

# Anthony Mathelier, Ph.D.

*Group Leader*

✉ anthony.mathelier@ncmm.uio.no | 📞 +47 228-40-561 | 🐦 @AMathelier

## 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 Medical Genetics, Oslo University Hospital, Oslo, Norway

## Employment

|         |                                                                                                                                                                                                               |
|---------|---------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| 2023-   | <b>Associate Directory</b> , Centre for Molecular Medicine Norway (NCMM), Nordic EMBL Partnership for Molecular Medicine, University of Oslo (UiO), Oslo, Norway                                              |
| 2023-   | <b>Professor II</b> , UiO, Oslo, Norway                                                                                                                                                                       |
| 2020-   | <b>Adjunct Researcher</b> , Dept. of Medical Genetics, Oslo University Hospital, Oslo, Norway                                                                                                                 |
| 2016-   | <b>Group Leader</b> , NCMM, UiO, Oslo, Norway                                                                                                                                                                 |
| 2016-19 | <b>Adjunct Researcher</b> , Dept. of Cancer Genetics, Institute for Cancer Research, Oslo University Hospital, Oslo, Norway                                                                                   |
| 2013-16 | <b>Deputy Group Leader</b> , Wasserman laboratory, Centre for Molecular Medicine and Therapeutics (CMMT), Child and Family Research Institute (CFRI), University of British Columbia (UBC), Vancouver, Canada |
| 2011-13 | <b>Postdoctoral fellow</b> , Wasserman laboratory, CMMT, CFRI, UBC, Vancouver, Canada                                                                                                                         |
| 2006-10 | <b>Teaching assistant</b> , Université Pierre et Marie Curie (UPMC), Paris, France                                                                                                                            |
| 2006-10 | <b>Ph.D. student</b> , Laboratory of Computational and Quantitative Biology, Analytical Genomics Group, UPMC, Paris, France                                                                                   |

## Education

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

## Grants and Awards

|           |                                                                                                                                                                                                                                                     |
|-----------|-----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| 2024-2028 | Open project call from the <b>Norwegian Cancer Society</b> (Pink Ribbon scheme): NOK 7.5 millions                                                                                                                                                   |
| 2024      | UiO:Life Science event support for the Sven Furberg Seminars in Bioinformatics and Statistical Genomics: NOK 20K                                                                                                                                    |
| 2021-2025 | Open call on <b>personalized breast cancer treatments from the Pink Ribbon scheme, Norwegian Cancer Society</b> : NOK 5 millions                                                                                                                    |
| 2021-25   | <b>Renewal package from the NCMM</b> after external evaluation to support the Computational Biology & Gene Regulation Group: NOK 13.5 millions                                                                                                      |
| 2020-2024 | Marie Skłodowska-Curie - University of Oslo Scientia Fellowship awarded to Dr. Vipin Kumar (postdoctoral fellow): NOK 163.8K                                                                                                                        |
| 2019      | <b>Young Research Talents grant from the Research Council of Norway</b> : NOK 7.352 millions                                                                                                                                                        |
| 2019      | <b>Open Call grant from the Norwegian Cancer Society</b> : NOK 1.968 millions                                                                                                                                                                       |
| 2018      | Åsgard Research Mobility Program. Funding for spending one week in France developing new scientific interactions: NOK 15.5K<br><br>UiO:Life Science event support for the Sven Furberg Seminars in Bioinformatics and Statistical Genomics: NOK 20K |
| 2017      | UiO:Life Science event support for the Sven Furberg Seminars in Bioinformatics and Statistical Genomics: NOK 49K                                                                                                                                    |
| 2016      | <b>Start-up package from the NCMM</b> to launch the Computational Biology & Gene Regulation Group: NOK 18 millions                                                                                                                                  |
| 2006      | Ministry of National Education, Research and Technology Ph.D. scholarship attributed by the President of Paris 6 University                                                                                                                         |

## Publications

Senior author 2024

K. Ferenc, I. Rauluseviciute, L. Hovan, V. Kumar, M.L. Kuijjer, **A. Mathelier**. Improving bioinformatics software quality through teamwork. *Bioinformatics*, doi:10.1093/bioinformatics/btae632.

I. Rauluseviciute, T. Launay, G. Barzaghi, S. Nikumbh, B. Lenhard, A.R. Krebs, J.A. Castro-Mondragon, **A. Mathelier**. Identification of transcription factor co-binding patterns with non-negative matrix factorization. *Nucleic Acids Research*, doi:10.1093/nar/gkae743.

