

## Rare genetic diseases give insight into normal human physiology

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Rare diseases play a key role in healthcare today, with far-reaching implications that have shaped medicine in the past few decades. Interestingly, the sheer volume of diseases (>5000) mean that cumulatively, there could be as many as 30 million Europeans and [25 million Americans affected](#).

Out of that, 80% are of genetic origin. Nevertheless, the actual burden of these diseases remain elusive due to scant or overall lack of epidemiological data, making diagnosis and management [all the more difficult](#).

Current research into pathophysiological mechanisms has not only shed light on normal and abnormal physiology, but also helped to elucidate underlying pathology behind common disorders. Besides that, the development of more efficacious diagnostic tools have broadened clinical perspectives and led to diverse applications in the treatment of other diseases too.

These would not have been possible if not for the immense collaborative partnerships formed between academia, pharmaceutical establishments, patient-driven organizations and regulatory authorities, underscoring the importance of cooperation. Undoubtedly, delving deeper into rare genetic diseases can provide valuable insights into our current understanding of medicine.

Unravelling the intricacies of the mechanisms leading to abnormal pathology holds the potential to understanding the opposite – the normal physiology of the body. They provide immense opportunities to decipher the multifarious complexities of the human body, through learning about the extremes of human pathology.

**Read full, original post:** [Using rare genetic diseases to understand medicine](#)