DPT

DEVELOPMENTAL PEDIATRICS TODAY

November 2020

Monthly e-Newsletter of IAP Chapter of Neurodevelopmental Pediatrics

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Editorial

Our Motto - "Preventing impairment from becoming a disability and preventing disability from becoming a handicap through early intervention at every stage."

Respected Seniors and dear friends,

I hope you all are well and celebrated the festivals as best as one can in the current circumstances. Most family meetings for festivals and birthdays have shifted to online Video platform. Marriages being live streamed to minimize the attendance.

The world is facing second Covid wave which seems to be deadlier than the first one for the western nations. As far as India is concerned the Ministry of Home affairs issued fresh guidelines on 26 November for "surveillance, containment and caution "in view of the surge in Covid 19 cases due to the ongoing festival season and the onset of winter. Further it said, states and Union Territories, based on their assessment of the situation may impose local restrictions with a view to contain the spread of Covid 19.

November is the Awareness month for Epilepsy, Prematurity and good nutrition. 17th November is world prematurity day. So, the quiz questions include topics of prematurity and epilepsy. Kindly send your answers for the quiz.

Long live IAP!

Dr. Lata Bhat
Chief Editor

Chairperson's Message

Dear Readers,

The advent of autumn reminds us to be prepared for winter and reflect on our year's harvest of performance, while shedding its leaves for a new appearance. I feel each one of us have so much to ponder and so has it been for the Neurodevelopmental Chapter too. We had to look at things with a new eye and one of them is delivering our services to these children in a new fashion, shedding the heavy cloak of centre-based rehabilitation. To my mind this has been a sigh of relief for the children who long to grow up in freedom. There are many amongst us who have been supporting and propagating homebased programme in natural settings, working on empowering parents. Autism brought a different dimension to teaching, as video modelling creeped in to as one of the modalities in shaping these children.

Today teleconsultation, ushered in by the pandemic, is what we have fallen back on. We are beginning to see the children more in the home environment where they can be best assessed and best nurtured for an applicable learning. This was always the need of the hour but of course we need to work on greater parental involvement.

This Newsletter upholds the importance of it. It anchors the above thoughts to ignite your minds for innovative thinking. We will certainly be happy to get a feedback of the contents of this issue.

Happy reading!

Dr. Shabina Ahmed MD, FIAP
National Chairperson
Neurodevelopmental Pediatrics Chapter of IAP

Snippets from the Secretary

'Sometimes real superheroes live in the hearts of small children fighting big battles.' - Anonymous

Dear seniors and friends,

Seasons greetings from the IAP Chapter of Neurodevelopmental Pediatrics.

Hope this issue of newsletter find you all and your families in good health.

The year is coming to an end and hopefully, so is the Covid-19 pandemic as we see a drop in the number of cases reported in the country and the hope of a vaccine in the near future brightening. Even then, the year 2020 has changed the way we look at things in our everyday life in ways more than one. The chapter conducted fellowship examination online in the last week of October which was a big learning experience for all those involved. The fresh batch of fellows have started their course at their respective centers and I take this opportunity to wish them a great learning journey in the coming year. The current issue of the newsletter also features few articles and journal scans received from the outgoing batch of fellowship students.

The chapter is planning its annual national conference in the last week of December on the online platform of dIAP with a host of eminent national and international speakers who will be speaking on a wide range of topics related to developmental pediatrics. Three workshops are planned ahead of the main conference with topics like - basics of neurodevelopment, early diagnosis of neuro-developmental disorders (NDDs) and living with NDDs to give the participants a good platform to learn and interact with experts in the field.

Wishing all the readers a merry Christmas and a happy 'n' prosperous new year in advance.

"Disability is a matter of perception." Martina Navratilova

Long live IAP.

Wg Cdr (Dr) KS Multani
National Secretary
IAP Chapter of Neurodevelopmental Paediatrics

Journal Scan

Neurological Aspects of Covid-19 in Children

Dr Priyanka Patil
Child Development Centre, Trivandrum

Only 2% of large cohort of COVID-19 cases from China were less than 19 years old.1 There are no published cohorts describing neurological complications of COVID-19 in children with exception of a few case reports/ series.2

Neurological Manifestations:

The neurological manifestations reported in few COVID-19 pediatric case reports include spectrum of manifestation. Most of the manifestations (except taste and smell impairment) were associated with severe COVID-19 in adults which are rare in children.

The striking absence of literature on cerebrovascular events in young children may probably be due to the presence of proactive anti-thrombotic factors and absence of comorbidities like atherosclerosis and hypertension.

Considerations in children with neurodevelopmental disorders (NDD)

Children with chronic neurodevelopmental disorders (NDD) are a vulnerable population due to limited understanding of the mode of spread of COVID-19, inevitable dependency on caregivers for personal hygiene and care and limited access to healthcare facilities.

Children with specific neurological disorders may require immunosuppressive therapy such as steroids which may act as a double-edged sword. There is a rising concern about immunosuppressive therapies like adrenocorticotrophic hormone (ACTH) or steroids used in children with infantile spasms, Duchenne muscular dystrophy (DMD), etc. Guidelines suggest continuing standard therapies with these drugs as per the concerned entity. Children with DMD, in case of acute illness the steroids can be converted to stress dose as per the clinical judgement.9

Journal Scan

Intravenous IVIg and azathioprine can be given under careful monitoring of lymphocyte counts in the case of azathioprine while rituximab can be initiated after careful risk-benefit ratio consideration.10

Children with disabilities and their families frequently require medical support as compared with typically developing children. Closure of special schools and early intervention centers, lockdown, restriction in mobility may further heighten their rehabilitation needs.

Telemedicine is a need of an hour for continual provision of medical services (including rehabilitation). Therefore, policy-making and resource allocation should aim at providing optimal care to children with NDDs.

Conclusion

A high index of suspicion and characterization of clinical features by the the frontline teams are key to diagnosis. For outpatient care and rehabilitation of children with NDD, teleconsultation may be a beneficial approach.

Points to Remember

• Neurological manifestations are reported in pediatric COVID-19 albeit in lower frequency than that in adults.

Reference article: Gulati, S. (2020). NEUROLOGICAL ASPECTS OF COVID-19 IN CHILDREN. Indian Journal of Practical Pediatrics, Vol.22 No.2(Apr. - Jun. 2020), issn 0972-9607, 144-146.;

Journal Scan

Home-based Sensory Interventions in Children with Autism Spectrum Disorder: A Randomized Controlled Trial

Padmanabha H, Singhi P, Sahu J, Malhi P.
The Indian Journal of Pediatrics. 2018;86(1):18-25.

Introduction

The interventions targeting the sensory problems in children with ASD are clinic-based, child-centered sensory integration therapy; followed by classroom-based, adult-directed sensory-based interventions; environmental enrichment, auditory integration based approaches, music therapy, massage. Some modest response has been seen with sensory integration therapy and environmental enrichment interventions. However, the applicability of these results has been limited due to lack of uniformity in the study population, assessment and outcome measures, intervention approaches, along with feasibility and financial constraints in a developing country. This study was done to determine the feasibility and efficacy of home-based sensory interventions in children with Autism spectrum disorder (ASD) with sensory processing abnormalities.

Methods

This was a 12-wk, parallel group, pilot, randomized controlled trial. During the study-period, 185 children with ASD between 3–12 y of age, with sensory processing abnormalities were screened for eligibility. Twenty-one children were randomly assigned to the sensory-intervention group and 19 to the standard-therapy group. Sensory-intervention group received homebased sensory interventions by the parents/caregivers plus standard therapy; standard-therapy group received speech therapy by the speech pathologists and applied behavior analysis by the child psychologist.

Results

The mean change in scores at baseline and 12 wk into intervention showed that children in sensory-intervention group (Mean = 9.33, SD = 3.52) scored significantly better on Parent Rated 10-item Likert Scale (PRILS-10), as compared to standard therapy group (Mean = 2.47, SD = 1.46), $t(36) = 8.16$, $p < 0.001$; $d = 2.54$. Marked improvement was noted especially in reduction of hyperactivity, motor-stereotypies and auditory sensitivity in those who underwent sensory interventions. The mean change in scores in sensory-intervention group on Children's Global Assessment Scale (CGAS) (Mean = −9.19, SD = 2.33, $p < 0.011$; $d = -1.75$) and Pediatric Quality of Life Inventory 4.0 (PedsQLTM) (M = −10.53, SD = 5.34, $p = 0.008$; $d = -0.88$) showed significant difference in the sensory-intervention group as compared to standard-therapy group. Overall, there was 32.3%, 18.1% and 15.8% improvement on PRILS-10, CGAS and PedsQLTM respectively in sensory-intervention group.

Conclusions

The present findings suggest that home-based sensory interventions are feasible in a developing country and are suggested to have a beneficial role in ASD.