2023 I. Rauluseviciute\*, R. Riudavets-Puig\*, R. Blanc-Mathieu, J.A. Castro-Mondragon, K. Ferenc, V. Kumar, R.B. Lemma, J. Lucas, J. Chèneby, D. Baranasic, A. Khan, O. Fornes, S. Gundersen, M. Johansen, E. Hovig, B. Lenhard+, A. Sandelin+, W.W. Wasserman+, F. Parcy+, **A. Mathelier+**. JASPAR 2024: 20th anniversary of the open-access database of transcription factor binding profiles. *Nucleic Acids Research*, doi:10.1093/nar/gkad1059.

2022 J.A. Castro-Mondragon, M. Ragle Aure, O.C. Lingjærde, A. Langerød, J.W.M. Martens, A.-L. Børresen-Dale, V. Kristensen, **A. Mathelier**. Cis-regulatory mutations associate with transcriptional and post-transcriptional deregulation of gene regulatory programs in cancers. *Nucleic Acids Research*, doi:10.1093/nar/gkac1143.

R.B. Lemma, T. Fleischer, E. Martinsen, M. Ledsaak, V. Kristensen, R. Eskeland, O. Stokke Gabrielsen, and **A. Mathelier**. Pioneer transcription factors are associated with the modulation of DNA methylation patterns across cancers. *Epigenetics and Chromatin*, doi:10.1186/s13072-022-00444-9.

2021 J.A. Castro-Mondragon\*, R. Riudavets-Puig\*, I. Rauluseviciute.\*, R. Berhanu Lemma, L. Turchi, R. Blanc-Mathieu, J. Lucas, P. Boddie, A. Khan, N. Manosalva Pérez, O. Fornes, T.Y. Leung, A. Aguirre, F. Hammal, D. Schmelter, D. Baranasic, B. Ballester, A. Sandelin+, B. Lenhard+, K. Vandepoele, W.W. Wasserman+, F. Parcy+, and **A. Mathelier+**. JASPAR 2022: the 9th release of the open-access database of transcription factor binding profiles. *Nucleic Acids Res.*, doi:10.1093/nar/gkab1113.

R. Riudavets Puig, A. Khan, P. Boddie, J.A. Castro-Mondragon, **A. Mathelier**. UniBind: maps of high-confidence direct TF-DNA interactions across nine species. *BMC Genomics*, doi:10.1186/s12864-021-07760-6.

2020 A. Khan, R. Riudavets Puig, P. Boddie, **A. Mathelier**. BiasAway: command-line and web server to generate nucleotide composition-matched DNA background sequences. *Bioinformatics*. doi:10.1093/bioinformatics/btaa928.

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*, doi:10.1093/nar/gkz1001.

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.

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.

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.

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.

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 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.

First author 2016

**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.

**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 **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. 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.

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.

#### Other co-authorships

2024 J. Ankill, Z. Zhao, X. Tekpli, E. H. Kure, V.N. Kristensen, **A. Mathelier**, T. Fleischer. Integrative pan-cancer analysis reveals a common architecture of dysregulated transcriptional networks characterized by loss of enhancer methylation. *PLoS Computational Biology*, in press.

P.-H. Hsieh, R.-X. Hsiao, K. Ferenc, **A. Mathelier**, R. Burkholz, C.-Y. ChShahi, G.K. Sandve, T. Belova, M.L. Kuijjer. CAVACHON: a hierarchical variational autoencoder to integrate multi-modal single-cell data. *arXiv*, doi:10.48550/arXiv.2405.18655.

P. Shahrouzi, F. Forouz, **A. Mathelier**, V.N. Kristensen, P.H.G. Duijf. Copy number alterations: a catastrophic orchestration of the breast cancer genome. *Trends in Molecular Medicine*, doi:10.1016/j.molmed.2024.04.017.

S.M. Lind, M. Sletten, M. Hellenes, **A. Mathelier**, X. Tekpli, M. Tinholt, N. Iversen. Coagulation factor V in breast cancer: A p53 regulated tumor suppressor and predictive marker for treatment response to chemotherapy. *Journal of Thrombosis and Haemostasis*, doi:10.1016/j.jtha.2024.02.008.

2023 O. Fornes, T.V. Av-Shalom, A.J. Korecki, R.A. Farkas, D.J. Arenillas, **A. Mathelier**, E.M. Simpson, W.W. Wasserman. OnTarget: in silico design of MiniPromoters for targeted delivery of expression. *Nucleic Acids Research*, doi:10.1093/nar/gkad375

2022 R. Rossini, V. Kumar, **A. Mathelier**, T. Rognes, and J. Paulsen. MoDLE: High-performance stochastic modeling of DNA loop extrusion interactions. *Genome Biology*, in press; preprint: doi:10.1101/2022.04.13.488157.

S. Bjørklund, M. Ragle Aure, J. Häkkinen, J. Vallon-Christersson, S. Kumar, K. Bull Evensen, T. Fleischer, J. Tost, **A. Mathelier**, G. Bhanot, S. Ganesan, X. Tekpli, V.N. Kristensen. Subtype and cell type specific expression of lncRNAs provide insight into breast cancer. *Communications Biology*, doi:10.1038/s42003-022-03559-7.

