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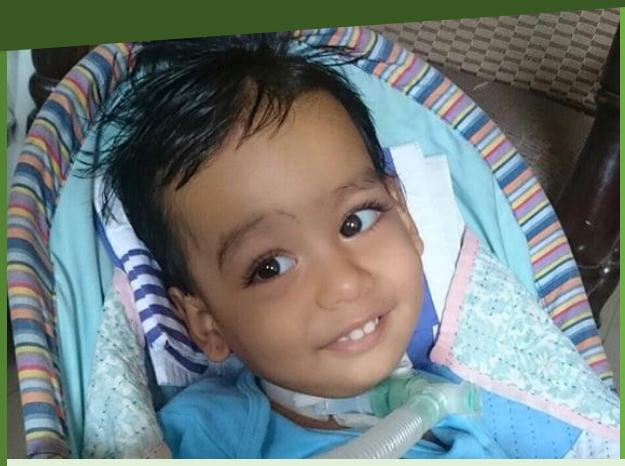

In this issue:

EDITORIAL

We are glad to present the September newsletter! We extend our heartfelt gratitude and thanks to every member for their invaluable contribution.

In this edition we have an article on rare diseases -'New treatment options in childhood rare diseases: The pediatrician as an advocate and messenger of hope' .We are in the era of personalized treatment of rare diseases, a new ray of hope for today's children for a better future.

Its CIAP Election time! Please exercise your franchise of voting to safeguard Mother IAP.

We look forward for interesting articles. Happy reading!

Warm regards,

Dr Nandeesh

Dr Priya Shivalli

New treatment options in childhood rare diseases: The paediatrician as an advocate and messenger of hope

Dr. Ann Agnes Mathew, Paediatric Neurologist

Introduction:

We are privileged to stand at a momentous stage of history where cure and treatment will soon be available for many monogenic disorders. Many genetic diseases are quite complex. The recognition of their symptomatology, the actual diagnoses and later on, the management of complications and sometimes end of life care becomes difficult for each rare disease, more so in the absence of guidelines and expertise.

The Role of the Paediatrician:

To my mind a Paediatrician is first and foremost an advocate for child welfare and child rights. We are always trained to put children first. All of us in the field can identify a time in our training and history, when we decided to take up this field. No matter how senior one gets or how experienced one becomes, rare diseases often confuse us. But one should not hesitate to reach out and seek help. Our children would be well served if we work together, with collaboration between the primary care paediatrician and the sub specialists with roles that can be well demarcated and later amended with the lead paediatrician taking on a more active role as he or she gets more comfortable with that particular rare disease.

This would always benefit the child and their family, as they will more often than not, be comfortable with the paediatrician. This of course would entail better communication, be it written or telephonic between all the specialists involved and often across various sectors, government and private.

Early diagnosis:

Having a high index of suspicion often helps in Paediatrics, as it invariably means an early pick up of problems. This in turn means, that our patients end up having better outcomes. This may mean a simple CPK test in any boy, for example a 2-year-old, who has a **speech delay**, as that might be the first sign of a Duchenne Muscular Dystrophy or looking for **CPK i**n a boy with tip toe walking in the absence of upper motor neuron signs. Picking up diseases like Spinal muscular atrophy in this day and age, in infants, especially the type 2 variety can often be a challenge. But the experienced clinician will notice that there is something about the discrepancy in the motor milestones of a baby who sat at 7 months but is yet to achieve even supported standing 5 months down the line, which needs looking into. This in turn might lead to an examination during routine vaccination, which might segue to the discovery that the said baby is unable to weight bear, which definitely bears further investigation. Like in most childhood diseases, we are custodians of growth and development and so monitoring normal growth, be it centile plotting or milestone recording is the key and will almost always bring us to the diagnosis.

Multidisciplinary Approach:

One of the key aspects of caring for a child with rare diseases, be it DMD or SMA or a genetic epilepsy or a metabolic disorder for that matter, is the fact that it often involves multiple organ systems and so by definition can not be under the purview of one specialist or clinician. Here is where the paediatrician emerges as the champion of the child. The child often meets specialists whom, families travel very far to meet and who might not be accessible most of the times for geographic reasons for an emergency or otherwise. Hence it would be useful to have an individual care plan for most children with rare diseases, with the paediatrician in the lead, but with adequate support from the paediatric subspecialist, be it a paediatric neurologist or a paediatric haematologist or a developmental paediatrician. This may initially mean some extra time during a routine visit to the Paediatric OPD. . But in the long run it means that hospitalizations for complications are streamlined and even diminished. So, a chest infection might get flagged sooner rather than later and a child might end up with a feeding tube, preventing further PICU

admissions and better quality of life over all or even end up being identified as someone who requires prophylactic antibiotics in the face of frequent chest infections.

