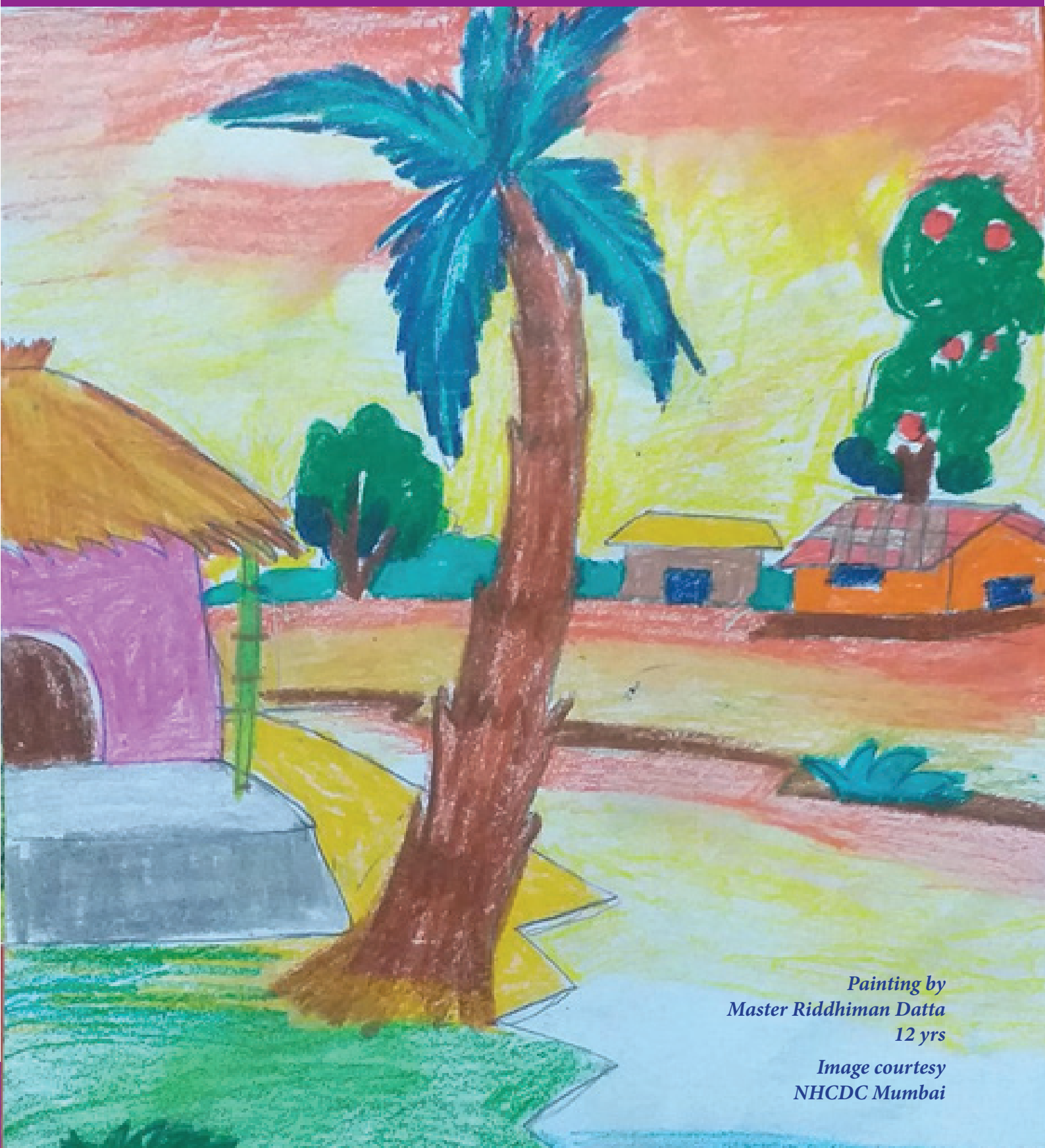
MARCH 2024

# DPT

## DEVELOPMENTAL PEDIATRICS TODAY

Your monthly booster for developmental and behavioural news!

*The Official Newsletter of the IAP Chapter of Neurodevelopmental Pediatrics*



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# CENTRAL IAP OFFICE BEARERS 2024


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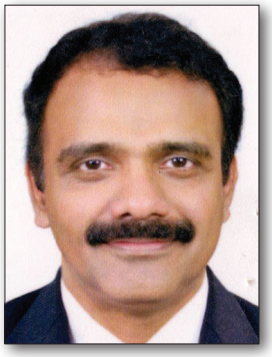

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| 2006-2010 | Dr Abraham K Paul | Dr SS Kamath |
| 2010-2013 | Dr SS Kamath | Dr Samir H Dalwai |
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| 2019-2023 | Dr Shabina Ahmed | Dr KS Multani |
| 2024 | Dr Leena Srivastava | Dr Shambhavi Seth |

# NATIONAL CONFERENCES OF THE CHAPTER

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| 1 | 2004 | Bhubaneswar | Dr J Sarangi, Dr Arabindo Mohanty |
| 2 | 2005 | Hyderabad | Dr Hanumantha Rao |
| 3 | 2006 | Cochin | Dr Abraham K Paul, Dr S S Kamath |
| 4 | 2007 | Bangalore | Dr Nandini Mundkur |
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| 17 | 2020 | eNCDP | Dr KS Multani |
| 18 | 2021 | eNCDP 2.0 | Dr KS Multani |
| 19 | 2022 | Kolkata | Dr Atanu Bhadra |
| 20 | 2023 | Guwahati | Dr Shabina Ahmed |

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**Dr Kawaljit Singh Multani**

**Dr Leena Srivastava**

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**Dr Anshuman Verma**

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# Interactions – The Editorial

Dr Samir H Dalwai

It is heartening to see Developmental Behavioural Pediatrics fast becoming one of the most popular sub specialties in the field of pediatrics in India today. The IAP and the IAP Chapter of Neurodevelopmental Pediatrics have played a stellar role in this progress. And the monthly newsletter of the Chapter, Developmental Pediatrics Today (DPT) has served an important role in taking this to the pediatricians.

Going ahead in 2024, yours truly has the responsibility of taking the DPT to 44,000 plus pediatricians of the IAP and I am grateful for the honour. While accepting this privileged task, one feels the need to adapt the DPT to stand out and compete for the pediatricians attention amidst a maze of information s/he is inundated with. And hence, the DPT will be more mindful of the content which will be specially curated for our pediatricians across the length and breadth of our country. You will be delighted to find an entirely new concept – your DPT will reinforce developmental and behavioural pediatrics in a witty, pleasant and an alluring manner. You will find interesting titles as well as interesting perspectives, news from the world of developmental pediatrics, reviews from the IAP Handbook of Developmental Behavioural Pediatrics, challenging interactions in the form of quizzes and contests, news from the Chapter members across the States, a parenting corner and enough space for you to contribute your thoughts and opinions. We welcome you to peruse this inaugural issue of 2024 and write in to us as a contributor. The DPT will be the voice of each and every pediatrician.

