Carlo Rivolta, Ph.D.

Tenured group leader (MER I)

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***Education**:*

1989-1994: Master of Science (Molecular Biology). University of Pavia, Italy. Score: 110/110 *cum laude*

1997: Twelve-month postgraduate course in bioinformatics at the Swiss Federal Institute of Technology Lausanne (EPFL)

1995-1999: Ph.D. in Science (Molecular Genetics) *cum laude* (mention). 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). Boston, USA

Positions held:

1994-1998: Collaborateur Scientifique (OFES), University of Lausanne

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) Department of Medical Genetics, University of Lausanne

Teaching (University of Lausanne):

2006-Present: Classes of "Medical Genetics" to 4th year medical students

2007-Present: Course of "Genetics & Genomics" to the 1st year of the M.Sc.program

2006-2007: Classes of Medical Genetics to 4th year medical students

2009-2010: Course of Ocular Genetics for the Ph.D. program

2010-present: Course of Genetics & Genomics for the Ph.D. program

2010-present: Course of Genomic Sequencing for the M.Sc. program

Scientific refereeing:

- The Wellcome Trust
- Swiss National Science Foundation
- Swiss League Against Cancer
- The Netherlands Organisation for Health Research and Development
- Agence Nationale de la Recherche (F)
- Czech Science Foundation
- Ad-hoc refereeing for several journals (J Clin Invest, Hum Mol Genet, Hum Genet, Science, BMC Mol Biol, etc.).

Grants:

2005-Present: Swiss National Science Foundation, Div 3 grant
2006: University of Lausanne. Cardiomet grant
2007-Present: European Union. FP7 grant
2008-Present: Synapsis Foundation grant
2008-2010: Swiss Multiple Sclerosis society
2009-Present: The Gebert Rüf Foundation

Publications (since 2008)

H-index = 15, in 14 years (1997-2011).
First or last author in 73% of all publications (87% over the last 5 years).

1. Rio Frio T, Wade NM, Ransijn A, Berson EL, Beckmann JS and **Rivolta C** (**2008**). Premature termination codons in *PRPF31* cause retinitis pigmentosa via haploinsufficiency due to nonsense-mediated mRNA decay. ***J Clin Invest***. 118: 1519-1531.

2. Rio Frio T, Civic N, Ransijn A, Beckmann JS* and **Rivolta C*#** (**2008**). Two trans-acting eQTLs modulate the penetrance of PRPF31 mutations. ***Hum Mol Genet***. 17: 3154-3165. (*equal contribution, #corresponding author).

3. Fukada T*, Civic N*, Furuichi T*, Shimoda S, Mishima K, Higashiyama H, Idaira Y, Asada Y, Kitamura H, Yamasaki S, Hojyo S, Nakayama M, Ohara O, Koseki H, Dos Santos HG, Bonafe L, Ha-Vinh R, Zankl A, Unger S, Kraenzlin ME, Beckmann JS, Saito I*, **Rivolta C***, Ikegawa S*, Superti-Furga A* and Hirano T *(**2008**). The zinc transporter SLC39A13/ZIP13 is required for connective tissue development; its involvement in BMP/TGF-beta signaling pathways. ***PLoS ONE***. 3: e3642. (*equal contribution).

4. Tanackovic G and **Rivolta C** (**2009**). PRPF31 alternative splicing and expression in human retina. ***Ophthalmic Genet***. 30: 76-83.

5. Butticaz C, Werge T, Beckmann JS, Cuenod M, Do KQ and **Rivolta C** (**2009**). Mutation screening of the glutamate cysteine ligase modifier (*GCLM*) gene in patients with schizophrenia. ***Psychiatr Genet***. 19: 201-208.

6. Rio Frio T, McGee TL, Wade NM, Iseli C, Beckmann JS, Berson EL and **Rivolta C** (**2009**). A single-base substitution within an intronic repetitive element causes dominant retinitis pigmentosa with reduced penetrance. ***Hum Mutat***. 30: 1340-1347.

7. Rio Frio T, Panek S, Iseli C, Di Gioia SA, Kumar A, Gal A and **Rivolta C** (**2009**). Ultra

high throughput sequencing excludes *MDH1* as candidate gene for RP28-linked retinitis pigmentosa. ***Mol Vis***. 15: 2627-2633.

8. Langmann T, Di Gioia SA, Rau I, Stöhr H, Maksimovic NS, Corbo JC, Renner AB, Zrenner E, Kumaramanickavel G, Karlstetter M, Arsenijevic Y, Weber BH, Gal A and **Rivolta C** (**2010**). Nonsense mutations in *FAM161A* cause RP28-associated recessive retinitis pigmentosa. ***Am J Hum Genet***. 87: 376-381.

9. Benaglio P and **Rivolta C** (**2010**). Ultra high throughput sequencing in human DNA variation detection: a comparative study on the *NDUFA3-PRPF31* region. ***PLoS ONE***. 5: e13071.

10. Lango Allen H, Estrada K, ... **Rivolta C** ... Frayling, TM and Hirschhorn JN **(2010)**. Hundreds of variants clustered in genomic loci and biological pathways affect human height. ***Nature***. 467:832-838.

11. Valsesia A, Rimoldi D, Martinet D, Ibberson M, Benaglio P, Gaillard M, Pidoux M, Rapin B, **Rivolta C**, Xenarios I, Simpson AJG, Antonarakis SE, Beckmann JS, Jongeneel CV, Iseli C, Stevenson BJ **(2011)**. Network-guided analysis of genes with altered somatic copy number and gene expression reveals pathways commonly perturbed in metastatic melanoma. ***PLoS ONE*** 6:e18369.

12. Benaglio P, McGee TL, Capelli LP, Harper S, Berson EL and **Rivolta C (2011)**. Next generation sequencing of pooled samples reveals new *SNRNP200* mutations associated with retinitis pigmentosa. ***Hum Mutat*** 32: E 2246-2258

13. Tanackovic G, Ransijn A, Thibault P, Abou Elela S, Klinck R, Berson EL, Chabot B and **Rivolta C (2011).** *PRPF* mutations are associated with generalized defects in spliceosome formation and pre-mRNA splicing in patients with retinitis pigmentosa. ***Hum Mol Genet*** 20:2116-21130.

14. Tanackovic G, Ransijn A, Ayuso C, Harper S, Berson EL and **Rivolta C (2011).** A missense mutation in PRPF6 causes impairment of pre-mRNA splicing and autosomal dominant retinitis pigmentosa. ***Am J Hum Genet*** 88:643-649.

15. Venturini G, Moulin AP, Deprez M, Uffer S, Bottani A, Zografos L and **Rivolta C (2011)**. Clinico-pathological and molecular analysis of a choroidal pigmented schwannoma in the context of a PTEN hamartoma tumor syndrome. ***Ophthalmology*** (in press).