A very good example is the management of fractures in neuromuscular disorders; especially DMD where, having a fracture in the lower limb and a routine immobilization for 6 weeks would almost definitely mean that the child would become non ambulant sooner and thus have a reduced life span with all its attendant complications. So the key here would be early mobilization with monitoring for callus formation at the fracture site, again with the help of all the experts involved.

End of life care:

One of the abiding pains that families often mention is that of not being understood and supported, oftentimes as their child enters an irreversible stage of the illness. This is where we can make a difference, with just our presence and understanding of the illness in helping them prepare to let go, while ensuring that we have given the best of care for their little one at all times. Innumerable times, it is often for this that families come back and thank us, in helping them with their difficult decisions, not making the decisions per se, but giving them the facts and allowing them to process. This might mean, knowing when not to ventilate a child with a deteriorating degenerative process and a pneumonia and no discernable end point. But we should also be able to differentiate this child from a child with a neuromuscular disease that is reasonably static with a preexisting undiagnosed sleep hypoventilation and a chest infection who will not get better after extubations, unless one takes the help of noninvasive ventilation such as BiPAP and cycling with self-ventilation instead of a direct wean off the ventilator as these children tire easily, can't clear secretions and often end up blocking off their air ways with its attendant atelectasis. So, anticipating this difficulty and predicatively acting on it might mean better outcomes and less morbidity and mortality, as these children are often salvageable if we are not too quick to wean off respiratory support.

Patient advocacy and the role of Patient support groups:

In rare diseases, unity and collaboration is our only strength. One of the organizations and resources that is available is a homegrown one and is called the ORDI- The Organization for rare diseases India, which even has a helpline. This is meant for both families and physicians. While in certain rare diseases like SMA there are well maintained PAGs (Patients Advocacy Groups) like Cure SMA, many rare diseases are not as fortunate and are less cohesive. This is where ORDI comes in as an often under-utilized resource and an umbrella entity. An example is what happened last year with West syndrome or Infantile Spasm, a devastating condition, more so if it is a manifestation of early tuberous sclerosis (TS). This as we know, is most responsive to Vigabatrin especially in TS, a drug

that is still not freely available in our country for various reasons. Recently when the availability hit rock bottom, many families were being held to ransom for the drug by unscrupulous pharmacists, mainly due to the changeover in GST policy by the government and various other practical issues. The ORDI stepped in as a bridge and helped the Tuberous Sclerosis alliance of India link with their international counterpart. For our part, we managed to obtain signatures from over a hundred paediatric neurologists from all over the country to persuade our leaders and policy makers to make things easier for these families. While this is still work in progress, things have improved for many of our families since this exercise.

Spinal Muscular Atrophy, an example of collaboration:

The recently held camps in Sagar Hospitals and Bangalore Baptist Hospital are a good example of collaborative work between various organizations. We have been chosen as the sole dispensing body in South India for the drug cure for SMA, Spinraza (Nusinersen) which costs roughly 70 lakhs per injection (so approximately 5 crores the first year and 2 crores or so thereafter) on a humanitarian basis. This is an example showcasing public private collaboration as the way forward. Many paediatricians and geneticists from Bangalore Baptist Hospital, Sagar Hospital, Kolkata, Rainbow children's Hospital, St. John's Medical College Hospital, St. Martha's hospital and elsewhere joined hands to make a difference. The end results being that we as a whole saw a total of 140 patients with Spinal Muscular Atrophy in just 7 days. This we know is just the beginning of the journey. We hope to have better registries and subsequently make our own "made in India" cures for these diseases as we both have the numbers and the intellectual resources for doing this. The era of personalized medicine is here and who best to understand this than we who are in the field of paediatrics and see all the rare diseases as they manifest themselves in childhood. For diseases like SMA, we are entering a world where, one day not too long into the future, when universal new born screening becomes a reality, we will end up diagnosing these babies before they are symptomatic. Then we could have infants with no discernible difference in their motor milestones in comparison to their normal counterparts. So, here is towards making a better future for our children, the wealth of our nation!

Dr. Ann Agnes Mathew

Paediatric Neurologist, with special Interest in Neuromuscular Disorders.
Sagar hospital and
Bangalore Baptist hospital
Bengaluru

PHOTO GALLERY

IAP Charity Day

Monthly free medical camp at BPS site, RR Nagar

Rheumatology meet at API Bhavan

IAP PG Quiz Inauguration

Rheumatology meet at API Bhavan

IAP UG Quiz inauguration

UPCOMING EVENTS

IAP NEOCON

(National Conference of Neonatology Chapter of IAP)

Date: **27,28,29 September, 2019** Venue: **JW Marriott Hotel, Bengaluru**

Registration details: www.iapneocon2019.com, rads.latha@gmail.com

October monthly meet: Haemato-Oncology, Date: October 13, 2019

CME on Vit D deficiency - CIAP program in **November**