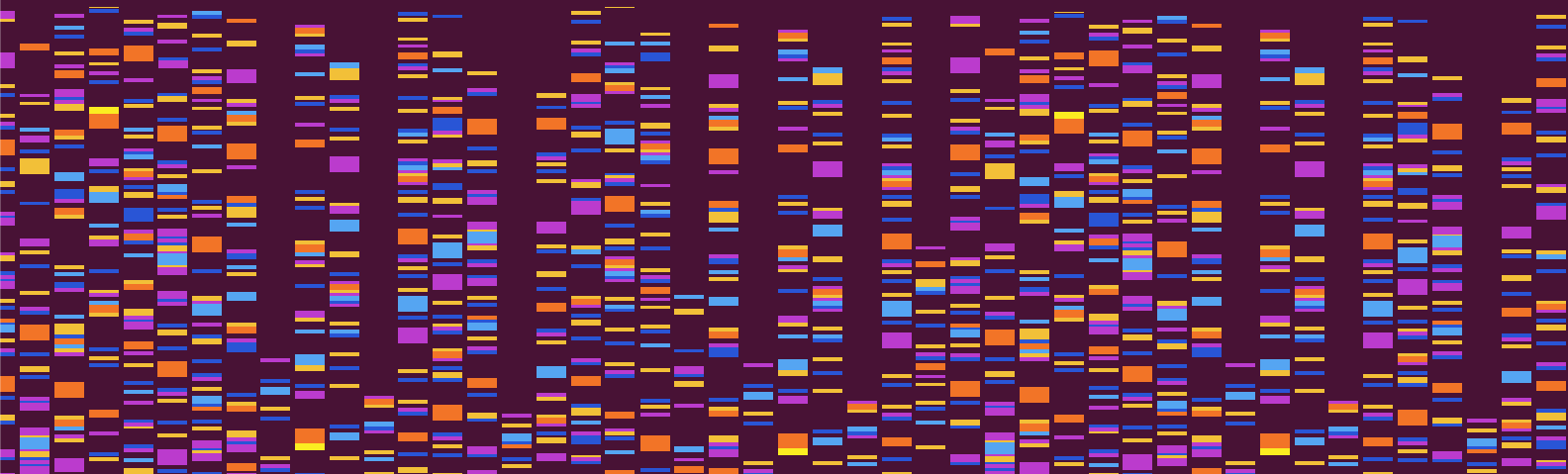# THE BIOETHICS OF GENOMICS AND JUSTICE

NEW DIVISION OF ETHICS
EXPLORES THE IMPLICATIONS
OF PRECISION MEDICINE

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When the Human Genome Project launched in 1990, the international collaboration to sequence and map every gene in a single human being ushered in a new era of scientific and medical research and laid the groundwork for precision medicine. But in unraveling the map of our genes, researchers unearthed more than a collection of As, Ts, Gs, and Cs. Along with groundbreaking knowledge came a host of sticky philosophical and ethical questions. "Genomic technology allows us to do many different things—and the ethics is *What should we do*?" says Wendy Chung, MD, the Kennedy Professor of Pediatrics (in Medicine). "That's a discussion we have to have in the public square, not just as doctors and scientists. Entire societies need to think about it because it affects everything from the criminal justice system to our legal system to our health system."

Thirty years later, precision medicine has arrived at a crossroads. With such projects as the NIH's "All of Us," researchers continue to unleash the power to treat, prevent, and cure disease. As the field accelerates, though, so too does the imperative to grapple with the social and ethical dimensions of emerging technology and the associated research infrastructure, so as not to perpetuate or create new inequities.

Engaging the ethical, legal, and social implications—ELSI—of precision medicine requires a 360-degree perspective. In 2019, VP&S reinforced its leadership in the ELSI field with the appointment of Sandra Soo-Jin Lee, PhD, as founding chief of a new Division of Ethics in the Department of Medical Humanities & Ethics. Trained as a medical anthropologist, Dr. Lee has long investigated the intersection of race, identity, and emerging technology articulated in such questions as, "How does genetics impact how we identify as humans and what counts as difference?"