Quiz

Dr. Lata Bhat
Director and Developmental Paediatrician
Palak Child Development Centre, Delhi

Qs.1) What are the brain lesions identified by cranial ultrasound in a preterm baby which are associated with Neurodevelopmental impairment :

a) IVH
b) Periventricular Hemorrhage
c) White matter damage
d) PVL

2) Which of the following Cranial USG findings in a preterm baby is the highest predictor of cerebral palsy:

a) PVL Grade 2
b) Cystic PVL
c) Hydrocephalus
d) IVH

3) Factors that contribute to the development of PVH-IVH in a preterm baby are:

a) Loss of Cerebral autoregulation
b) Abrupt alterations in Cerebral blood flow
c) Abrupt alterations in Cerebral blood pressure
d) All the above

4) What percentage of people with Epilepsy develop it before the age of 18 years:

a) 70-80%
b) 50%
c) 20%
d) 40%

5) What percentage of Seizure disorder has no known cause i.e., Idiopathic:

a) 10%
b) 50%
c) 75%
d) 30%

6) Causes of Seizure disorder are:

a) Trauma to fetus during pregnancy or birth,Poisoning (Lead, environmental contaminants)
b) Viral or Bacterial meningitis, alteration in blood sugar levels
c)Alcohol/drug abuse, brain tumors/stroke
d) All the above

Quiz

7) Which of the following is true about Lendau Kleffner Syndrome :

a) Encephalopathy with Status epilepticus in sleep
b) Age of onset is 2-8 years
c) There is Language regression
d) Premorbid Development is normal

8) What all screening/assessment should be done in a preterm baby born < 32 weeks:

a) ROP
b) BERA
c) Growth and Developmental assessment
d) Neurological

9) Regarding Hypoglycemia in a baby, which of the following results in high risk stratification for Neurodevelopmental Delay, according to the NNF guidelines of risk stratification for follow up of high risk babies :

a) Transient hypoglycemia
b) Blood sugar < 25mg/dl for > 3 days
c) Hypoglycemia resulting in seizures
d) All the above

10) WHO defines prematurity as a birth before 37 weeks of completed gestation or birth before 259 days from the first day of Woman's LMP.

Which of the following is correct regarding Preterm terminology?

a) Very Preterm – Born before 32 weeks of gestation
b) Moderate Preterm – Born from 32 weeks to 33 weeks plus 6 days of gestation
c) Late Preterm – Born from 34 weeks to 36 weeks plus 6 days gestation
d) All the above

Please send answers to lata2207@gmail.com / Kawaljit000@gmail.com before 30 November 2020. Correct answer will be published in next issue

Answers - OCTOBER

1. a
2. d
3. c
4. d
5. a
6. d
7. d
8. d
9. a, b, c, d
10. a, b, c, d

Autism Spectrum Disorder

Dr G Swetha
Child Development Centre, Trivandrum

Autism Spectrum Disorder (ASD) is a Neurodevelopmental Disorder characterized by the presence of marked impairment in social interaction, communication and abnormally restricted activities and presence of repetitive behaviors. ASD prevalence as per CDC is 18.5 per 1000 population, 1 in 54 children aged 8 years. Atypical sensory experiences are a defining feature of autism. (1) Kanner's original description of autism referred to negative reactions to sensory stimuli, "loud noises or moving objects, which are therefore reacted to with horror or panic". Asperger also described children with ASD to show hypersensitivity in some circumstances and either ignoring (appearing hyposensitive) or seeking out response to particular stimuli.

Sensory processing is described to "the way in which the central and peripheral nervous systems manage incoming sensory information from the sensory organs. The process includes the reception, modulation, integration, discrimination, organization of sensory stimuli, and the behavioral responses to sensory input. Sensory Processing disorder (SPD) is neurophysiological condition in which sensory input either from the environment or from one's own body is poorly detected, modulated or interpreted and /or to which atypical responses are observed. The nosology of SPD includes three main types: 1) Sensory Modulation i.e sensory over-responsivity/sensory under-responsivity/ sensory craving /sensory seeking 2) Sensory Discrimination - recognition of qualitative and quantitative sensory features and 3) Sensory-based Motor Disorders- posture disorder, and dyspraxia. Dr. Jean Ayres, was the first person to use the term "Sensory integration dysfunction" related it to "neurological traffic jam. Sensory processing abnormalities in ASD are reported across all ages(2) and levels of symptom severity.

In the early years of autism, sensory features were considered a key aspect of the autism phenotype. However, as more studies were conducted on the cognitive and social aspects of autism, a more social cognition view of autism became predominant. The prevalence of clinically significant sensory processing abnormalities in children with ASD is 40-88%.(3) Sensory processing disorders are increasingly recognized not just as a part of symptomatology in ASD but has been speculated to be directly associated with its core features. Strong associations between sensory symptoms and core features like verbal status and repetitive, restricted behaviors were found. (he high prevalence of these sensory symptoms led to the inclusion of sensory symptoms as part of the ASD diagnosis in the new edition of the Diagnostic and Statistical Manual of Mental Disorders (DSM-V). Under Symptom B the new autism criteria for DSM-

5 include 'Hyper- or hypo reactivity to sensory input or unusual interests in sensory aspects of the environment. A better understanding of sensory processing in Autism, including domains will improve our understanding of what persons with autism experience every day and how their sensory experience may shape their behaviors and their response to the world. Sensory Processing Disorder has not received adequate importance by the Pediatricians and Development Pediatricians, who are the first resort to children with Neurodevelopmental disorders. Collective interests in SPD has been from the discipline of Occupational Therapy so far.

Functional impairment due to sensory dysfunction:

Children with evidence of sensory processing dysfunction often have difficulty regulating responses to sensations and specific stimuli and may use self-stimulation to compensate for limited sensory input or to avoid overstimulation. Behaviors such as stereotypic motor movements, aimless running, aggression, and self-injurious behaviors have been correlated with sensory processing abnormalities. Attention and arousal impairments have been due to impairments in modulating sensory input.(4) Miller outlined five functional impairments associated with SPD namely- social participation, decreased length/ frequency/complexity of adaptive responses, impaired self-esteem, poor daily life skills and diminished sensory- motor development. (5) Sensory dysfunction causes difficulty in completing everyday tasks or activities of daily living (ADL)- bathing, dressing, brushing, feeding, toilet care and leisure activities, ability to engage in daily occupations and diminished functional capabilities, including his sense of purposefulness, social interactions, participation in activities and significant care giver strain. Variable response to similar stimuli makes the child's behavior unpredictable.

Role in causing the other core features:

The various manifestations of core symptoms of ASD can root from the sensory disturbances. Boyd showed there were significant associations between hyper-responsive and repetitive behaviors, and sensory seeking and ritualistic/ sameness behavior. (6) In addition to sensory-motor problems such as excessive rocking and spinning, sensory dysfunction leads to preoccupations with personal interests, sensitivity to certain foods or clothes, and a strong attachment to certain objects. Difficulties with modulating visual input lead to avoidance of eye contact and inefficient use of eye gaze and play in an unusual way with their peripheral vision (hands, moving objects). Abnormal emotional regulation and tantrums are due to frustration caused by hypo/hyperresponsiveness.

Early Sensory disturbances as tell – tale signs of future ASD:

Sensorimotor features of social touch aversion and excessive mouthing of objects, as well as delayed response to name and decreased affect, hypo reactivity (e.g., diminished response to name) has been an early diagnostic consideration.

A thorough evaluation of sensory processing abilities to determine whether that child's sensory processing is compromised has to be done alongside the initial screening and diagnostic test. This would help these children affected with sensory dysfunction to integrate and function better in their activities of daily living. The

interventions targeting the sensory problems in children with ASD include clinic-based, child-centered sensory integration therapy; followed by classroom-based, adult-directed sensory-based interventions; environmental enrichment, auditory integration-based approaches, music therapy, massage.

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Naturalistic Developmental Behavioural Interventions for Autism: An Amalgam of Developmental and Behavioural Science

Dr. Rangan Srinivasaraghavan, Dr. Reeba Roshan
Dr. Beena Koshy, Dr. Samuel Philip Oommen
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Introduction:

Autism spectrum disorder (ASD) is a life-long developmental disorder characterized by qualitative impairments in social and communication behaviour and a restricted range of activities and interests. Although initially autism was considered to have poor prognosis, over the past three decades research has shown significant proportion of children receiving early intensive behavioural intervention have been proven to have optimal outcomes. Treatment for children with ASD has advanced considerably over the past three decades. Currently the two primary approaches to treating autism include a behavioural approach and the developmental-focused approach. The behavioural approach focussed mainly on changing the behaviours associated with autism. It was proven to be effective in teaching skills to autism and was promoted as the 'gold standard' for treatment of autism. Advances in the fields of child development, and developmental psychopathology, have contributed to the development of many child-development centred intervention models which have been called as the ''Naturalistic Developmental Behavioural Interventions (NDBI)''. Evidence on the effectiveness of NDBI in younger children with Autism is emerging recently. This article describes the evolution of therapy in Autism, development of NDBIs, common features, evidence base and their applicability to India.

