

# Welcome to Advances in Rare Diseases

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## Abstract

Rare diseases represent a broad spectrum of low-prevalence human pathologies. They are primarily genetic in origin, and they profoundly impact on the health of more than 300 million people worldwide. They therefore present a large medical challenge, which is exacerbated by a lack of FDA-approved therapies for the vast majority of rare diseases. Major advances in gene sequencing technologies have, however, led to greatly increased understanding of the genetic mutations that underlie these pathologies, and these findings are increasingly being translated into clinical outcomes.

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## Editorial

Although common conditions, such as diabetes and cardiovascular disease, present substantial socioeconomic burdens on society, rare diseases arising due to genetic, infectious and degenerative mechanisms have nonetheless garnered significant attention due to their high rates of morbidity and mortality. Since the discoveries late last century that diseases such as cystic fibrosis and muscular dystrophies linked to specific genetic alterations, the list of rare genetic diseases that affect humans has swollen to several thousand clinically recognised conditions [1]. Although the prevalence of these rare genetic diseases can be very low, sometimes only affecting a minute proportion of the population, their cumulative medical burden is nonetheless substantial. There is, therefore, a pressing need for improved treatments for these diseases, particularly since they disproportionately affect children, and since they are often fatal.

While rare infectious and environmental diseases (e.g., SARS, ARDS) can be of acute relevance to human health, several rare genetic and degenerative conditions have proven to also be of considerable

academic interest. In addition to the relevance of prion diseases (such as the variant Creutzfeldt Jakob disease in humans) to several aspects of biomedical science [2], studies of Rett syndrome (MeCP2 mutations) have shed new light on neuroplasticity [3]. Aside from their direct impact of human health, rare diseases - regardless of their aetiology - therefore represent a clear and diverse area of biomedical interest.

Although the low prevalence rate of these diseases has limited knowledge progress and investment by the pharmaceutical industry in the search for new treatments, marked acceleration and accessibility of exome sequencing in recent years has greatly increased the understanding of the specific genetic alterations that often underlie rare diseases. Spearheaded largely by grass-roots initiatives [4], this has led to important treatment breakthroughs for at least some of these conditions, while additional new therapeutics comprising both small molecules drugs and biologics are progressing through early stages of development and clinical testing (e.g. [5]). Perhaps one of the most striking aspects of the progress made with the treatment of rare diseases is the high rate of clinical success. Indeed, since these rare diseases are

often due to only a single gene mutation, the identification and development of new and effective treatments are often achievable in record time compared to common pathologies that tend to have much more complex genetic aetiologies. A notable early example of this is phenylketonuria, which, following its genetic characterization, is now routinely screened for in neonates and treated very effectively through dietary changes. More recent examples of clinical successes for rare genetic diseases include straightforward drug repurposing for Dopamine Responsive Dystonia [4].

Despite a multitude of challenges that still need to be addressed and overcome, the rare disease arena can hence be seen as one of the more promising areas of medical research, with clear examples of clinical success and ample potential for further progress in terms of both understanding and treatment of the diverse pathologies.

In light of the very substantial advances in the understanding of the molecular aspects of rare diseases, and the prospect of an increasingly productive drug pipeline for clinical studies, we are pleased to announce the inaugural launch of *Advances in Rare Diseases*. Rare diseases are often referred to as Orphan diseases; a classification that emphasizes the lack of attention and resources allocated to this area of medicine. Our mission, therefore, is to provide biomedical researchers and clinicians further opportunity to disseminate their findings regarding rare diseases in humans. The principal aim of this journal is to publish articles from all areas of the full spectrum of rare diseases, ranging from molecular genetic studies to clinical trial results. All submitted articles will be screened initially by a member of the journal's Editorial Board and if found to be suitable will be sent out for peer-review to internationally recognised experts in the field of rare disease research.

Accepted articles will be of the highest quality and can take the form of original research articles, reviews and commentaries providing pertinent, cutting edge up-dates in a focused area of research. These can be in any field of research related to rare diseases, from identification of novel mechanisms and rare genetic factors associated with human

pathologies, in vitro and in vivo disease models and results from clinical trials.

*Advances in Rare Diseases* welcomes your submissions, and hopes you will visit the website often to stay up-to-date with the latest articles.

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