J.M. Strømme, B. Johannessen, S.G. Kidd, M. Bogaard, K.T. Carm, X. Zhang, A. Sveen, **A. Mathelier**, R.A. Lothe, U. Axcrone, K. Axcrone, and R.I. Skotheim. Expressed prognostic biomarkers for primary prostate cancer independent of multifocality and transcriptome heterogeneity. *Cancer Gene Therapy*, doi:10.1038/s41417-022-00444-7.

2021 M. Ragle Aure, T. Fleischer, S. Bjørklund, J. Ankill, J.A. Castro-Mondragon, OSBREAC, A.-L. Børresen-Dale, K.K. Sahlberg, **A. Mathelier**, X. Tekpli, V.N. Kristensen. Crosstalk between microRNA expression and DNA methylation drive the hormone-dependent phenotype of breast cancer. *Genome Medicine*, doi:10.1186/s13073-021-00880-4.

E. Mileti, K.H.M. Kwok, D.P. Andersson, **A. Mathelier**, A. Raman, J. Bäckdahl, J. Jalkanen, L. Massier, A. Thorell, H. Gao, P. Arner, N. Mejhert, C.O. Daub, M. Rydén. Human white adipose tissue displays selective insulin resistance in the obese state. *Diabetes*, doi:10.2337/db21-0001.

A.J. Korecki, J.L. Cueva-Vargas, O. Fornes, J. Agostinone, R.A. Farkas, J.W. Hickmott, S.L. Lam, **A. Mathelier**, M. Zhou, W.W. Wasserman, A. Di Polo, E.M. Simpson. Human MiniPromoters for ocular-rAAV expression in ON bipolar, cone, corneal, endothelial, Müller glial, and PAX6 cells. *Gene Therapy*, doi:10.1038/s41434-021-00227-z.

- 2020 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. *Communications Biology*, doi:10.1038/s42003-020-0884-6.
- B. Fromm, D. Domanska, E. Hoye, 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*, doi:10.1093/nar/gkz885.
- 2019 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.
- 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 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.
- 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.
- D. Sugiaman-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 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.
- 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.
- 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.
- 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.

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.

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.

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.

## 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.

## Speaker at Conferences / Symposiums / Workshops / Seminars (last 5 years)

2024 ELIXIR Norway All hands meeting, **invited speaker** (Trondheim, Norway)  
National Breast Cancer Network meeting, **invited speaker** (Stavanger, Norway)

2023 CARE symposium, Centre de Recherches en Cancérologie de Toulouse, **invited speaker** (Toulouse, France)

2022 Cancer Genomics and Epitranscriptomics: from the bench to the clinic, **speaker** selected from abstracts (Salamanca, Spain)

2021 Nanotumor Bioinformatics webinar series, **invited speaker** (Virtual)  
Centre for Integrative Genetics seminar, **invited speaker** (Ås, Norway; Virtual)  
Symposium "Where are we in the search for DNA motifs involved in transcriptional regulation?" at the JOBIM conference, **invited speaker** (Virtual)  
Workshop "Genomic Regulation: Focus on Cancer", **invited speaker** (Virtual)

2019 EMBL conference - Perspectives in Translational Medicine, **invited speaker** (Barcelona, Spain)  
Institute for Cancer Research seminar, **invited speaker** (Oslo, Norway)

## Supervision

Supervised **4 PhD students** (2 completed), **7 postdoctoral/research fellows**

## Academic activities

Conference / Workshop /

Seminar organizer

2018-now Co-chair, Regulatory and Systems Genomics (RegSys) track at the ISMB and ISMB/ECCB conferences.

2017-now Co-chair, Sven Furberg Seminars in Bioinformatics and Statistical Genomics (Oslo, Norway).

2022 Norwegian Bioinformatics Days conference (Oslo, Norway)  
ISMB Regulatory and Systems Genomics, ISMB 2022 conference (Madison, USA)



|      |                                                                                                                                                                                                 |
|------|-------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
|      | EMBL partnership meeting (Heidelberg, Germany)                                                                                                                                                  |
| 2021 | ISMB Regulatory and Systems Genomics, ISMB/ECCB 2021 conference (virtual).                                                                                                                      |
| 2020 | ISMB Regulatory and Systems Genomics, ISMB 2020 conference (virtual).                                                                                                                           |
| 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) |