Evolution of Behavioural treatment approaches to Autism:

- Early 1960s: Autism was considered untreatable.
- 1961-62: The idea of using operant conditioning for teaching new skills to children with autism was demonstrated by Ferster and DeMyer.
- 1960s till early 1980s: Operant teaching applied for teaching various skills like language, social, play, self-help, academic skills for children with Autism. Studies showed that they were effective. New field of "applied behaviour analysis (ABA)"- operant skills to reduce the occurrence of ''interfering'' or challenging behaviours.
- 1987: Lovaas' autism treatment study was published [1]. The method used was Discrete Trial Training, which was an ABA based intervention approach. This paper brought about a paradigm shift in management of Autism in children. ABA based approaches gained popularity.
- Mid- to late-1980s: There was a flourish of research on highly structured interventions such as DTT. Studies showed the followed advantages and disadvantages.
  - Advantage of ABA based approach: The strategy was effective in teaching skills among children with Autism.

o Disadvantages of ABA based approach:

1. Children had problem in generalization of acquired skills across multiple environments and circumstances
2. Many children started having challenging behaviours aimed at task escape/ task avoidance.
3. Lack of spontaneity and overdependence on prompts was observed often.

Developmental approaches to Autism therapy:

- 1980s and 1990s: Research on early developmental learning processes involved in communication, language, and social learning emerged. It was clearly demonstrated that young children with Autism were also found to follow the same developmental trajectory as typically developing child. Discrepancy between the highly structured teaching strategies used in DTT and the early childhood development and learning was demonstrated.
- Emphasis on incorporation of developmental principles and sequences in early autism treatment: Studies that focussed on building the pre-linguistic skills like joint attention, imitation, social engagement were done and it was realised that they were pivotal for language development. Role of positive affect and a conducive social environment for developing the foundations of communication was realised.
- Further research showed children learn best when they are engaged as active participants, in developmentally appropriate learning experiences, and in contexts meaningful to the child. This led to the advent of Naturalistic Developmental Behavioural Interventions
- It is well-known that children follow regular developmental sequences in virtually all developmental domains and learn most easily the skills that are just beyond their present knowledge. Research showed that rather than arbitrary goals, choosing targets from the zone of proximal development helped the child master a skill better and in a more meaningful manner.

Characteristics of NDBIs [2]:

- NATURALISTIC: Learning is incorporated into everyday life. This facilitates children's adaptive functioning in natural contexts and environments. Rewards used are natural and child-preferred- eg: saying car is rewarded by allowing access to a toy car rather than giving a cookie
- DEVELOPMENTAL: NDBI is firmly grounded in principles and science of learning. After assessing the present skills, targets in the zone of proximal development are chosen.
- BEHAVIOURAL: Interventions fully meet criteria as ABA techniques including (1) intervention protocols that are composed of operant teaching techniques; (2) intervention goals that are socially significant; and (3) intervention results are analyzed objectively by assessing a child's progress before, during and after the intervention. The three-part contingency, antecedent-behaviour-consequence, is utilized in all NDBIs.
- Constructivist approach is taken- child is actively engaged and new skills are built on existing knowledge. Child initiative and spontaneity are fostered and rewarded- affectively rich social interactions involving play with both people and objects
- Clear procedures are carefully described in the intervention manuals. Fidelity of implementation assessments are to ensure integrity of treatment implementation.
- Interventions follow the child's lead- child-chosen or child-preferred activity or familiar routine. The environment set in such a way that the child must initiate or interact with the adult in order to access the preferred materials or participate in a preferred routine.

Core components of NDBI [2]:

- Nature of learning targets: skills are NOT taught in isolation- integration of across

developmental domains is ensured

- o Eg: learning a new word is linked simultaneously with using the word in social context
- Nature of learning contexts: activities contain emotionally meaningful social interactions
- o Eg: Adult-child engagement activities involving play
- Nature of development enhancing strategies: strategies are built around child's everyday routines- success in these motivates the child. Behavioural strategies like modelling, shaping, chaining, prompting, and differential reinforcement used
- o Eg: routine of wearing a shirt combined with tickle play or to improve language by engaging in social commenting (describing the activity playfully to the child)

Examples of NDBIs:

- Incidental teaching
- Pivotal response teaching
- Early Start Denver Model
- SCERTS- Social Communication Emotional Regulation Transactional Support
- JASPER- Joint Attention Symbolic Play Engagement and Regulation
- Project ImPACT (Improving Parents as Communication Teachers)
- Social ABCs- parent-mediated intervention for toddlers

NDBIs- relevance to India:

The knowledge of Western psychiatry and psychology first arrived in India with British colonialism [3]. The first report of a "child showing schizophrenic behaviour" was first mentioned in 1959 [4]. Autism-specific research has substantially increased since then and at present, there is good awareness of autism among the medical, political, and legal circles in India [3]. The average age of diagnosis of ASD in India is around 3–6 years and there is a time delay of 2 years from symptom recognition to treatment initiation in Indian children [3]. In the western world, autism is diagnosed at earlier ages which provides us with both an opportunity and a challenge, the opportunity to begin intervention much earlier in life and the challenge to design and adapt our interventions to very young children in order to achieve optimal outcomes. In the Low- and Middle-Income Countries (LMIC), where the health care resources are limited, disorder-specific interventions are delivered by trained personnel only in specialist child development centres. Majority of the patient population have access only to non-specific interventions or no treatment at all, at the primary care settings [5]. The lack of acceptable, culturally appropriate intervention modules and trained personnel for service delivery necessitates employing alternate strategies for intervention in LMIC settings. Adapting the existing intervention modules available in the high-income countries and adapting them for non-specialist delivery has been attempted and found to be effective [6].

Parents of children with ASD experience higher levels of stress in the personal and social spheres when compared to parents of typically developing children and parents of children with other neurodevelopmental disorders [3,7]. Empowering parents and including them in the affectively rich intervention strategies in NDBIs may have a positive impact on the parents and reduce the stress by enhancing coping and perceived competence.

It is in this context that NDBIs may have a wider role in LMICs. The fact that NDBIs involve home-based, parent-mediated teaching of developmentally appropriate skills in a naturalistic environment, using family-friendly behavioural strategies [2], make it probably a cost-effective strategy for early intervention for children with ASD. Home delivered parent-mediated interventions can help in addressing the delay in initiation of interventions and prepare the family for more comprehensive centre-based interventions. In times as this when the pandemic has hampered delivery of centre-based interventions, tele-health based delivery of parent mediated NDBIs would serve as a useful tool until access to comprehensive services are available.

Table showing salient aspects of some NDBIs:

Intervention	Age grp studied	Salient aspects
ESDM Early Start Denver Model **Rogers and Dawson, 2010**	12-48 months	• Based on Infant toddler learning • Targets social-emotional, cognitive, language skills • Principles of ABA used to teach goals in a play way- targets chosen on the ESDM checklist based on the developmental level of the child • Interventions: delivered at home or centre • Prescribed duration: 15-20 hrs per week therapist led in addition to parents practicing skills at home
DIR/ Floortime Development- Individual differences- Relationship model **Greenspan et al, 2006**	All age groups	• Emotion is critical to the growth of the mind and brain. Following the child's lead is the key • Nine step assessment- six core developmental capacities, and three more advanced levels of reflective thinking • Interventions: play based and delivered at home • Prescribed duration: 6-10 times a day for 20 minutes or more per day
SCERTS model **Prizant, Wetherby, Rubin & Laurent, 2007**	All age groups	• Principle: build social communication, emotional regulation and transactional support-interpersonal support embedded in the natural environment to foster socialization and emotions • helping a child become a competent and confident social communicator, while preventing problem behaviours that interfere with learning and the development of relationships • Incorporate practices from ABA (e.g., Pivotal Response Treatment, LEAP), TEACCH, Floortime, RDI, Hanen, and Social Stories
JASPER approach Joint Attention, Symbolic Play, Engagement & Regulation (JASPER) **Kasari et al, 2008**	1-8 yrs	• Joint attention, symbolic play, engagement and regulation- considered the four main targets • increase the rate and complexity of social communication • implemented by parents, teachers, and professionals • Strategies used: Modelling, Prompting, Imitating, Expanding, pacing adult language to match the child's language, adjusting play routines based on the child's interests

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Telemedicine in COVID-19 Pandemic: A New Approach in Neurodevelopmental Pediatrics

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Child Development and Guidance Clinic (CDGC)
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This year, 2020, is marked by one of the most historic pandemics the world has faced. COVID-19 has taken toll over various sectors of our society consisting schools, workplaces including daily wage jobs, hospitals, healthcare services, transport services, grocery stores etc. Globally, various modalities have been formulated and followed to combat the disease progression and its ill effects. While emergency services in hospitals have been available including many exclusive COVID-19 dedicated hospitals, a major chunk of population has faced limitation in regular care and follow up including habilitation and rehabilitation services for neurodevelopmental disorders which were previously a part of routine hospital services before the pandemic. These services initially curtailed to urgency and priority of the services required, but now they are gradually sprouting to their previous pace and capacities with advancement of precautionary advisories. To avoid risk of infection and continue providing unbarred routine medical care and follow up, the government declared guidelines for practicing telemedicine, in whichever cases it is possible [1].