# Joint Attention - From the Office

Greetings of the new year to all!

As we embark on this journey for the new year with the members of the chapter we have one common focus and that is steps towards better more comprehensive care for our children with developmental and behavioral problems. Another aspect we hope to include is preventive aspects and prophylaxis in developmental pediatrics too.

As the chair of the new committee steering the chapter ahead a sense of responsibility and enthusiasm to excel is in our minds in planning our academic agenda for the year. The salient points of the plan will include-

Academic updates and research opportunity

Better awareness in all strata- health care workers to the community

Congenial environment for communication and participation amongst the chapter members

The glorious past of the chapter under stalwarts of the field has sown the seeds to help the chapter grow and pave the way ahead for academic excellence and advances in screening and interventional practices customised to our cultural context . The knowledge and practice needs to be in sync to give way to indigenous best practices in this field of developmental and behavioral pediatrics where ambiguity of approach and therapies should not be an excuse to poor outcomes.

This year and ahead the chapter hopes to bring together Pediatricians working in this field to create a unified and strengthened knowledge basis of child development and it's atypical patterns to help achieve better health both physical and mental for our children.

The academic milestone with the official journal of developmental and behavioural paediatrics that has been initiated recently. Being the first in the specialty in this part of the world it would give an opportunity to many to publish good indigenous research in related topics.

The DPT has been a newsletter of the chapter for quite a few years now and has received lot of positive feedback for keeping the members abreast of the updates in the field along with relevant activities across the country. January's health awareness theme is 'Birth defect prevention' and 'Thyroid awareness' both powerful topics finally contributing to better developmental outcomes.Thus this month's DPT is also dedicated to the same.

We hope the year ahead is promising and successful for all of you. We as a chapter hope to move in the right direction working towards enhancing the care contributing to better and positive outcomes in the development and behaviour of our children.

***Coming together is a beginning, keeping together is progress , working together is success...***
***- Henry Ford***

**Dr Leena Srivastava**

# Joint Attention
# Message from Secretary

The Indian Academy of Pediatrics (IAP) chapter of neurodevelopmental pediatrics has been bustling with exciting activities and initiatives. New team has recently taken up responsibilities, bringing fresh perspectives and enthusiasm to the chapter's endeavors. Their dedication and expertise promise a vibrant future for the field. As Secretary of Chapter for 2024-25, I thank the outgoing team for their exemplary leadership, dedication, and contributions.

One of the highlights of the chapter's recent activities was the successful organization of a symposium and General Body Meeting (GBM) at Pedicon in Kochi. The symposium provided a platform for professionals to exchange knowledge, discuss the latest developments in neurodevelopmental pediatrics. The GBM served as a forum for members to deliberate on important issues, share updates, and chart the course for future initiatives.

The chapter has also been actively involved in enhancing education and training in neurodevelopmental pediatrics. Collaborating with our 17 fellowship centers across the country, we have developed a module-based teaching program that offers comprehensive and structured learning experiences for aspiring professionals on weekly online teaching platform.

In a significant step towards strengthening their outreach, the chapter has appointed state coordinators and established five zonal coordinators. This decentralized approach enables them to engage with local pediatric communities and understand their unique challenges. This grassroots-level engagement is crucial for raising awareness, promoting early intervention, and advocating for the needs of children with developmental disorders.

As secretary of chapter of neurodevelopmental I congratulate Dr. Samir Dalwai and his editorial team for their outstanding efforts in revamping DPT (Developmental Pediatrics Today) newsletter. Their dedication and vision have revitalized this important communication platform, providing valuable insights, updates, and resources to the community.

Best Wishes !

**Dr Shambhavi Seth**
Secretary
IAP Chapter of Neurodevelopmental pediatrics
2024-25

# Social Communication
# From the States, State Coordinators


World Down syndrome Day celebrated on 21 March 2024 by nationwide awareness activities organized by chapter in more than 15 states. (Pictures follow)


Down's Syndrome Day
21st March 2024
KOLHAPUR ACADEMY OF PEDIATRICS
IAP Branch Kolhapur


SILIGURI ACADEMY OF PEDIATRIC
MOTHER CARE CENTRE & YOUNG INDIANS
celebrates
WORLD DOWN SYNDROME DAY
Theme : END THE STEREOTYPES


Parents Support Group Meeting
on the occasion of
World Down Syndrome Day
21 March 2024


Down's Syndrome Day
21st March 2024
KOLHAPUR ACADEMY OF PEDIATRICS
IAP Branch Kolhapur 2024


DEPARTMENT OF PEDIATRICS
MALDA MEDICAL COLLEGE & HOSPITAL
WORLD DOWN SYNDROME DAY
MARCH 21, 2024
Shot on OnePlus


WORLD DOWN SYNDROME DAY

# Speak up! Interesting Cases

## - Dr Samir H Dalwai

***A 2 month baby bitten by brother 2 years old brother, how would you counsel the parents and brother? Should Pediatricians refer the older sibling; if so, to who?***
***(Contributed by Dr Sankhe, Senior Pediatrician, Tarapur)***

As rightly said, besides managing this baby (wound management, vaccination, etc), it is vital to look at the sibling and parents- not too difficult for a pediatrician at all.

The approach could be :

1. Who has done this? Parent's may say that it's the sibling - there won't be any evidence unless there was a camera installed in the room. (No one will take videos while this is happening) but Battered Baby or inadvertent fall, etc are a possibility to keep in mind. Was there a history of suspicious injury in the older sibling ever? Is there a nanny in the house?

2. If adults ruled out, History of the older sibling. Pre natal, natal, post natal, early milestones esp social behaviour and communication domain, behaviour (esp temper tantrums, soothing, aggressive acts, self harm, with peers ) will give straight forward idea into the child's developmental status and situation

3. most imp- Parenting patterns - how do they handle the older child's behaviour, does he get time with parents, screen time violence, domestic violence.. also to rule out child abuse in the older sibling.

In most of the above, speaking to the parents will help. Guiding the parents to spend more time with the child and being more patient is the most important advice. Follow up if they don't get it the first time.

If they don't listen to you, unlikely "child counselling" or "sensory integration therapy" will help the child. The outcome won't change.

Pediatricians know the family the best and all of this is entirely within their domain. Sub specialists will find something "abnormal" in their field and we will be under a false impression that the problem is solved.

