

Anthony Mathelier, Ph.D.

Group Leader

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Currently

Group Leader, 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**, Dept. of Cancer Genetics, Oslo University Hospital, Oslo, Norway.

Employment

- 2016- **Group Leader**, Centre for Molecular Medicine Norway (NCMM), Oslo, Norway
- 2016- **Adjunct Researcher**, Dept. of Cancer Genetics, Institute for Cancer Research, Oslo University Hospital, Oslo, Norway
- 2013-16 **Deputy Group Leader**, 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**, Wasserman laboratory, CMMT, CFRI, UBC, Vancouver, Canada
- 2006-10 **Teaching assistant**, Université Pierre et Marie Curie (UPMC), Paris, France
- 2006-10 **Ph.D. student**, 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 **Young Research Talents grant from the Research Council of Norway** for the project “Characterisation of cis-regulatory variations that dysregulate driver microRNAs in cancer” with NOK 7.352 millions over 3 years
- Open Call grant from the Norwegian Cancer Society** for the project “Characterisation of cis-regulatory variations that dysregulate driver microRNAs in cancer” with NOK 1.968 millions over 4 years
- Åsgard Research Mobility Program. Funding for spending one week in France developing new scientific interactions (NOK 15.5K)
- UiO:Life Science event support for the Sven Furberg Seminars in Bioinformatics and Statistical Genomics (NOK 20K)
- 2017 NCMM Programme for networking with Associate Investigators and Young Associate Investigators. NOK 480K funding a project on “The molecular mechanism and impact of transition from diploid to aneuploid cells in cancer” in collaboration with Dr. Hege Russnes
- UiO:Life Science event support for the Sven Furberg Seminars in Bioinformatics and Statistical Genomics (NOK 49K)
- 2016 **Start-up package from the NCMM** to launch the Computational Biology & Gene Regulation Group with NOK 18 millions 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 1 million over 3 years
- 2006-09 Ministry of National Education, Research and Technology Ph.D. scholarship attributed by the President of Paris 6 University

Publications

Preprints

A.V. Pladsen, G. Nilsen, O.M. Rueda, M.R. Aure, Ø. Borgan, K. Liestøl, V. Vitelli, A. Frigessi, A. Langerød, OSBREAC, **A. Mathelier**, O. Engebråten, D.C. Wedge, P. Van Loo, C. Caldas, A.-L. Børresen-Dale, H.G. Russnes, O.C. Lingjærde. Copy number motifs expose genome instability type and predict driver events and disease outcome in breast cancer. *bioRxiv*, 2019, doi:10.1101/769356

Peer-reviewed

- 2019 O. Fornes*, J.A. Castro-Mondragon*, A. Khan*, R. van der Lee, X. Zhang, P.A. Richmond, B.P. Modi, S. Correard, M. Gheorghe, D. Baranašić, W. Santana-Garcia, G. Tan, J. Chèneby, B. Ballester, F. Parcy, A. Sandelin+, B. Lehnard+, W.W. Wasserman+, and **A. Mathelier+**. JASPAR 2020: update of the open-access database of transcription factor binding profiles. *Nucleic Acids Research*, 2019, in press.
- S. Salvatore, K.D. Rand, I. Grytten, E. Ferkingstad, D. Domanska, L. Holden, M. Gheorghe, **A. Mathelier**, I. Glad, G.K. Sandve. Beware the Jaccard: the choice of metric is important and non-trivial in genomic colocalisation analysis. *Briefings in Bioinformatics*, 2019, doi:10.1093/bib/bbz083.
- B. Fromm, D. Domanska, E. Høy, V. Ovchinnikov, W. Kang, E. Aparicio-Puerta, M. Johansen, K. Flatmark, **A. Mathelier**, E. Hovig, M. Hackenberg, M.R. Friedlander, K.J. Peterson. MirGeneDB2.0: the curated microRNA Gene Database. *Nucleic Acids Research*, 2019, doi:10.1093/nar/gkz885.

T.J. Ha, P.G.Y. Zhang, R. Robert, J. Yeung, D.J. Swanson, **A. Mathelier**, W.W. Wasserman, S. Im, M. Itoh, H. Kawaji, T. Lassmann, C.O. Daub, E. Arner, The FANTOM Consortium, P. Carninci, Y. Hayashizaki, A.R.R. Forrest, and D. Goldowitz. Identification of novel cerebellar developmental transcriptional regulators with motif activity analysis. *BMC Genomics*, doi:10.1186/s12864-019-6063-9

H.M. Itkonen, A. Urbanucci, S.E.S. Martin, A. Khan, **A. Mathelier**, B. Thiede, S. Walker, I.G. Mills. High OGT activity is essential for MYC-driven proliferation of prostate cancer cells. *Theranostics*, doi:10.7150/thno.30834.