## Referee activities

|                             |                                                                                                                                                                              |  |
|-----------------------------|------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|--|
| Ph.D. defense and committee |                                                                                                                                                                              |  |
|                             |                                                                                                                                                                              |  |
| 2025                        | Opponent for the Ph.D. defense of Endre Bakken Stovner (supervisor Pal Saetrom), Norwegian University of Science and Technology (Trondheim, Norway)                          |  |
| 2024                        | Opponent for the Ph.D. defense of Yuna Landais (supervisor Céline Vallot), Curie Institute (Paris, France).                                                                  |  |
|                             | Opponent for the Ph.D. defense of Delphine Balaramane (supervisor Anaïs Bardet), Institute of Genetics, Molecular, and Cellular Biology (Strasbourg, France).                |  |
| 2022                        | Opponent for the Ph.D. defense of Claudia Arnedo (supervisor Nuria Lopez-Bigas), Institute for Research in Biomedicine (Barcelona, Spain).                                   |  |
|                             | Member of the Ph.D. evaluation committee of Weikaixin Kong (supervisor Tero Aittokallio), University of Helsinki (Helsinki, Finland).                                        |  |
|                             | Opponent for the Ph.D. defense of Adnan Niazi (supervisor Eivind Valen), University of Bergen (Bergen, Norway).                                                              |  |
| 2021                        | Opponent for the Ph.D. defense of Raphael Romero (supervisor Charles-Henri Lecellier), Institut de Génétique Moléculaire de Montpellier (Montpellier, France).               |  |
|                             | Opponent for the Ph.D. defense of Ina Eilertsen (supervisors Ragnhild Lothe and Anita Sveen), Institute for Cancer Research, Oslo University Hospital (Oslo, Norway).        |  |
|                             | Opponent for the Master thesis defense of Lloyd Andreassen (supervisor Anagha Joshi-Michael), University of Bergen (Bergen, Norway).                                         |  |
| 2020                        | Opponent for the Ph.D. defense of Axel Thieffry (supervisor Albin Sandelin), Bioinformatics Centre, Copenhagen University (Copenhagen, Denmark).                             |  |
|                             | Committee member of the mid-term Ph.D. evaluation of Ping-Han Hsieh (supervisor Marieke Kuijjer), Centre for Molecular Medicine Norway, University of Oslo (Oslo, Norway).   |  |
| 2019                        | Opponent for the Ph.D. defense of Bram Van de Sande (supervisor Stein Aerts), VIB (Leuven, Belgium).                                                                         |  |
| 2018                        | Opponent 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), Institute for Cancer Research, 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                        | Opponent for the Ph.D. defense of Rezvan Ehsani (supervisor Finn Drablos), Norwegian University of Science and Technology (Trondheim, Norway).                               |  |
|                             | Opponent for the Ph.D. defense of Bjarne Johannessen (supervisor Rolf I. Skotheim), Institute for Cancer Research, Oslo University Hospital (Oslo, Norway).                  |  |

|                       |       |                                                                                                                                                                            |
|-----------------------|-------|----------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| Recruitment committee | 2024- | Member of the Data Driven Life Science Advisory Committee (Sweden)                                                                                                         |
|                       | 2019  | Member of the selection committee for the recruitment of a Group Leader at NCMM                                                                                            |
|                       | 2018  | Member of the selection committee for the recruitment of a Group Leader at NCMM                                                                                            |
|                       | 2013  | Member of the Canada Research Chairs Tier 2 / Associate Professor recruitment committee in Statistical Genetics at the University of British Columbia (Vancouver, Canada). |

## Journals and conferences

Associate Editor for the journal *Bioinformatics*, Oxford University Press since 2019

Member of reviewing committee for [ISMB conference \(2020, 2022\)](#), [ISMB/ECCB conference \(2019, 2021\)](#), [RECOMB conference on Regulatory & Systems Genomics \(2020\)](#), [Journées Ouvertes Biologie, Informatique et Mathématique \(2020\)](#), [ISMB Regulatory and Systems Genomics \(2019, 2020\)](#)

## Teaching

|          |                                                                                                                                                                                                                                                                                                                        |
|----------|------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| Ph.D.    | Multi-omics data analysis and integration for precision medicine, international course, Faculty of Medicine, UiO (Oslo, Norway) - 2022<br><br>Systems medicine approaches in cancer, national NCMM Molecular Medicine Research Course, UiO (Oslo, Norway) - since 2016                                                 |
| Master   | Statistics in bioinformatics and algorithms on sequences, UPMC (Paris, France)                                                                                                                                                                                                                                         |
| Bachelor | Recursive programming in Scheme, UPMC (Paris, France)<br><br>Imperative programming and data structures in C, UPMC (Paris, France)<br><br>Discrete structures, UPMC (Paris, France)<br><br>Imperative programming in Visual Basic, UPMC (Paris, France)<br><br>Types and data structures in Caml, UPMC (Paris, France) |

## Links



anthony.mathelier@ncmm.uio.no

@AMathelier



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## Footer

Last updated: Oct. 24th 2024