones en el Gen de la Conexina 26 (GJB2).* **Doctoral Thesis**

Estivill X and **Rabionet R**. (2000) *Molecular basis of deafness due to mutations in the connexin26 gene (GJB2)*. En: Peracchia C (ed) **Gap Junctions. Molecular basis of cellular communication in health and disease**. Academic Press, San Diego, Ca, USA, pp 483-508.

Lopez-Bigas N, Govea N, **Rabionet R**, Arbones M, Estivill X. (2000) *Genética y genómica de la sordera no sindrómica*. En: **Tratado de otorrinolaringología pediátrica.** Ponencia oficial de la SEORL 2000.

R Rabionet and X Estivill (1998) *Genes y herencia: sordera progresiva de herencia materna*. **FIAPAS** (Federación Española de Asociaciones de Padres y Amigos de los Sordos) 60: 45-46.

R Rabionet and X Estivill (1998) *Genes y herencia: Sordera congénita de herencia recesiva*. **FIAPAS** 62: 40-42.

R Rabionet and X Estivill (1998) *Identificación de genes de sordera.* **ACAPPS** (Asociación Catalana para la Promoción de las Personas Sordas).

For more details on publications, see profile at researchgate, Publons or ORCID:
https://www.researchgate.net/profile/Raquel_Rabionet
https://publons.com/researcher/1277561/raquel-rabionet/
https://orcid.org/0000-0001-5006-8140

Selected presentations at meetings and conferences:

Oral presentations:

EORTC CLTF 2018 Meeting Cutaneous Lymphoma, St Gallen Suiza. **Oral presentation**. *Methylation analysis in Sézary syndrome and integration of exome and transcriptome data.*

Fundació Sant Joan de Déu series of seminars 2017-18, Barcelona, april 2018. Invited oral presentation: *Aproximacions NGS en l'estudi de malalties complexes: estudi d'associació de variants rares en el trastorn obsessiu-compulsiu*.

FEBS3+ meeting of the Spanish, Portuguese and French societies of biochemistry and molecular biology. Barcelona, Spain, November 2017**. Invited oral presentation**: *Next Generation Sequencing in Mendelian and Complex disorders*

Academia de ciencies Mèdiques. Cicles de formació de dermatologia. **Invited oral presentation***:*

450k Methylation Array Analysis Workshop, at UCL, London, UK, April 2012. **Oral presentation:** *DNA methylation profile of Alzheimer's disease brain areas*

IPATIMUP Genomics symposium, Porto, Portugal, July 2011. **Invited oral presentation**: *Application of Next Generation Sequencing Technologies to the understanding of human disease*

CIBERER workshop DNA-DAY, Madrid, Spain, April 2011. **Oral presentation:** *Secuenciación del exoma en una familia consanguínea.*

7th workshop of biomedical genomics and proteomics, in Barcelona, Spain, December 2010. **Oral presentation:** *Using homozygosity mapping and exome sequencing to search for a gene causing a rare articular disease*

IX CRG annual symposium, "Medical Genome Sequencing: Understanding the Genomes of Disease", in Barcelona, Spain, October 2010. **Oral presentation:** *Using homozygosity mapping and exome sequencing to search for a gene causing a rare articular disease*

TECHGENE scientific workshop, organized by the TECHGENE consortium, in Nijmegen, The Netherlands, March 2010. **Oral presentation:** *CNV detection performance of HTS and array-CGH: comparison on 20cases of mental retardation*

Annual meeting of the Sociedad Española de Neurología, in Barcelona, Spain, November 2009. **Oral presentation:** *Asociación de variantes de número de copia (CNVs) e ictus*

IMFAR meeting, in Sacramento, CA, USA, May 2004, organized by the society for autism research. **Oral presentation:** *Family history may play a role in the association of serotonin transporter gene (SLC6A4) polymorphisms in autism. ***was not able to present, substituted by co-author****

IMFAR meeting, in Orlando, FL, USA, May 2002, organized by the society for autism research. **Oral presentation:** *analysis of candidate genes in autD linkage region of chromosome 2q*

HEAR meeting in Bibione, Italy, March 1999, organized by the European Workgroup on Genetics of Hearing Impairment. **Oral presentation**: *Mutations in GJB2 in Italian and Spanish families with deafness*.

