

Curriculum Vitae

Jean MULLER

Born the 3rd may 1979 in Wissembourg (France)
Married, 3 children (2006, 2008 et 2012)



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Laboratoire de Diagnostic Génétique
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Languages : English and German

Education

- 1996 - 1997 **Baccalauréat**/High school diploma (biology option).
1998 - 1999 **DEUG**- General University Diploma Biology with honors (UdS, Strasbourg University).
1999 - 2000 **Degree** in Biology with honors (UdS).
2000 - 2001 **Master** in Biochemistry (Molecular biology) with honors (UdS).
2001 - 2002 **Post-master** in advanced specialized studies (DESS) - Complementary Skills in Informatics with honors (Fifth year professional degree) (UdS).
Thesis: « Development of an integrated program dedicated to the comparative analysis of groups of proteomes and genomes: application to pathogenic bacterial responsible for human diarrhea »
2002 - 2006 PhD in Bioinformatics co-directed by Dr Evelyne Friederich (CRP-Santé, Luxembourg) and Dr Olivier Poch (IGBMC, Strasbourg) with honors
Thesis: « high throughput Comparative genomics and transcriptomic analysis of cytoskeletons »
2007 - 2009 PostDoc at the European Molecular Biology Laboratory (Heidelberg, Germany) EMBL (Heidelberg) in Peer Bork's Computational Group.
2009 - **Maître de Conférences des Universités - Praticien Hospitalier** des Hôpitaux Universitaires et de la Faculté de Médecine de Strasbourg.

Personal authorization to perform human genetics tests or identification of human markers in medicine (20/12/2011 Agence de BioMédecine). extended to prenatal diagnosis (11/06/2014 Agence Régionale de la Santé).

Elected member of the Scientific Comity from the University of Strasbourg (since 2013).

Technical skills

Informatics and Bioinformatics:

Programming languages (C, Java, Tcl/Tk), Unix/Linux and Windows systems, network and databases. Knowledge and practices in sequences databases (nucleotide, protein and structure), in sequence analysis packages, homology searches, multiple alignments and phylogeny.

Comparative genomics.

Sequence annotation.

Microarray analysis and development (home made program for probe design).

3D structure analysis.

Biologist training during common projects.

Molecular biology:

Analysis and interpretation of Sanger sequencing and Next Generation Sequencing.

Analysis and interpretation of various molecular biology methods such as High Resolution Melt, Quantitative Multiplex PCR of Short Fragments and QPCR.

Analysis and interpretation of genotyping data including SNP chips.

Other Activities

Reviewing: Briefings in Bioinformatics, Clinical Genetics, Gene, Genome Research, Journal of Human Genetics, Molecular Biology of Evolution, Orphanet Journal of Rare Diseases, PLOS Genetics.

Grant Reviewing: Agence de Biomédecine «AMP, diagnostic prénatal et diagnostic génétique», Contrat Programme de Recherche Clinique CHU Nancy.

Grant obtained: Agence de Biomédecine «AMP, diagnostic prénatal et diagnostic génétique» (2011 : BBS, 2012 : Déficience intellectuelle, 2014 : Rétinopathies pigmentaire), PHRC-i 2014 RP.

Conference/Meeting Organization: Symposium Club Génétique de l'EST (2014, Strasbourg, France), European Tcl/Tk User Meeting (2008, 2009 et 2010 Strasbourg, France), FEBS/ESF workshop Integrated Approaches in Cytoskeleton Research (2005, Luxembourg, Luxembourg).

Teaching : Master Architectures et Fonctions du Vivant : Bioinformatique et Biologie Structurale, UE Transcriptomes et Protéomes (3H) (2009) ; ESBS 2ème année, « Structure et Annotation des Génomes » (8H + 6H TD + 3 weeks TP) (2009) ; PAES1 45H TD 2009- ; Master Physiopathologie cellulaire et moléculaire, UE Génétique Humaine (4H+CR Bibliography) (2009-) ; DES Biologie Médicale (2H) (2012-) ; DU Séquençage haut-débit et maladies génétiques à Dijon (7H) (2013-).

