August 14th 2015

National Institutes of Health 5-year Planning Strategy Request for Information

Potential benefits, drawbacks/challenges, and areas of consideration for the current framework

Making the research opportunities presented by rare diseases a priority is a move in the right direction. There are over 7,000 rare diseases affecting more than 30 million Americans, and 95% of these diseases lack an FDA approved treatment. While industry has made significant progress in the fight against rare diseases, we believe the government can and should play a central and more prominent role in supporting rare disease research. While we applaud the efforts of this framework to make rare disease treatments a reality, we encourage this planning committee to include specific rare disease references in more of the Areas of Opportunity.

Specifically, under the Health Promotion/Disease Prevention subheading, we feel that advances in early diagnosis/detection should explicitly reference the desperate need for improvements in this area for rare disease patients. Currently, many rare disease patients are forced to take part in the so-called “patient’s odyssey”, a years-long struggle to find a concrete diagnosis that takes a steep toll on each patient’s finances and health, as undiagnosed diseases typically remain unchecked and continue to progress during this period. Rare disease patients deserve better than this, and the NIH can galvanize their support by adding specific reference to rare disease patients in the “Advances in early diagnosis/detection” language.

Research into rare diseases often provides insight into other diseases and contributes to a more comprehensive understanding of the human body overall. This allows for increased speed and nimbleness when attending to all manner of medical needs, and continues to build the groundwork for advancing precision medicine.

The 7,000 rare diseases are central to the NIH’s historic mission: breaking down barriers and advancing science to find fundamental knowledge about the nature and behavior of living systems and the application of that knowledge to enhance health, lengthen life, and reduce illness and disability.

Compatibility of the framework with the broad scope of the NIH mission

As with the NIH’s effort to map out the human genome, the NIH’s focus moving forward should be in developing a robust, comprehensive understanding of the human body and the diseases affecting it. Discoveries in areas of rare disease research are often beneficial to other areas of research, and contribute to a greater overall understanding of the human body. We can never be certain where the most profound discoveries in medical sciences will come from, and research into the largely unknown and untouched fields within the rare disease space can provide innumerable insights into how the human body works.
Additional concepts in ICO strategic plans that are cross-cutting and should be included in this trans-NIH strategic plan

Nowhere in this strategic plan is the importance of translational research mentioned, and we feel that this is a missed opportunity. The NIH’s own National Center for Advancing Translational Sciences is working to create methods for effectively incorporating much of the research done by the NIH into broader medical fields, and it is critical that the NIH works to make its discoveries as accessible as possible to all stakeholders. Not only will this ensure that Americans are receiving the safest, most advanced medical care in the world, but it also ensures that the incredible work that the NIH is supporting ultimately has an impact on improving health outcomes for patients.

Comprehensive trans-NIH research themes that have not been captured in the Areas of Opportunity that Apply Across Biomedicine

The goal of resolving unmet medical needs is critical for improving the living conditions of rare disease patients who currently lack solutions to even the most basic problems a debilitating illness can cause. The NIH has the unique opportunity to step into this uncharted territory and develop meaningful insights that can spur future solutions. We encourage the NIH to incorporate meeting unmet medical needs across its ICOs, allowing for collaborative research that can result in unconventional solutions to patient problems.

Future opportunities or emerging research needs

As discussed above, the private sector has made significant progress in the fight against rare disease. However, more could be done leveraging public support to effectively de-risk various areas of scientific research that pertain to rare diseases. NIH funding, for example, could play an integral role in elucidating some of the basic pathophysiology of rare diseases and in doing so, lay the needed groundwork for the development of targeted therapies. This research would go a long way in de-risking and accelerating the rare disease drug development process, which is a top priority for patients in the rare disease community.