Universität Basel
Medizinische Fakultät

UNIVERSITY OF LEICESTER
College of Life Science
Leicester Precision Medicine Institute

NCCR RNA & Disease

Curriculum Vitae

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***Summary of qualifications**:*

Medical geneticist, with specific expertise in the fields of genomics, large-scale experimental approaches, and Next-Generation Sequencing in relationship to ophthalmic conditions. Strong background and skills in bioinformatics, statistics, and computer programming, in addition to competences in molecular biology and classic human genetics.

***Research interests**:*

Molecular genetics of hereditary retinal degenerations and genetic conditions with elevated allelic heterogeneity. Computational approaches to genomics of monogenic to complex diseases, with specific interest on quasi-Mendelian conditions, modifier genes, and oligogenic inheritance.

***Education**:*

1989-1994:	Master of Science (Molecular Biology) *cum laude*. University of Pavia, Italy
1997:	Twelve-month postgraduate course in bioinformatics at the Swiss Federal Institute of Technology Lausanne (EPFL)
1994-1999:	Ph.D. in Science (Molecular Genetics) *cum laude*. University of Lausanne,

Postdoctoral training:

1999-2003:	Postdoctoral fellowship in the group of Prof. Thaddeus Dryja at Harvard Medical School, Massachusetts Eye and Ear Infirmary (MEEI/MGH), Boston, USA

Academic career:

2003-2004:	Instructor (Junior Faculty) of the Medical School, Harvard
2004-2008:	Junior Group Leader (Maître Assistant), University of Lausanne
2008-present:	Tenured Group Leader (Maître d'Enseignement et de Recherche I), Department of Medical Genetics, University of Lausanne
2012:	Privat Dozent (University of Lausanne)
2017-present:	Full Professor, Chair of Medical Genetics, Department of Genetics and Genome Biology, University of Leicester, UK
2019-present:	Full Professor, Chair of Ophthalmic Genetics, IOB and University of Basel

Formal Teaching (University of Lausanne):

2006-2007:	Classes of "Medical Genetics", to medical students, 4th year
2009-2010:	"Ocular Genetics", Ph.D. program in Neurosciences
2010-2011:	Course of Genomic Sequencing, Master MLS program
2007-present:	"Human Molecular Genetics", Master MLS program
2010-present:	"Human Molecular Genetics", Ph.D. (all UNIL's Ph.D. programs)
2012-present:	"Write a review", Master MLS program
2015-present:	Course "Restore vision: from bench to bedside" (Genetics of retinal dystrophies module), to all Ph.D. programs
2015-present:	"Genes and vision", to medical students, 4th year

Other duties (Lausanne):

- Ph.D. fellowship, FBM, member of the board (2006-present)
- Member of the "Conseil de l'Ecole Doctorale", FBM (2009-2014)
- Member of the "Bâtiment Bugnon 27" committee (2006-2010)
- Member of the "Commission de la recherche", FBM (2009-2010)
- UNIL representative for the SNF Div. 3 at SNF Research day (2006)
- Departmental head for biosafety (2005-present)
- Elected member of the Medicine and Biology Faculty Council (2013-present)

Other duties (Switzerland):

- Principal investigator for Genetic Research Days (federal program for the education of the general public) (2007-present)
- Member of the Forum Genforschung (FoGeFo), Swiss Academy of Sciences (SCNAT), Bern (2015-2016)
- Member of the Working Group “Personalized Health” (FoGeFo, SCNAT), Bern (2016-present)

Other duties (Leicester):

- Member of the Leicester Precision Medicine Institute (LPMI) (2017-present)
- Member of the Genomics England Clinical Interpretation Partnership (GeCIP, hearing and sight) (2018-present)

Editorial and reviewing activities:

- Member of the Editorial Board of: Journal of Human Genetics (Nature PG), Vision (MDPI, Basel), Scientific Reports (Nature PG)
- Reviewer for (Journals): Nature Genetics, American Journal of Human Genetics, Journal of Medical Genetics, BMC Medical Genetics, Human Molecular Genetics, Human Mutation, Journal of Clinical Investigations, Nature Biotechnology, Genetics in Medicine, European Journal of Medical Genetics, Nature Neuroscience, etc.
- Reviewer for (Funding Bodies): The Wellcome Trust, Swiss National Science Foundation, Swiss League Against Cancer, Netherlands Organization for Health Research and Development, Agence Nationale de la Recherche (F), Czech Science Foundation, etc.

