

Anthony Mathelier, Ph.D.

Group Leader

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Currently

Group Leader of the Computational Biology & Gene Regulation Group at the Centre for Molecular Medicine Norway (NCMM), Nordic EMBL Partner for Molecular Medicine, University of Oslo, and **Adjunct Researcher** at the Dept. of Cancer Genetics, Oslo University Hospital, in Oslo, Norway.

Employment

- 2016- **Group Leader** at the Centre for Molecular Medicine Norway (NCMM), Oslo, Norway
- 2016- **Adjunct Researcher** at the Dept. of Cancer Genetics, Institute for Cancer Research, Oslo University Hospital, Oslo, Norway
- 2013-16 **Deputy Group Leader** at the Wasserman laboratory, Centre for Molecular Medicine and Therapeutics (CMMT), Child and Family Research Institute (CFRI), University of British Columbia (UBC), Vancouver, Canada
- 2011-13 **Postdoctoral fellow** at the Wasserman laboratory, CMMT, CFRI, UBC, Vancouver, Canada
- 2006-10 **Teaching assistant** at the Université Pierre et Marie Curie (UPMC), Paris, France
- 2006-10 **Ph.D. student** at the Laboratory of Computational and Quantitative Biology, Analytical Genomics Group, UPMC, Paris, France

Education

- 2006-10 **Ph.D.** in Computer Science and Bioinformatics (supervisor: Dr. Alessandra Carbone), UPMC, Paris, France
- 2004-06 **M.Sc.** in Computer Science, UPMC, Paris, France

Grants and Awards

- 2018 Åsgard Research Mobility Program. Funding for spending one week in France developing new scientific interactions.
- 2017 NCMM Programme for networking with Associate Investigators and Young Associate Investigators. 480K NOK funding a project on "The molecular mechanism and impact of transition from diploid to aneuploid cells in cancer."
- 2016 Start-up package from the NCMM to launch the Computational Biology & Gene Regulation Group with 18M NOK over 5 years.
Participation in the successful grant application "OnTarget: Big Data Informed Software for the Design of cis-Regulatory Regions Controlling Human Gene Expression" of the Wasserman lab selected by Genome Canada with funding of CAD 250K over 2 years.
- 2013 Participation in the successful grant application "Applied Bioinformatics of *Cis*-regulation for Disease Exploration (ABC4DE)" of the Wasserman lab selected by Genome Canada with funding of CAD 1M over 3 years
- 2012 Child and Family Research Institute Trainee Travel Award
European Conference on Computational Biology 2012, conference fellowship funded by the Swiss Institute of Bioinformatics
- 2011 Child and Family Research Institute Trainee Travel Award
- 2009 ISMB/ECCB Travel Fellowship from the International Society for Computational Biology funded by BioSapiens
- 2006-09 Ministry of National Education, Research and Technology Ph.D. scholarship attributed by the President of Paris 6 University

Publications

Preprints

- 2018 M. Gheorghe, G.K. Sandve, A. Khan, J. Chèneby, B. Ballester, and **A. Mathelier**. A map of direct TF-DNA interactions in the human genome. *bioRxiv*, doi:10.1101/394205.
- B. Fromm, D. Domanska, M. Hackenberg, **A. Mathelier**, E. Høy, M. Johansen, E. Hovig, K. Flatmark, and K.J. Peterson. MirGeneDB2.0: the curated microRNA Gene Database. *bioRxiv*, doi:10.1101/258749.

Peer-reviewed journals

- 2018 O. Fornes*, M. Gheorghe*, P.A. Richmond, D.J. Arenillas, W.W. Wasserman+, and **A. Mathelier**. MANTA2, update of the Mongo database for the analysis of transcription factor binding site alterations. *Scientific Data*, doi:10.1038/sdata.2018.141.
- E. Simpson, A.J. Korecki, O. Fornes, T.J. McGill, J.L. Cueva-Vargas, J. Agostinone, R.A. Farkas, J.W. Hickmott, S.L. Lam, **A. Mathelier**, L.M. Renner, J. Stoddard, M. Zhou, A. Di Polo, M. Neuringer, W.W. Wasserman. New MiniPromoter Ple345 (NEFL) Drives Strong and Specific Expression in Retinal Ganglion Cells of Mouse and Primate Retina. *Human Gene Therapy*, doi:10.1089/hum.2018.118.
- R.A.G. De Souza*, N. Kosior*, S.B. Thomson, **A. Mathelier**, A.W. Zhang, K. Becanovic, W.W. Wasserman, and B.R. Leavitt. Computational Analysis of Transcriptional Regulation Sites at the HTT Gene Locus. *Journal of Huntington's Disease*, doi:10.3233/JHD-170272.
- C.-H. Lecellier+, W.W. Wasserman, and **A. Mathelier**+. Human enhancers harboring specific sequence composition, activity, and genome organization are linked to the immune response. *Genetics*, doi:10.1534/genetics.118.301116.