Students : *PhD:* Redin C. (UdS) 2010-14, *DUT :* Maeder M. 2014, Freytag D. 2012, *Master 1 :* Acker A. 2012, Diedhiou B. 2014, *Ingénieur/Master 2 :* Schwartz A. (ENSPS) 2014, Pizot C. (INSA) 2011, Seifert C. (UdS) 2009, Szklarczyk D. (EMBL) 2008.

Phd Jury: A. Doran, National University of Ireland Maynooth, 2014 ; E. Elinati, Université de Strasbourg 2012 ; C. Fossard, Faculté de Pharmacie de Strasbourg 2012

Prize : Prix de thèse 2007 de la Société de Biologie De Strasbourg (prix de l'association AREMANE).

Scientific Experience

Les auteurs ayant contribué de manière égale sont indiqués par *.

Journal article

1. Redin C., Gérard B., Lauer J., Herenger Y., **Muller J.**, Quartier A., Masurel-Paulet A., Willems M., Lesca G., El-Chehadeh S., Le Gras S., Vicaire S., Philipps M., Dumas M., Geoffroy V., Feger C., Haumesser N., Alembik Y., Barth M., Bonneau D., Colin E., Dollfus H., Doray B., Delrue MA., Drouin-Garraud V., Flori E., Fradin M., Francannet C., Goldenberg A., Lumbroso S., Mathieu-Dramard M., Martin-Coignard D., Lacombe D., Morin G., Polge A., Sukno S., Thauvin-Robinet C., Thevenon J., Doco-Fenzy M., Genevieve D., Sarda P., Edery P., Isidor B., Jost B., Olivier-Faivre L., Mandel JL., Piton A. **Efficient strategy for the molecular diagnosis of intellectual disability using targeted high-throughput sequencing.** J Med Genet. 2014
2. Huckert M., Mecili H., Laugel-Haushalter V., Stoetzel C., **Muller J.**, Flori E., Laugel V., Maniere M., Dollfus H., Bloch-Zupan A. **A novel mutation in the ROGD1 gene in a patient with Kohlschütter-Tönz Syndrome.** Molecular Syndromology 2014.
3. Renaud M.*, Anheim M.*, Kamsteeg E.J., Mallaret M., Mochel F., Vermeer S, Drouot N., Pouget J., Redin C., Salort-Campana E., Kremer H.P.H, Corien C., Verschuuren-Bemelmans C.C., Muller J., Scheffer H., Durr A., Tranchant C., Koenig M. **ARCA3 due to ANO10 mutations: delineation and genotype/phenotype correlation study.** JAMA Neurology 2014
4. Braun JJ.*, Noblet V.*, Durand M., Scheidecker S., Zinetti-Bertschy A., Foucher J., Marion V., **Muller J.**, Riehm S., Dollfus H., Kremer S. **Olfaction evaluation and correlation with brain atrophy in Bardet-Biedl syndrome.** Clinical Genetics. 2014 Mar 29.
5. Piton A.*, Poquet H.*, **Redin C.**, Masurel A., Lauer J., **Muller J.**, Thevenon J., Herenger Y., Chancenotte S., Bonnet M., Pinoit JM., Huet F., Thauvin-Robinet C., Jaeger AS., Le Gras S., Jost B., Gérard B., Peoc'h K., Launay JM., Faivre L.*, Mandel JL*. **20 ans après: a second mutation in MAOA identified by targeted high-throughput sequencing in a family with altered behavior and cognition.** European Journal Human Genetics. 2013 Oct 30.
6. Scheidecker S.*, Etard C.*, Pierce NW.*, Geoffroy V., Schaefer E., **Muller J.**, Chennen K., Flori E., Pelletier V., Poch O., Marion V., Stoetzel C., Strahle U., Nachury MV., Dollfus H. **Exome sequencing of Bardet-Biedl syndrome patient identifies a null mutation in the BBSome subunit BBIP1 (BBS18).** Journal of Medical Genetics. 2013 Sep 11.
7. Bonnefond A.*, Philippe J.*, Durand E., **Muller J.**, Saeed S., Arslan M.D, Martinez S., De Graeve F., Dhennin V., Rabearivelo I., Polak M., Cavé H., Castano L., Vaxillaire M., Mandel JL., Sand O. & Froguel P. **Highly sensitive diagnosis of 43 monogenic forms of diabetes or obesity, through one step PCR-based enrichment in combination with next-generation sequencing.** Diabetes Care 2013.
8. Fradin M., Merklen-Djafri C., Perrigouard C., Aral B., **Muller J.**, Stoetzel C., Frouin E., Flori E., Doray B., Dollfus H., Lipsker D. **Long-Term Follow-Up and Molecular Characterization of a Patient with a RECQL4 Mutation Spectrum Disorder.** Dermatology 2013 Jul 26.
9. Piton A.*, **Redin C.***, Mandel JL. **XLID-Causing Mutations and Associated Genes Challenged in Light of Data From Large-Scale Human Exome Sequencing.** American Journal of Human Genetics. 2013 Jul 18.