No one knows the child better than pediatricians. Unfortunately, these aspects were ignored during our Residency training for infectious diseases neonatology intensive care etc and rightly so because that burden of disease was far higher then. Now it is reversing and we just need to pay more attention to these issues.

# Rolling Over

## The Other Side - News that is Path Breaking (Leena Srivastava)

# Pincer Grasp – Journal Scan

# JOURNAL SCAN

**Dr Ritika Chaudhry,**

**Assistant Professor, Fellow in Developmental and Behavioural Pediatrics,**

**Bharati Vidyapeeth Medical College and Deemed University, Pune**

**Title: Systematic Review and Meta-analysis: Efficacy of Pharmacological Interventions for Irritability and Emotional Dysregulation in Autism Spectrum Disorder and Predictors of Response**

**Authors: Gonzalo Salazar de Pablo et al**

**Journal: Journal of the American Academy of Child & Adolescent Psychiatry, volume 2**

**Date of publication: February 2023**

Citation: Salazar de Pablo G, Pastor Jordá C, Vaquerizo-Serrano J, Moreno C, Cabras A, Arango C, Hernández P, Veenstra-VanderWeele J, Simonoff E, Fusar-Poli P, Santosh P, Cortese S, Parellada M. Systematic Review and Meta-analysis: Efficacy of Pharmacological Interventions for Irritability and Emotional Dysregulation in Autism Spectrum Disorder and Predictors of Response. J Am Acad Child Adolesc Psychiatry. 2023 Feb;62(2):151-168. doi: 10.1016/j.jaac.2022.03.033. Epub 2022 Apr 22. PMID: 35470032.

This study is a systematic meta-analysis which aims at assessing the efficacy of a broad range of pharmacological interventions for emotional regulation and irritability in ASD as well as the predictors of response which have previously not been evaluated by any meta-analysis. Emotional dysregulation and/or irritability have been defined as the failure to regulate emotions appropriately and effectively and a state of reduced control over temper

or an excessive response to stimuli, respectively and are often considered overlapping constructs. Both are common in ASD and may manifest as aggression, tantrums, rapidly changing moods, or self-injurious behaviours etc which profoundly impair functioning and lead to a substantial burden on families and health care services.

Methodology: Following a preregistered protocol (PROSPERO: CRD42021235779) the researchers systematically searched multiple databases until January 1, 2021 and included only placebo-controlled randomized controlled trials (RCTs) for the meta-analysis. Heterogeneity was assessed using Q statistics. Sub analyses and meta-regressions were conducted to identify the predictors of response. The primary effect size was the standardized mean difference. Quality of studies was assessed using the Cochrane Risk of Bias Tool (RoB2).

Most frequently provided pharmacological groups were antipsychotics, neuropeptides, antidepressants, mood stabilizers, medications used to treat ADHD, fatty acids, glutamatergic blockers, opioid antagonists, diuretics and others. Most studies (93%) used the Aberrant Behavioral Checklist–Irritability (ABC-I) subscale to evaluate emotional dysregulation and irritability.

## Results

Compared to placebo, antipsychotics (standardized mean difference = 1.028, 95% CI = 0.824-1.232) and medications

used to treat attention-deficit/hyperactivity disorder (ADHD) (0.471, 0.061-0.881) were significantly better than placebo in improving emotional dysregulation and irritability, whereas evidence of efficacy was not found for other drug classes ($p > .05$). Within individual medications, evidence of efficacy was found for aripiprazole (1.179, 0.838-1.520) and risperidone (1.074, 0.818-1.331). Increased rates of comorbid epilepsy ($\beta = -0.049$, $p = .026$) were associated with a lower efficacy.

## Conclusion

Some pharmacological interventions (particularly risperidone and aripiprazole) have proved efficacy for short-term treatment of emotional dysregulation and irritability in ASD and should be considered within a multimodal treatment plan, taking into account also the tolerability profile and families' preferences.

**Title: Congenital Hypothyroidism and the Deleterious Effects on Auditory Function and Language Skills: A Narrative Review**

**Authors: Andrade CLO et al**

**Journal: Frontiers in Endocrinology**

**Date of publication: 10 Aug 2021**

Citation: Andrade CLO, Alves CAD, Ramos HE. Congenital Hypothyroidism and the Deleterious Effects on Auditory Function and Language Skills: A Narrative Review. Front Endocrinol (Lausanne). 2021 Aug 10;12:671784. doi: 10.3389/fendo.2021.671784. PMID: 34447350; PMCID: PMC8382885.

This article is a mini review article which highlights the role of thyroid hormones in development and optimal functioning of the auditory system and the audiological problems seen in patients with congenital hypothyroidism (CH). The reported incidence of hearing loss (HL) in individuals with CH is around ~20%. It can be isolated or associated with vertigo and tinnitus with usual characteristics being sensorineural, bilateral, and symmetrical with degrees varying from mild to moderate.

Thyroid hormones act in both systems (peripheral and central) in the auditory system, and they are responsible for forming key structures of the inner ear, such as the cochlear duct, organ of Corti, and tectorial membrane and hence thyroid deficiency can lead to structural as well as maturational deficits in addition to the deficits in central auditory processing. The article describes the possible underlying genetic basis for the same from review of animal model studies. It describes the role of differential expression of thyroid hormone receptor alpha (THRa) and thyroid hormone receptor beta (THRb) and precise signalling necessary for proper THR-dependent differentiation events, comprising complete inner sulcus, sensory epithelium, spiral ganglion, cochlea, and auditory nerve maturation. It also describes the possible association of the expression of the SLC26a5 gene, which encodes the prestin protein which is considered to be the outer hair cells (OHC) engine in the cochlear amplification process and the gene expression encoding the K+ channels, KCNQ4 responsible for endolymphatic potential formation. These ion channels are significantly reduced and poorly distributed in conditions of thyroid hypo function.

The authors conclude by stating that CH can be considered a potential risk factor for changes in acoustic signals' processing mechanisms along the auditory pathway, which manifests itself as cognitive, language, and socio-emotional delays. The risk of hearing loss may be associated with the severity of CH and small hearing changes can be observed in individuals with CH even after early treatment is initiated.

# Congenital Hypothyroidism

**Dr Rahul Jahagirdar** (MD, PDCC (Ped Endo), ESPE Fellowship, UK)
**Dr Ruma Deshpande** (MD, PDCC (Ped Endo), ESPE Fellowship, UK)
Department of Pediatric Endocrinology
Bharati Vidyapeeth University Medical College and Hospital, Pune

Congenital Hypothyroidism (CH) is one of the most common preventable causes of mental retardation. The worldwide incidence is 1:4000 live births and the estimated incidence in India is 1:2500-2800 live births.

Thyroid dysgenesis is the commonest cause accounting for 75-80% of all cases of CH.

