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Development of orphan medical products

Martine Zimmermann
Alexion Pharmaceuticals, Inc., USA

Overview: This track will discusses opportunities and challenges associated with developing drugs for rare and ultra-rare diseases, and the use of novel approaches to bridge these challenges and successfully bring important lifesaving therapies to patients in need.

Context/Details: Between 5,000 and 8,000 districts, rare diseases exist; affecting around 27 million to 36 million people in the European Union. Collectively, these diseases represent a significant proportion of the population, and a growing healthcare concern since many of these rare disorders are serious conditions with no approved treatments. Since roughly 80% of these diseases have identified genetic origin, our scientific understanding of and ability to target these diseases is growing. Technological advances and our increased understanding of underlying disease biology has aided in the development of several groundbreaking therapies over the past ten years. In addition, heightened public policy support and push to find treatments for patients with rare and devastating diseases has helped in the adoption of legislative vehicles and regulatory programs that compliment scientific discovery and provide important development and financial incentives to companies to bring new targets from bench to bedside. Among them include expedited development and approval programs that offer enhanced regulatory dialogue and support for developers from early stages of clinical development through to approval. More and more we see appreciable regulatory support for innovative program design and increased regulator openness in applying regulatory flexibilities. All these factors have aided in the approval of important medicines for ultra-rare and devastating diseases. Yet challenges do persist in the development process; chief among them is the absence of knowledge about the disease itself, which is needed to inform important aspects of clinical development. Natural history for rare diseases is often poorly described or missing altogether given the small affected populations who themselves have variable phenotypes and clinical courses. For developers, it is important, early in the development process, to assess the depth and quality of this information so that a parallel study might better inform the development process. In this track, we will discuss the pathway to the development of rare diseases, starting with this most critical first step of assessing and acquiring knowledge of the disease itself through natural history studies. We will discuss endpoint selection, innovative design and leveraging expedited programs to help with small data sets. We will discuss ways in which frequent and early communication is essential. Finally, we will bring all these points together by using a real life example/case study.

martine.zimmermann@alexion.com