"Inherited disorders and their genes in different European populations" meeting in Acquafreda di Maratea, Italy, February 1998, organized by the European Science Foundation. **Oral presentation***: Sporadic and inherited sensorineural deafness is mainly due to mutations in the connexin-26 gene in different European populations*.

Selected poster presentations (presenting author):

World Congress of Psychiatric Genetics (ISPG) in Los Angeles, CA, USA, October 2019. **Poster presentation**: *Changes in the Stool and Oropharyngeal Microbiome in Obsessive-Compulsive Disorder*

Annual meeting of the European society of Human Genetics in Barcelona, Spain, May 2016. **Poster presentation:** *A novel truncating LACC1 mutation in patients with rheumatoid factor-negative polyarticular juvenile idiopathic arthritis*

"The Biology of Genomes" meeting in Cold Spring Harbor, NY, USA, May 2015. **Poster presentation**: *Identification of genes involved in functional recovery after stroke through exome sequencing of extreme phenotypes*

Annual Meeting of the American Society of Human Genetics, in San Diego, USA, October 2014. **Poster presentation**: *Deconstructing obsessive-compulsive disorder (OCD) by whole exome sequencing and integration of clinical endpoints and cognitive domains*

Annual Meeting of the American Society of Human Genetics, in San Diego, USA, October 2014. **Poster presentation**: *Identification of genes involved in functional recovery after stroke through exome sequencing of extreme phenotypes*

Annual Meeting of the American Society of Human Genetics, in Boston, USA, October 2013. **Poster presentation**: *Novel intellectual disability genes identified by exome sequencing*

Annual Meeting of the American Society of Human Genetics, in Boston, USA, October 2013. **Poster presentation**: *Whole genome analysis in fibromyalgia suggests a role for the central nervous system in disease susceptibility*

European Human Genetics Conference, in Paris, France, June 2013. **Poster presentation:** *Whole genome analysis of single nucleotide polymorphisms and copy number variants in fibromyalgia suggest a role for the central nervous system*

Annual Meeting of the American Society of Human Genetics, in San Francisco, USA, October 2012. **Poster presentation**: *Novel intellectual disability genes identified by exome sequencing*

International Congress of Human Genetics meeting in Montreal, Canada, October 2011. **Poster presentation**: *Exome sequencing in a consanguineous family segregating familiar juvenile polyarthritis.*

"The biology of genomes" meeting in Cold Spring Harbor, NY, USA, June 2011, organized by Cold Spring Harbor Laboratory. **Poster presentation**: *PESV-Fisher: a pipeline for somatic structural variant identification from high throughput sequencing data*

"The biology of genomes" meeting in Cold Spring Harbor, NY, USA, June 2010, organized by Cold Spring Harbor Laboratory. **Poster presentation**: *A CNV in chromosome 5 generating a chimaeric gene is a common protective variant for stroke*

"The biology of genomes" meeting in Cold Spring Harbor, NY, USA, June 2009, organized by Cold Spring Harbor Laboratory. **Poster presentation**: *Copy Number variation in the late cornified envelope cluster in autoimmune disorders*

American Society of Human Genetics meeting in Honolulu, HI, USA, October 2009. **Poster presentation**: *A CNV in chromosome 5 generating a chimaeric gene is a common protective variant for stroke*

American Society of Human Genetics meeting in Philadelphia, PA, USA, October 2008. **Poster presentation**: *Genomic structural variation in patients with multiple sclerosis*.

"The biology of genomes" meeting in Cold Spring Harbor, NY, USA, June 2008, organized by Cold Spring Harbor Laboratory. Poster presentation: Genomic structural variation in patients with multiple sclerosis

European Society of Human Genetics meeting (ESHG) in Barcelona, Spain, June 2008. Poster presentation: Genomic structural variation in patients with multiple sclerosis

World Congress of Psychiatric Genetics, in Dublin, Ireland, October 2004. Poster presentation: Fine mapping and association analysis of chromosome 19 and autism

American Society of Human Genetics in Los Angeles, CA, USA, October 2003. Poster presentation: Analysis of methyl binding domain (MBD) genes in autism risk.