CH can be permanent or transient. Thyroid dysgenesis is the commonest cause of permanent CH affecting 1 in 4000 live births. It is usually sporadic with a 2:1 female to male preponderance. The cause is largely unknown but maternal cytotoxic antibodies and genetic mutations causing inactivation of thyroid receptor are sometimes found.

Thyroid hormone synthetic defects account for 10% of all cases. These are inherited as autosomal recessive disorders. The defect can lie in iodide trapping or organification, iodotyrosine coupling or deiodination and thyroglobulin synthesis or secretion.

Hypothalamic- pituitary hypothyroidism has an incidence of 1 in 100,000. It may be isolated or associated with concomitant deficiency of other pituitary hormones and present with hypoglycaemia and micro phallus.

Transient hypothyroidism due to trans placental transfer of TSH binding inhibitory immunoglobulins (TBII) from mothers with autoimmune thyroid disease is seen in 1: 50,000 births. Their effect wanes off by 3-6 months in the majority but may last up to 9 months. Exposure to iodine in sick preterm infants (e.g., application of povidone iodine for skin disinfection) or intake of iodine containing expectorants by pregnant mothers can lead to transient hypothyroidism.

## Clinical description

The clinical features of congenital hypothyroidism are often subtle and many new-born infants remain undiagnosed at birth. Up to one third have a birth weight greater than the ninetieth percentile.

On initial examination, the most common signs are umbilical hernia, macroglossia and cold or mottled skin. Thyroid hormone is also important in the formation and maturation of bone this can lead to a wide posterior fontanel of greater than 5 mm. This, along with persistent jaundice and poor feeding are the most striking clinical features.

A few infants with congenital hypo-thyroidism may have a palpable goitre. This is usually found in thyroid dyshormonogenesis where there is a defect in thyroid hormone production.

The typical appearance of a hypothyroid infant is shown in Figure 1.

Features include jaundice, a puffy face and a wide posterior fontanelle with open sutures. The nasal bridge is flat and the eyes exhibit pseudohypertelorism. The mouth may be slightly open revealing macroglossia.

Further examination would reveal bradycardia and a protuberant abdomen with a large umbilical hernia.

Neurologic examination findings include hypotonia with delayed reflexes. Skin may be cool to touch and mottled in appearance reflecting circulatory compromise

Figure 1

## Investigations:

Early detection of congenital hypothyroidism (CH) through new born screening (NBS) and prompt treatment can prevent morbidities. Indian infants are at higher risk of CH (approximately 1 in 1000) because of ethnicity, increased survival of very low birth weight (VLBW) neonates, and endemic iodine deficiency belts. However, countrywide NBS has not yet been established.

The new guideline suggests CH-NBS by measuring primary thyroid stimulating hormone (TSH) in dried blood spot (DBS), preferably at 48-72 hours of age in all neonates. In the Indian setup, considering earlier hospital discharge of term healthy neonates and simultaneous screening of other metabolic disorders, DBS may be preferred over cord sample and sampling timing may be preferred at 48-72 hour rather than after 72 hour of age.

Mentioned below in the algorithm is the approach and investigation for a new born with hypothyroidism based on TSH and FT4 values as recommended by ISPAE

## TREATMENT OF CH

Term as well as preterm infants with low T4 and elevated TSH should be started on L-thyroxine as soon as the diagnosis is made. The initial dose of L-thyroxine should be 10-15µg/ kg/ day with the aim to normalize the T4 level at the earliest.

***Monitoring of Therapy:***

T4 should be kept in the upper half of normal range (10-16 µg/dL) or free T4 in the 1.4 - 2.3

ng/dl range with the TSH suppressed in the normal range.

T4 and TSH levels should be checked according to the following schedule:

0-6 months: every 6 weeks

6 months-3 years: every 3 months

> 3 years: 6 monthly

T4 and TSH should also be checked 6-8 weeks after any dosage change.

It is equally important to avoid over treatment. Adverse effects of over treatment include premature fusion of cranial sutures, acceleration of skeletal maturation and problems with temperament and behaviour

The best outcome occurs with L-thyroxine therapy started by 2 weeks of age at 9.5µg/kg or more per day, compared with lower doses or later start of therapy. Residual defects can include impaired visuospatial processing and selective memory and sensorimotor defects.