Dr. Chung has long wrestled with such questions in her clinical work as chief of the Division of Clinical Genetics in the Department of Pediatrics. Chief among the challenges is the lack of diversity in precision medicine data: Roughly 80% of genetic samples are derived from people of European ancestry, who comprise just 20% of the world's population.

Sandra Soo-Jin Lee

Such discrepancies can produce skewed results that aren't relevant to most of the world's population. "I can tell you unequivocally that my patients of Latino ancestry or Black ancestry don't get as much out of genetic evaluations," says Dr. Chung, "because I cannot as readily interpret the data to help them make important decisions about managing risk for cancer or heart disease or what medication might be safest for them."

Yet solving biomedical research's diversity problem involves more than just recruiting historically underrepresented groups to provide samples. It requires examining what Dr. Lee calls "upstream decisions"—how individuals and groups are defined, measured, and compared; how practices to recruit, engage, and retain research participants promote or compromise goals of diversity and inclusion; and how researchers, themselves, engage communities in a long-term research relationship. She's particularly interested in tracing the cascading effects of early decisions about how populations are defined and how researchers make comparisons among populations.

Consider, for example, the downstream effects of how samples are collected and categorized in terms of race and ethnicity. "As we're building infrastructure for a biobank, for example, where the samples are already identified using racial categories, it's easy then to just continue using those categories as a stand-in for genetics," says Dr. Lee. "The downstream effects could be that researchers use race as a biological variable instead of looking at the socio-cultural aspects of those categories."

Dr. Lee's work often involves "studying the studies" to better understand the ELSI of precision medicine research. In her National Human Genome Research Institute-funded study, "The Ethics of Inclusion: Diversity in Precision Medicine Research," Dr. Lee and her team observe and interview investigators, funders, and participants and examine research materials. Through this ethnographic research and analysis, the teams evaluate how those upstream decisions affect such downstream outcomes as engagement, retention, and participation. Ultimately, she hopes to produce a set of recommendations and guidance for precision medicine research going forward.

"For many years, there have been people like Sandra who have made issues related to fair representation in genomics part of their research agenda, but there's no question it's become more

salient since the inception of 'All of Us' under President Obama," says Paul Appelbaum, MD, who chaired the search committee for the new division chief. While Columbia has hubs of ELSI scholarship, including the Department of Psychiatry's Center for Research on the ELSI of Psychiatric, Neurologic, and Behavioral Genetics founded by Dr. Appelbaum, those efforts have been scattered across departments. "There was no single locus around which it coalesced. We wanted someone who could bring people together, who had the skills to be a builder and a leader, and who brought research skills that would augment the growing focus at Columbia on precision medicine," he says. He sees Dr. Lee as a catalyst for interdisciplinary collaboration and scholarship across the University and around the world.

Already, Dr. Lee has begun to nourish a broader ELSI community. In September 2019, she launched the Center for ELSI Resources and Analysis (CERA), a multi-institutional partnership among Stanford University, the Hastings Center, and Harvard University, funded by a $7.1 million grant from the National Human Genome Research Institute. It's the first international center to house ELSI-related research, study instruments, policies and guidance, and a directory of ELSI researchers intended to promote collaboration. Columbia was set to host the fifth ELSI Congress meeting in June, but the event was postponed until June 2022 because of the COVID-19 pandemic. Instead, Dr. Lee, who co-directs the Biennial ELSI Congress, hosted an abbreviated virtual forum with more than 1,000 registrants from 46 countries. The discussion ranged from new models for the implementation of artificial intelligence in precision medicine to use of DNA databases by law enforcement and frameworks for research collaboration with indigenous communities.

Dr. Lee says she was drawn to Columbia's institutional commitment to ethics and humanities, particularly within a medical school setting. "Support for dialogue across the humanities and social sciences is unique," she says. Equally important, says Dr. Lee, is the focus on social justice. "This was an opportunity to build a program that addresses issues of structural racism and inequities and to bring social science to bear on bioethical questions in ways that could be creative, generative, and ultimately impactful." ❖

"HOW DOES GENETICS IMPACT HOW WE IDENTIFY AS HUMANS AND WHAT COUNTS AS DIFFERENCE?"