World Health Organization defines telemedicine as

"The delivery of health-care services, where distance is a critical factor, by all health-care professionals using information and communication technologies for the exchange of valid information for diagnosis, treatment and prevention of disease and injuries, research and evaluation, and the continuing education of health-care workers, with the aim of advancing the health of individuals and communities." [2]

Telemedicine consultation can be provided by following methods [3]:

1. Synchronous: This includes real-time telephone or live audio-video interaction typically with a patient using a smartphone, tablet, or computer. In some cases, peripheral medical equipment (e.g., digital stethoscopes, otoscopes, ultrasounds) can be used by another health care professional (e.g., nurse, medical assistant) physically with the patient, while the consulting medical professional conducts a remote evaluation.
2. Asynchronous: This includes "store and forward" technology where messages, images, or data are collected at one point in time and interpreted or responded later. Patient portals can facilitate this type of communication between professional and patient through secure messaging.
3. Remote patient monitoring: This allows direct transmission of a patient's clinical measurements from a distance (may or may not be in real time) to their healthcare professional.

Advantages of Telemedicine [2,3,4,5,6]

1. It increases timely access to appropriate interventions along with convenience with faster access and access to services that may not otherwise be available.

2. It is cost effective and reduces efforts especially of rural patients, as they need not travel long distances for obtaining consultation and treatment.
3. Telemedicine helps in covering large geographical distances.
4. It reduces the inconvenience/impact on family and caregivers especially in case of those patients who are debilitated.
5. It can reduce the burden on the secondary and tertiary hospitals by triaging and thereby reducing overcrowding.
6. It has higher likelihood of maintenance of records and documentation. Hence, reduces the likelihood of missing out advice from the doctor or other health care staff.
7. Written documentation increases the legal protection of both parties.
8. It provides patient's safety, as well as health workers safety especially in cases of contagious outbreaks or natural disasters by aiding social distancing.
9. Unnecessary and avoidable exposure of the people involved in requiring healthcare can be avoided using telemedicine and patients can be screened remotely.
10. It makes extra working hands available to provide physical care at respective health institutions.
11. Where clinically appropriate, telemedicine is a safe, effective and a valuable modality to support patient care.
12. It helps in transmission of the summary of the patient's complaints and supplementary data including images, lab reports and/or radiological investigations between the medical professionals and patients. Such data can be forwarded to different parties at any point of time and thereafter accessed as per convenience/need.
13. No separate infrastructure is required.
14. Privacy is ensured.
15. There is real-time interaction.
16. It saves time required to assess or review the case history of the patient for in-person follow up later. Thereby reducing the time of exposure of both medical professional and patient.
17. It is useful especially in the management of chronic disorders, like autism spectrum disorder, cerebral palsy etc. After making a diagnosis and setting an intervention plan along with anticipatory guidance, any changes in the intervention plan can be easily applied.
18. If a diagnosis has not yet been made, telemedicine can still prove useful in medical specialities where external signs are important in identifying the problem, such as autism spectrum disorder, attention deficit hyperactivity disorder, behavioural problems etc. Anticipatory guidance to caregivers can be given based upon basic evaluation and intervention plan with investigations, if required.

Drawbacks of Telemedicine [3, 4, 5, 6]

1. It requires an initial financial investment in equipment, software, and telecommunications.
2. Inability to perform physical examination especially in conditions which require in-person consultation.
3. Low-quality images, videos and image sharing through communication media can significantly reduce the picture quality. Thereby, creating doubts in a finding suspected.
4. An error in communication network can create hindrance in advice given by the medical professional or some important history provided by the caregiver may be missed.
5. The need to address sensitive topics, especially if there is patient discomfort or concern for privacy.
6. Limited access to technological devices (e.g., smartphone, tablet, computer) due to poverty or lack of technical knowledge needed for telemedicine consultation by the

patient and caregiver is a major barrier.

7. Level of comfort with technology for medical professional and patients or caregivers might be an issue.

In times of outbreaks or pandemics, telemedicine is a boon for patients by reducing the risk of infection and hospital visits and aiding in continuing intervention plan and follow ups in neurodevelopmental paediatrics with support for the families from the Developmental Pediatrician and his/her team. The advantages do outweigh the drawbacks of telemedicine. Telemedicine can be an upcoming approach to deal with patients having neurodevelopmental disorders but guidelines have to be formulated further to overcome the drawbacks to provide better medical services and it might be a beginning of a new era in developmental and behavioural pediatrics.

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Clinical Case Letter
A rare cause of vision impairment in a child

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INTRODUCTION

Osteopetrosis is a genetic condition of increased bone mass, which is caused by defects in osteoclast formation and function. Both autosomal recessive and autosomal dominant forms exist. Autosomal recessive osteopetrosis present in early childhood and is more severe. It can present with a variety of clinical features; diagnosis is, therefore, often delayed, or incorrect. Patients present in childhood with complaints of bone pains, failure to thrive and growth retardation. Other clinical findings include severe anemia, hepatosplenomegaly, lymphadenopathy and thrombocytopenia. The dense, extremely brittle bones fracture easily. Involvement of the cranium can lead to optic nerve atrophy with blindness or other cranial nerve defects. The diagnosis can easily be made in most patients by radiography combined with targeted molecular genetic analysis. Hematopoietic stem cell transplantation (HSCT) can be curative in the autosomal recessive forms if given in early life before neurological involvement. We report a case of a 5-year-old girl, who presented with blindness since the age of 6 months, in whom a diagnosis of osteopetrosis was made.

CASE REPORT

5-year-old girl was brought with concerns of vision impairment noticed since infancy. She was the second child born to non-consanguineous parents. Her perinatal period was uneventful. She was noticed to have vision concerns from 6 months of age when she was responding well to parents' voices but not looking at them well. She was not reaching out for toys. She had delayed attainment of all the milestones- started walking at 2 years and started speaking bisyllables after 1.5 years of age. At presentation, she was able to walk independently inside her home, indicate her toilet needs consistently, socialize well with siblings, and speak sentences in her native language. She had never been sent to school thus far. There was no history of seizures or regression of acquired milestones in the past. There were no concerns with her sleep and behaviour. She was shown to an ophthalmologist and was detected to have optic atrophy. She was referred for MRI brain.

On examination, she was underweight and stunted. She had marked pallor. The child had proptosis with frontal bossing. She had vision impairment with searching nystagmus, and right sided lower motor neuron facial palsy. Although she was answering questions, her localization of sound was inconsistent. Some words that she spoke were unclear. Examination of the motor system was normal. Examination of the abdomen showed firm hepatosplenomegaly. The other systems were within normal limits. In view of vision impairment with organomegaly, storage disorder was considered. But lower motor neuron facial palsy did not fit in with storage disorder. The other possibility considered was Osteopetrosis, which could explain the multiple cranial nerve palsies, pallor and organomegaly. Radiographs of the long bones confirmed the diagnosis of osteopetrosis. The child had thickening and sclerosis of the skull base, increased density of the bones, bone within a bone appearance, pathological fractures, and Erlenmeyer flask deformity of the femora (Figure

1). Fundus examination showed optic atrophy. The family was given the option of genetic testing and Hematopoietic stem cell transplantation; due to economic constraints they did not opt for it. Hearing assessment showed mild hearing loss in both ears. For the vision impairment, she was started on rehabilitation measures.

DISCUSSION:

This case report highlights that Osteopetrosis can present as vision impairment. There are two major forms of Osteopetrosis based on their mode of inheritance: autosomal dominant osteopetrosis (ADO, formerly known as Albers-Schönberg disease), is usually considered an adult-onset, more benign form; whereas autosomal recessive osteopetrosis (ARO), also termed malignant infantile osteopetrosis, presents soon after birth, is often severe and fatal if left untreated [1].

The most important function of the osteoclast is to dissolve the bone mineral and degrade the bone matrix using specialized enzymes. Cell polarization and formation of the ruffled border and sealing zone is crucial to this function. Hydrochloric acid is actively secreted into the resorption lacuna thus formed, resulting in the dissolution of bone mineral hydroxyapatite [2]. Most forms of osteoclast-rich osteopetrosis are caused by defects in gene products involved in the acidification machinery. Acid secretion is dependent on two key molecules, which facilitate proton transport: the proton pump vacuolar ATPase (V-ATPase) and the chloride-specific ion channel, chloride channel 7 (CLCN-7). Homozygous mutations in the genes encoding the a3 subunit of V-ATPase (TCIRG1) and the CLCN-7 produce severe malignant osteopetrosis phenotypes. TCIRG1 mutations are responsible for autosomal recessive osteopetrosis in more than 50% of affected individuals [3]. Mutations in TCIRG1 and CLCN7 together account for nearly 70% of all patients with ARO.

Children present with a myriad clinical manifestation (Table 1). They can present with anemia, infection, hemorrhage due to marrow failure from obliteration of the marrow spaces, and organomegaly due to extra-medullary hematopoiesis. Bones are fragile and fracture easily. Dentition may be delayed. Cranial nerve entrapment neuropathies occur due to inadequate formation of the skull foramina [2]. Choanal stenosis causing obstructive sleep symptoms is also associated. ARO can be misdiagnosed as idiopathic Chiari malformation, hereditary optic atrophy, rickets, or even neonatal leukemia [1]. Thus, it is important that pediatricians, ophthalmologists, ENT surgeons and neurosurgeons who might be the first point of contact for patients with ARO, to have an adequate knowledge of the disease.