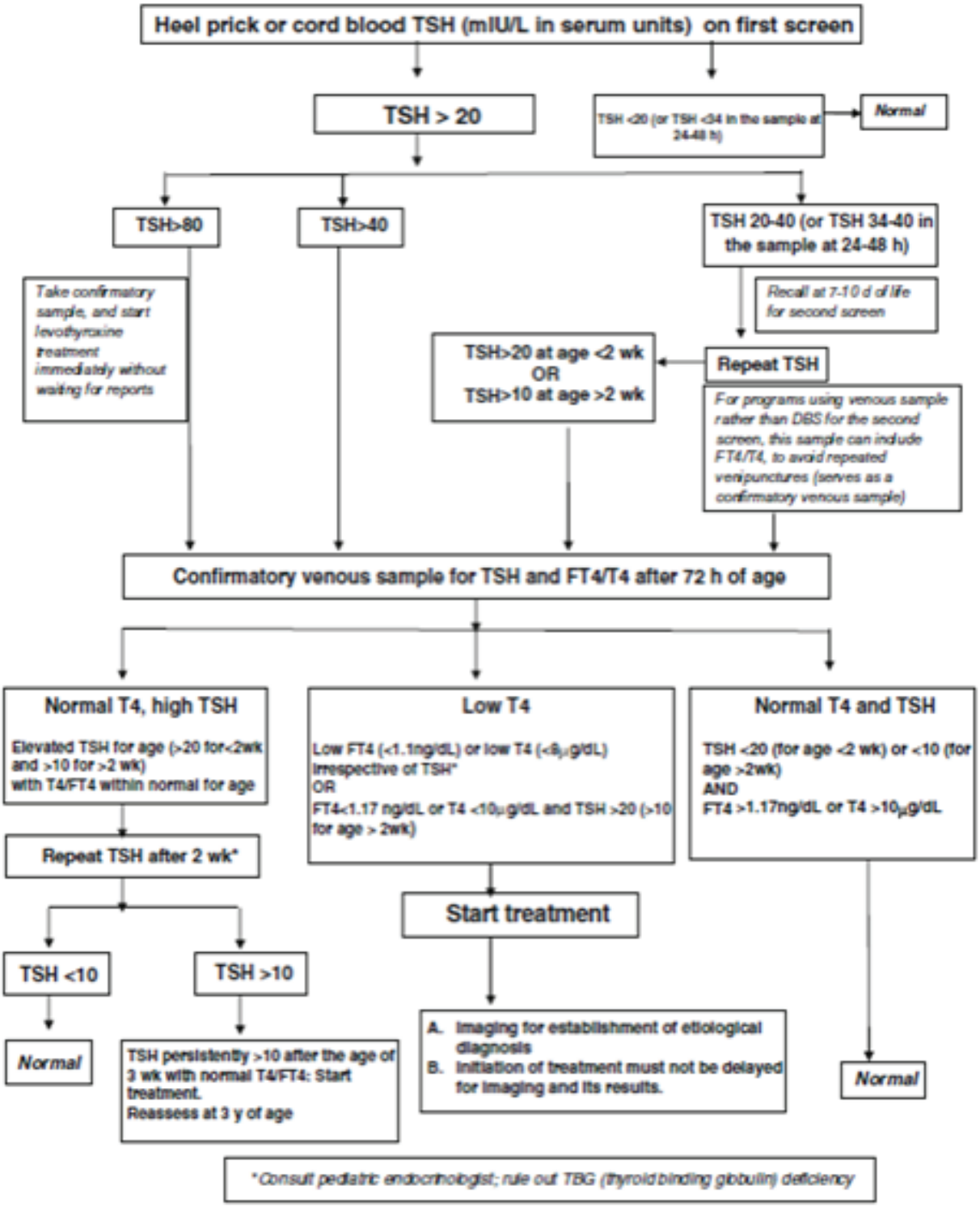


**Fig. 1** Algorithm for screening and detection of congenital hypothyroidism (CH)

More than 80% of infants given replacement therapy before three months of age have an IQ greater than 85 but show signs of minimal brain damage, including impairment of

arithmetic ability, speech, or fine motor coordination in later life. When treatment is started between 3-6 months late, the mean IQ is 71 and when delayed to beyond 6 months, the mean IQ drops to 54 as reported by studies.

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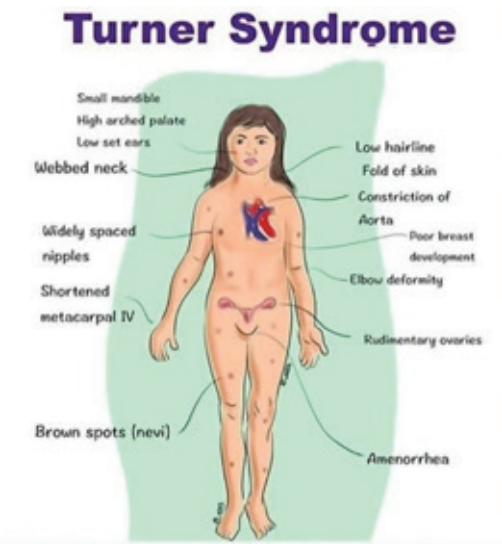
# Response to Name Call – Eponyms

Dr Kawaljit Singh Multani




Turner syndrome is a chromosomal disorder due to complete or partial monosomy for the X chromosome associated with short stature and primary ovarian failure in phenotypic females. The eponym derives from a study published in 1938 by Henry Turner describing seven women with short stature, sexual immaturity, neck webbing, low posterior hairline, and cubitus valgus. Several years earlier, Otto Ullrich had described an 8-year-old girl with short stature, lymphedema of the hands and feet, neck webbing, high arched palate, low-set auricles, and several other featuresnow associated with Turner syndrome. Ullrich later recognized that his patient and those of Turner appeared to have the same condition and called attention to the work of Bonnevie, who described cervical distention and malformations of the ears, face, and limb buds in mice secondary to dissection of subcutaneous fetal tissues by fluid. Ullrich suggested that fetal lymphatic obstruction may cause neck webbing and other superficial features of Turner syndrome and proposed the eponym Bonnevie-Ullrich to describe this constellation of anomalies. Ullrich's contributions gave rise to the eponym Ullrich-Turner syndrome sometimes used in Europe. Endocrine and pathology studies of the 1940s revealed primary ovarian failure in women with Turner syndrome, associated with elevated gonadotropins,reduced estrogen and "streak" ovaries consisting of connective tissue depleted of germ cells. These early studies also discovered an extraordinary incidence of hypertension and aortic disease in young women with Turner syndrome. The first link between Turner syndrome and sex chromosome anomaly was provided in 1954 by Polani and colleagues, who reported three patients with Turner syndrome and coarctation of the aorta who were sex chromatin negative. Soon thereafter advances in cytogenetic identification of specific chromosomes revealed that Turner syndrome was associated with the presence of a single X chromosome (X monosomy).6 These observations were paradigm shifting in our understanding of the role of the human sex chromosomes in sex determinatio, and the designation Turner or Ullrich-Turner syndrome is more specific than gonadal dysgenesis.