- A. Khan, A. Montenegro-Montero, and **A. Mathelier**. Put science first and formatting later. *EMBO Reports*, doi:10.15252/embr.201845731.
- D. Sugiama-Trapman, M. Vitezic, E.-M. Jouhilahti, **A. Mathelier**, G. Lauter, S. Misra, C.O. Daub, J. Kere, and P. Swoboda. Characterization of the human RFX transcription factor family by regulatory and target gene analysis. *BMC Genomics*, doi:10.1186/s12864-018-4564-6.
- 2017 A. Khan and **A. Mathelier**. JASPAR RESTful API: accessing JASPAR data from any programming language. *Bioinformatics*, doi:10.1093/bioinformatics/btx804.
- A. Khan*, O. Fornes*, A. Stigliani*, M. Gheorghe, J.A. Castro-Mondragón, R. van der Lee, A. Bessy, J. Chèneby, S.R. Kulkarni, G. Tan, D. Baranasic, D.J. Arenillas, A. Sandelin+, K. Vandepoele, B. Lenhard+, B. Ballester, W.W. Wasserman+, F. Parcy, and **A. Mathelier**+. JASPAR 2018: update of the open-access database of transcription factor binding profiles and its web framework. *Nucleic Acids Research*, doi:10.1093/nar/gkx1126.
- T. Fleischer*, X. Tekpli*, **A. Mathelier**, S. Wang, D. Nebdal, H.P. Dhakal, K.Kleivi Sahlberg, E. Schlichting, Oslo Breast Cancer Research Consortium (OSBREAC), A.-L. Børresen-Dale, E. Borgen, B. Naume, R. Eskeland, A. Frigessi, J. Tost, A. Hurtado, and V.N. Kristensen. DNA methylation at enhancers identifies distinct breast cancer lineages. *Nature Communications*, doi:10.1038/s41467-017-00510-x.
- J. Chèneby, M. Gheorghe, M. Artufel, **A. Mathelier**, and B. Ballester. ReMap 2018: an updated atlas of regulatory regions from an integrative analysis of DNA-binding ChIP-seq experiments. *Nucleic Acids Research*, doi:10.1093/nar/gkx1092.
- D. de Rie, ..., **A. Mathelier**, ..., M.J.L. de Hoon (70 authors). An integrated expression atlas of miRNAs and their promoters in human and mouse. *Nature Biotechnology*, doi:10.1038/nbt.3947.
- A. Khan and **A. Mathelier**. Intervene: a tool for intersection and visualization of multiple gene or genomic region sets. *BMC Bioinformatics*, doi:10.1186/s12859-017-1708-7.
- 2016 C.-Y. Chen, W. Shi, B.P. Balaton, A.M. Matthews, Y. Li, D.J. Arenillas, **A. Mathelier**, M. Itoh, H. Kawaji, T. Lassmann, Y. Hayashizaki, P. Carninci, A.R.R. Forrest, C.J. Brown, and W.W. Wasserman. YY1 binding association with sex-biased transcription revealed through X-linked transcript levels and allelic binding analyses. *Scientific Reports*, doi:10.1038/srep37324.
- M. Lizio, J. Harshbarger, I. Abugessaisa, S. Noguchi, A. Kondo, J. Severin, C. Mungall, D. Arenillas, **A. Mathelier**, Y.A. Medvedeva, A. Lennartsson, F. Drablos, J.A. Ramilowski, O. Rackham, J. Gough, R. Andersson, A. Sandelin, H. Ienasescu, H. Ono, H. Bono, Y. Hayashizaki, P. Carninci, A.R.R. Forrest, T. Kasukawa+ and H. Kawaji+. Update of the FANTOM web resource: high resolution transcriptome of diverse cell types in mammals. *Nucleic Acids Research*, doi: 10.1093/nar/gkw995.
- A. Mathelier**, B. Xin, T.-P. Chiu, L. Yang, R. Rohs, and W.W. Wasserman. DNA Shape Features Improve Transcription Factor Binding Site Predictions In Vivo. *Cell Systems*, doi:10.1016/j.cels.2016.07.001.
- J.W. Hickmott, C.-Y. Chen, D.J. Arenillas, A.J. Korecki, S.L. Lam, L.L. Molday, R.J. Bonaguro, M. Zhou, A.Y. Chou, **A. Mathelier**, S.L. Boye, W.W. Hauswirth, R.S. Molday, W.W. Wasserman, and E.M. Simpson. PAX6 MiniPromoters drive restricted expression from rAAV in the adult mouse retina. *Molecular therapy - Methods & Clinical Development*, doi:10.1038/mtm.2016.51.