Table 1: Table showing clinical features according to the organ system

Organ system	Some common clinical features in Osteopetrosis
Endocrinology	Osteopetrorickets, Hypocalcemia
Ophthalmology	Papilledema, Ptosis, Strabismus, Optic nerve atrophy, Proptosis, Nystagmus, Retinal degeneration, nasolacrimal duct obstruction
Dental	Delay/failure of tooth eruption, caries, Osteomyelitis of the mandible
Orthopedics	Skeletal deformities, Spondylolisthesis, Fractures of long bones
Neurology/ neurosurgery	Compressive cranial neuropathies (II, VII and VIII common), hydrocephalus, Arnold–Chiari I malformation, developmental delay/regression, seizures (OSTM1 mutation), Calcifications of the basal ganglia, thalami (CAII deficiency)
Hematology	Thrombocytopenia, Anemia, Leukopenia with frequent infections, Hepatosplenomegaly, Transfusion dependence
Otolaryngology	Conductive hearing loss, Recurrent otitis media, Choanal atresia, Obstructive sleep apnea
Nephrology	Renal tubular acidosis, nephrocalcinosis, nephrolithiasis (CAII deficiency)

The mainstay of diagnosis is clinical and largely depends on the radiographic appearance of the skeleton. The classic radiological features of osteopetrosis comprise:

- Diffuse sclerosis, affecting the skull, spine, pelvis and appendicular bones
- Bone modelling defects at the metaphyses of long bones- funnel-like appearance ("Erlenmeyer flask" deformity) especially of the distal femoral and humeral metaphyses, and characteristic lucent bands
- Increased bone mineral density giving the "bone-in-bone" appearance particularly in iliac wings, the vertebrae and phalanges
- Focal sclerosis of the skull base, pelvis and vertebral end plates – "sandwich" vertebrae and "rugger-jersey" spine

Radiological features usually are diagnostic [2,4]. Pathological fractures after minor trauma occur mainly after the infantile period, and poor bone healing has been reported. Age of onset, inheritance pattern and the presence of associated features, such as neurodegeneration, mental retardation, skin and immune system involvement, or renal tubular acidosis may point to particular subtypes of osteopetrosis. Bone biopsy can distinguish between osteoclast-poor and osteoclast-rich subtypes of ARO; however, this is invasive and rarely performed [2].

Next-generation sequencing methods, especially exome sequencing, play a great role in the identification of the genetic defect in all patients with ARO. Targeted molecular genetic analysis helps in choosing the most appropriate therapy and aids ineffective genetic counselling.

Hematopoietic stem cell transplantation (HSCT) can be a curative therapy and has a high success rate in patients with osteoclast-intrinsic disease who lack neurological involvement [5].

Table 2: Classification, genetics and clinical manifestations of human ARO*

Gene	Usually presents at (years)	Haematological system	Vision impairment	CNS involvement	Life expectancy (years)	Incidence
TCIRG1	<1	Severe	Mild to severe	None to moderate (hydrocephalus)	0–10	51–53%
CLCN7	<1	Mild to severe	Mild to severe	None to severe (hydrocephalus, neurodegeneration)	0–3	13–16%
OSTM1	<1	Mild to severe	Mild to severe	Severe (neurodegeneration)	0–2	2–6%
SNX10	<1	Severe	Severe	None to moderate (hydrocephalus)	0–22	4%
PLEKHM1	1–10	None	None	None	14	2 cases
TNFRSF11A	<1	Mild	Mild to severe	None to moderate (hydrocephalus)	1–10	<1–4%
TNFSF11	<1	Mild	Mild to severe	None	1–16	<1–3%

*- Modified from Sobacchi et al [1]

HSCT using HLA-identical donors results in 73% 5-year disease-free survival. Complications include rejection, delayed hematopoietic reconstitution, venous occlusive disease, pulmonary hypertension and hypercalcemic crisis. HSCT does not necessarily reverse complications: retrospective report of HSCT in osteopetrosis showed that only 7% of survivors experienced improvement in vision, whereas 69% had no further deterioration and 25% experienced further deterioration [6]. Outcomes were better with earlier transplantation, particularly before the age of 3 months. Patients with RANKL-related ARO and those with neurodegenerative disease (all patients with OSTM1-related ARO and some with CLCN7-related ARO) are not candidates for HSCT. Life-threatening hypercalcemia, a common complication after successful HSCT in patients with ARO, can be managed with denosumab, a monoclonal antibody that inhibits RANKL and potentially normalize calcium levels [1]. Pharmacological administration of synthetic RANKL could benefit patients with RANKL-dependent ARO [5]. The severe infantile forms of osteopetrosis are associated with decreased life expectancy, with most untreated children dying in the first decade as a complication of bone marrow suppression [2].

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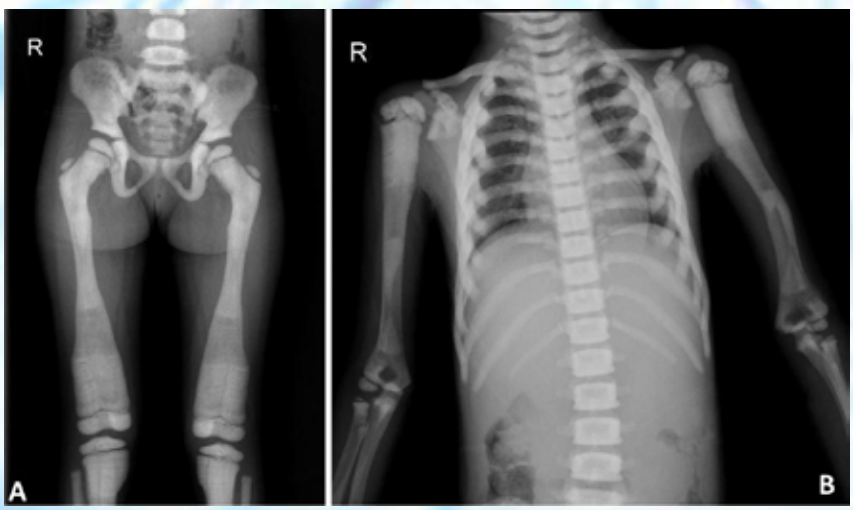

LEGEND

Fig. 1. (A) and (B) Radiographs of the long bones show increased bone mineral density, bone in bone appearance and pathological fracture of left humerus (blue arrow). In Figure 1A, the Femora show Erlenmeyer flask deformity (yellow arrow).

A Case pf Tuberous Sclerosis with Behavioral Problems and Poor Scholastic Performance

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INTRODUCTION

Tuberous sclerosis complex (TSC) is an autosomal dominant neurocutaneous syndrome with variable and heterogeneous expression affecting multiple organs and with a prevalence of 1 in 6,000 newborns [1] affecting both sexes equally [2]. Spontaneous genetic mutations occur in 65% of the cases [1]. TSC1 gene, located on chromosome 9q34 which encodes protein hamartin and TSC2 gene, located on chromosome 16p13 which encodes protein tuberin, are two identified foci for TSC. In 85% of TSC, 31% have TSC1 mutation (hamartin gene) and 69% have TSC2 mutation (tuberin gene) [1, 2]. TSC1 and TSC2 genes are tumor-suppressor genes, either of which when lost causes formation of numerous benign tumors called hamartomas [1, 3].

Although there has been great progress in the identification and treatment of many of the physical features of TSC, including subependymal giant cell astrocytoma (SEGA), angiomyolipoma (AML), and epilepsy, the neuropsychiatric manifestations remain highly underidentified and undertreated [4].

CASE PRESENTATION

An 8 years old, male, studying in second grade of an English medium school, only child born of a non- consanguineous marriage delivered at term/vaginally delivered/birth weight of 1.5 kg (intra-uterine growth restricted)/cried at birth with NICU stay for 12 days for low birth weight care and neonatal hyperbilirubinemia, was brought by his parents with complaints of behavioral issues in the form of inattention and hyperactivity reported from school and home settings with poor scholastic performance since beginning of schooling. No history of seizures was reported. Family history was not significant.

Developmental History

1. Gross motor development: Age appropriate
2. Fine motor development: Age appropriate
3. Cognitive development: Was cooperative, understood complex commands
4. Social communication development: Age appropriate
5. Speech and language: Initial delay reported; Could narrate with unclear articulation
6. Academics: Concerns reported since early schooling in comprehension and writing, needed repetition of instructions
7. Behavior: Poor attention span with hyperactivity reported from home and school settings

Examination

- His anthropometry was between 3rd to 50th percentile according to IAP growth charts.
- General examination revealed multiple (08) hypomelanotic macules over his body with normal systemic examination.

Summary of Psychoeducational Assessments

- His intelligence quotient (IQ) was 88 indicating low average intelligence. His verbal IQ was 82 and performance IQ was 93.

Verbal Scale		Performance Scale	
Domain	IQ	Domain	IQ
Information	80	Picture completion	70
General comprehension	90	Block design	125
Arithmetic	80	Object assembly	62
Analog and similarity	90	Coding	94
Digit span	67	Mazes	113
Vocabulary	85		

- Social maturity by Vineland Social Maturity Scale was adequate (Social Quotient=95).
- He could not perform corresponding to his grade in battery of reading and spelling, graded reading and listening comprehension tests, graded mathematical test and expressive language.