A.J. Korecki, J.W. Hickmott, S.L. Lam, L. Dreolini, **A. Mathelier**, O. Baker, C. Kuehne, R.J. Bonaguro, J. Smith, C.-V. Tan, M. Zhou, D. Goldowitz, J.M. Deussing, A.F. Stewart, W.W. Wasserman, R.A. Holt, and E.M. Simpson. Twenty-Seven Tamoxifen-Inducible iCre-Driver Mouse Strains for Eye and Brain; Including Seventeen Carrying a New Inducible-First Constitutive-Ready Allele. *Genetics*, doi:10.1534/genetics.119.301984.

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. *Nucleic Acids Research*, doi:10.1093/nar/gky1210.

A. Khan, **A. Mathelier**, X. Zhang. Super-enhancers are transcriptionally more active and cell type-specific than stretch enhancers. *Epigenetics*, doi:10.1080/15592294.2018.1514231.

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. Sugjaman-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-Mondragon, 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*, 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*, 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*, 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 (last 5

years)

2019 EMBL conference - Perspectives in Translational Medicine, **invited speaker** (Barcelona, Spain)

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

French Bioinformatics Conference (JOBIM) (Marseille, France)

Anniversary RSAT / GINsim workshop, **invited speaker** (Paris, France)

2017 Oslo Epigenetics Mini Symposium, **invited speaker** (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)

Invited seminar talks

(last 5 years)

2019	Institute for Cancer Research seminars (Oslo, Norway)
2018	Curie Institute (Paris, France) Molecular Genetics Institute of Montpellier (Montpellier, France) 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 Maladie Génétique (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 Génétique (IMAGINE) (Paris, France) Laboratory Computational and Quantitative Biology (Paris, France) Institut de Recherche Clinique de Montréal (Montréal, Canada)

Supervision

Current	Rafael Riudavets Puig (Ph.D. student since 2019) Marius Gheorghe (Ph.D. student since 2016) Roza Berhanu Lemma (Postdoctoral fellow since 2019) Jaime Castro-Mondragon (Postdoctoral fellow since 2017) Aziz Khan (Postdoctoral fellow since 2016) Solveig Klokkerud (Master student since 2019) Peter Arnesen (Master student since 2019)
Previous	Arnaud Stigliani (visiting Ph.D. student, 3 months) Kübra Altinel (Master student, 10 months) Victor Laigle (Master student, 3 months) Eleftherios Pavlos (Master student, Erasmus plus programme, 3 months)

Referee activities

Ph.D. defense and

committee

2019	Opponent and reviewer for the Ph.D. defense of Bram Van de Sande (supervisor Stein Aerts), VIB (Leuven, Belgium).
2018	Opponent and reviewer for the Ph.D. defense of Chloé Bessière (supervisor Charles-Henri Lecellier), Institut de Génétique Moléculaire de Montpellier (Montpellier, France).
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. defense of Rezvan Ehsani (supervisor Finn Drablos), Norwegian University of Science and Technology (Trondheim, Norway). First opponent for the Ph.D. defense of Bjarne Johannessen (supervisor Rolf I. Skotheim), Oslo University Hospital (Oslo, Norway).

Recruitment committee

2013	Member of the Canada Research Chairs Tier 2 / Associate Professor recruitment committee in Statistical Genetics at the University of British Columbia (Vancouver, Canada).
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Journals and conferences

63 reviews for 17 international peer-reviewed journals
Associate Editor for the journal Bioinformatics, Oxford University Press

Teaching activities

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

Academic activities

Conference / Workshop /

Seminar Series organizer	2017-	Sven Furberg Seminars in Bioinformatics and Statistical Genomics (Oslo, Norway).
	2019	ISMB Regulatory and Systems Genomics, ISMB/ECCB 2019 conference (Basel, Switzerland)
	2018	Nordic EMBL Partnership meeting (Oslo, Norway)
	2012	European Conference on Computational Biology, Workshop "Detecting transcription factor binding sites with ChIP-seq data and predicting damaging cis-regulatory variations" (Basel, Switzerland)

Links



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Footer

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