

# JOELLE MBATCHOU PH.D.

- Proficient and innovative researcher with 9 years of research experience in developing cutting-edge methods to help improve understanding of genetic variation and its relationship to human health and disease
- Avid and charismatic learner who continuously strives for excellence by always meeting deadlines, providing timely solutions, and staying attentive to new developments in the field
- Eager to assist biotech companies in discovering novel targets and indications by building scalable and efficient association analysis tools and pipelines
- Developed a C++ tool which decreased computation costs at the Regeneron Genetics Center by over 50X
- Top Skills: Analytical, Highly Proficient in C++ and R, Pipeline development with WDL & docker, Project Management



## EDUCATION

2019	● <b>PhD., Statistics</b> University of Chicago	📍 Chicago, IL
2011	● <b>BSc Biology &amp; Mathematical Sciences</b> DePaul University	📍 Chicago, IL

## INDUSTRY EXPERIENCE

Current   2024	● <b>Senior Manager, Statistical Genetics</b> Regeneron Genetics Center	📍 Tarrytown, New York
2023   2022	● <b>Manager, Statistical Genetics</b> Regeneron Genetics Center	📍 Tarrytown, New York

• Leading and mentoring a team member in the development of an analytical pipeline for time-to-event data.

• Routinely carry out statistical analyses using cloud-based computing platforms on large-scale and high-dimensional human genetics datasets containing millions of genetic variants and 100,000s of individuals

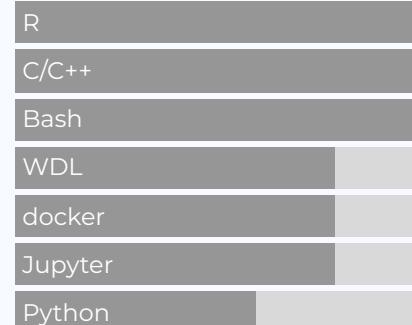
• Develop statistical methods and computational tools geared for large-scale genetic and genomics studies

• Build WDL pipelines for data sets with 100,000s of individuals from whole-exome and whole-genome sequencing data

## CONTACT

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👤 US Citizen  
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🔗 [github.com/joellesophya](https://github.com/joellesophya)  
🔗 [joellesophya.github.io](https://joellesophya.github.io)

## TECHNICAL SKILLS



2021  
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2019

### ● Senior Statistical Geneticist

Regeneron Genetics Center

📍 Tarrytown, New York

- Developed a computational efficient whole genome regression method REGENIE for large-scale genetic association analyses which can be more than 100x faster than current state-of-the-art methods and can handle population structure and imbalanced binary traits
- Implemented REGENIE into a C++ software which was publicly released on Github
- Published the REGENIE method as first author in Nature Genetics where it was applied to UK Biobank data (>100 phenotypes, >400K individuals and >10M genetic variants).

## 💻 RESEARCH EXPERIENCE

2019  
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2013

### ● Graduate Student Researcher

Department of Statistics

📍 University of Chicago

- Developed a computationally fast method JASPER to assess significance for a general class of association tests, including tests for high dimensional phenotypes and gene-based tests, adjusting for population structure and family relatedness
- Designed a permutation-based testing procedure BRASS for assessing significance with binary traits in structured samples for association tests with unknown exact/asymptotic distributions.
- Built C/C++ software to evaluate JASPER and BRASS through simulation studies & real data applications.

## 💡 TEACHING EXPERIENCE

2023  
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2022

### ● Instructor

Summer Institute in Statistical Genetics

📍 University of Washington

- Taught the association mapping module on genome-wide association studies and sequencing (120 students)
- Designed coursework as well as hands-on practical exercises using software such as PLINK, REGENIE and R packages GWASTools and bigsnpr.
- Built a website to host the course materials using workflowr R package.

2019  
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2012

### ● Teaching Assistant

Department of Statistics

📍 University of Chicago

- Assisted in undergraduate courses: Statistical Methods and Applications, Statistical Models/Methods, Applied Regression Analysis and Analysis of Categorical Data
- Created introductory material for R and STATA through weekly computer sessions
- Organized weekly office hours where students expanded their understanding of course materials

2018	<ul style="list-style-type: none"> <li>● <b>Course Instructor</b> Department of Statistics</li> </ul>	 University of Chicago
	<ul style="list-style-type: none"> <li>• Taught an introductory course on statistical methods (STAT 234: Statistical Models/Methods).</li> <li>• Designed course materials and delivered bi-weekly lectures to a class of 36 students.</li> <li>• Assisted students in office hours with learning statistical concepts as well as R programming skills.</li> </ul>	
2017   2016	<ul style="list-style-type: none"> <li>● <b>Statistics Collaborative Learning Team Leader</b> Department of Statistics</li> </ul>	 University of Chicago
	<ul style="list-style-type: none"> <li>• Facilitated the design of a new program based on collaborative learning.</li> <li>• Designed practical exercises based on class mateirals and supervised weekly hands-on workshops for students.</li> </ul>	