Memberships:

The Association for Research in Vision and Ophthalmology (ARVO), Swiss Society of Ophthalmology (SSO), Swiss Society of Medical Genetics (SSMG), International Society for Eye Research (ISER), the American Society of Human Genetics (ASHG), the European Retinal Disease Consortium (ERDC), National Center for Competences in Research (NCCR) RNA and disease

Mentoring:

Approximately 40 people so far, including M.Sc. students, M.D. students, Ph.D. students, and postdocs

Current lab:

Ten people: 5 Ph.D. students, 3 postdocs, 1 intern, 1 lab manager

Publications:

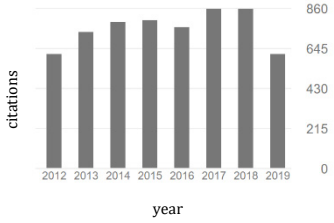
90 peer-reviewed articles, of which 36 during the last 5 years

Competitive extramural funding:

Approximately $ 8,800,000 as a group leader

Citations:

11,283 (4,683 during the last 5 years), as of September 2019.



Publications

1. Moye AR, Bedoni N, Cunningham JG, Sanzhaeva U, Tucker ES, Mathers P, Peter VG, Quinodoz M, Paris LP, Coutinho-Santos L, Camacho P, Purcell MG, Winkelmann AC, Foster JA, Pugacheva EN, **Rivolta C***, Ramamurthy V*. **(2019)**. Mutations in ARL2BP, a protein required for ciliary microtubule structure, cause syndromic male infertility in humans and mice. ***PLoS Genet***. 15(8):e1008315. *equal contribution and correspondence. Impact factor: 5.54

2. Peter VG, Quinodoz M, Pinto-Basto J, Sousa SB, Di Gioia SA, Soares G, Ferraz Leal G, Silva ED, Pescini Gobert R, Miyake N, Matsumoto N, Engle EC, Unger S, Shapiro F, Superti-Furga A*, **Rivolta C***, Campos-Xavier B. **(2019)**. The Liberfarb syndrome, a multisystem disorder affecting eye, ear, bone, and brain development, is caused by a founder pathogenic variant in the PISD gene. ***Genet Med***. (in press). *correspondence. Impact Factor: 9.94

3. Royer-Bertrand B, Tsouni P, Mullen P, Campos Xavier B, Mittaz Crettol L, Lobrinus AJ, Ghika J, Baumgartner MR, **Rivolta C**, Superti-Furga A, Kuntzer T, Francklyn C, Tran C. **(2019)**. Peripheral neuropathy and cognitive impairment associated with a novel monoallelic HARS variant. ***Ann Clin Transl Neurol***. 6: 1072-1080. Impact Factor: 4.66

4. El Zaoui I, Bucher M, Rimoldi D, Nicolas M, Kaya G, Pescini Gobert R, Bedoni N, Schalenbourg A, Sakina E, Zografos L, Leyvraz S, Riggi N, **Rivolta C**, Moulin AP. **(2019)**. Conjunctival Melanoma Targeted Therapy: MAPK and PI3K/mTOR Pathways Inhibition. ***Invest Ophthalmol Vis Sci***. 60: 2764-2772. Impact Factor: 3.81

5. Nikopoulos K, Cisarova K, Quinodoz M, Koskiniemi-Kuendig H, Miyake N, Farinelli P, Rehman AU, Khan MI, Prunotto A, Akiyama M, Kamatani Y, Terao C, Miya F, Ikeda Y, Ueno S, Fuse N, Murakami A, Wada Y, Terasaki H, Sonoda KH, Ishibashi T, Kubo M, Cremers FPM, Kutalik Z, Matsumoto N, Nishiguchi KM, Nakazawa T, **Rivolta C. (2019)**. A frequent variant in the Japanese population determines quasi-Mendelian inheritance of rare retinal ciliopathy. ***Nat Commun*** 10: 2884. Impact Factor: 12.35