10. Schaefer E.*, Lauer J.*, Durand M., Pelletier V., Obringer C., Clausmann A., Braun JJ., **Redin C.**, Mathis C., **Muller J.**, Schmidt-Mutter C., Flori E., Marion V., Stoetzel C., Dollfus H. **Mesoaxial Polydactyly is a Major Feature in Bardet-Biedl Syndrome (BBS) patients with LZTFL1 (BBS17) Mutations.** *Clinical Genetics*. 2013 May 21
11. M'Hamdi O.*, **Redin C.***, Stoetzel C.*, Ouertani I., Chaabouni M., Faouzi M., M'rad R., Mandel JL., Dollfus H., **Muller J.**, Chaabouni H. **Clinical and Genetic Characterization of Bardet-Biedl Syndrome in Tunisia: Defining a Strategy for Molecular Diagnosis.** *Clinical Genetics*. 2013 Feb 21.
12. Ellnati E., **Muller J.**, Viville S. **Autosomal mutations and human spermatogenic failure.** *BBA Molecular Basis of Disease*, 2012 July 27.
13. **Redin C.**, Le Gras S., Mhamdi O., Geoffroy V., Stoetzel C., Vincent MC., Chiurazzi P., Lacombe D., Ouertani I., Petit F., Till M., Verloes A., Jost B., Chaabouni H.B., Dollfus H., Mandel JL., **Muller J.** **Targeted High-Throughput Sequencing for Diagnosis of Genetically Heterogeneous Diseases: Efficient Mutation Detection in Bardet-Biedl and Alström Syndromes.** *Journal of Medical Genetics*, 2012 July 7.
14. Elinati E.*, Kuentz P.*, **Redin C.**, Jaber S., Vanden Meerschaut F., Magarian J., Koscinski I., H Nasr-Esfahani M., Demirol A., Gurgan T., Louanjli N., Iqbal N., Bisharah M, Carré Pigeon F., Gourabi H., De Briel D., Brugnion F., Gitlin S.A., Grillo JM., Deemeh X., Ghaedi K., Tanhee X., Heindryckx B., Benkhalifa M., Nikiforaki D., Oehninger S.C., De Sutter P., **Muller J.**, Viville S. **Globozoospermia is mainly due to DPY19L2 deletion via non allelic homologous recombination involving two recombination hotspots.** *Human Molecular Genetics*. 2012 May 31
15. Bricker DK.*, Taylor EB. *, Schell JC. *, Orsak T. *, Boutron A., Chen YC., Cox JE., Cardon CM., Van Vranken JG., Dephoure N., **Redin C.**, Boudina S., Gygi SP., Brivet M., Thummel CS., Rutter J. **A Mitochondrial Pyruvate Carrier Required for Pyruvate Uptake in Yeast, Drosophila, and Humans.** *Science*. 2012 May 24
16. Luu TD., Rusu A., Walter V., Linard B., Poidevin L., Ripp R., Moulinier L, **Muller J.**, Raffelsberger W., Wicker N., Lecompte O., Thompson J.D., Poch O. , Nguyen H. **KD4v: Comprehensive Knowledge Discovery System For Missense Variant.** *Nucleic Acid Research* 2012 May 27.
17. Vasli N., Böhm B., Le Gras S., **Muller J.**, Pizot C., Jost B., Echaniz-Laguna A., Laugel V., Tranchant C., Plewniak F., Vicaire S., Jean-Louis Mandel JL. , Biancalana V., Laporte J. **Next generation sequencing for molecular diagnosis of neuromuscular diseases.** *Acta Neuropathologica* 2012.
18. Cowling B., Toussaint A., **Muller J.**, Laporte J. **Defective Membrane Remodeling in Neuromuscular Diseases: Insights from Animal Models.** *PLoS Genetics*. 2012.
19. Luu TD., Rusu AM., Walter V., Ripp R., Moulinier L, **Muller J.**, Torsel T., Thompson J.D., Poch O., Nguyen H. **MSV3d: Database of human MisSense Variants mapped to 3D protein structure.** *Database* 2012.
20. Bloch-Zupan A., Jamet X., Etard C., **Muller J.**, Geoffroy V., Strauss JP., Pelletier V., Marion V., Poch O., Strahle U., Stoetzel C. and Dollfus H. **Homozygosity Mapping and Candidate Prioritization Identify Mutations, Missed by Whole-Exome Sequencing, in SMOX2, Causing Major Dental Developmental Defects.** *American Journal of Human Genetics*. 2011 Dec 9.

