Shortening the Path to Diagnosis in Rare Disease

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Harnessing technology to pave the way toward faster diagnoses

With 10,000 known rare diseases defined today, the path to diagnosis can often resemble a puzzle, with physicians as detectives searching for clues. Many physicians never encounter a rare disease beyond the pages of their medical school textbooks. Moreover, many symptoms can be varied and non-specific, which can lead to a time-consuming process of excluding more common diseases.

These complexities can further delay answers and place a heavy burden on patients and caregivers, as they endure a multitude of tests and appointments in search of an accurate diagnosis. This quest can place a strain on families and add expenses, requiring long-distance travel for specialist visits, missed workdays and childcare. This experience, referred to as the diagnostic odyssey, is often marked by frustration, misdiagnosis and prolonged uncertainty–and it is far too common within the rare disease community.

Watch the video to hear more from Alexion's R&D leaders about promising areas of diagnostic innovation that are bringing new hope to millions of people impacted by rare diseases.

Fortunately, there is momentum building around efforts to shorten the diagnostic odyssey. Promising developments are being made to use genetic testing to accelerate accurate diagnosis, particularly as it relates to children, who make up 50% of rare disease patients. By identifying rare diseases early in life through blood test-based newborn screening, families and physicians may have new tools to help make better-informed decisions about a child's treatment plan, including eligibility for clinical trials when approved treatments may not yet exist.

Advanced analytics tools, such as machine learning and artificial intelligence, also hold promise. These technologies are making it possible for physicians to interpret existing rare disease health information and diagnostic criteria more rapidly, which may allow them to reach answers faster. With the help of these powerful algorithms, industry, academia and health systems are identifying new ways to diagnose rare diseases sooner.

By harnessing these technologies and broadening access to them for diagnostics, we have the potential to create better outcomes for children and their families and a more sustainable and equitable healthcare system.

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