Translational proteomics of rare diseases and the journey towards precision medicine

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Rare diseases have their own diagnostic and therapeutic challenges that genetic information alone cannot solve entirely. Proteomics, the global study of proteins, offers dynamic insights into protein expression, modifications, and interactions hence contributing to the understanding of disease mechanism.

Translational proteomics in particular, play a pivotal role in patients' stratification to enhance diagnostic accuracy and prognostic assessment. It also contributes to understanding the cellular pathways altered in the disease and to elucidate the relationship between phenotype and genotype.

In this talk, through the results of translational proteomics studies in different rare disease, Cystinuria, Cystic Fibrosis, Cystinosis and Idiopathic Nephrotic Syndrome, I will highlight the significant contribution of translational proteomics to the long journey towards precision medicine.

Despite challenges like data complexity and limited sample availability, translational proteomics holds promise in advancing precision medicine, contributing to personalized and targeted therapies for rare diseases.