## 🏆 AWARDS

2022	<ul style="list-style-type: none"> <li>● <b>Selected as one of 35 innovators under 35<sup>1</sup></b> MIT Technology Review</li> </ul>	
2022	<ul style="list-style-type: none"> <li>● <b>Selected as one of 17 Rising Stars in Health Tech<sup>2</sup></b> Fierce Health Care</li> </ul>	
2021	<ul style="list-style-type: none"> <li>● <b>Selected for Reviewers' Choice (top 10% scoring abstracts)</b> American Society of Human Genetics</li> </ul>	
2013	<ul style="list-style-type: none"> <li>● <b>Department of Education GAANN Fellowship Recipient</b> University of Chicago</li> </ul>	
2010	<ul style="list-style-type: none"> <li>● <b>Departmental Award for Outstanding Performance in Organic Chemistry</b> DePaul University</li> </ul>	

## ☰ PUBLICATIONS

2023	<ul style="list-style-type: none"> <li>● <b>Genotyping, sequencing and analysis of 140,000 adults from Mexico City<sup>3</sup></b> Nature           <ul style="list-style-type: none"> <li>• Authors: Ziyatdinov, A., Torres, J., Alegre-Díaz, J., Backman, J., <b>Mbatchou, J.</b>, Turner, M., et al.</li> </ul> </li> </ul>	
2023	<ul style="list-style-type: none"> <li>● <b>BRASS: Permutation methods for binary traits in genetic association studies with structured samples<sup>4</sup></b> PLoS Genetics           <ul style="list-style-type: none"> <li>• Authors: <b>Mbatchou, J.</b>, Abney, M., &amp; McPeek, M. S.</li> </ul> </li> </ul>	

- 2023
- **Rare coding variants in CHRNB2 reduce the likelihood of smoking.<sup>5</sup>**  
Nature Genetics
    - Authors: Rajagopal, V. M., Watanabe, K., **Mbatchou, J.**, Ayer, A., Quon, P., et al.
- 2022
- **Common and rare variant associations with clonal haematopoiesis phenotypes.<sup>6</sup>**  
Nature
    - Authors: Kessler, M. D., Damask, A., O'Keeffe, S., Banerjee, N., Li, D., Watanabe, K., Marcketta, A., Van Meter, M., Semrau, S., Horowitz, J., Tang, J., Kosmicki, J. A., Rajagopal, V. M., Zou, Y., Houvras, Y., Ghosh, A., Gillies, C., **Mbatchou, J.**, et al.
  - **Genome-wide analysis provides genetic evidence that ACE2 influences COVID-19 risk and yields risk scores associated with severe disease.<sup>7</sup>**  
Nature Genetics
    - Authors: Horowitz, J. E., Kosmicki, J. A., Damask, A., Sharma, D., Roberts, G. H. L., Justice, A. E., Banerjee, N., Coignet, M. V., Yadav, A., Leader, J. B., Marcketta, A., Park, D. S., Lanche, R., Maxwell, E., Knight, S. C., Bai, X., Guturu, H., Sun, D., Baltzell, A., Kury, F. S. P., Backman, J. D., Girshick, A. R., O'Dushlaine, C., McCurdy, S. R., Partha, R., Mansfield, A. J., Turissini, D. A., Li, A. H., Zhang, M., **Mbatchou, J.**, et al.
- 2022
- **Germline Mutations in CIDEB and Protection against Liver Disease.<sup>8</sup>**  
New England Journal of Medicine
    - Authors: Verweij, N., Haas, M. E., Nielsen, J. B., Sosina, O. A., Kim, M., Akbari, P., De, T., Hindy, G., Bovijn, J., Persaud, T., Miloscio, L., Germino, M., Panagis, L., Watanabe, K., **Mbatchou, J.**, et al.
- 2021
- **Sequencing of 640,000 exomes identifies GPR75 variants associated with protection from obesity.<sup>9</sup>**  
Science
    - Authors: Akbari, P., Gilani, A., Sosina, O., Kosmicki, J. A., Khrimian, L., Fang, Y. Y., Persaud, T., Garcia, V., Sun, D., Li, A., **Mbatchou, J.**, et al.
  - **Exome sequencing and analysis of 454,787 UK Biobank participants.<sup>10</sup>**  
Nature
    - Authors: Backman, J. D., Li, A. H., Marcketta, A., Sun, D., **Mbatchou, J.**, et al.
- 2021
- **Pan-ancestry exome-wide association analyses of COVID-19 outcomes in 586,157 individuals.<sup>11</sup>**  
American Journal of Human Genetics
    - Authors: Kosmicki, J. A., Horowitz, J. E., Banerjee, N., Lanche, R., Marcketta, A., Maxwell, E., Bai, X., Sun, D., Backman, J. D., Sharma, D., Kury, F. S. P., Kang, H. M., O'Dushlaine, C., Yadav, A., Mansfield, A. J., Li, A. H., Watanabe, K., Gurski, L., McCarthy, S. E., Locke, A. E., Khalid, S., O'Keeffe, S., **Mbatchou, J.**, et al.