21. Creevey C.J.*, **Muller J.***, Doerks T., Thompson J. D., Arendt D., Bork P. **Single copy orthologs in Metazoa**. PLoS Computational Biology 2011 Dec.
22. Powell S., Szklarczyk D., Trachana K., Roth A., Kuhn M., **Muller J.**, Arnold R., Rattei T., Letunic I., Doerks T., Jensen L.J., von Mering C., Bork P. **eggNOG v3.0: orthologous groups covering 1133 organisms at 41 different taxonomic ranges**. Nucleic Acids Res. 2011 Nov 16.
23. Trachana K., Larsson TA., Powell S., Chen WH., Doerks T., **Muller J.** and Bork P. **Orthology prediction methods: A quality assessment using curated protein families**. Bioessays. 2011 Aug 19.
24. Luu TD., Nguyen NH., Friedrich A., **Muller J.**, Moulinier L., Poch O. **Extracting Knowledge from a Mutation Database Related to Human Monogenic Disease Using Inductive Logic Programming**. In *International Conference on Bioscience, Biochemistry and Bioinformatics; Singapore 2011*. IEEE Catalog Number: CFP1134M-PRT. ISBN: 978-1-4244-9388-3.
25. Schaefer E. Zaloszc A., Lauer J., Durand M., Stutzmann F., Perdomo-Trujillo Y., Redin C., Bennouna Greene V., Toutain A., Perrin L., Gérard M., Caillard S., Bei X., Lewis R.A., Christmann D., Letsch J., Kribs M., Mutter C., **Muller J.**, Stoetzel C., Fischbach M., Marion V., Katsanis N., Dollfus H. **Mutations in SDCCAG8/NPHP10 Cause Bardet-Biedl Syndrome and Are Associated with Penetrant Renal Disease and Absent Polydactyly**. Molecular Syndromology 2011
26. The MetaHIT Consortium, incluant Arumugam M., Raes J., Yamada T., Mende DR., **Muller J.** and Bork P. **Enterotypes of the human gut microbiome**. Nature April 2011.
27. Kosciński I.*, Elinati E. *, Fossard C., Redin C., **Muller J.**, Velez de la Calle J., Schmitt F., Khelifa M.B., Ray P., Kilani Z., Barratt CL.R., Viville S. **DPY19L2 deletion as a major of cause of globozoospermia**. American Journal of Human Genetics. 2011.
28. Colbourne JK., Pfender ME., Gilbert D., Thomas WK., Tucker A., Oakley TH., Tokishita S., Aerts A., Arnold GJ., Basu MK., Bauer DJ., Caceres CE., Carmel L., Casola C., Choi JH., Detter JC., Dong Q., Dusheyko S., Eads BD., Frohlich T., Geiler-Samerotte KA., Gerlach D., Hatcher P., Jogdeo S., Krijgsveld J., Kriventseva EV., Kultz D., Laforsch C., Lindquist E., Lopez J., Manak JR., **Muller J.**, Pangilinan J., Patwardhan1 RP., Pitluck S., Pritham EJ., Rechtsteiner A., Rho M., Rogozin IB., Sakarya O., Salamov A., Schaack S., Shapiro H., Shiga Y., Skalitzky C., Smith Z., Souvorov A., Sung W., Tang Z., Tsuchiya D., Tu H., Vos H., Wang M., Wolf YI., Yamagata H., Yamada T., Ye Y., Shaw JR., Andrews J., Crease TJ., Tang H., Lucas SM., Robertson HM., Bork P., Koonin EV., Zdobnov EM., Grigoriev IV., Lynch M. and Boore JL. **The Ecoresponsive Genome of Daphnia pulex**. Science. February 2011.
29. Szklarczyk D., Franceschini A., Kuhn M., Simonovic M., Roth A., Minguéz P., Doerks T., Starck M., **Muller J.**, Bork P., Jensen L.J. and von Mering C. **The STRING database in 2011: functional interaction networks of proteins, globally integrated and scored**. Nucleic Acids Research, 2010 Nov 3.
30. Fradin M., Stoetzel C., **Muller J.**, Koob M., Christmann D., Debry C., Kohler M., Isnard M., Astruc D., Desprez P., Zorres C., Flori E., Dollfus H., Doray B. **Osteosclerotic bone dysplasia in siblings with a Fam20C mutation**. Clinical Genetics. 2010 Jul 23.
31. **Muller J.***, Stoetzel C.*, Vincent MC., Leitch CC., Laurier V., Danse JM., Hellé S., Marion V., Bennouna-Greene V., Vicaire S., Megarbane A., Kaplan J., Drouin-Garraud V., Hamdani M., Sigaudy S., Francannet C., Roume J., Bitoun P., Goldenberg A., Philip N., Odent S., Green J.,