Test	Result	Inference
Schonell's Graded Word Reading Test	Reading Age (RA)= 6 years 10 months	1 year 2 months below chronological age (CA)
Schonell's Graded Word Spelling Test	Spelling Age (SA)= 5 years	3 years below CA
Graded Reading Comprehension Test	Below the level of grade I	Not grade appropriate
Graded Listening Comprehension Test	Below the level of grade I	Not grade appropriate
Expressive Language	Not grade appropriate	Not grade appropriate
Graded Math Achievement Test	Not grade appropriate	Not grade appropriate

- Conners-3 ADHD rating scale reported by parents showed moderate to severe dysfunction in domains of Inattention, Hyperactivity/Impulsivity, Learning problems, Executive functioning, Defiance/Aggression with normal scores in peer relations. Teachers rating report could not be taken in view of schools being closed and was planned to be taken later.

Other Investigations

- Ophthalmology reference-Within normal limits
- Audiology evaluation-Within normal limits
- Magnetic Resonance Imaging (MRI) was done in view of the hypopigmented macules to look for TSC and the findings were consistent with findings suggestive of TSC.
- A genetic diagnostic testing was offered to the patient but they could not do it for economic reasons.

Management

A multidisciplinary intervention plan with goals was counseled to parents constituting remedial teaching for his academic issues, behavioral therapy to be carried out with the child and the parents (medication plan for ADHD/behavioral issues had been kept for later plan), speech and language therapy for misarticulation, occupational therapy for inattention and graphomotor issues. Regular follow up with Developmental Pediatrician (to monitor and review goals for the intervention 3 monthly), Pediatrician, Neurologist and Ophthalmologist (to follow up and monitor features and progression for the TSC) was advised.

DISCUSSION

TSC was first observed by Von Recklinshausen in 1862, but the first clear description of TSC was given by Desire-Magloire Bourneville in 1880, who recognized the pathological features of white tumors or tubers and areas of sclerosis of cerebral gyri at post-mortem in patients with epilepsy and mental retardation [2, 5].

The hallmark of TSC is the involvement of central nervous system. The cognitive and behavioral problems are of greatest concern to parents and caregivers. Approximately 50% of the individuals

diagnosed with TSC present with epilepsy, cognitive impairment and developmental psychopathologies including autism spectrum disorder. Those with normal intellectual abilities are also at high risk of specific neuropsychologic deficits and behavioral, learning, and other psychiatric disorders [1, 4]. An IQ in the normal range of intellectual ability is present in 40–50% patients with TSC [4]. TSC during infancy may present with infantile spasms. The characteristic brain lesion is a cortical tuber. Subependymal nodules are found along the wall of lateral ventricles where they later calcify and project into ventricular cavity, producing a candle-dripping appearance which can later grow into subependymal giant cell astrocytomas (SEGAs). These tumors can grow and block the circulation of cerebrospinal fluid around the brain causing hydrocephalus [1].

Retinal lesions consist of hamatomas and white depigmented patches found in 50% to 80% patients [5]. Other retinal findings include retinal pigmentary disturbance ranging from hyperpigmented areas to "punched out" hypopigmented areas at the posterior pole or mid periphery. Non-retinal findings include angiofibromas of the eyelids, coloboma of the iris, lens and choroid, strabismus, poliosis of eyelashes, papilloedema and sector iris depigmentation [5].

Typical hypomelanotic patches over trunk and extremities are found on 90% of patients. Shagreen patch located mostly in lumbosacral region is also a hallmark of TSC. During adolescence or later, 15-20% of TSC cases may develop small fibromas or nodules around fingernails or toenails. Facial angifibromas develop by 4 to 6 years of age which are small red nodules over the nose and cheeks [1].

Approximately 50% children with TSC have cardiac rhabdomyomas which may cause congestive cardiac failure or arrhythmias. In children older than 10 years of age, angiomyolipomas may develop in kidneys in 75-80% cases, which by third decade may cause lumbar pain, hematuria or rarely, retroperitoneal bleeding [1].

Definite TSC is diagnosed when at least two major or one major plus two minor features are present [1, 5].

Major Features of TSC	Minor Features of TSC
1. Cortical tuber	1. Cerebral white matter migration lines
2. Subependymal nodule	2. Multiple dental pits
3. Subependymal giant cell astrocytoma SEGA)	3. Gingival fibromas
4. Facial angiofibroma or forehead plaque	4. Bone cysts
5. Ungual or periungual fibroma (non-traumatic)	5. Retinal achromatic patch
6. Hypomelanotic macules (>3)	6. Confetti skin lesions
7. Shagreen patch	7. Non-renal hamartomas
8. Multiple retinal hamartomas	8. Multiple renal cysts
9. Cardiac rhabdomyoma	9. Hamartomatous rectal polyps
10. Renal angiomyolipoma	
11. Pulmonary lymphangioleiomyomatosis	

Our patient had three major features of TSC with low average IQ, behavioral issues and academic difficulties.

CONCLUSION

TSC affects many organ systems other than skin and brain including the heart, kidneys, eyes, lungs and bones. Cardiac, renal and cerebral pathologies are major causes of mortality. Sudden deaths may be seen following cardiac arrhythmias, intractable epilepsy, intracranial hemorrhages, obstructive hydrocephalus, aneurysm rupture and spontaneous pneumothorax. Subependymal tubers may convert into giant cell astrocytoma in brain causing obstructive hydrocephalus [1, 2]. Treatment for TSC is mainly symptomatic.

Multiple systemic involvement renders regular follow ups and screening namely by developmental pediatrician, neurologist, ophthalmologist and dermatologist along with brain MRI in every 1-3 years, renal imaging in every 1-3 years and neurodevelopmental testing annually with regular follow ups to monitor progress [1, 2].

In view of this case presenting with academic concerns and behavioral issues in a developmental clinic it is essential that a detailed neurological examination be carried out in each and every patient presenting with poor scholastic performance and/or behavioral concerns.

The surveillance and management recommendations for TSC (2012) advise to screen and assess for the behavioral and neuropsychiatric symptoms under the terminology TAND.

TAND is a terminology proposed to describe the interrelated functional and clinical manifestations of brain dysfunction common in TSC, including aggressive behaviors, autism spectrum disorders, intellectual disabilities, psychiatric disorders, and neuropsychological deficits as well school and occupational difficulties [6]. These include the behavioral level (observed behaviors such as sleep problems or aggressive behaviors), the psychiatric level (DSM/ICD defined psychiatric disorders such as autism spectrum disorders (ASD) or attention deficit hyperactivity disorder (ADHD), the intellectual level [intellectual ability as defined by intelligence quotient (IQ)-type tests], the academic level (learning disorders, e.g., reading or mathematics difficulties), and the psychosocial level (e.g., self-esteem, family difficulties) [4].

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'My Child was Born Small Hence is Still Small'

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Primordial dwarfism (PD) is a disease in which severely impaired fetal growth persists throughout postnatal development and results in stunted adult size. The condition is highly heterogeneous clinically, but the presence of certain phenotypic aspects such as head circumference and facial appearance differentiates between them. These disorders share similar characteristics including skeletal malformation (dysplasia), growth deficiency before birth (intrauterine growth retardation) and during infancy and childhood, ultimately resulting in varying degrees of short stature. This group of disorders currently includes five disorders: Seckel syndrome, ear-patella-short stature (Meier-Gorlin) syndrome; Russell-Silver syndrome; Majewski osteodysplastic bird-head dwarfism type I/III; and Majewski osteodysplastic bird-headed dwarfism type II.

This is a case of 5 years 8 months years old male child, first born to second degree consanguineous parents who presented with development delay and poor weight gain. She complaints that child was always slow in development and was not gaining adequate weight despite normal eating.

Antanatal Risk: Gestational HTN was noted at 7th month – controlled on medication. Growth scan- severe growth restriction (30 weeks). Reversal of end diastolic flow (36 weeks).

He was delivered Late Preterm(36 wk) / Emergency LSCS (Doppler abnormality) / 1.4 Kg cried at birth/ HC- 29.5cm Length- 37 cm Ponderal index- 2.7 (Symmetrical IUGR)

Postnatal events: He was admitted in NICU for 5 days (LBW)- no respiratory difficulty/ feeding issues (suck was good)/ seizures/ abnormal activity. Child was discharged on mixed feeding (formula and DBF). No feeding issues / excess sleep/ lethargy

Child did not thrive well even with adequate feeding

Mother had noticed that the child was not smiling at her at 4 months of age, further all the milestones were delayed. History was suggestive of Global Development Delay with current Gross motor age (3 years), Fine motor age (2.5 years), Social age (2.5 years) and language development age (3 years). He has no difficulty watching TV / happy to watch/ dances in response

Play: Child has imaginative play. Group play with peers- involving running- but slow in running/ No frequent falls/ abnormal gait. Does not understand game rules

Child attended Anganwadi for 3 years (from1 -4 years) where the child mostly played by himself. Currently, child is not sent to school as mother perceives that going to school would make him lose more weight.

Activities of daily living: Child cannot self feed/ dress/attend toilet needs but cooperates.

Child was always shorter and leaner than peers. Child has not caught up growth after being born small. No history suggestive of difficult feeding malabsorption/ chronic infection/ abnormal neurological activity

Mother perceived that child was being small because he was born small and will catch up.