- W. Shi, O. Fornes, **A. Mathelier**, and W.W. Wasserman. Evaluating the impact of single nucleotide variants on transcription factor binding. *Nucleic Acids Research*, doi: 10.1093/nar/gkw691.
- D.J. Arenillas, A. Forrest, H. Kawaji, T. Lassman, the FANTOM consortium, W.W. Wasserman+, and **A. Mathelier**+. CAGED-oPOSSUM: motif enrichment analysis from CAGE-derived TSSs. *Bioinformatics*, doi: 10.1093/bioinformatics/btw337.
- S. Klein*, L.C. Dieterich*, **A. Mathelier**, C. Chong, A. Sliwa-Primorac, Y.-K. Hong, J.W. Shin, M. Lizio, M. Itoh, H. Kawaji, T. Lassmann, C.O. Daub, E. Arner, the FANTOM consortium, P. Carninci, Y. Hayashizaki, A.R.R. Forrest, W.W. Wasserman, and M. Detmar. DeepCAGE transcriptomics identify HOXD10 as transcription factor regulating lymphatic endothelial responses to VEGF-C. *Journal of Cell Science*, doi:10.1242/jcs.186767.
- R.A.G. De Souza, S.A. Islam, L.M. McEwen, **A. Mathelier**, A. Hill, S.M. Mah, W.W. Wasserman, M.S. Kobor, and B.R. Leavitt. DNA methylation profiling in human Huntington's disease brain. *Human Molecular Genetics*, doi:10.1093/hmg/ddw076.
- A. Mathelier**, O. Fornes, D.J. Arenillas, C.-Y. Chen, G. Denay, J. Lee, W. Shi, C. Shyr, G. Tan, R. Worsley-Hunt, A.W. Zhang, F. Parcy, B. Lenhard, A. Sandelin, and W.W. Wasserman. JASPAR 2016: a major expansion and update of the open-access database of transcription factor binding profiles. *Nucleic Acids Research*, database issue, doi:10.1093/nar/gkv1176.
- 2015 L.C. Dietrich*, S. Klein*, **A. Mathelier**, A. Primorac, Q. Ma, Y.-K. Hong, J.W. Shin, M. Hamada, M. Lizio, M. Itoh, H. Kawaji, T. Lassman, C.O. Daub, E. Arner, P. Carninci, Y. Hayashizaki, the FANTOM5 consortium, A.R.R. Forrest, W.W. Wasserman, and M. Detmar. The transcription factor MAFB links VEGFR-3 signaling with PROX1 expression in lymphatic endothelial cells. *Cell Reports*, doi:10.1016/j.celrep.2015.10.002.
- A. Mathelier**, C. Lefebvre, A.W. Zhang, D.J. Arenillas, J. Ding, W.W. Wasserman, and S.P. Shah. Cis-Regulatory Somatic Mutations and Gene-Expression Alteration in B-cell Lymphomas. *Genome Biology*, doi:10.1186/s13059-015-0648-7.
- A. Mathelier**, W. Shi, and W.W. Wasserman. Identification of altered cis-regulatory elements in human diseases. *Trends in Genetics*, doi:10.1016/j.tig.2014.12.003.
- 2014 A. Saumet, **A. Mathelier**, and C.H. Lecellier. The potential of microRNAs in personalized medicine against cancers. *Biomed Research International*, doi:10.1155/2014/642916.
- R. Worsley-Hunt, **A. Mathelier**, L. Del Peso, and W.W. Wasserman. Improving detection and analysis of transcription factor binding sites within ChIP-seq data based on topological motif enrichment. *BMC Genomics*, doi:10.1186/1471-2164-15-472.
- A.R. Forrest, ..., **A. Mathelier**, ..., Y. Hayashizaki, The FANTOM5 Consortium (~275 authors). A promoter level mammalian expression atlas. *Nature*, doi:10.1038/nature13182.
- L. Yang, T. Zhou, I. Dror, **A. Mathelier**, W.W. Wasserman, R. Gordan, and R. Rohs. TFBSshape: a motif database for DNA shape features of transcription factor binding sites. *Nucleic Acids Research*, database issue,

