

The Night May Be Dark, but the Morning Will Be Brighter!

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The year 2020 has been a strange and frustrating year. Everyone of us, whether doctors or the society as a whole, is dealing with the COVID-19 pandemic, though the scenario has changed considerably from the time the initial COVID-positive cases were detected in India to the present time when we have crossed millions of cases. Now we are more confident about the disease and management options and gradually the dismay has subsided with the large-scale use of face masks and hand hygiene. Lockdown, social distancing and the COVID-19 scare greatly affected patient-hospital interactions. Patients with cancer, thalassemia and rare metabolic diseases who need to visit the hospital on a regular basis for their treatment have been particularly affected.

As we all know, adversities bring new opportunities and mankind has always excelled and achieved new milestones whenever faced with doom. Historically, mankind has overcome many such pandemics and international crises, whether social or economic. One of the significant outcomes of this pandemic has been the e-OPD initiative started by many hospitals wherein online interaction is done with patients through email/telephone/ WhatsApp etc. which has significantly decreased patient discomfort. This concept should be given due consideration as an important entity of patient management as it is cost effective as well. Travel expenses decrease, it enables effective time management and even patients from remote areas can consult doctors at a tertiary centre. If required they are asked to visit at an appointed time and specific interventions can be planned well in advance.

As medical geneticists we come across a lot of families and patients with rare debilitating disorders and witness the challenges they face, as most of these disorders are either incurable or the available treatments have constraints like cost and limited accessibility. But the situation is

changing as more advanced diagnostic techniques (like whole genome sequencing) are becoming available and a lot of new therapies for rare disorders are knocking at the door.

The month of August is celebrated by national and international forums working in the field of spinal muscular atrophy (SMA) as SMA awareness month. We have started treating some children with SMA with Spinraza (Nusinersen) since late last year as part of a humanitarian access program. It started with an enrolment process with around hundred of them approaching our centre with a hope of getting this wonder drug. It was an online selection process with stringent inclusion criteria and only a few children could get access to the drug. This whole process of selection was done by international experts with adequate steps to avoid any biased selection. During this time, we came across most of these bright children and were really astonished to see their talents, whether the sister-brother duo of AR and RR from Varanasi or the talented AC (her paintings and artwork are prodigious) or ZS from Kashmir valley. Though this neuromuscular disease impairing motor neurons is an obstacle for them and their families, they still smile and most of them have a very good intellect. Seeing them and walking the path of their journey as clinicians, our conscience awakens and inspires us to strive further and hope that most of them get treatment.

Coming back to ZS; she hails from the Kashmir valley and is one of the lucky few to get the drug. This drug is given as an intrathecal injection, initially three loading doses 2 weeks apart, then the 4th loading dose 30 days after the third dose followed by maintenance therapy every 4 months. The family was compliant to therapy despite travel restrictions etc. in Kashmir. Her last maintenance dose got delayed due to COVID-19 related travel restrictions but was finally given last month. We assess the motor function of patients of

SMA with the Hammersmith Infant Neurological Examination (HINE-2) and Children's Hospital of Philadelphia Infant Test of Neuromuscular Disorders (CHOP-INTEND) scores. As seen in other centres, the children in our treatment cohort are also showing favourable response to nusinersen therapy with improvement in HINE-2 and CHOP-INTEND scores. Similar improvement was observed in ZS as well. Apart from the clinical improvement in the child, we also observed significant improvement in the interaction with her parents. Her father used to be very hesitant in interacting with us and we could sense his anxiety which has gradually changed over time and now he is far more transparent and communicative. A good doctor-patient-family relationship is a prerequisite for disease management, more so for chronic conditions such as genetic disorders and for associated genetic counselling. We felt gratified when he shared videos of ZS performing on a song with active movements of her upper limbs and torso which was surely a gain considering her previous history of paucity of movements. We could see tears of hope in the father's eyes. This hope and optimistic attitude allow us to carry on, whatever adversities are there. *'Tough times do go away and don't last, but tough people do'*—this

attitude is the need of the moment as we are dealing with the COVID-19 pandemic; *'this too shall pass'*.

Other newer therapies for SMA like gene therapy (onasemnogene abeparvovec) and the oral *SMN2*-splicing modifier Risdiplam are also now accessible to some children in India, largely due to efforts of the families and some dedicated doctors. Hope the scenario changes in the future and policy-makers start paying more heed to medical expenditure and research. The COVID-19 pandemic has taught everyone that health is indispensable and the verse which we learnt in primary school days needs to be altered a bit, i.e. *"If wealth is lost, nothing is lost; if character is lost, all is lost but if health is lost, the world is lost"*.

We doctors have to follow certain ethics and duties and apart from being good clinicians we should also develop empathy and good interpersonal relations with patients and their families. Medicine is considered a noble profession and such incidents reiterate the fact that it is more than a job. The satisfaction which you get from the joy you bring in someone's life is more than any materialistic gain.

"The nights are dark, the soul quivers in fright; don't worry O creature, the morning will be bright".