NEWS IN BRIEF

Registry for Rare Diseases

The Indian Council of Medical Research has recently set up a Registry of Rare Diseases. A rare disease is defined as a disease with a prevalence of less than 1 in 2500 population. Research in rare disorders is important because it helps to clarify mechanisms of disease, which may have unexpected windfalls in other wider areas. For example, the discovery of statins was due to research into primary hypercholesterolemia. The finding that patients with Laron's syndrome are immune from cancer has opened the floodgates to research in the role of insulin-like growth factor 1 (IGF1) in cancer. Many of the rare disorders are due to mutations in single genes unlike common disorders which are due to a complex interplay of multiple genes and environment. Hence rare disorders break new frontiers in the basic understanding of medicine.

The scope of this registry will evolve over time. It will begin as a community-building effort to clarify basic understanding of patients and disease characteristics, and grow slowly to a supportive mechanism for research funding and attracting healthcare providers for these orphan diseases. It intends to comprehensively cover the spectrum of rare and ultra-rare disorders prevalent in the country but initially it shall only gather data of conditions which have an established treatment available in India or globally. With time, many of the other diseases would also be incorporated.

The classes of diseases initially targeted are lysosomal disorders, hemoglobinopathies, skeletal dysplasias, primary immunodeficiencies, bleeding disorders and neuromuscular diseases. More than 70 million people in India suffer from various rare disorders, and both diagnosis and medical care is a challenge for them. The aim of the project is to monitor incidence, prevalence and natural history of disease; to provide access to innovations in diagnosis and therapy; and to support research innovations. Rare diseases are part of the romance of science, and this passion often fuels great breakthroughs. (*http://bmi.icmr.org.in/irdr/index.php, The Hindu 18 June 2017*)

Online Survey of Stress in Doctors

An all India survey by the Indian Medical Association reveals that a whopping 82.7% feel stressed out in their profession. The chief cause of stress was fear of violence in 46.3% followed by the fear of being sued in 24.2%. The online survey included 1681 participants, including physicians, surgeons, gynecologists and super-specialists working in diverse settings ranging from private hospitals to governmental set-ups. The fear of violence has reached an all time high with majority having considered hiring security in their premises.

Maximum violent outbursts were faced by doctors working in emergency care settings. According to 90% of the doctors surveyed, patients' relatives often subject doctors to unruly behavior, verbal abuse and physical assault post-surgery. Violence is largely unreported either due to fear or concern for the patient's distress. Why is violence at the work place rising? Why are we silent about it? What can be done? Questions that haunt every doctor today can't be easily swept under the carpet. Rome is burning and we cannot nonchalantly play the fiddle. (*The Hindu 3 July 2017*)

Genomic Vaccines

A revolution in on the way. Genomics is set to transform vaccine development. The traditional route to vaccine development takes anywhere between 7-15 years, and costs $200 to $600 million. Genomic vaccines promise to cut down both the money and time. There are many key areas of vaccine development that can be handled by genomics. Normally identifying the key antigens of the pathogen to be incorporated in the vaccine is a herculean task. High-throughput methods, in which multiple antigens can be screened simultaneously, could dramatically speed the process.

A new technique, reverse vaccinology, begins with the sequenced genome of a pathogen, and then uses statistical analysis to identify the genes that are most likely to influence the pathogen's ability to infect the host. The proteins that these genes code for become the target antigens, and a vaccine is created from this information. Genomic advances may also make animal models obsolete. If scientists can use genomic data to engineer human tissue in the laboratory, they may be able to study host-pathogen interactions and vaccine efficacy directly in human tissue, and eliminate the guesswork of translating results from animal to human models.

Genomic vaccines will be made from DNA or RNA which enter the cell and produce the required protein. Compared with manufacturing proteins in cell cultures or eggs, producing the genetic material should be simpler and less expensive. A single vaccine can include the coding sequences for multiple proteins, and it can be changed readily if a pathogen mutates or properties need to be added.

Genomics also enables a new twist on a vaccination approach known as passive immune transfer, in which antibodies are delivered instead of antigens. Scientists can now identify people who are resistant to a pathogen, isolate the antibodies that provide that protection and design a gene sequence that will induce a person's cells to produce those antibodies.

Genetics is the flavor of the day and vaccines are not exempt from it. (*Scientific American 26 June 2017*)

Gouri Rao Passi
gouripassi@hotmail.com