6. Del Pozo-Valero M, Martin-Merida I, Jimenez-Rolando B, Arteche A, Avila-Fernandez A, Blanco-Kelly F, Riveiro-Alvarez R, Van Cauwenbergh C, De Baere E, **Rivolta C**, Garcia-Sandoval B, Corton M, Ayuso C. **(2019)**. Expanded phenotypic spectrum of retinopathies associated with autosomal recessive and dominant mutations in PROM1. ***Am J Ophthalmol*** S0002-9394(19)30244-2. Impact Factor: 4.79

7. Peter VG, Nikopoulos K, Quinodoz M, Granse L, Farinelli P, Superti-Furga A, Andréasson S, **Rivolta C (2019).** A novel missense variant in IDH3A causes autosomal recessive retinitis pigmentosa. ***Ophthalmic Genet*** 40: 177-181. Impact Factor: 1.57

8. Verbakel SK, van Huet RAC, den Hollander AI, Geerlings MJ, Kersten E, Klevering BJ, Klaver CCW, Plomp AS, Wesseling NL, Bergen AAB, Nikopoulos K, **Rivolta C**, Ikeda Y, Sonoda KH, Wada Y, Boon CJF, Nakazawa T, Hoyng CB, Nishiguchi KM. **(2019)**. Macular Dystrophy and Cone-Rod Dystrophy Caused by Mutations in the RP1 Gene: Extending the RP1 Disease Spectrum. ***Invest Ophthalmol Vis Sci*** 60:1192-1203 Impact Factor: 3.39

9. Zhao M, Mantel I, Gelize E, Li X, Xie X, Arboleda A, Seminel M, Levy-Boukris R, Dernigoghossian M, Prunotto A, Andrieu-Soler C, **Rivolta C**, Canonica J, Naud MC, Lechner S, Farman N, Bravo-Osuna I, Herrero-Vanrell R, Jaisser F, Behar-Cohen F **(2019)**. Mineralocorticoid receptor antagonism limits experimental choroidal neovascularization and structural changes associated with neovascular age-related macular degeneration. ***Nat Commun*** 10:369 Impact Factor: 12.35

10. Fregni G, Quinodoz M, Möller E, Vuille J, Galland S, Fusco C, Martin P, Letovanec I, Provero P, **Rivolta C**, Riggi N, Stamenkovic I. **(2018)**. Reciprocal modulation of mesenchymal stem cells and tumor cells promotes lung cancer metastasis. ***EBioMedicine*** 29:128-145. Impact Factor: 6.18

11. Khateb S, Kowalewski B, Bedoni N, Damme M, Pollack N, Saada A, Obolensky A, Ben-Yosef T, Gross M, Dierks T*, Banin E*, **Rivolta C***, Sharon D*. **(2018)**. A homozygous founder missense variant in arylsulfatase G abolishes its enzymatic activity causing atypical Usher syndrome in humans. ***Genet Med.*** (in press). Impact Factor: 8.23. *co-senior authors. Impact Factor: 8.23

12. Quinodoz M, Royer-Bertrand B, Cisarova K, Di Gioia SA, Superti-Furga A, **Rivolta C. (2017).** DOMINO: using machine-learning to predict genes associated with dominant disorders. ***Am J Hum Genet***. 101:623-629. Impact Factor: 10.79

13. Volpi S, Yamazaki Y, Brauer PM, van Rooijen E, Hayashida A, Slavotinek A, Sun Kuehn H, Di Rocco M, **Rivolta C**, Bortolomai I, Du L, Felgentreff K, Ott de Bruin L, Hayashida K, Freedman G, Marcovecchio GE, Capuder K, Rath P, Luche N, Hagedorn EJ, Buoncompagni A, Royer-Bertrand B, Giliani S, Poliani PL, Imberti L, Dobbs K, Poulain FE, Martini A, Manis J, Linhardt RJ, Bosticardo M, Rosenzweig SD, Lee H, Puck JM, Zúñiga-Pflücker JC, Zon L, Park PW, Superti-Furga A, Notarangelo LD. **(2017)**. *EXTL3* mutations cause skeletal dysplasia, immune deficiency, and developmental delay. ***J Exp Med***. 214:623-637. Impact Factor: 11.24

