Approved by the Regents
May 16, 2019

PROMOTION RECOMMENDATION
THE UNIVERSITY OF MICHIGAN
MEDICAL SCHOOL
DEPARTMENT OF HUMAN GENETICS

Stephanie Bielas, Ph.D., assistant professor of human genetics, Department of Human Genetics, Medical School, is recommended for promotion to associate professor of human genetics, with tenure, Department of Human Genetics, Medical School.

Academic Degrees:

Ph.D.	2007	University of California, San Diego
B.A.	1998	Andrews University
B.S.	1997	Andrews University

Professional Record:

2018–present	Adjunct Professor, Manipal University, Manipal, India
2012–present	Assistant Professor of Human Genetics, University of Michigan

Summary of Evaluation:

Teaching: Dr. Bielas' teaching occurs primarily in the laboratory setting. She is a mentor to graduate students, postdoctoral trainees, andundergraduate and masters' students. She has served on six Ph.D. thesis committees for students outside her lab; three have successfully defended their Ph.D.s. Dr. Bielas is an active member of the Cell and Molecular Biology Training Program, the Michigan Pre-doctoral Training Program in Genetics, and the Neuroscience Training Program. She has been a strong and influential contributor to classroom instruction as is evidenced by development and instruction of a new class, Human Genetics 480/580, Neurobiology of Developmental Disorders. Dr. Bielas is the co-director of the Cell and Molecular Biology Student Seminar and is involved in global outreach of teaching and training in human genetics. She began a collaboration in India to establish an inaugural genetic counseling training program at Manipal University. At that institution, Dr. Bielas co-developed and teaches courses that define the clinical indications for new medical genomic testing options, variant annotation, test result interpretation, and delivery of test results to families.

Research: Dr. Bielas has established national and international recognition through study of the genetic diagnosis of human neuro-developmental disorders, including microcephaly, CHARGE syndrome, Bainbridge-Ropers syndrome and other autism spectrum disorders. Her research follows the identification of candidate genetic causes with detailed examination of the mechanisms through which the mutations impact cortical development. Her studies involve large patient cohorts, whole genome sequencing efforts, and complicated analytic methods. Dr. Bielas has been consistently well-funded for her research; currently she is the principal investigator of two NIH R01 grants, two foundation grants and an institutional grant, as well as a co-investigator of an NIH R01 grant. She is the recipient of a Directors' Award from the Simons Foundation for Autism. At the University of Michigan, Dr. Bielas received the Elizabeth Caroline Crosby Award to address global disparities in genetic diagnosis. She has published 32 peer-reviewed articles, and has presented her research, by invitation, on 19 occasions regionally, nationally and internationally.

Recent and Significant Publications:

Srivastava A, Srivastava KR, Hebbar M, Galada C, Kadavigere R, Su F, Cao X, Chinnaiyan AM, Girisha KM, Shukla A, Bielas SL: Genetic diversity of NDUFV1 dependent Mitochondrial complex I deficiency. *Eur J Hum Genet.* Nov 26(11):1582-1587, 2018.

Moccia A, Srivastava A, Skidmore JM, Bernat JA, Wheeler M, University of Washington Center for Mendelian Genomics, Chong JX, Nickerson D, Bamshad M, Hefner MA, Martin DM, Bielas SL: Genetic analysis of CHARGE syndrome identifies overlapping molecular Biology. *Genet Med.* Sep 20(9): 1022-1029, 2018.

KC R, Srivastava A, Wilkowski JM, Burke DT, Bielas SL: Detection of nucleotide-specific CRISPR/Cas9 modified alleles using multiplex ligation-detection. *Sci Rep.* 25;6:32048, 2016.

Harding B, Moccia A, Soukarieh O, Drunat S, Chitty L, Verloes A, Gressens P, El Ghouzzi V, Joriot S, Passemard S, Martins A, Di Cunto F, Bielas SL: Mutations in Citron-Kinase cause recessive micro-lissencephaly with multinucleated neurons. *Am J Hum Genet.* 4;99(2):511-20, 2016.

Srivastava A, KC R, Tsan YC, Liao R, Su F, Cao X, Hannibal MC, Keegan CE, Chinnaiyan AM, Martin DM, Bielas SL: De Novo dominant ASXL3 mutations alter H2A deubiquitination and transcription in Bainbridge-Ropers syndrome. *Hum. Mol. Genet.* 25(3):597-608, 2015.

Service: Dr. Bielas is the chair of the Scientific Strategy Committee in the Department of Human Genetics. In that capacity, she is the leader for a committee of five faculty members and one staff support that address department-wide initiatives and issues in research and works on team science initiatives, such as developing "U" and "P" program grant proposals to the NIH, the University Biosciences Initiative, and the Precision Health Initiative. Dr. Bielas serves on the scientific board of directors for Leo's Lighthouse and the ASXL Related Research Endowment. These are private foundations organized by affected families to support clinical and translational research on Bainbridge-Ropers Syndrome and ASXL disorders. Dr. Bielas has served as an invited manuscript reviewer for 19 scientific journals and as an ad hoc grant reviewer. Her grant reviews have included both national and international funding agencies, including the NIH, as an ad hoc panel member and a Special Emphasis review panel on the neurological developmental challenges of Zika virus, the Action Medical Research Foundation, and the Sir Henry Dale Fellowships offered by the United Kingdom Royal Society and the Wellcome Trust. She is a member of the American Society of Human Genetics, the American Neuroscience Society, the International Society of Stem Cell Research, and the American Society of Cell Biology.

External Reviewers:

Reviewer A: "Stephanie Bielas has made important research contributions to understanding the genetic and molecular basis of human neurodevelopmental disorders. She has also shown a strong commitment to the Medical School community and to solving deeper problems in improving genetic diagnostics in disadvantaged settings."

Reviewer B: "She has a productive research program that dedicated to the discovery and understanding of the genetic basis of neurodevelopmental disorders. She has funding for her work, …has a good

publication record and trajectory, and has shown dedication to her field through service to her local, national and international community and teaching genetics at many levels."

Reviewer C: "Her papers reflect considerable creativity, reporting genetic and molecular developmental causes of neurodevelopmental disorders accompanied by considerable methods development. She is becoming known for her careful work in cellular models of human disease and her focus on chromatin modification in neurodevelopmental disorders. The quality and quantity of her work is excellent and highly appropriate for someone in her field and at her career stage."

Reviewer D: "...I find Dr. Bielas to be an extremely well-rounded candidate for promotion with substantial contributions in teaching, independent scientific research, service within and outside the University of Michigan, and in basic clinical research...Dr. Bielas has made important contributions to the field of human genetics."

Reviewer E: "...my comments are based on my knowledge of the field, review of her CV, and knowledge of her published work. On that basis, Stephanie fully deserves immediate promotion to the rank of the Associate Professor with Tenure (Instructional Track), and would likely be promoted here at [my institution]...Stephanie is already among those poised to lead the field in the coming years and I endorse her nomination enthusiastically."

Summary of Recommendation:

Dr. Bielas is a nationally and internationally recognized scholar and scientist studying human neurological developmental disorders and the control of neurological development. Since joining the University of Michigan, she has built a strong and sustainable research program and has obtained consistent and significant funding to support her work. She has significant service contributions, is well-funded and has established an international partnership in India. I am pleased to recommend Stephanie Bielas, Ph.D. for promotion to associate professor of human genetics, with tenure, Department of human Genetics, Medical School.

Marschall S. Runge, M.D., Ph.D.
Executive Vice President for Medical Affairs
Dean, Medical School

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