RDDC™
RARE DISEASE DIVERSITY COALITION

July 15, 2025

Dr. Jay Bhattacharya
Director
National Institutes of Health
Department of Health and Human Services (HHS)
9000 Rockville Pike
Bethesda, Maryland 20892

Re: Request for Information on NIH Artificial Intelligence Strategic Plan Submitted by the Rare Disease Diversity Coalition (RDDC)

Dr. Jay Bhattacharya:

The Rare Disease Diversity Coalition (RDDC) appreciates the opportunity to submit comments on the National Institutes of Health's (NIH) efforts to develop an Artificial Intelligence (AI) Strategy. This initiative presents a critical opportunity to embed equity, transparency, and inclusivity into the future of biomedical innovation and ensure that AI improves and enables equitable health outcomes for all patients.

In the rare disease community, patients frequently experience delayed diagnoses, limited treatment options, and barriers to clinical trial participation. These challenges are compounded for patients from underrepresented communities. While AI can help fill knowledge and access gaps, it also carries a risk of amplifying existing disparities. We strongly recommend that the NIH AI Strategy be intentionally designed to be inclusive and transparent, and grounded in the lived experiences of diverse patients. An impactful strategy would also lay the ground work for continued engagement with patients, their communities, and patient-advocacy stakeholders. We respectfully submit the following recommendations to ensure NIH's AI Strategy prioritizes inclusion, safeguards against harm, and delivers measurable benefits to diverse patients across the rare disease continuum.

I. About RDDC

RDDC was launched in 2020 to address the extraordinary challenges faced by faced by historically underrepresented rare disease patients as encompassed by social determinants of health (SDOH). The Coalition brings together rare disease experts, patients, health care professionals, diversity advocates, and industry leaders to bring about evidence-based solutions that alleviate the disproportionate burden of rare diseases on marginalized populations.

II. AI Models must be trained on disaggregated data to ensure insights are grounded in experiences of diverse patients.

RFI Question 1: Bullet 1: Foundational themes (e.g., Data Readiness, Trust, Translation, Workforce) and pillars that should anchor the plan.

AI has the potential to revolutionize healthcare by improving diagnosis, treatment, and operational efficiency. However, the effectiveness of AI in healthcare heavily relies on the quality, diversity, and representativeness of the data it is trained on. In the development of its AI strategy, NIH must create clear, institutional policies to ensure that datasets used to train AI models are disaggregated by specific characteristics such as race, ethnicity, age, gender, socioeconomic status, and geographic location. Without such granularity, AI models may systematically overlook or misclassify patients from historically underserved populations. These risks are especially concerning for rare diseases, where the patient population is already small and often excluded from traditional research.

We recommend that NIH support equity audits, fund model transparency and explainability tools, and require disaggregated performance metrics to ensure fairness across population subgroups, including by race, ethnicity, gender identity, age, socioeconomic status, and geographic location. These steps will improve clinical utility, promote accountability, and support better outcomes for marginalized communities affected by rare conditions, where sample sizes are often limited.

III. NIH should establish formal mechanisms to collaborate with rare disease and diversity-focused organizations across the AI development process.

RFI Question 7: Bullet 1: Preferred modalities for NIH to collaborate with other federal agencies, state/local partners, international bodies, patient organizations, industry, and/or philanthropic entities.

Active and long-term engagement with patients and patient advocacy organizations is essential for ethical and effective AI development. Engagement with the people and communities impacted by diseases provides critical insight into disease progression, diagnostic barriers, community trust, and patient priorities. Patient perspectives are especially important in rare diseases, where expert clinical knowledge may be limited.

NIH should formalize partnerships with these organizations and ensure they are meaningfully engaged across the entire lifecycle of AI development. This includes research design, data stewardship, risk assessment, model validation, and community dissemination. Sustained collaboration will help ensure AI tools reflect patient experiences and meet the needs of diverse communities.

RDDC applauds NIH's leadership in advancing AI to improve health and urges the agency to prioritize equity and inclusion in rare disease research. A commitment to diverse patient representation is essential not only for fairness but also for the scientific rigor and success of NIH's AI initiatives.

We look forward to continued collaboration with NIH to ensure AI technologies serve all communities – especially those too often left behind.

Jenifer Waldrop
Executive Director
Rare Disease Diversity Coalition (RDDC)