doi:10.1093/nar/gkt1087.

A. Mathelier*, X. Zhao*, A.W. Zhang, F. Parcy, R. Worsley-Hunt, D.J. Arenillas, S. Buchman, C-Y. Chen, A. Chou, H. Ienasescu, J. Lim, C. Shyr, G. Tan, M. Zhou, B. Lenhardt, A. Sandelin, and W.W. Wasserman. JASPAR 2014: an extensively expanded and updated open-access database of transcription factor binding profiles. *Nucleic Acids Research*, database issue, doi:10.1093/nar/gkt997.

2013 **A. Mathelier** and W.W. Wasserman. The next generation of transcription factor binding site prediction. *PLOS Computational Biology*, doi:10.1371/journal.pcbi.1003214.

A. Mathelier and A. Carbone. Large scale chromosomal mapping of human microRNA structural clusters. *Nucleic Acids Research*, doi:10.1093/nar/gkt112.

2012 J.-F. Schmouth, K.G. Banks, **A. Mathelier**, C.Y. Gregory-Evans, M. Castellarin, R. Holt, K. Gregory-Evans, W.W. Wasserman and E.M. Simpson. Retina restored and brain abnormalities ameliorated by single-copy knock-in of Human NR2E1 in null mice. *Molecular and Cellular Biology*, doi:10.1128/MCB.06016-11.

2010 **A. Mathelier** and A. Carbone. MIRENA: finding microRNAs with high accuracy and no learning at genome scale and from deep sequencing data. *Bioinformatics*, doi:10.1093/bioinformatics/btq329.

A. Mathelier and A. Carbone. Chromosomal periodicity and positional networks of genes in *Escherichia coli*. *Molecular Systems Biology*, doi:10.1038/msb.2010.21.

Collections 2008

A. Carbone and **A. Mathelier**. Environmental and physiological insights from microbial genome sequences. *Elements of Computational Systems Biology*, Huma Lodhi and Stephen Muggleton (eds.), Wiley Book Series in Bioinformatics. Pages 325-339.

Presentations

Conference / Symposium /

Workshop talks

2018 *NuGOweek 2018 - Mitochondria, Nutrition, and Health*, **keynote speaker** (Newcastle, UK).

French Bioinformatics Conference (JOBIM) (Marseille, France).

Anniversary RSAT / GINsim workshop (Paris, France).

2017 *Oslo Epigenetics Mini Symposium* (Oslo, Norway).

2016 *European Conference on Computational Biology*, **highlight talk** (The Hague, The Netherlands).

2015 *Basel Computational Biology Conference*, **highlight talk** (Basel, Switzerland).

Basel Computational Biology Conference, Workshop "Beyond position weight matrices" (Basel, Switzerland).

2012 *FANTOM5 Time-course meeting* (Yokohama, Japan).

European Conference on Computational Biology, Workshop "Detecting transcription factor binding sites with ChIP-seq data and predicting damaging cis-regulatory variations" (Basel, Switzerland).

2011 *Rocky Mountain Bioinformatics Conference* (Aspen, USA).

Invited seminar talks

2018 *Institute for Cancer Research seminars* (Oslo, Norway).

EMBL Heidelberg blue seminars (Heidelberg, Germany).

2017 *Gene Regulation Ensemble Effort for the Knowledge Commons* meeting (Lisbon, Portugal)

VIB - KU Leuven seminars (Leuven, Belgium).

2016 *Department of Cancer Genetics*, Oslo University Hospital (Oslo, Norway).

Furberg CLS seminar series (Oslo, Norway).

2015 *Young Investigator Research Symposium*, Institute for Research in Immunology and Cancer (Montréal, Canada).

Institut des Maladies Génétiques (IMAGINE) (Paris, France).

Institut de Génétique et de Biologie Moléculaire et Cellulaire (IGBMC) (Strasbourg, France).

Centre for Molecular Medicine Norway (Oslo, Norway).

Webinar on JASPAR BioPython module and MANTA database.

2014 *Istituto Toscano Tumori* (Florence, Italy). Webinar.