American Society of Human Genetics meeting in San Diego, CA, USA, October 2001. Poster presentation: Identification and functional analysis of amino acid variants in the connexin26 (GJB2) gene.

American Society of Human Genetics meeting in San Diego, CA, USA, October 2001. Poster presentation: A mutation in MYO6, the human homologue of the gene causing deafness in Snell's waltzer mice, is associated with hearing loss

American Society of Human Genetics meeting in San Diego, CA, USA, October 2001. Poster presentation: Mutations in the TMPRSS3 gene are a rare cause of childhood non-syndromic deafness in Caucasian patients.

American Society of Human Genetics meeting in Denver, CO, USA, October 1997. Poster presentation: Mutations in the connexin-26 (GJB2) gene in Italian and Spanish patients with congenital deafness.

Selected poster and oral presentations (not first, presenting, author):

Annual meeting of the European society of Human Genetics in Vienna, Austria, May 2022. Poster presentation: Unravelling the effects of germline missense variants in TRAF7

Annual meeting of the European society of Human Genetics in Vienna, Austria, May 2022. Poster presentation: Truncating mutations in MAGEL2 cause alterations in A beta 1-40 levels and gene expression in fibroblasts

Annual meeting of the European society of Human Genetics in Vienna, Austria, May 2022. Poster presentation: Genetic influences on functional outcome after stroke

Annual meeting of the European society of Human Genetics in Vienna, Austria, May 2022. Poster presentation: A missense mutation in VAV3 in a familial case of high bone mass

Annual meeting of the European society of Human Genetics in Copenhagen, Denmark, May 2017. Poster presentation: Next generation sequencing diagnostic yield in intellectual disability

Annual meeting of the European society of Human Genetics in Copenhagen, Denmark, May 2017. Poster presentation: Allele balance based variant callability score for whole exome sequencing

Annual meeting of the European society of Human Genetics in Barcelona, Spain, May 2016. Poster presentation: Deconstructing obsessive-compulsive disorder by whole exome sequencing and rare variant association study

Annual meeting of the European society of Human Genetics in Barcelona, Spain, May 2016. Poster presentation: Analysis of the role of copy number variation in Obsessive-compulsive disorder

Annual meeting of the European society of Human Genetics in Barcelona, Spain, May 2016. Poster presentation: Germline mutational landscape of Chronic Lymphocytic Leukemia

Annual meeting of the European society of Human Genetics in Barcelona, Spain, May 2016. Poster presentation: GEUVADIS European Exome Variant Server: Variant allele frequency aggregation from multicentre, access-restricted data

Annual meeting of the European society of Human Genetics in Barcelona, Spain, May 2016. Poster presentation: Association between INADL polymorphisms and ischemic stroke functional outcome through genome wide association meta-analysis. GODS Project

Annual European Congress of Rheumatology in Berlin, Germany, June 2012. Oral presentation: Cluster analysis of clinical data identifies fibromyalgia subgroups

Direction and participation in research projects:

A) As Principal Investigator

Project title: BBModels: Modelos 2D y 3D de cerebro y hueso y su aplicación a la investigación de la patofisiología de las enfermedades
Funding organism: MICINN
Reference: PID2022-141461OB-I00 **Duration:** 2023-2025
Principal Investigator/s: Susanna Balcells & Raquel Rabionet

Project title: Grup de recerca en genètica molecular humana
Funding organism: AGAUR
Reference: 2022SGR-1093 **Duration:** 2022-2024
Principal Investigator/s: Raquel Rabionet

Project title: GENIUS: influències genètiques en el resultat funcional de l'ictus
Funding organism: Fundació La Marató de TV3
Reference: **Duration:** 2018-2019
Principal Investigator/s: Raquel Rabionet

Project title: GODS project: genetic contribution to functional outcome and disability after stroke
Funding organism: Fundació la Marató de TV3
Reference: 112031 **Duration:** 2011-2015
Principal Investigator/s: Xavier Estivill & Raquel Rabionet

