Rare Disease Treatments Aren't So Rare Any More

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Over 350 million people worldwide suffer from a rare disease. One out of every two of those diagnosed with a rare disease is a child. Eighty percent of rare diseases are caused by a faulty gene. Ninety-five percent of rare diseases have no FDA-approved treatments.[1] The need to find cures and therapies for these afflictions is overwhelming, and the potential for our industry to have an impact is staggering.

While the number of treatments has grown, the path to evidence generation, approval, and access remains uniquely challenging.

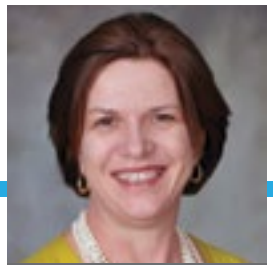
- A widely divergent spectrum of diseases are potential targets for life-changing therapies, however, many of these are characterized by an absence of widespread clinical knowledge of disease natural history, consensus on clinical endpoints, and development and regulatory precedent.
- Widely dispersed patient populations, who often have a high disease burden and significant medical challenges, must be identified, accessed, and engaged to design and execute successful clinical development and access programs.
- Clinical programs can be data intensive and logistically complicated, and are often conducted at research naïve sites, presenting significant management hurdles which must be proactively identified and addressed.
- And finally, because children and adolescents represent at least half of patients affected by rare diseases, development programs must address the specific needs and protections specific to pediatric patients.

Despite these challenges, there are a myriad of innovative and tailored solutions being developed that can help to generate the evidence needed for successful approval and access. In this issue of *The Evidence Forum*, our thought leaders provide insights on many of the important issues and novel solutions affecting the path to market for these special treatments, such as: the changing regulatory landscape (e.g., the US Food and Drug Administration's recent updated draft guidance on rare diseases); incorporating patient-perspective; choosing the right clinical outcome assessments (COAs); the implications of the new EU-HTA process; and, the application of innovative data collection methods.

As this segment of the industry continues to grow, success will depend on keeping an open mind and adapting to the shifting landscape as knowledge increases, new discoveries are made, and expectations and requirements evolve. Now more than ever it is critical to plan early, develop an evidence strategy that considers both regulatory approval and market access, and commit to understanding the nuances of this dynamic space. As Marcel Proust said: "The real voyage of discovery consists not in seeking new landscapes, but in having new eyes." ■

REFERENCES

1. Global Genes. RARE Facts. Available at: https://globalgenes.org/rare-facts/. Accessed February 28, 2019.



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