- 2021
- **Computationally efficient whole-genome regression for quantitative and binary traits.<sup>12</sup>**  
Nature Genetics
    - Authors: Mbatchou, J., Barnard, L., Backman, J., Marcketta, A., Kosmicki, J. A., Ziyatdinov, et al.
- 2015
- **Retrospective Association Analysis of Binary Traits: Overcoming Some Limitations of the Additive Polygenic Model.<sup>13</sup>**  
Human Heredity
    - Authors: Jiang, D., Mbatchou, J. & McPeek, M. S.
- 2011
- **New insights into relationships of lichen-forming Dothideomycetes.**  
Fungal Diversity
    - Authors: Nelsen, M. P., Lücking, R., Mbatchou, J. S., Andrew, C. J., Spielmann, A. A. & Lumbsch, H. T.
- 2010
- **Heiomasia, a new genus in the lichen-forming family Graphidaceae (Ascomycota: Lecanoromycetes: Ostropales) with disjunct distribution in Southeastern North America and Southeast Asia.**  
The Bryologist
    - Authors: Nelsen, M. P., Lücking, R., Plata, E. R. & Mbatchou, J. S.
- 2009
- **A class-wide phylogenetic assessment of Dothideomycetes.**  
Studies in Mycology
    - Authors: Schoch, C. L., Crous, P. W., Groenewald, J. Z., Boehm, E. W., Burgess, T. I., Gruyter, J. de, Hoog, G. S. de, Dixon, L. J., Grube, M., Gueidan, C., Harada, Y., Hatakeyama, S., Hirayama, K., Hosoya, T., Huhndorf, S. M., Hyde, K. D., Jones, E. B., Kohlmeyer, J., Kruys, A., Li, Y. M., Lucking, R., Lumbsch, H. T., Marvanova, L., Mbatchou, J. S., et al.

## PROFESSIONAL ACTIVITIES

- **Manuscript reviewer**  
Nature Genetics, Genetic Epidemiology, Bioinformatics.

## ORAL PRESENTATIONS AND POSTERS

- 2023
- **Using protein language model annotations to improve the power of exome-wide association studies**  
The American Society of Human Genetics Annual Meeting
    - Poster presentation
- 2023
- **Powerful gene-based testing for exome-wide association studies**  
University of Washington Statistical Genetics Symposium
    - Invited Talk

- 2022
- **REGENIE v3: more efficient analysis of rare genetic variation with an extended set of gene-based tests**  
The American Society of Human Genetics Annual Meeting
    - Poster presentation
- 2022
- **Computationally efficient whole-genome regression framework for association analyses in large-scale biobanks.**  
International Common Disease Alliance
    - Virtual Scientific Plenary
- 2021
- **Gene-burden tests, gene-environment interactions and time-to-event data analysis within an efficient whole genome regression framework for large-scale biobanks.**  
The American Society of Human Genetics Annual Meeting
    - Poster presentation
- 2021
- **Fast and robust methods to detect gene-environment interactions in large-scale biobanks.**  
International Genetic Epidemiology Society Conference
    - Lightning talk
- 2020
- **Computationally efficient whole-genome regression for quantitative and binary traits.**  
The American Society of Human Genetics Annual Meeting
    - Platform talk
- 2017
- **A generalized permutation testing method for binary trait association in structured samples.**  
The American Society of Human Genetics Annual Meeting
    - Poster presentation
- 2017
- **Genetic association analysis of binary traits in structured samples.**  
Eastern North American Region Spring Meeting
    - Poster presentation

## 🔗 LINKS

- 1: <https://www.technologyreview.com/innovator/joelle-mbatchou/>
- 2: <https://www.fiercehealthcare.com/health-tech/rising-stars-health-tech>
- 3: <https://doi.org/10.1038/s41586-023-06595-3>
- 4: <https://doi.org/10.1371/journal.pgen.1011020>
- 5: <https://doi.org/10.1038/s41588-023-01417-8>
- 6: <https://doi.org/10.1038/s41586-022-05448-9>
- 7: <https://doi.org/10.1038/s41588-021-01006-7>
- 8: <https://doi.org/10.1056/NEJMoa2117872>

- 9: <https://doi.org/10.1126/science.abf8683>
- 10: <https://doi.org/10.1038/s41586-021-04103-z>
- 11: <https://doi.org/10.1016/j.ajhg.2021.05.017>
- 12: <https://doi.org/10.1038/s41588-021-00870-7>
- 13: <https://doi.org/10.1159/000446957>