- Cossée M., Davis EE., Katsanis N., Bonneau D., Verloes A., Poch O., Mandel JL., Dollfus H. **Identification of 28 novel mutations in the Bardet-Biedl syndrome (BBS) genes: the burden of private mutations in an extensively heterogeneous disease.** Human Genetics. 2010 Feb 23.
32. Friedrich A.*, Garnier N.*, Gagnière N., Nguyen H., Albou LP., Biancalana V., Bettler E., Deléage G., Lecompte O., **Muller J.**, Moras D., Mandel JL., Toursel T., Moulinier L., Poch O. **SM2PH-db: an interactive system for the integrated analysis of phenotypic consequences of missense mutations in proteins involved in human genetic diseases.** Human Mutation. 2010 Feb.
 33. The Nasonia Genome Working Group, incluant **Muller J.**, Yamada T. and Bork P. **Functional and Evolutionary Insights from the Genomes of Three Parasitoid Nasonia Species.** Science. January 2010.
 34. **Muller J.***, Creevey CJ.*, Thompson JD., Arendt D., Bork P. **AQUA: Automated quality improvement for multiple sequence alignments.** Bioinformatics. 2009 Nov 19.
 35. **Muller J.**, Szklarczyk D., Julien P., Letunic I., Roth A., Kuhn M., Powell S., von Mering C., Doerks T., Jensen L.J. and Bork P. **eggNOG v2.0: extending the evolutionary genealogy of genes with enhanced non-supervised orthologous groups, species and functional annotations.** Nucleic Acids Research 2009.
 36. Vetter G*, Le Béchech A*, **Muller J.**, Muller A, Moes M, Yatskou M, Altanoury Z, Poch O, Vallar L, Friederich E. **Time-resolved analysis of transcriptional events during SNAI1-triggered epithelial to mesenchymal transition.** Biochem Biophys Res Commun. 2009 May 11.
 37. Foerstner K. U., Doerks T., **Muller J.**, Raes J., Bork P. **A Nitrile Hydratase in the Eukaryote *Monosiga brevicollis*** PLoS ONE 2008.
 38. Jensen L.J.* , Kuhn M.* , Stark M., Chaffron S., Creevey C., **Muller J.**, Doerks T., Julien P., Roth A., Simonovic M., Bork P. and von Mering C. **STRING 8: a global view on proteins and their functional interactions in 630 organisms.** Nucleic Acids Research. 2008.
 39. Becker J.A.J., Befort K., Blad C., Filliol D., Ghate A., Dembele D., Thibault C., Koch M., **Muller J.**, Lardenois A., Poch O. and Kieffer BL. **Transcriptome analysis identifies genes with enriched expression in the mouse central extended amygdala.** Neuroscience 2008 Aug 14.
 40. Befort K, Filliol D, Ghate A, Darcq E, Matifas A, **Muller J.**, Lardenois A, Thibault C, Dembele D, Le Merrer J, Becker JA, Poch O, Kieffer BL. **Mu-opioid receptor activation induces transcriptional plasticity in the central extended amygdala.** European Journal of Neuroscience. 2008 Jun.
 41. Befort K., Filliol D., Darcq E., Ghate A, Matifas A., Lardenois A., **Muller J.**, Thibault C., Dembele D., Poch O. and Kieffer B.L. **Gene Expression Is Altered in the Lateral Hypothalamus upon Activation of the mu Opioid Receptor** Annals of New York Academy of Sciences. 2008.
 42. Wicker N., **Muller J.**, Kiran Reddy Kalathur R. and Poch O. **A maximum likelihood approximation method for Dirichlet's parameter estimation** Computational Statistics & Data Analysis 2008 Jan 1.
 43. Jensen LJ*, Julien P*, Kuhn M, von Mering C, **Muller J.**, Doerks T, Bork P. **eggNOG: automated construction and annotation of orthologous groups of genes.** Nucleic Acids Research. 2007 Oct 16