14. Coppieters F, Ascari G, Dannhausen K, Nikopoulos K, Peelman F, Karlstetter M, Xu M, Brachet C, Meunier I, Tsilimbaris MK, Tsika C, Blazaki SV, Vergult S, Farinelli P, Van Laethem T, Bauwens M, De Bruyne M, Chen R, Langmann T, Sui R, Meire F, **Rivolta C**, Hamel CP, Leroy BP, De Baere E. **(2016).** Isolated and Syndromic Retinal Dystrophy Caused by Biallelic Mutations in RCBTB1, a Gene Implicated in Ubiquitination. ***Am J Hum Genet***. 99:470-80. Impact Factor: 10.79

15. Royer-Bertrand B, Torsello M, Rimoldi D, El Zaoui I, Pescini-Gobert R, Raynaud F, Cisarova K, Zografos L, Schalenbourg A, Speiser D, Nicolas M, Vallat L, Klein R, Leyvraz S, Ciriello G, Riggi N, Moulin AP, **Rivolta C. (2016)**. Comprehensive genetic landscape of uveal melanoma by whole-genome sequencing. ***Am J Hum Genet***. 99:1190-8. Impact Factor: 10.79

16. Bedoni N, Haer-Wigman L, Vaclavik V, Tran V, Farinelli P, Balzano S, Royer-Bertrand B, El-Asrag M, Bonny O, Ikonomidis C, Litzistorf Y, Nikopoulos K, Yioti GG, Stefaniotou MI, McKibbin M, Booth AP, Ellingford JM, Black GC, Toomes C, Inglehearn CF, Hoyng CB, Bax N, Klaver CCW, Thiadens AA, Murisier F, Schorderet DF, Ali M, Cremers FPM, Andreasson, S Munier FL, **Rivolta C. (2016)**. Mutations in the polyglutamylase gene TTLL5, expressed in photoreceptor cells and spermatozoa, are associated with cone-rod degeneration and reduced male fertility. ***Hum Mol Genet.*** 25:4546-4555. Impact Factor: 5.98

17. Nikopoulos K, Farinelli P, Giangreco B, Tsika C, Royer-Bertrand B, Mbefo MK, Kjellstrom U, El Zaoui I, Di Gioia SA, Balzano S, Cisarova K, Messina A, Decembrini S, Plainis S, Mplazaki S, Khan MI, Michael S, Boldt K, Ueffing M, Moulin AP, Cremers FPM, Roepman R, Arsenijevic Y, Tsilimbaris MK, Andreasson S, **Rivolta C. (2016)**. Mutations in *CEP78* cause cone-rod dystrophy and hearing loss associated with primary cilia defects. ***Am J Hum Genet***. 99:770-6. Impact Factor: 10.79

18. Kiper POS, Saito H, Gori F, Unger S, Hesse E, Yamana K, Kiviranta R, Solban N, Liu J, Brommage R, Boduroglu K, Bonafé L, Campos-Xavier B, Dikoglu E, Eastell R, Gossiel F, Harshman K, Nishimura G, Girisha KM, Stevenson BJ, Takita H, **Rivolta C**, Superti-Furga A*, Baron R* **(2016)**. Cortical-Bone Fragility--Insights from sFRP4 Deficiency in Pyle's Disease. ***N Engl J Med***. 374:2553-62. Impact Factor: 59.55. *shared contribution

19. van Karnebeek CD, Bonafe L, Wen XY, Tarailo-Graovac M, Balzano S, Royer-Bertrand B, Ashikov A, Garavelli L, Mammi I, Turolla L, Breen C, Donnai D, Cormier V, Heron D, Nishimura G, Uchikawa S, Campos-Xavier B, Rossi A, Hennet T, Brand-Arzamendi K, Rozmus J, Harshman K, Stevenson BJ, Girardi E, Superti-Furga G, Dewan T, Collingridge A, Halparin J, Ross CJ, Van Allen MI, Rossi A, Engelke UF, Kluijtmans LA, van der Heeft E, Renkema H, de Brouwer A, Huijben K, Zijlstra F, Heisse T, Boltje T, Wasserman WW, **Rivolta C**, Unger S, Lefeber DJ, Wevers RA and Superti-Furga A. **(2016)**. NANS-mediated synthesis of sialic acid is required for brain and skeletal development. ***Nat Genet***. 48:777-84. Impact Factor: 29.35

