Milken Institute 2020 Future of Health Summit

Easing the Burdens of Rare Disease: Accelerating Diagnosis

One in 10 people in the United States has a rare disease. Although tremendous progress has been made to identify and understand the 7,000 known rare diseases, more than 95 percent of rare diseases still have no FDA-approved treatment. And even when treatments exist, delays in diagnosis lead to unnecessary delays in treatment.

At a 2020 Milken Institute Future of Health Summit session, experts in the field discussed how to accelerate a rare disease diagnosis.

The Challenges

It can take an average of five years from the time a patient or caregiver first reports a concern to a primary care doctor to a confirmed diagnosis. During this time, patients undergo invasive and unnecessary testing and take on significant out-of-pocket costs. It is a financially burdensome time not only for patients and their families, but also for the healthcare system. All the while, the disease continues to progress.

Meeting participants discussed some of the challenges to rare disease diagnosis, including the following:

- Many frontline providers are not prepared to consider a rare disease diagnosis due to a lack of sufficient training.
- The signs and symptoms of a rare disease can be disparate, making it difficult for providers to connect the dots.
- There is a lack of genetics professionals outside of academic medical centers.
- Coverage and reimbursement for diagnostic tests are variable.

Policy Recommendations

Meeting participants agreed the following is needed to accelerate time to diagnosis for people with rare diseases so that promising therapies reach the newborns and patient communities for whom they are intended:

- **Strengthening health system integration and coordination.** Many rare diseases cut across multiple specialties. Patients can visit multiple doctors across multiple healthcare facilities

before receiving the right diagnosis. To minimize the burden on the patients, coordination of care is essential. Advances in technology and new digital tools can support better care coordination by allowing providers share information and discuss care plans.

- **Supporting primary care physicians in recognizing a rare disease.** Low rare disease knowledge among providers contributes to late and mis-diagnoses of rare diseases. With 7,000 rare diseases, a healthcare professional cannot diagnose every rare disease. As such, participants discussed the need to create new tools to support primary care providers in more quickly assessing a patient and knowing that a particular symptom or set of symptoms might be a sign of a rare disease. Examples include creating diagnostic protocols for common rare diseases and tools that leverage AI to identify a potential rare disease. Participants also called for more training for medical students and providers about rare disease symptoms and diagnostics early as well as more clinical exposure to rare diseases in order to empower more physicians to consider a rare disease diagnosis.
- **Expanding adoption of teleheath.** Changes to reimbursement for telehealth and the ability to offer telehealth across state lines due to the COVID-19 pandemic allowed providers to see more patients during this period. Participants agreed there is great potential to build on the infrastructure enabled by COVID-19. However, participants also acknowledged that telehealth approaches must be properly evaluated to better understand what works and where such approaches can widen disparities due to differential access to technology or preferences.
- **Improving access to genetic screening.** Most rare diseases are genetic or have a genetic component. Participants highlighted the success of the newborn screening program, under which almost every one of the 4 million infants born in the United States each year are screened, and one in 300 are found to have a potentially serious medical condition. Advances in diagnostic technologies will continue to contribute to improvements in diagnosis and screening, but there needs to be greater clarity on their coverage and reimbursement.
- **Providing more mental health support to patients.** Living with an undiagnosed disease can take a tremendous toll on the emotional well-being of patients and families. Participants agreed that policies must consider the holistic needs of those who may be living with an undiagnosed condition, especially mental health support.

FasterCures is grateful to those who participated in this session of the Future of Health Summit for their contributions to the ideas and recommendations summarized in this brief. We would especially like to thank Alexion Pharmaceuticals and Horizon Therapeutics for their partnership on this session.