Risdiplam - Another ray of hope for SMA patients

Spinal muscular atrophy is an autosomal recessive degenerative disease of motor neurons. Caused by homozygous deletion of survival motor neuron 1 (SMN1) gene on chromosome 5q, characterized by the progressive denervation of muscles leading progressive loss of motor function. SMA is divided into different subtypes based on age of onset and maximum function achieved.

At present the focus of research is on strategies to increase the body’s production of the SMN protein. In 2019, FDA approved first gene replacement therapy to be used in SMA (Zolgensma), which acts by replacing the mutated non-working SMN gene with a working copy of the gene. The cost of one time intravenous dose for treatment of patients with SMA costs approximately 1.6 crores rupees.

A team of researchers studied the safety and efficacy of Risdiplam, which is an orally administered molecule that increases the level of functional SMN protein by modifying SMN2 pre–messenger RNA splicing. Use of risdiplam resulted in higher percentages of SMA affected infants showing improvement of motor functions and attainment of motor milestones compared to the control group, after 12 months of therapy.

Hopefully this will act as a game changer for infants affected with SMA and their families, as this will bring down the cost of treatment and increase the accessibility.

(Vaccine strategy for the COVID-19 exposed)

Since the detection of the first case in China in December, 2019, COVID-19 has spread around the world and infected approximately 200 million people and caused 4.2 million deaths globally. Apart from all this, it had affected the lives of much more people by affecting their livelihood and access to the adequate nutrition. Now, with development of vaccine against the COVID-19, a hope has started to rise that situation will improve. But the cost of vaccine and the availability are the two factors affecting the coverage.

A team from Rush University, Chicago studied the levels of SARS-CoV-2 spike immunoglobulin (Ig) G antibody levels after 1 and 2 BNT162b2 doses in previously infected individuals compared with those without previous infection. Their observations showed that higher levels SARS-CoV-2 spike IgG antibody levels in previously infected individuals after 1 dose of BNT162b2 vaccine compared with infection-naive individuals after two doses. In previously infected individuals, after the second dose, the rise in SARS-CoV-2 spike IgG levels was not significant compared with the first dose, suggesting that one dose may be acceptable in this group. But they also cautioned that a positive PCR diagnosis alone was not sufficient to take off the requirement of second dose of the vaccine.

These findings might redefine the current vaccination strategy, especially for previously infected individuals, and could free up of millions of additional doses thus decreasing the burden on the economies and increase the availability for larger masses.

(Spectral power of EEG: Classify HIE in real time)

Birth asphyxia is a major cause of neonatal mortality worldwide, contributing to up 24% of all neonatal deaths and 11% of under-5 deaths globally. A significant proportion of these babies survive with life-long morbidities. Picking up the moderate and severe forms of the hypoxic ischemic encephalopathy (HIE) is relatively easier compared to the milder form due to lack of definition and evolution of the clinical signs over a period.

Researchers from the Dallas, Texas studied the electroencephalographic (EEG) power as an objective biomarker of the evolution of clinical signs of encephalopathy in newborns with various forms of HIE. Their findings revealed that the delta power and total power are sensitive real time markers for tracking the evolution of HIE from one form to another and can help in picking up the mild forms also. This can help in monitoring and timely beginning of the interventions in order to improve the overall outcome of such babies.

(Adolescent mental wellbeing: Time to act)

Mental health is one of the important aspects of health, which is started getting attention over last few decades. But with the easy availability of internet and round the clock access to it, more and more youngsters are getting addicted to the screens and spending most of their time indoors with limited physical activity. This is not only affecting their physical health but also causing a decline in their mental wellbeing.

Recently, in an international multi-centric observational study, involving 577,475 adolescents, researchers studied the gender-stratified relationships between screen time, mental wellbeing and physical activity. Results showed that increased screen time, exceeding 1 hour per day is detrimental for the mental wellbeing of adolescents, on the contrary, increased physical activity levels are beneficial for the mental wellbeing. Duration of the screen time and physical activity are the two main factors which have a dose dependent association with life satisfaction and mental wellbeing in the adolescents.

Thus, it is high time to plan and implement some public health strategies in order to save our younger generations from the ill effects of the technology.