Project title: Detección de secuencias víricas en muestras de síndrome de fatiga crónics mediante captura por hibridación y posterior secuenciación
Funding organism: Fundación para la fibromialgia y el sindrome de fatiga crónica
Reference: V premio FF2011 **Duration:** 2010-2011
Principal Investigator/s: Xavier Estivill, Raquel Rabionet, Elisa Docampo

Project title: Evaluación de CNVs en la etiología de las neoplasias linfoides
Funding organism: CIBERESP
Reference: **Duration:** 2008
Principal Investigator/s: Raquel Rabionet &Sílvia Sanjosé

Project title: Identificación de genes de susceptibilidad del autismo en la población española
Funding organism: Ministerio de economía y competitividad
Reference: RyC04 Raquel Rabionet **Duration:** 2005-2010
Principal Investigator/s: Raquel Rabionet

B) As a team investigator

Project title: Estudios genéticos y funcionales hacia aproximaciones terapéuticas para varias enfermedades óseas y síndromes de discapacidad intelectual.
Funding organism: Ministerio de ciencia, Innovación y Universidades
Reference: PID2019-107188RB-C21 **Duration:** 2020-2023
Principal Investigator/s: Susanna Balcells

Project title: Biomarkers and clinical predictors of long-term course in obsessive-compulsive disorder: a prospective cohort study.
Funding organism: Ministerio de economía y competitividad
Reference: PI18/00856 **Duration:** 2019-2021

Principal Investigator/s: Mª Pino Alonso

Project title: Estudios funcionales y de identificación de estrategias terapéuticas en discapacidad intelectual: *TRIM28, TRAF7* y *MAGEL2*.
Funding organism: CIBERER intramural
Reference: ACCI **Duration:** 2018-2020
Principal Investigator/s: Roser Urreizti Frexedas

Project title: Sinergias en el estudio genético y la búsqueda de terapias para el síndrome de Opitz C, la fractura femoral atípica, la osteoporosis y enfermedades lisosomales.
Funding organism: Ministerio de ciencia e Innovación
Reference: SAF2016-75948-R **Duration:** 2016-2020
Principal Investigator/s: Susanna Balcells

Project title: PanCanRisk: Personalized bioinformatics for global cancer susceptibility identification and clinical management
Funding organism: European Comission
Reference: 635290 **Duration:** 2015-2018
Principal Investigator/s: Ángel Carracedo (consortium)/Xavier Estivill (participant)

Project title: DECOCD: decodificando la complejidad biológica del trastorno obsesivo compulsivo: hacia una medicina de precisión en una enfermedad mental
Funding organism: Ministerio de economía y competitividad
Reference: SAF2013-49108-R **Duration:** 2013-2016
Principal Investigator/s: Xavier Estivill

Project title: European sequencing and genotyping structure (ESGI)
Funding organism: European comission
Reference: 262055_ESGI **Duration:** 2011-2015
Principal Investigator/s: Xavier Estivill

Project title: Technological innovation of high throughput molecular diagnostics of clinically and molecularly heterogeneous genetic disorders (Techgene): application to mental retardation
Funding organism: Ministerio de economía y competitividad
Reference: PI10/01702 **Duration:** 2011-2013
Principal Investigator/s: Xavier Estivill

Project title: Technological innovation of high throughput molecular diagnostics of clinically and molecularly heterogeneous genetic disorders (Techgene)
Funding organism: European comission
Reference: techgene_223143 **Duration:** 2009-2013
Principal Investigator/s: Xavier Estivill

Project title: European network for genetic and genomic epidemiology (ENGAGE)
Funding organism: European comission
Reference: Engage_201413 **Duration:** 2008-2012
Principal Investigator/s: Xavier Estivill

Project title: CIBER en epidemiología y salud pública
Funding organism: Ministerio de ciencia e innovación
Reference: CB06/02/0058 **Duration:** 2006-2016
Principal Investigator/s: Xavier Estivill