Institut des Maladies Génétiques (IMAGINE) (Paris, France).

Laboratory Computational and Quantitative Biology (Paris, France).

Institut de Recherche Clinique de Montréal (Montréal, Canada).

2013 *TGIF Seminar Series, Centre for Molecular Medicine and Therapeutics* (Vancouver, Canada).

2011 *TGIF Seminar Series / Centre for Molecular Medicine and Therapeutics* (Vancouver, Canada).

2010 *Laboratoire de Recherche en Informatique / Laboratoire d'Informatique de l'Ecole Polytechnique* (Orsay, France).

Interdisciplinary working group on computation and simulation (Paris, France).

2009 *Workshop "Interactions between bioinformatics and marine biology"* (Paris, France).

Paris 6 Computer Science Ph.D. Students Seminar (Paris, France).

Supervision

Current Jaime Castro-Mondragón (Postdoctoral fellow since 2017)

	Aziz Khan (Postdoctoral fellow since 2016)
	Marius Gheorghe (Ph.D. student since 2016)
2017	Arnaud Stigliani (visiting Ph.D. student, 3 months)
	Kübra Altinel (Master student, 10 months)
2017	Victor Laigle (Master student, 3 months)
2016	Eleftherios Pavlos (Master student, Erasmus plus programme, 3 months)
2011-15	7 Master's rotations (4 months each)

Referee activities

Ph.D. defence and committee	2017	Committee member of the mid-term Ph.D. evaluation of Jonas Meier Stromme (supervisor Rolf Skotheim), Oslo University Hospital (Oslo, Norway).
		Committee member of the mid-term Ph.D. evaluation of Fatima Heinicke (supervisor Benedicte Lie), University of Oslo (Oslo, Norway).
	2016	First opponent for the Ph.D. defence of Rezvan Ehsani (supervisor Finn Drablos), Norwegian University of Science and Technology (Trondheim, Norway).
		First opponent for the Ph.D. defence of Bjarne Johannessen (supervisor Rolf I. Skotheim), Oslo University Hospital (Oslo, Norway).
Recruitment committee	2013	Member of the Canada Research Chairs Tier 2 recruitment committee in Statistical Genetics at the University of British Columbia (Vancouver, Canada).
Journals and conferences		44 reviews for 14 international peer-reviewed journals.

Teaching activities

Ph.D.	<i>Systems medicine approaches in cancer</i> , NCMM Molecular Medicine Research Course (Oslo, Norway).
Master	<i>Statistics in bioinformatics and algorithms on sequences</i> , UPMC (Paris, France).
Bachelor	<i>Recursive programming in Scheme</i> , UPMC (Paris, France).
	<i>Imperative programming and data structures in C</i> , UPMC (Paris, France).
	<i>Discrete structures</i> , UPMC (Paris, France).
	<i>Imperative programming in Visual Basic</i> , UPMC (Paris, France).
	<i>Types and data structures in Caml</i> , UPMC (Paris, France).

Academic activities

Workshop / Seminar Series	organizer	2018	<i>Nordic EMBL Partnership meeting</i> (Oslo, Norway)
		2017-	<i>Sven Furberg Seminars in Bioinformatics and Statistical Genomics</i> (Oslo, Norway).
		2012	<i>European Conference on Computational Biology</i> , Workshop "Detecting transcription factor binding sites with ChIP-seq data and predicting damaging <i>cis</i> -regulatory variations" (Basel, Switzerland)
	Poster judge	2017	<i>European Conference on Computational Biology</i> (Prague, Czech Republic).
		2016	<i>European Conference on Computational Biology</i> (The Hague, The Netherlands).
		2014	<i>Child and Family Research Institute Summer Student Research Program Poster Day</i> (Vancouver, Canada).
			<i>Bioinformatics Training Program, Genome Science and Technology, and the Interdisciplinary Oncology Program Annual Research Day</i> (Vancouver, Canada).
		2012	<i>Medical Genetics Research Day, University of British Columbia</i> (Vancouver, Canada).
		2011	<i>Medical Genetics Research Day, University of British Columbia</i> (Vancouver, Canada).
	Miscellaneous	2013	<i>Summer Student Research Program, Child and Family Research Institute</i> , moderator (Vancouver, Canada).
		2011	<i>High School Tour of the International Gairdner Symposium, Centre for Molecular Medicine and Therapeutics</i> , spokesman (Vancouver, Canada).

Links



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