Clinical examination:

Child is alert/ active/ playful. He is an easy child, secure attachment with mother. Participated in conversation- spoke in words/ Speech is clear

Anthropometry:

Wt- 9.7 kg(< 3rd)

Ht- 78 cm(<3rd).

US: LS= 1.3

HC- 41.5 cm(< -3 SD)

Poorly built with proportional short stature and microcephaly

Dysmorphic facial features - Microcephaly with prominent metopic suture

With forehead recession

AF closed – no sutural ridging

Prominent eyes- Rt divergent squint/ no cataract/ no facial asymmetry

Low set ears

Prominent nose- sharp beaked

Microtia- rounded mouth/ micrognathia/ Retrognathia

Genitals – normal / male – penile length appears normal. B/l testis normal

Upper limbs – clinodactyly of B/L little fingers

Lower limbs - normal

Systemic examination

CNS: Mild hypertonia (Grade 1+) in all limbs with brisk reflexes and plantar withdrawal response. Power normal. Rest systemic examination is normal

Development assessment:

TDSC – delay (3-6 yrs chart- no items performed)

(0-3 yrs chart- 7 items not performed)

LEST- delay

DDST: suspect all domains

IQ- could not be assessed

VSMS- 2 y 6m (SQ= 48)

Global developmental delay with Primordial dwarfism- Proportionate short stature (IUGR) with microcephaly with dysmorphism

Hence genetic cause was suspected and genetic analysis was done

Karyotyping – 46 XY

Whole exome sequencing- SCKL1 (ATR gene mutation on chromosome 3q 23)- suggestive of Seckel's Syndrome

Management:

Immediate:

Calorigenic diet/ deworming

Vision and hearing testing

Thyroid profile

Neurologist opinion

MRI- to look for cranial anomalies (Lissencephaly)

Long term:

Training for Activities of Daily Living

Therapy for gross motor

Speech therapy

Squint- ophthalmologist opinion and therapy

Inclusive education

ID certification

Antenatal screening of mother and growth monitoring in further pregnancy- risk of recurrence was explained pertaining

Yearly check on CBP (Aplastic anemia surveillance)

Discussion:

Seckel's syndrome is also known as Bird-Headed Dwarfism, Seckel Type

Microcephalic Primordial Dwarfism, Nanocephalic Dwarfism, Seckel Type Primordial Dwarfism

Seckel syndrome is an extremely rare autosomal recessive disorder characterized by prenatal and postnatal growth retardation and delayed bone maturation. Craniofacial abnormalities are microcephaly, varying degrees of mental retardation, a hypoplastic face with a prominent nose, and low-set and/ or malformed ears "beak-like" nose, large eyes, a narrow face, malformed ears, micrognathia. Skeletal abnormalities include radial dislocation, dysplasia) of the hip, kyphoscoliosis, clubfoot 11 12 pairs of ribs. Approximately 25% of patients have aplastic anemia or malignancies. Lissencephaly is the most commonly associated brain anomaly. There is broad genetic heterogeneity comprising 7 classifiable types: SCKL1, ATR mutation; SCKL2, RBBP8 mutation; SCKL3, maps to 14q21-q22; SCKL4, CENPJ mutation; SCKL5, CEP152 mutation; SCKL6, CEP63 mutation; and, SCKL7, NIN mutation.

Syndromes with IUGR + Short stature + low IQ

1) Cornelia de Lange IUGR - Failure to thrive (rule)/ Retarded osseous maturation / Hypertonia /Thick eye lashes/ Nasal bridge depressed

2) Russell Silver Syndrome- IUGR/Triangular facies/ Relative macrocephaly (prominent head)

3)Seckel syndrome- IUGR/ Postnatal growth decline/ Receding forehead with prominent beaking nose

Conclusion:

Seckel syndrome has proportionate short stature, which differentiates it with short-limb dwarfism. In this case, antenatal reports showed Gestational Hypertension at 32 weeks and REDF at 36 weeks. Retarded intrauterine growth was noted at 30 weeks. Child had on early onset fetal growth reduction due to the syndrome and probably late onset reduction due to Gestation HTN (as indicated by REDF). Genetic counselling explaining the risk of occurrence in future pregnancy, prenatal diagnostic techniques and the role of ADL skill training are of utmost significance.

Take home message: Genetic disease has to be considered as a differential diagnosis to early onset IUGR. Role of serial growth monitoring has a great role.

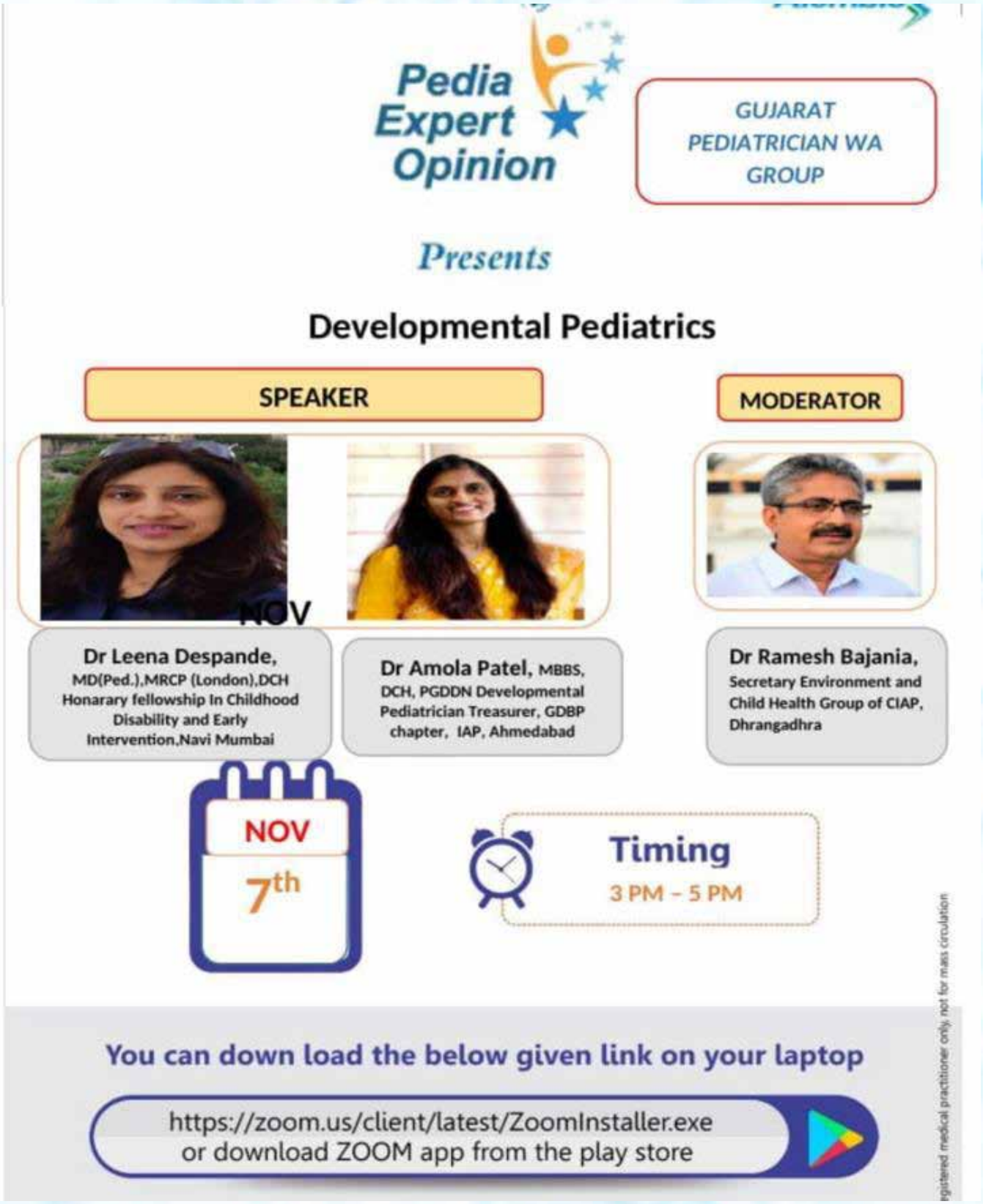
Month in pics

Celebrating seven years of our parent based intervention program which has growl in leaps and bound giving us enormous amount of knowledge andoppurtunity to look after these children

Month in pics

Sangamitra continues with online sessions. But we have not missed out the fun we had, we still continue... glimpse@ todays fancy Friday child and mom together with their creative ideas

Month in pics



Month in pics

cloudnine
#WeKnowMoms

WEBINAR on

Cerebral Palsy and Autism

Date: Thursday, 5th November, 2020 | Time: 5 pm - 6 pm

TIME	TOPIC
5 pm - 6 pm	Cerebral Palsy and Autism

MODERATOR

Moderator -
Dr. Ritu Jain
Obstetrics & Gynaecology

PANELISTS

Panelist -
Dr. Chetna Jain
Obstetrics & Gynaecology

Panelist -
Dr. Sanjay Wazir
Neonatologist &
Pediatrician

Panelist -
Dr. Shreyasi Sharma
Foetal Medicine Specialist

Panelist -
Dr. Suboohi Rizvi
Obstetrics & Gynaecology

Panelist -
Dr. Promila Malik
Obstetrics & Gynaecology

Panelist -
Dr. Puja Kapoor
Pediatric Neurology

Panelist -
Dr. Gopal Agarwal
Neonatologist &
Pediatrician

Panelist -
Dr. Mohd Aamir
Neonatologist &
Pediatrician

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Month in pics

" KNOW US WELL-CHILDREN with DOWNS SYNDROME "
Initiative by IAP Delhi in association with IAP North Delhi & DSPS (Downs Syndrome Parents Society)