# Non verbal Communication

## SPOTTER-1 Q

Dr Anoop Verma

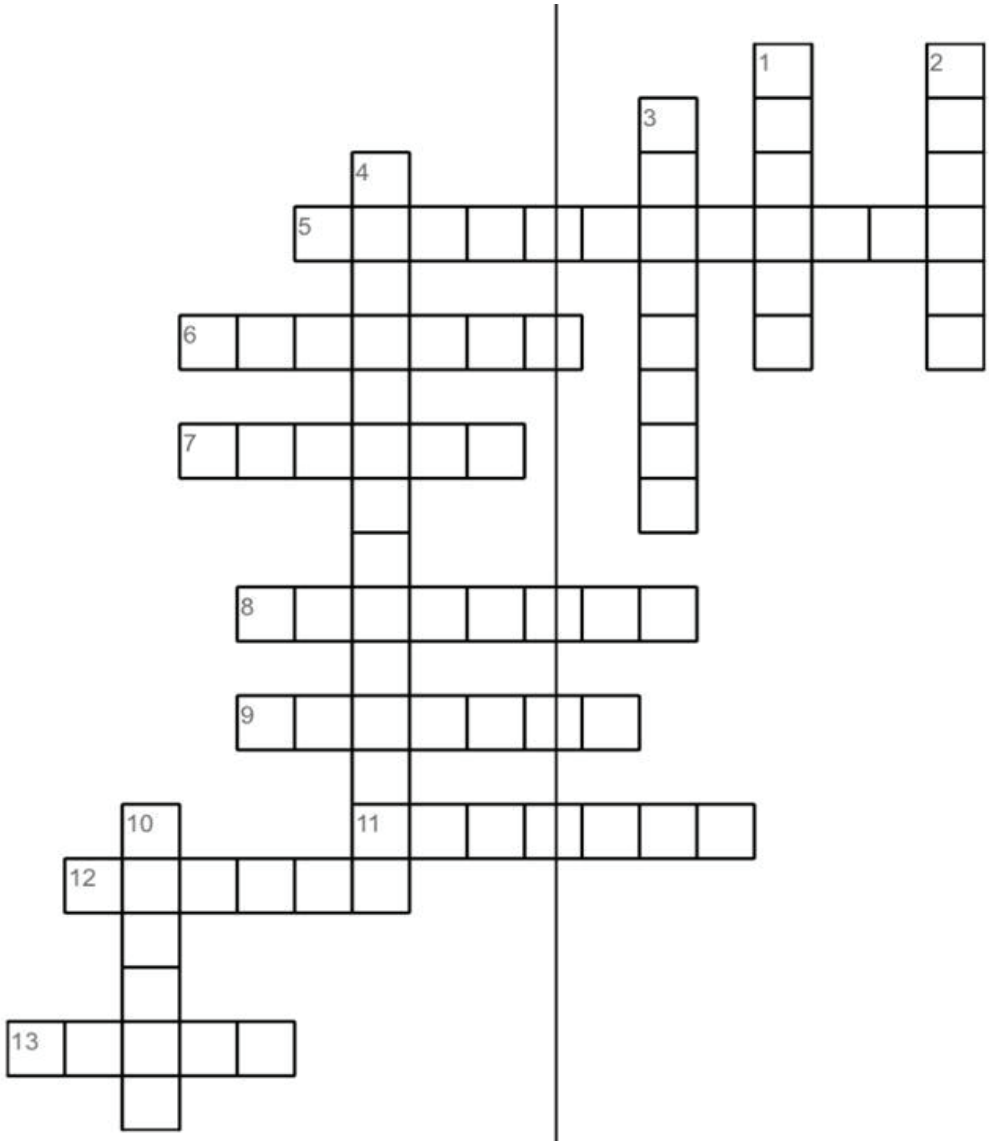

**5 yr old,** walks with parents
support poor social communication
speech delay, poor cognition, on Antiepileptic
drugs. No Birth asphyxia, No h/o NNJ

1. **What is your tentative diagnosis"**
2. **What is GDD/MR?**
3. **What a pediatrician can do or Can't do?**
4. **What are red flag signs?**

*(See page 31 for answer)*

# The Meaningful Word - Quiz

Dr Anshuman Verma




**Across**

5 Maturational theory
6 Operant conditioning theory
7 Ethology theory
8 Multiple intelligence theory
9 Social learning theory
11 Psychosocial theory
12 Information processing theory
13 Psychosexual theory

**Down**

1 Classical conditioning theory
2 Attachment theory
3 Sociocultural theory
4 Ecological systems theory
10 Cognitive development theory

# Sudden Bursts of Laughter

Dr Leena Deshpande


## Live ones!!

Saw a child in the clinic. She is a fisherman's daughter. She is doing well now on medicines. Today after the consultation, the father asked me 'Do you eat fish?' The conversation ended with me telling him that I did not but my family did.

So he said 'At the next visit, I will bring fish.' He took some names that sounded Greek and Latin to me. He continued his monologue and finally in a flash of inspiration said ' I will bring CRABS.' While I was telling him that all that was unnecessary, the way he said the last line made me very suspicious. I asked him 'You mean LIVE crabs?!'

He replied 'Of course. They can then cook fresh ones at home.' I hope my face reflected my incredulity as I asked him 'And how do you think I will carry these live crab back home? You really expect me to carry live crab in my car?' He replied, matter of fact, 'I will have tied them in a bag and they won't escape. That will be easy.' A shudder went up my spine. Imagine one of them got loose and then escaped in the car. What a nightmare! Thinking about it made me break out into cold sweat.

Not trusting him at all, I emphatically told him 'DO NOT BRING LIVE CRABS!!' I repeated that 3 to 4 times to make sure that I did not experience encounters with crusteceans!! This is the stuff nightmares are made of. Imagine the crab escaping and at a traffic signal, I feel a tap on my shoulder! Yuck!

*Excerpt from the book "Kids, Kidney, Kidding" by Dr Pankaj Deshpande*

# Babbling and Cooing – From our Fellows

Fellowship Activity for the year 2024 has take off with rigor. Academic program has been divided into different modules – Neuromotor, Neurogenetic, Neurobehaviour , Neurosesnory , Neurocognitive etc

Across the 17 fellowship centers the online zoom activity has started on Tuesdays at 2pm.


FEBRUARY 2024

| Date | Topic | Presenter | Moderator |
|---|---|---|---|
| 6.02.24 | Neuromotor assessment | CDC TRIVANDRUM Dr.Nagee G/ Dr. Hashma Md P | Dr.Leena Srivastava |
| 13.02.24 | Tone disorders in children | SGRH Delhi. Dr.Jyoti Prajapati | Dr.Deepa Bhaskaran |
| 20.02.24 | Approach to Hypotonia: a case based discussion. | BVP Pune Dr.Amola Bhavesh Khandwala | Dr.Praveen Suman |

TUESDAY 2-3 PM , ON ZOOM

| Date | Topic | Presenter | Moderator |
|---|---|---|---|
| 5.03.23 | Genetic testing in clinical practise. | NIMS TRIVANDRUM, Dr.Sneha Balasundaram | St. John's,Bangalore,Dr .Maria Lewis |
| 19.03.24 | Approach to genetic testing in NDD. | St.Johns,Bangalore ,Dr.Shweta Srinivasan V.V. | NIMS ,Dr.MKC Nair. |
| | | | |

NEURO GENETICS

# Birth Defects - Screening and Prevention

**Dr. Chaitanya A. Datar**
**Consultant Clinical & Metabolic Geneticist**
**Bharati Hospital, Pune**

It is estimated that ~3-5% neonates born worldwide have a birth defect (Source- CDC, USA). These defects can be classified as- Minor defects- those of cosmetic significance only and do not have any functional consequences eg- ear tags, isolated polydactyly, clinodactyly etc. and Major defects- which if left uncorrected can lead to significant functional impairment eg. Cleft lip/ palate, congenital heart defect, anal atresia etc.

Single minor defects may be noted in ~10-15% of newborns and these are usually of no functional significance. Single major defects may be noted in ~3% newborn and these may need some intervention, while multiple congenital anomalies have an incidence of ~0.7% of all newborns.

Causes of birth defects are diverse and Genetics, in-utero environment, and an interaction between genes & environment i.e. epigenetics play a crucial role. A definite genetic etiology may be identified in ~40-60% cases with multiple congenital anomalies while the environmental factor may be implicated in ~10% cases. The etiology of many birth defects is still obscure.

Genetic etiology could involve aberrations at the level of chromosomes, chromosomal segments or at a gene level, and hence genetic syndromes causing MCA can be divided as-

- chromosomal aneuploidies eg. Down, Edward or Patau syndromes which are diagnosed by a karyotype

- chromosomal microdeletions eg Williiam syndrome, Di George syndrome that can be picked up by FISH or chromosomal microarray or

- single gene syndromes eg- Noonan syndrome, Treacher Collins syndrome, Orofaciodigital syndrome etc. that can be diagnosed by exome sequencing.