20. Sanchez-Alcudia R, Garcia-Hoyos M, Lopez-Martinez MA, Sanchez-Bolivar N, Zurita O, Gimenez A, Villaverde C, Rodrigues-Jacy da Silva L, Corton M, Perez-Carro R, Torriano S, Kalatzis V, **Rivolta C**, Avila-Fernandez A, Lorda I, Trujillo-Tiebas MJ, Garcia-Sandoval B, Lopez-Molina MI, Blanco-Kelly F, Riveiro-Alvarez R, Ayuso C. **(2016)**. A Comprehensive Analysis of Choroideremia: From Genetic Characterization to Clinical Practice. ***PLoS One***. 11: e0151943. Impact Factor: 3.23

21. Bonafé L, Kariminejad A, Li J, Royer-Bertrand B, Garcia V, Mahdavi S, Bozorgmehr B, Lachman RL, Mittaz-Crettol L, Campos-Xavier B, Nampoothiri S, Unger S, **Rivolta C**, Levade T, Superti-Furga A. **(2016)**. Peripheral osteolysis in adults linked to ASAH1 (acid ceramidase) mutations: A new presentation of Farber disease. ***Arthritis Rheumatol***. 68:2323-7. Impact Factor: 7.76

22. Sharon D, Kimchi A, **Rivolta C. (2016)**. OR2W3 sequence variants are unlikely to cause inherited retinal diseases. ***Ophthalmic Genet***. 18:1-3. Impact Factor: 1.45

23. Rose AM, Shah AZ, Venturini G, Krishna A, Chakravarti A, **Rivolta C**, Bhattacharya SS. **(2016)**. Transcriptional regulation of PRPF31 gene expression by MSR1 repeat elements causes incomplete penetrance in retinitis pigmentosa. ***Sci Rep***. 6:19450 Impact Factor: 5.58

24. Nikopoulos K, Butt GU, Farinelli P, Mudassar M, Domènech-Estévez E, Samara C, Kausar M, Masroor I, Chrast R, **Rivolta C***, Siddiqi S*. **(2016)**. A large multiexonic genomic deletion within the ALMS1 gene causes Alström syndrome in a consanguineous Pakistani family. ***Clin Genet*** 89: 510-511. *equal contribution. Impact Factor: 3.89

25. **Rivolta C,** Royer-Bertrand B, Rimoldi D, Schalenbourg A, Zografos L, Leyvraz S, Moulin A. **(2015)**. UV light signature in conjunctival melanoma; not only skin should be protected from solar radiation. ***J Hum Genet***. 61:361-2. Impact Factor: 2.46

26. Royer-Bertrand B, Castillo-Taucher S, Moreno-Salinas R, Cho TJ, Chae JH, Choi M, Kim OH, Dikoglu E, Campos-Xavier B, Girardi E, Superti-Furga G, Bonafé L, **Rivolta C**, Unger S, Superti-Furga A. **(2015)**. Mutations in the heat-shock protein A9 (HSPA9) gene cause the EVEN-PLUS syndrome of congenital malformations and skeletal dysplasia. ***Sci Rep*** 5:17154. Impact Factor: 5.58

27. Winkler TW, Justice AE, Graff M, Barata L,… **Rivolta C** … , Borecki IB, Kutalik Z, Loos RJ. **(2015)**. The Influence of Age and Sex on Genetic Associations with Adult Body Size and Shape: A Large-Scale Genome-Wide Interaction Study. ***PLoS Genet*** 10:e1005378. Impact Factor: 8.51

28. Nikopoulos K, Avila-Fernandez A, Corton M, Lopez-Molina MI, Perez-Carro R, Bontadelli L, Di Gioia SA, Zurita O, Garcia-Sandoval B, **Rivolta C***, Ayuso C*. **(2015)**. Identification of two novel mutations in CDHR1 in consanguineous Spanish families with autosomal recessive retinal dystrophy. ***Sci Rep*** 5:13902. *equal contribution. Impact Factor: 5.58

