ACMG Foundation for Genetic and Genomic Medicine
Better Health Through Genetics™

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Nara Sobreira, MD, PhD is the Recipient of the 2023 Dr. Michael S. Watson Genetic and Genomic Medicine Innovation Award from the ACMG Foundation for Genetic and Genomic Medicine

BETHESDA, MD – March 15, 2023| Nara Lygia de Macena Sobreira, MD, PhD is the recipient of the ACMG Foundation for Genetic and Genomic Medicine's **2023 Dr. Michael S. Watson Genetic and Genomic Medicine Innovation Award**—the "Watson Award"—named for the American College of Medical Genetics and Genomics first and longstanding executive director, Michael S. Watson, MS, PhD, FACMG.

"I am honored and grateful for receiving the 2023 Dr. Michael S. Watson Genetic and Genomic Medicine Innovation Award. Since the beginning of my medical genetics residency in Brazil, I have been blessed to work with amazing mentors and colleagues in innovative projects that benefited many patients around the world. It has been a privilege to study and practice genetics in the last 20 years and witness and benefit from the rapid development of genomics. I hope that genetics and genomics continue to become more accessible to students, clinicians, researchers and patients around the world and that my work in clinic and research can help with that process," said Dr. Sobreira.

"Dr. Sobreira was the chief architect of GeneMatcher, a system with which anyone involved in genome sequencing for diagnosis of rare diseases will be familiar. It has been a game-changer in rare disease diagnosis that has helped clinicians, scientists, and patients around the world," said Bruce R. Korf, MD, PhD, FACMG, president of the ACMG Foundation.

Dr. Sobreira is currently Associate Professor in the Departments of Genetic Medicine and Pediatrics at Johns Hopkins University. Dr. Sobreira was the chief analyst for the Baylor-Hopkins Center for Mendelian Genomics and was instrumental in the development of tools that have transformed clinical and research genetics. She developed the analysis module of PhenoDB, a tool for the collection, storage and analysis of phenotypic and genotypic information geared to the identification of novel disease genes. PhenoDB is the platform used for analysis by the BHCMG but has also been downloaded and installed in many academic and clinical centers to be used for genome analysis. It is also used in the Gabriella Miller Kids First Initiative.

Dr. Sobreira graduated from the University of Pernambuco School of Medicine, then completed a Clinical Genetics Residency at Paulista School of Medicine (UNIFESP). In 2007, she started her PhD in Human Genetics followed by a one-year postdoc and a two-year

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Clinical Genetics Fellowship at Johns Hopkins School of Medicine. Dr. Sobreira and colleagues recognized that many excellent candidate genes were only present in one family, they then developed GeneMatcher, the most widely used resource to facilitate case matching in rare disease. Dr. Sobreira next developed VariantMatcher, a tool for investigating genotype-phenotype relationships in large cohorts of patients with rare diseases. Dr. Sobreira's efforts are currently focused on developing a federated one-sided matching system to expand the reach beyond VariantMatcher. This is a coordinated effort with the Global Alliance for Genomics and Health.

Dr. Sobreira is also passionate about educating and spreading the utility of genome sequencing across health care and the world. In addition to developing tools that facilitate genome analysis and discovery, Dr. Sobreira has developed both in-person and virtual educational programs that teach genome analysis using PhenoDB. These courses are taught at Johns Hopkins through a week-long intensive course open to third- and fourth-year medical students, as well as a required rotation for genetics residents and fellows. She also teaches this to students at UNIFESP-Brazil and at the McKusick Short Course in Human and Mammalian Genetics and Genomics taught at the Jackson Laboratory in Bar Harbor, Maine every year. She is Board Certified by the Brazilian Board of Medical Genetics, the American Board of Medical Genetics and Genomics. She is also a member of the American Society of Human Genetics. She has taught genomic medicine courses to students around the world.

Dr. Sobreira has contributed to 102 peer-reviewed publications, 18 as first or last author. She is particularly interested in Ollier disease and Maffucci syndrome and serves on the medical advisory board of the patient organization. She is performing clinical studies and translational research to understand the pathogenesis and potential treatments for these rare disorders.

"Dr. Sobreira has demonstrated remarkable creativity in advancing medical genomics through innovations in informatic tools over a span of many years. Her work in leading the development of the GeneMatcher tool has been instrumental in fostering collaborations between investigators around the world toward the identification of countless genes that are causal to numerous genetic disorders, and which has borne tremendous fruit in the lives of the families and communities we serve," said Robert Best, PhD, MS, FACMG, interim CEO of the ACMG. "

About the ACMG Foundation for Genetic and Genomic Medicine

The ACMG Foundation for Genetic and Genomic Medicine, a 501(c)(3) nonprofit organization, is a community of supporters and contributors who understand the importance of medical genetics and genomics in healthcare. Established in 1992, the ACMG Foundation supports the American College of Medical Genetics and Genomics (ACMG) mission to "translate genes into health." Through its work, the ACMG Foundation fosters charitable giving, promotes training opportunities to attract future medical geneticists and genetic counselors to the field, shares information about medical genetics and genomics, and sponsors important research. To learn more and support the ACMG Foundation mission to create "Better Health through Genetics" visit www.acmgfoundation.org.

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