Known environmental factors that lead to birth defects are known as teratogens. These could include- viruses (TORCH, Parvovirus, Zika, chicken pox etc.), radiation exposure, drugs (eg. Valparin, phenytoin, retinoic acid, isotretinoin etc.), high grade fever in first trimester, uncontrolled maternal diabetes etc. History of exposure to any of these agents must be taken for all cases with congenital anomalies. However, it must be remembered that the exact teratogenic effect on the fetus will depend on the dose and duration of exposure to these teratogens and the period of gestation when the exposure happened.

However, as mentioned above, it is likely that many birth defects occur due to a complex interaction between the genes and environment. This includes epigenetic modifications that are happening during embryogenesis and fetal development. The best example is that of neural tube defects where some genetic predispositions are known, but deficiency of folic acid and Vitamin

B12 seem to be more crucial factors. These deficiencies in the one carbon metabolism is postulated to affect the methylation process that controls the gene expression in-utero.

Even in the current scenario, neural tube defects are the possibly the most commonly encountered congenital anomalies followed by congenital cardiac anomalies and cleft lip/ palate. A definite genetic etiology may be identified in ~ 20% Neural tube defect cases and ~40% of cases with cleft lip/ palate. In congenital cardiac anomalies, a genetic case may be noted in ~10-40% cases. Defects of limbs eg congenital talipes deformity is also a common anomaly which may occur due to external factors such as oligoamnios or due to genetic neuromuscular or arthrogryposis disorders.

Hence when evaluating birth defects, it is necessary to discuss genetic as well as non-genetic factors in most of the cases while counselling.

Primary prevention of birth defects is difficult. However good periconceptional practice of taking adequate folic acid, Vitamin B12, multivitamin, iron and calcium intake will certainly help to reduce the incidence. Adequate management of thyroid disorders and diabetes is also essential. Appropriate caution must be exercised during pregnancy while taking any unprescribed medications or while undergoing procedures. Rubella and varicella vaccines can be taken prior to planning a pregnancy if there has been no prior exposure. Prompt treatment of infections and proper nutrition are crucial factors in reducing the risk of birth defects.

Screening for birth defects is possible with serial ultrasound scans. NT scan at 12 weeks and anomaly scans at 19 weeks, Fetal Echocardiography and neurosonogram (if necessary) at 23 weeks are likely to pick up most of the major anomalies described above because of improved ultrasound techniques and expertise. A first trimester maternal serum double marker test (checking levels of PAPPA and beta hCG) at 12-13 weeks will screen for Down, Edward, Patau and Turner syndromes with ~85-90% accuracy. Alpha feto protein (AFP) checked in maternal serum between 15-20 weeks screens for neural tube defects. Non invasive prenatal screening (NIPS) done on cell free fetal DNA in maternal blood has a good screening accuracy for Down syndrome (~96-97%). NIPS can be done after documenting a normal NT scan at 12 weeks instead of a double marker test if the patient wants better screening accuracy.

Appropriate genetic counselling by trained personnel is necessary to understand implications of birth defects when identified in the fetal stage or when encountered postnatally. Appropriate investigations may be needed if a genetic etiology is suspected. A multidisciplinary approach involving the primary Pediatrician, Developmental Pediatrician, Pediatric surgeon, Cardiologist, Geneticist, Neurologist, therapists, etc. is required to be able to discuss the prognosis of the given birth defects whenever identified, antenatally or postnatally. Referring the family to disorder specific patient support groups also helps in allaying some of their anxiety regarding the outcomes. Genetic counselling for couples having a previous child with birth defects must ideally be done in the pre-conceptional period to be able to complete the necessary investigations and discuss the recurrence risks, this will greatly reduce their anxiety.

Overall, birth defects cannot be altogether prevented, but measures as discussed above can be taken to reduce the risks of the same and a proper counselling will help the family cope with the anxiety associated with the condition.

# Read a Book – Review of a Chapters from the IAP Handbook of DBP

## Developmental Concerns in Pediatric Practice

### Dr YK Amdekar

1. Pediatricians should be Health Providers and not just Disease Managers
2. Health includes Physical (growth), Mental (Development), Social (Behaviour) well being; all three need to be monitored closely in the first two years of life.
3. Developmental delay may leave a permanent disability if not picked up early.
4. Development monitoring is thus an important part of Pediatric Practice.
5. Every Pediatrician should 'Develop' the habit of observing and monitoring deviations in developement of the child at every visit to the Clinic.
6. Frequent visits in the first two years of life, offer the Pediatrician this opportunity
7. All children must be screened at all ages in all domains of development by simple actions in the clinic, like his response to smiling, picking a toy, waving bye-bye etc.
8. Pediatricians should be familiar with some simple motor, fine motor and other milestones at various ages and refer when concerned about deviations.
9. Look out for signs of Autism Spectrum Disorder, ADHD, Learning Disability at earliest and do the needful.
10. This habit will lead to Excellence in Practice and Satisfaction for the Doctor and the Patient's Family.

***Dr Anjali Gokarn, Dr Jayshree Deshpande***

# SPOTTER-1 A

Dr Anoop Verma


**Intellectual disability** (ID) is a commonly acknowledged developmental disorder . It has replace the notorious term **"mental retardation".**

1. **Intellectual disability** (ID) is a NDD that has ***impairment in the cognitive*** (intellectual) ability and ***adaptive*** function of the individual in the environment.
2. ID begins ***before 18 years*** and affects 1% of population. It presents as failure of the child to progressively achieve skills to ***receive, analyse*** and ***process*** information function adequately and age-appropriately in ones environment
3. As a result learning, reasoning , problem solving and abstract thinking are affected. These children may presents as dysmorphic facies and this is termed as ***"Syndromic ID"***
4. The diagnosis is made around age 5 yrs after comprehensive standardised testing, till then the term GDD may be provisional be used

**Approach-Elaborate History and examination helps**

**1. History**

1. 3 generation pedigree
2. Prenatal , perinatal & postnatal issues
3. Assess development, behaviour, medical, family, social, educational

**2. Examination**

1. Anthropometry
2. Dysmorphism
3. Cutaneous clues
4. Neurodevelopment assessment
5. Sensory screening- Fundus & audiometry

1. **Assessment-** screening for development & behaviour are recommended for early recognition. If screening test is positive comprehensive development evaluation is warranted.
2. ***A diagnosis of ID requires impaired intellectual and adaptive function in at least one of the three domains*** (conceptual, social, practical) and includes standardised assessments.