44. **Muller J**, Mehlen A, Vetter G, Yatskou M, Muller A, Chalmel F, Poch O, Friederich E, Vallar L. **Design and evaluation of Actichip, a thematic microarray for the study of the actin cytoskeleton.** BMC Genomics. 2007 Aug 29.
45. Stoetzel C*, **Muller J***, Laurier V, Davis EE, Zaghoul NA, Vicaire S, Jacquelin C, Plewniak F, Leitch CC, Sarda P, Hamel C, de Ravel TJ, Lewis RA, Friederich E, Thibault C, Danse JM, Verloes A, Bonneau D, Katsanis N, Poch O, Mandel JL, Dollfus H. **Identification of a Novel BBS gene (BBS12) Highlights the Major Role of a Vertebrate-Specific Branch of Chaperonin-Related Proteins in Bardet-Biedl Syndrome.** American Journal of Human Genetics. Jan 2007.
46. Dollfus H., **Muller J.**, Stoetzel C., Laurier V., Bonneau D., Mégarbané A., Poch O., Mandel JL. **Bardet-Biedl syndrome: a unique family for a major gene (BBS10).** Médecine Sciences 2006.
47. Laurier V.*, Stoetzel C.*, **Muller J.**, Thibault C., Corbani S., Jalkh N., Nabiha S., Chouery E., Poch O., Danse JM., Amati-Bonneau P., Bonneau D., Mégarbané A., Mandel JL. and Dollfus H. **Pitfalls of homozygosity mapping: an extended consanguineous Bardet–Biedl syndrome family with two mutant genes (BBS2, BBS10), three mutations, but no triallelism.** European Journal of Human Genetics. Jul 5 2006.
48. Stoetzel C*, Laurier V*, Davis EE*, **Muller J***, Rix S, Badano JL, Leitch CC, Salem N, Chouery E, Corbani S, Jalk N, Vicaire S, Sarda P, Hamel C, Lacombe D, Holder M, Odent S, Holder S, Brooks AS, Elcioglu NH, Da Silva E, Rossillion B, Sigaudy S, de Ravel TJ, Alan Lewis R, Leheup B, Verloes A, Amati-Bonneau P, Megarbane A, Poch O, Bonneau D, Beales PL, Mandel JL, Katsanis N, Dollfus H. **BBS10 encodes a vertebrate-specific chaperonin-like protein and is a major BBS locus.** Nature Genetics. Apr 2 2006.
49. Perrodou, E., Deshayes, C., **Muller, J.**, Schaeffer, C., Van Dorsselaer, A., Ripp, R., Poch, O., Reyrat, JM. and Lecompte, O. **ICDS database: interrupted CoDing sequences in prokaryotic genomes.** Nucleic Acids Research 2006.
50. **Muller, J.**, Oma, Y., Vallar, L., Friederich, E., Poch, O., and Winsor, B. **Sequence and Comparative Genomic Analysis of Actin-related Proteins.** Molecular Biology Cell. 2005.
51. Chalmel, F., Lardenois, A., Thompson, J.D., **Muller, J.**, Sahel, J.A., Leveillard, T., and Poch, O. **GOAnno: GO annotation based on multiple alignment.** Bioinformatics .2005.
52. Plewniak, F., Bianchetti, L., Brelivet, Y., Carles, A., Chalmel, F., Lecompte, O., Mochel, T., Moulinier, L., Muller, A., **Muller, J.**, Prigent, V., Ripp, R., Thierry, J.C., Thompson, J.D., Wicker, N., and Poch, O. **PipeAlign: A new toolkit for protein family analysis.** Nucleic Acids Research 2003.