Project title: Aneuploidy: understanding gene dosage imbalance in human health using genetics, functional genomics and systems biology
Funding organism: European comission
Reference: LSHG-CT-2006-037627 **Duration:** 2006-2010
Principal Investigator/s: Xavier Estivill

Project title: Novel variation in disease (NOVADIS)
Funding organism: Ministerio de ciencia e innovación
Reference: SAF2008-00357 **Duration:** 2009-2014
Principal Investigator/s: Xavier Estivill

Project title: Estudios de genotipación para el proyecto "identificación de factores de susceptibilidad genética para la fibromialgia y síndrome de fatiga crónica"
Funding organism: Fundación desarrollo investigación genómica y proteómica
Reference: CENIT'07 Fibromialgia Estivill **Duration:** 2008-2013
Principal Investigator/s: Xavier Estivill

Project title: Molecular and genetic epidemiology of autism
Funding organism: National institute of health (NIH)
Reference: 2R01NS036768-06 **Duration:** 1997-2006
Principal Investigator/s: Margaret Pericak-Vance

Project title: Análisis molecular y funcional de mutaciones en el DNA mitocondrial implicadas en sordera progresiva y diabetes
Funding organism: Fundació la Marató de TV3
Reference: **Duration:** 2000-2002
Principal Investigator/s: Xavier Estivill

Project title: Desenvolupament d'un model murí per a teràpia gènica de la sordesa congénita hereditària deguda a mutacions en el gen de la connexina-26
Funding organism: Fundació la Marató de TV3
Reference: **Duration:** 1999-2001
Principal Investigator/s: Maria Lourdes Arbonés

Project title: Identificación de los genes implicados en la sordera de la población de Cantabria
Funding organism: Fundación Marcelino Botín
Reference: 2R01NS036768-06 **Duration:** 1999-2000
Principal Investigator/s: Xavier Estivill

Awarded Fellowships and prizes:

2010	V Premio Fundación FF y ciencia
2010	pre-approved for the I3 MICINN programme
2005-2010	Awarded Ramón y Cajal Investigator contract
1998-2002	BEFI fellowship (Instituto de Salud Carlos III) for PhD students.
1996	ERASMUS fellowship (CEE) to perform a stage at INSERM's U-363, Paris, France.

Teaching experience:

2020: **Associate professor at the University of Barcelona** (UB), Department of Genetics, Microbiology and Statistics.

2019: **Lecturer professor at the University of Barcelona** (UB), Department of Genetics, Microbiology and Statistics

2015-18: **Associate lecturer at the University of Barcelona** (UB), Department of Genetics, Microbiology and Statistics

2015, 2018: Organization and teaching at the I and II workshops *curso de formación y actualización en genética de las enfermedades complejas* organized by the Genestroke consortium.

2015: Seminar on structural variations at the "Malalties Genètiques Mendelianes i Complexes" master course (UB)

2013-16: Organization and teaching (2-5h) of the yearly bioinformatics module of the CRG courses Advanced Seminars on Biomedical Research (CRG)

2013-14: Organization and teaching (3h) in two editions of the computational analysis of exome-sequencing data course (courses@CRG)

2011-12: Seminar at the master course "Advances in Human Genetics" (UB).

2007-16: Variable number of sessions (2-8h) teaching theory and problem based learning of the Multifactorial Genetics Module at the biannual Máster in Genetic Counseling (UPF).

2006-07: Seminar at the master course "Advances in Human Genetics" (UB).

Supervision of PhD students' dissertations:

- Juan Diego Gutiérrez (ongoing)
- Juan David Patiño (ongoing)
- Estefania Alcaide (ongoing)
- Mónica Centeno (ongoing)
- *Analysis of genetic factors in neuropsychiatric disorders* by Laura Domènech Salgado. Defended at Universitat Pompeu Fabra, December 2018, excellent *cum laude*
- *Analisi de factors genetics implicats en fibromialgia* by Elisa Docampo Martínez, defended at University of Barcelona, December 2013, apto *cum laude*
- *Estudio de factores epigenéticos y variantes estructurales del genoma en patología neurológica* by Susana Iraola Guzmán, defended at University of Barcelona, april 2013, apto *cum laude*