29. Safka Brozkova D, Deconinck T, Beth Griffin L, Ferbert A, Haberlova J, Mazanec R, Lassuthova P, Roth C, Pilunthanakul T, Rautenstrauss B, Janecke AR, Zavadakova P, Chrast R, **Rivolta C**, Zuchner S, Antonellis A, Beg AA, De Jonghe P, Senderek J, Seeman P, Baets J. **(2015)**. Loss of function mutations in HARS cause a spectrum of inherited peripheral neuropathies. ***Brain***. 138: 2161-2172. Impact Factor: 9.19

30. Garcia Segarra N, Ballhausen D, Crawford H, Perreau M, Campos-Xavier B, van Spaendonck-Zwarts K, Vermeer C, Russo M, Zambelli PY, Stevenson B, Royer-Bertrand B, **Rivolta C**, Candotti F, Unger S, Munier FL, Superti-Furga A, Bonafé L **(2015)**. NBAS mutations cause a multisystem disorder involving bone, connective tissue, liver, immune system, and retina. ***Am J Med Genet A*** 167:2902-12. Impact Factor: 2.05

31. Di Gioia SA, Bedoni N, von Scheven-Gête A, Vanoni F, Superti-Furga A, Hofer M, **Rivolta C. (2015)**. Analysis of the genetic basis of periodic fever with aphthous stomatitis, pharyngitis, and cervical adenitis (PFAPA) syndrome. ***Sci. Rep*** 5:10200. Impact Factor: 5.58

32. Saqib MA, Nikopoulos K, Ullah E, Sher Khan F, Iqbal J, Bibi R, Jarral A, Sajid S, Nishiguchi KM, Venturini G, Ansar M, **Rivolta C. (2015)**. Homozygosity mapping reveals novel and known mutations in Pakistani families with inherited retinal dystrophies. ***Sci. Rep*** 5:9965. Impact Factor: 5.58

33. Di Gioia SA, Farinelli P, Letteboer SJ, Arsenijevic Y, Sharon D, Roepman R and **Rivolta C. (2015).** Interactome analysis reveals that FAM161A, deficient in recessive retinitis pigmentosa, is a component of the Golgi-centrosomal network. ***Hum Mol Genet*** 15: 3359-3371. Impact Factor: 6.39

34. Bertelli C, Aeby S, Chassot B, Clulow J, Hilfiker O, Rappo S, Ritzmann S, Schumacher P, Terrettaz C, Benaglio P, Falquet L, Farinelli L, Gharib WH, Goesmann A, Harshman K, Linke B, Miyazaki R, **Rivolta C**, Robinson-Rechavi M, van der Meer JR, Greub G. **(2015).** Sequencing and characterizing the genome of Estrella lausannensis as an undergraduate project: training students and biological insights. ***Front Microbiol*** 6:101. Impact Factor: 4.00

35. Zhao M, Andrieu-Soler C, Kowalczuk L, Paz Cortés M, Berdugo M, Dernigoghossian M, Halili F, Jeanny JC, Goldenberg B, Savoldelli M, El Sanharawi M, Naud MC, van Ijcken W, Pescini-Gobert R, Martinet D, Maass A, Wijnholds J, Crisanti P, **Rivolta C** and Behar-Cohen F. **(2015).** A new CRB1 rat mutation links Müller glial cells to retinal telangiectasia. ***J Neurosci*** 35: 6093-6106. Impact Factor: 6.75

36. Benaglio P, San Jose PF, Avila-Fernandez A, Ascari G, Harper S, Manes G, Ayuso C, Hamel C, Berson EL and **Rivolta C**. **(2014)**. Mutational screening of splicing factor genes in cases with autosomal dominant retinitis pigmentosa. ***Mol Vis*** 20: 843-851. Impact Factor: 2.24

37. Hoggart CJ, Venturini G, Mangino M, … **Rivolta C**, Loos RJ and Kutalik Z. **(2014)**. Novel approach identifies SNPs in SLC2A10 and KCNK9 with evidence for parent-of-origin effect on body mass index. ***PLoS Genet*** 10: e1004508. Impact Factor: 8.51