Oral Communications

High Throughput Sequencing Applications in Diagnostics and Variant Ranking

BinGI days (rencontre laboratoire et plateforme bioinformatique) (Strasbourg, France), avril 2014.

Atelier Agilent : Applications diagnostiques du séquençage ciblé par capture dans des pathologies génétiquement hétérogènes.

7^{èmes} Assises de Génétique Humaine et Médicale, (Bordeaux, France), 29-31 janvier 2014.

Multi-gene resequencing for molecular diagnosis of obesity.

New genomics toward personalized medicine Lille Genomics European symposium (Lille, France), 10 octobre 2013.

Bases de données de mutations et de polymorphismes disponibles sur le web et leurs pièges.

Réunion des généticiens moléculaires de France – ANPGM, 7 septembre 2012.

Capture ciblée et séquençage à haut débit pour le diagnostic de maladies génétiquement très hétérogènes: Bardet-Biedl et autres ciliopathies

Journée Génomique Clinique, Agilent Technologies, « Outils génomiques pour l'activité clinique et hospitalière », 28 juin 2012.

***In silico* predictions tools.**

35th European Cystic Fibrosis Conference (Dublin, Irlande), Juin 2012.

Syndrome de Bardet-Biedl : description clinique et génétique.

2^{ème} journée Société Française d'Endocrinologie Diabétologie Pédiatrique (Paris, France), Mai 2012.

Séquençage à haut débit et diagnostic des maladies rares.

Mardi du CNRS, (Strasbourg, France) 17 avril 2012.

Etat des lieux, résultat de l'enquête nationale Groupe de travail « Plateformes nationales de laboratoires de diagnostic approfondi ».

Assemblée Générale ANPGM (Paris, France) 12 mars 2012.

Capture ciblée et séquençage à haut débit pour le diagnostic de maladies génétiquement très hétérogènes: Bardet-Biedl et autres ciliopathies.

6^{èmes} Assises de Génétique Humaine et Médicale (Marseille, France), 2-4 février 2012

Atelier SFE 9 : Le séquençage à haut débit du génome entier (C. Dodé et J. Muller).

28^{ème} congrès Société Française d'Endocrinologie (Clermont-Ferrand, France), Octobre 2011.

Bioinformatics and Genetic Diseases.

Rencontre des Laboratoires de Biologie Structurales IGBMC/IBMC (Obernai, France), Mars 2010.

An unusual family of non-lethal Raine Syndrome with Fam20C mutation contributes to a new classification of the Fam20 genes.

BinGI days (rencontre laboratoire et plateforme bioinformatique) (Strasbourg, France), Janvier 2010.

eggNOG v2.0: evolutionary genealogy of genes Non-supervised Orthologous Groups.

Quest for Orthologs, Wellcome Trust Conference Centre, Hinxton, Cambridge, UK, Juillet 2009.

Comparative high-throughput genomics to unravel genetic diseases: Application to the Bardet-Biedl Syndrome.

Workshop on Bioinformatics and modeling in biomedicine: From genes to biological systems, (Luxembourg, Luxembourg), Octobre 2006.