Supervision of master students' dissertations:

- *Genetic influences on functional outcome after stroke* by Estefanía Alcaide defended at University of Barcelona 2021
- *Analysis of post-translational modifications of TRIM28 mutations involved in intellectual disability* by Irene Higueras, defended at University of Barcelona 2020
- *Functional analysis of TRIM28 mutations involved in intellectual disability* by Álvaro Garcés, defended at University of Barcelona 2019
- *Integration of methylation and mutation data in Sézary syndrome* by Mar García Valero, defended at University of Barcelona 2018
- *Functional analysis of TRIM28 mutations involved in intellectual disability* by Isabel Hinarejos Martínez, defended at University of Barcelona 2018
- *Novel intellectual disability genes identified by exome sequencing* by Laura Domenech Salgado, defended at University of Barcelona in 2013

- *Genomic structural variation in multiple sclerosis* by Elisa Docampo Martínez, defended at university of Barcelona in 2008

Supervision of Final Degree Project students:

- *Validación funcional de genes implicados en la recuperación del ictus, by* Cristina Esteban, defended at University of Barcelona 2022
- *Identification of genetic variants involved in stroke recovery: TJP1, TEK, VASH2* by Claudia Zamora, defended at University of Barcelona 2020
- *Identification of genetic variants involved in stroke recovery: ANGPT2, CSMD1, PATJ,* by Estefania Alcaide, defended at University of Barcelona 2020

Supervision of internship students:

- Various undergraduate summer students within the "estius de Genètica" program: Andreu Pujolar, Sira Gardela, Marta Espina, Mar Haro, from 2016 to 2023
- Undergraduate research project *Estudi dels factors genetics implicats en fibromyalgia mitjançant l'analisi d'exomes* by Maria Bellet Coll, presented at the Univeristy of Girona in November 2013- February 2014
- Undergraduate internship *Causas genéticas de enfermedad: retraso mental* by María Becker Loyola, with the Univeristy of Navarra in July- August 2012
- High-school final research project *Validation of SNPs in Chronic lymphocytic leukemia through targeted Sanger sequencing* by Clara Roig Arsequell at Aula escola europea, in July 2014
- Undergraduate summer internship by Maja Gehre, with Freie Universität Berlin (FU Berlin), in June-July 2012
- Undergraduate summer internship by Caty Carrera, June-September 2011

Organization of workshops and meetings:

Co-organizer of the "7th workshop of biomedical genomics and proteomics", Societat Catalana de Biologia, Barcelona, Spain, December 2010

Collaborator on the organization and teaching of the workshop "I curso de formación y actualización en genética de las enfermedades complejas" by the Genestroke consortium, Barcelona, Spain, October 2015

Participation in outreach activities:

Collaboration with Fundació Alicia in the design of the exhibit "Materia condensada. Cuina i ciència" shown at Centre d'Arts Santa Mònica, July - November 2010

Volunteer at the PRBB Open Day 2013, 2014, 2015

Radio interview @ rtve: "Además, les ofrecemos un reportaje con **los actuales Gustavo Adolfo Bécquer y Rodrigo Vivar** y otro sobre los **genes que se heredan** con la investigadora **Raquel Rabionet**." www.rtve.es/alacarta/audios/viaje-al-centro-de-la-noche/viaje.../1680315

Language knowledge:

Spanish, native language. ; **Catalan**, native language.
English, advanced oral and written level.
French, good oral and written. ; **Dutch**, intermediate oral level, low written level.

Other merits:

Development and maintenance, util June 2002, of the website "The Connexin-deafness Web-site" (http://www.crg.es/deafness), a website that contained all information on deafness-related mutations in the connexin 26 and other deafness related connexin genes.

Scritping language knowledge: Python

Member of the committee at various doctoral thesis defenses (since 2008)

Reviewer for various journals, including Pain, Gene, Journal of the European Association of Dermatology and Venereology, BMC medical genetics, Molecular Biology Reports, Prenatal Diagnosis, Clinical Genetics and International Journal of Audiology

AQU accreditations for researcher (agregat) and lecturer (lector).