21 st November;2020; 2 pm to 5 pm

Organising Chairperson : Dr RK Nabh

Organising Secreteriat :

Dr Smrita Mishra, Dr Manish Gupta, Dr Dinesh Mittal, Dr Shalini Goel

Timmings	Talk	Speaker	Chairperson
2.-2.15 pm	Inauguration	TD Dhariyal Dr Rekha Ramachandran Dr Piyush Gupta	Dr RK Nabh Dr Smrita Mishra IAP Delhi
2.15-2.30 pm	Overview-Children with Downs Syndrome	Dr Manish Gupta Pedaitrician,JGH	Dr GP Kaushal Ambedkar Hosp
2.30 -2.55 pm	Antenatal Diagnosis & Counseling	Dr Ratna Dua Puri Genetics Specialist SGRH	Dr PS Narang Max Hosp
2.55-3.20 pm	Children with Downs – Parents Corner	Rashmi Joshi ,DSPS Deepti Mathur ,DSPS	
3.20-3.45 pm	Cardiac Issues –Optimal care	Dr Smrita Mishra Paed Cardiologist Manipal Hosp	Dr Poonam Sidana Max Hosp
3.45-4.10	Intellectual Disabilities in Children with special focus to Downs Syndrome	Dr Shefali Gulati HOD;AIIMS Pead.Neuorolgy	Dr Arun Garg Swastik Hosp
4.10-4.35 pm	Early intervention- Need of the Children	Dr Lata Bhat Developmental Peadiatrician	Dr RK Nabh President IAP Delhi
4.35-5 pm	Panel Discussion-Orthopaedics, Endocrinology, Neurology, Cardiac issues	Dr Anurag Agarwal, Dr Nargesh Agrawal Dr Ravinder Dr Smrita Mishra Dr Ratna Dua Puri Dr Shefali Gulati Dr Lata Bhat	
5 pm	Vote of Thanks		

Dr Lata Bhat gave talk on

" Early Intervention in Down Syndrome "

in a workshop on Down Syndrome organised by Delhi IAP

in association with IAP North Delhi on 21St November 2020

Month in pics

SILVER JUBILEE CELEBRATION OF CHILD CARE CENTRE & DISABILITY WEEK CELEBRATION

29-11-2020 6:30 pm

Talk on

Attention Defecit Hyperactivity Disorder

Speaker
Dr Chhaya S Prasad
Developmental & Behavioral Pediatrician
Director ASHA, Chandigarh

Moderator
Dr Jeeson C Unni
HOD Pediatrics Aster Medcity

Zoom
Meeting ID: 845 7847 0947
Passcode: IAPCOCHIN

Dr Pramod Wariyar
President IAP Cochin

Dr M Narayanan
President IAP Kerala

PROTECT CHILD RIGHTS · IAP KERALA 2020

Dr Sajith John
Secretary IAP Cochin

Dr Balachandar D
Secretary IAP Kerala

Month in pics

IAP CCB

Cordially invites you to join a webinar on

Child Development & Behaviour Issues

Date : 29/11/2020

Time : 4 pm - 6 pm

Dear Members,

GREETINGS GROM IAP CCB!

Join us for an enriching discussion on Child Development & Behaviour Issues.

TOPICS & EXPERTS

1. Drawing the line... Normal vs Abnormal Hyperactivity/Speech delay/Stuttering/Behavioural issues
2. A panel discussion on Challenges in convincing Parents for ASD management
3. Challenges in managing kids during lockdown -
4. Brush up on the new guidelines
   a) Sleep hygiene
   b) Screen hygiene

Moderator

Dr. P Sudhakar
Developmental Paediatrician

Speaker

Dr. A. Somasundaram
Developmental Paediatrician

Panelist

Dr. S. Subramanian
Developmental Paediatrician

Panelist

Dr. Venkateswaran
MD FICP (CMC,Vellore)
Consultant Child and Adolescent Psychiatrist

Panelist

Ms. Gita Srikanth
Founder Director of WeCAN,Chennai

Office Bearers

Dr. Annamalai Vijayaraghavan
President IAP, CCB

Dr. Sudhakar
Secretary, CCB

Dr. S. P. Karamath
Treasurer, CCB

Touch the link below to access the webinar:

zoom https://microlabs.zoom.us/j/98040353170 | https://youtu.be/elulHUWGUWg YouTube

Month in pics

CALL FOR PAPERS & POSTERS FOR ENCDP 2020

CALL FOR PAPERS & POSTERS FOR ENCDP 2020

CALL FOR PAPERS & POSTERS FOR ENCDP 2020

PAPER/POSTER PRESENTATION eNCDP 2020

Abstracts of scientific papers are invited for the e-NCDP 2020 (29-30 Dec 2020).

Authors need to send only the abstracts initially to eNCDP2020@gmail.com.

Paper category- Oral paper/ poster

The papers will be considered for awards under 2 sections according to the researchers/ presenting authors-

1. Pediatricians and Pediatric Postgraduate students
2. Allied health professionals

The presenting author must register for the conference.

Page 1 (Title page of the abstract) should include -

- Full name of the presenting author
- Age of the presenting author
- Registration No. of presenting author
- Department & Institution with address
- Designation
- Email
- Mobile No. and Other Contact Nos
- Title of the paper
- Author List (Give full names of all authors)
- Category-Oral Paper/poster
- Section-Pediatrician or PG/Allied health professionals
- Abstract Word Count

CALL FOR PAPERS & POSTERS FOR ENCDP 2020

Main abstract (Page 2)

- Abstract topic should be based on /related to "Developmental Pediatrics".
- Word limit- The main body of the abstract should not exceed 250 words. It should be typed with single line spacing in Times New Roman (font size 12) in word format. Abstract should not include tables/graphs/images.
- The abstract should be structured as far as possible in the following manner (Except Case Reports):
  - (a) Introduction
  - (b) Aims & Objectives
  - (c) Material & Methods (including statistical methods where relevant)
  - (d) Results
  - (e) Conclusions.
- Mode of submission: e-mail the abstract to eNCDP2020@gmail.com as an attached word document. No other forms of submission like hard copies, fax etc. shall be accepted
- The last date for submission of the abstracts is **20th Dec 2020**. This deadline will be strictly observed.
- The abstracts will be accepted as oral papers/poster paper depending on its merits. Case reports will be accepted as Posters ONLY. The Expert Committee will review the abstracts and decide the mode of presentation- poster or oral. The decision of the committee will be final in deciding the mode of presentation. Notification of acceptance of abstract submitted will be sent by e mail only to the presenting author on the e mail as given by the authors. Presentations: All presentations must be in English. Detailed instructions for poster/ oral paper presenters will be sent after the selection process is complete.

CALL FOR PAPERS & POSTERS FOR ENCDP 2020

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- Section-Pediatrician or PG/Allied health professionals
- Abstract Word Count

Membership Form

Indian Academy of Pediatrics
Chapter of Neuro Developmental Pediatrics
Membership Application Form

(Please fill in CAPITAL LETTERS; All Information Mandatory; Pl do not leave any blank spaces)

1. Surname: ..... First Name: ..... Middle Name: .....
2. Date of Birth .....
3. Central IAP Membership Number (For Pediatricians Only) : .....
4. Permanent address .....
   .....
   .....
5. Office Address .....
   .....
   .....
6. Email: ..... Landline Telephone: .....
7. Mobile Phone Number (1) .... (2) .....
8. Present Work Status: Private ..... Govt. ..... Medical College ..... Voluntary Agency .....
9.

Qualifications	Name of University	Year of Passing
MBBS		
MD Pediatrics		
DCH		
DNB Pediatrics		
Others		

10. Areas of Interest of Work .....

P.T.O.

Membership Form

11. Membership Subscription:
    a) Life Membership for Central IAP Members – Rs 1500
    b) Life Associate Membership for Doctors other than Pediatricians – Rs 1500
    c) Life Affiliate Membership for All Other Professionals – Rs 1500
12. On online transfer please e-mail the scanned form with transfer details to cdgiap@gmail.com with cc to kawaljit000@gmail.com

NAME OF ACCOUNT – **IAP CHAPTER OF NEURO DEVELOPMENTAL PEDIATRICS**

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ACCOUNT NUMBER 16860100040046

IFSC CODE – FDRL0001686

Signature of the Applicant with date:

For Office Use Only

Membership No .....

Particulars of the receipt: Cheque / D.D No ..... Bank .....

Amount ..... Date .....

Mailing address:

Dr K.S. Multani
Hon' Secretary
IAP chapter of Neuro Developmental Pediatrics Department of Pediatrics
9 Air Force Hospital
Air Force Station Halwara, Ludhiana (Punjab) 141106
Cell no : 8472087960
Email: kawaljit000@gmail.com