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Drug Development for Rare Diseases: Challenges and Regulatory Initiatives

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It is estimated that over 350 million individuals worldwide are affected by one of over 7000-8000 rare diseases. International definitions of rare diseases vary; the definition in the European Union is a disorder that affects fewer than 5 in 10,000 of the population, and in the USA it is one that affects fewer than 200,000 individuals. Collectively these diseases represent a significant global health burden; the majority is chronically debilitating and life-threatening, and is without specific treatments. A number of unique challenges are associated with the clinical development of therapeutics for rare diseases, including geographically dispersed small patient populations, phenotypic heterogeneity, incomplete knowledge of the disease pathophysiology or natural history, limited numbers of experienced clinical investigators worldwide, regulatory uncertainties and an absence of prior clinical studies. Additionally, the selection of clinical trial endpoints, which assess the efficacy of an intervention, and if met will result in a positive result for the therapy and trial, can be an arduous process, particularly if validated endpoints appropriate for the disease are unavailable. Therapeutics developed for rare diseases are referred to as orphan drugs because under normal market conditions the pharmaceutical industry would have little interest in developing drugs intended for a small target population. A number of countries have introduced orphan drug legislations through which commercial incentives are provided to the pharmaceutical industry to make rare disease drug development financially viable; incentives include marketing exclusivity, protocol assistance and fee reductions. Today, over 380 orphan designated drugs are commercially available on the global market and more than 800 are undergoing clinical development. To expedite the access of patients with unmet needs to novel therapies, regulatory initiatives exist which have the potential to accelerate market approval. The USA Food and Drug Administration (FDA) permits the approval of drugs based on an efficacy evaluation using surrogate endpoints which are likely to predict clinical benefit [1]. The European Medicines Agency (EMA) has initiatives such as conditional marketing authorization, approval under exceptional circumstances and accelerated assessment [2-4]. In 2016, two collaborative cluster working groups were established between the EMA and USA FDA. The focus of the rare disease cluster is to provide information exchange on topics such as the selection and validation of trial end points, trial designs in small populations, pre-clinical data evaluation to support trials, the design of post-marketing studies in the context of early access initiatives, and risk management strategies for long-term safety issues [5]. This initiative will complement the patient engagement cluster which incorporates patient's involvement and viewpoints in the drug development process [6]. These collaborative enterprises have the potential to drive collaborations on a global basis to address the key challenges that face orphan drug development and provide treatment accessibility to the rare disease population globally.

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