GeNeDit

Advanced Approaches for Rare Diseases

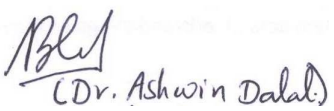
Editorial

Throughout history, humanity has grappled with various diseases, each presenting unique challenges. As medical advancements continue to evolve, our focus shifts accordingly. Thanks to breakthroughs in vaccines and antibiotics, we have triumphed over several infectious diseases and continue to combat others. Consequently, attention is increasingly directed towards non-communicable diseases (NCDs). India, too, is experiencing this transition, with a notable rise in NCDs. Among these, rare genetic disorders emerge as a significant concern. Rare diseases often pose significant challenges due to limited understanding, diagnosis, and treatment options. Patients and their families frequently endure long journeys marked by misdiagnoses, lack of effective therapies, and social isolation.

Rare Disease Day, observed annually on the last day of February, is a global initiative aimed at raising awareness about rare diseases and the challenges faced by those affected by them. This day serves as a platform to amplify the voices of individuals living with rare diseases, as well as their families and caregivers, highlighting the need for greater research, access to treatment, and support networks. This year, the Rare Disease Day falls on 29th February, which is itself a rare occurrence, occurring once in 4 years. Rare diseases often present unique and complex medical, social, and financial burdens, making it crucial to foster understanding and solidarity within communities worldwide. Through education, advocacy, and research, we all need to collectively strive to promote inclusivity, empowerment, and progress towards improved diagnosis, treatment, and ultimately, a better quality of life for patients impacted by rare diseases. A number of such events were conducted in association with SIAMG recently and few glimpses are present in the GenEvent section.

Technological advances in DNA sequencing have revolutionized the field of genetic diagnostics. It is very easy to obtain an exome or genome sequence of an individual in order to detect disease-causing variants even in individuals with atypical phenotypes. This is exemplified by the case reports in this issue on detection of a homozygous *TH* variant in a child presenting with hypotonia, and a homozygous *SOD1* variant in a child with infantile-onset motor regression. However, these high throughput technologies have led to new challenges with respect to interpretation of the pathogenic potential of the variants identified. There is a need for high throughput approaches to functionally characterize the variants in disease-causing genes so that the results can be interpreted with confidence and use for genetic counselling, prenatal diagnosis, and therapy. The GenExpress in this issue deals with few of such high throughput platforms for functional validation of genetic variants.

Recently, we heard the sad news of the passing of Dr I C Verma, a true stalwart in the field of rare diseases. Dr Verma dedicated his life to advancing our understanding and knowledge of rare diseases, leaving an indelible mark on the medical community and countless lives. His unwavering commitment, pioneering research, and tireless advocacy have inspired generations of clinicians, researchers, and patients alike. He leaves behind a legacy of compassion, resilience, and unwavering dedication that will continue to guide and inspire us all. We would like to dedicate this issue to his memory that we will cherish for a very long time.



Dr Ashwin Dalal
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