38. Miyazaki R, Bertelli C, Benaglio P, Canton J, De Coi N, Gharib WH, Gjoksi B, Goesmann A, Greub G, Harshman K, Linke B, Mikulic J, Mueller L, Nicolas D, Robinson-Rechavi M, **Rivolta C**, Roggo C, Roy S, Sentchilo V, Siebenthal AV, Falquet L and van der Meer JR. **(2014)**. Comparative genome analysis of Pseudomonas knackmussii B13, the first bacterium known to degrade chloroaromatic compounds. ***Environ Microbiol***. 17: 91-104. Impact Factor: 6.24

39. Nishiguchi KM, Avila-Fernandez A, van Huet RA, Corton M, Perez-Carro R, Martin-Garrido E, Lopez-Molina MI, Blanco-Kelly F, Hoefsloot LH, van Zelst-Stams WA, Garcia-Ruiz PJ, Del Val J, Di Gioia SA, Klevering BJ, van de Warrenburg BP, Vazquez C, Cremers FP, Garcia-Sandoval B, Hoyng CB, Collin RW, **Rivolta C*** and Ayuso C*. **(2014)**. Exome sequencing extends the phenotypic spectrum for ABHD12 mutations: from syndromic to nonsyndromic retinal degeneration. ***Ophthalmology*** 121: 1620-1627. *equal contribution. Impact Factor: 5.56

40. Rahmioglu N, Macgregor S, Drong AW, Hedman AK, Harris HR, Randall JC, Prokopenko I, Nyholt DR, Morris AP, Montgomery GW, [**Rivolta C** as a group author] Missmer SA, Lindgren CM and Zondervan KT. **(2014)**. Genome-wide enrichment analysis between endometriosis and obesity-related traits reveals novel susceptibility loci. ***Hum Mol Genet*** 24: 1185-1199. Impact Factor: 7.54

41. Venturini G, Koskiniemi-Kuendig H, Harper S, Berson EL and **Rivolta C**. **(2014)**. Two specific mutations are prevalent causes of recessive retinitis pigmentosa in North American patients of Jewish ancestry. ***Genet Med*** 17: 285-290. Impact Factor: 6.43

42. Winkler TW, Day FR, Croteau-Chonka DC, Wood AR, Locke AE, Magi R, Ferreira T, Fall T, Graff M, Justice AE, Luan J, Gustafsson S, [**Rivolta C** as a group author] Randall JC, Vedantam S, Workalemahu T, Kilpelainen TO, Scherag A, Esko T, Kutalik Z, Heid IM and Loos RJ. **(2014)**. Quality control and conduct of genome-wide association meta-analyses. ***Nat Protoc*** 9: 1192-1212. Impact Factor: 7.78

43. Venturini G, Di Gioia SA, Harper S, Weigel-DiFranco C, **Rivolta C***† and Berson EL **(2014)***. Molecular genetics of FAM161A in North American patients with early-onset retinitis pigmentosa. ***PLoS ONE*** 9: e94479. *equal contribution, †correspondence. Impact Factor: 3.73

44. Rose AM, Shah AZ, Venturini G, **Rivolta C**, Rose GE, Bhattacharya SS **(2014)**. Dominant PRPF31 mutations are hypostatic to a recessive CNOT3 polymorphism in retinitis pigmentosa: a novel phenomenon of "linked trans-acting epistasis". ***Ann Hum Genet***. 78:62-71. Impact Factor: 2.21

45. Nishiguchi KM, Tearle RG, Liu YP, Oh EC, Miyake N, Benaglio P, Harper S, Koskiniemi-Kuendig H, Venturini G, Sharon D, Koenekoop RK, Nakamura M, Kondo M, Ueno S, Yasuma TR, Beckmann JS, Ikegawa S, Matsumoto N, Terasaki H, Berson EL, Katsanis N and **Rivolta C. (2013)**. Whole genome sequencing in patients with retinitis pigmentosa reveals pathogenic DNA structural changes and *NEK2* as a new disease gene. ***Proc Natl Acad Sci U S A***. 110:16139-16144. Impact Factor: 9.73

46. Azzedine H, Zavadakova P, Plante-Bordeneuve V, Vaz Pato M, Pinto N, Bartesaghi L, Zenker J, Poirot O, Bernard-Marissal N, Arnaud Gouttenoire E, Cartoni R, Title A, Venturini G, Medard JJ, Makowski E, Schols

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