**Standardised test for diagnostic assessment of ID**

**1. Cognitive**

1. Below 2 yr- BSID-III (2005)=, Griffiths III (2015)
2. 2-5 yrs- WPPSI-IV ( 2012)
3. Above 5 y- WISC-V (2014)

**2. Adaptive**

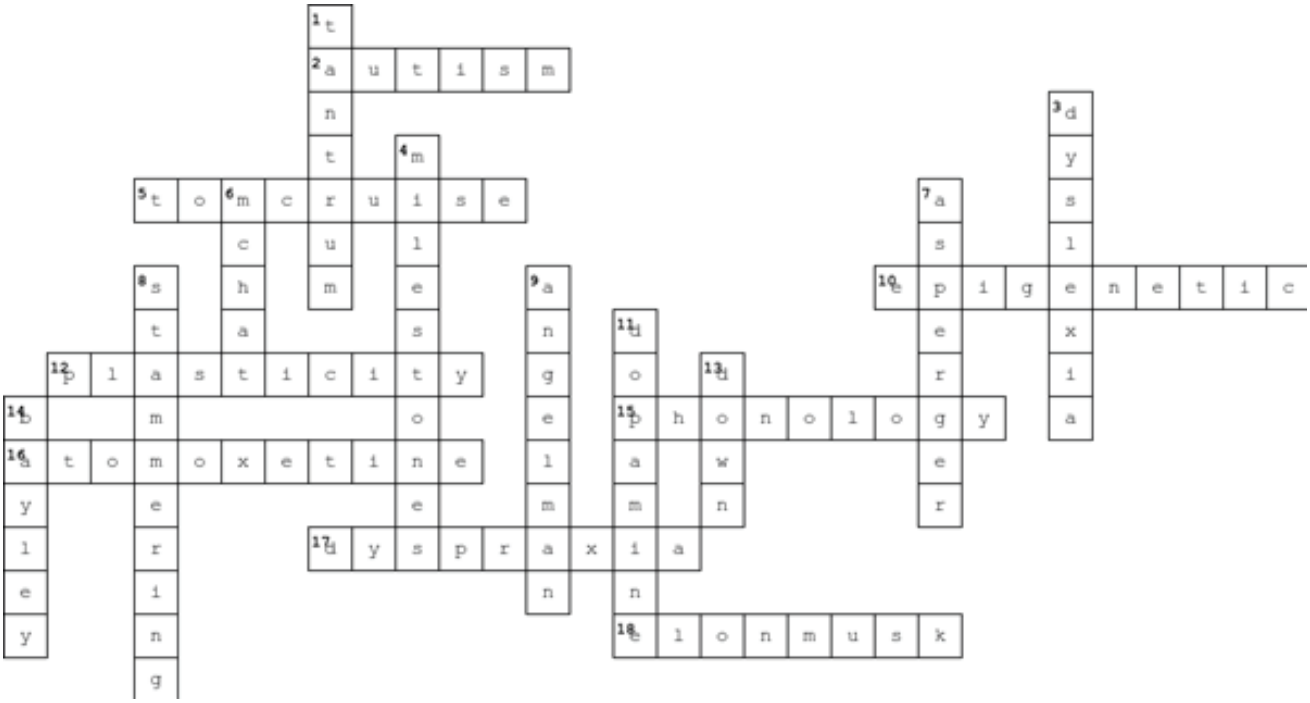
1. Below 2 ys- Vineland-III (2016), ABAS-III (2015)
2. 2-5 yr- Vineland-III (2016)

**Management**

1. Early diagnosis
2. Intervention
3. Appropriate support

Samir H Dalwai- IAP Handbook of Developmental & Behavioral Paediatrics 2022: Intellectual Disability 64-68.

# Answers to The Meaningful Word



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**
PAYABLE AT ERNAKULAM
FEDERAL BANK LTD
ERNAKULAM / KATHRUKADAVU
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……**

# IAP HANDBOOK OF DEVELOPMENTAL AND BEHAVIORAL PEDIATRICS

# OFFICIAL JOURNAL OF THE IAP CHAPTER OF NEURODEVELOPMENTAL PEDIATRICS

# NCDP

## National Conference of Developmental Pediatrics

21st National Conference of IAP Chapter of Neurodevelopmental Pediatrics

**Theme: Innovate, Inspire, Ignite for Developmental and Behavioural Excellence**

**Date:** Workshop - August 30th 2024
Conference - August 31st, September 1st 2024

Venue: JN Tata Auditorium IISC, Bengaluru

6 Credit Hours Awaited

**Co Hosts**

IAP BPS Bangalore

Rajarajeswari Medical College & Hospital
Recognised by National Medical Commission (NMC) & Govt, of India
A Constituent Institution of Dr. M.G.R Educational and Research Institute
(Deemed to be University)

Developmental and behavioural Pediatrics
Role of Environmental issues.

Co-sponsored by

Divecha Centre for Climate Change
IISc Bangalore

# WELCOME TO THE VIBRANT CITY OF BENGALURU!

With open arms and boundless enthusiasm, we invite you to join us for a remarkable intellectual gathering, the NCDP Bengaluru 2024, happening from August 30th to September 1st, 2024. This event, themed "Innovate, Inspire, Ignite for Developmental and Behavioral Excellence," promises to be an unforgettable experience set against the dynamic backdrop of our bustling city.

In collaboration with esteemed experts and organizations in the field, including the NCDP, we are thrilled to present a conference that embodies the pinnacle of developmental and behavioral research and practice. This partnership adds a profound dimension to our event, amplifying our commitment to advancing the well-being of individuals across the lifespan.

The NCDP Bengaluru 2024 is more than just a gathering of minds; it is a celebration of our collective dedication to innovation and excellence. Through a diverse array of sessions, workshops, and interactive discussions, we aim to facilitate learning, networking, and professional growth.

Join us as we delve into cutting-edge research, share inspiring insights, and ignite transformative ideas. Together, let us embark on a journey of discovery and collaboration, as we strive to innovate, inspire, and ignite positive change in developmental and behavioral sciences.

Welcome aboard this extraordinary voyage of knowledge and innovation!

And remember, in the heart of Bengaluru, it's not just about the conference—it's about soaking up the spirit of "Namma Bengaluru" and experiencing the vibe of

**"Mast Majaa Maadi!"**

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## SCIENTIFIC HIGHLIGHTS

1. Lifespan approach to neurodevelopmental disorders
2. Neuromotor, neurocognitive, neurogenetic - what is new
3. Vaccine and neurodevelopmental disorders
4. Impact of global climate change on child development
5. Behavioural issues in adolescence
6. Acute care of children with developmental disorders

## CONFERENCE REGISTRATION DETAILS

| CONFERENCE REGISTRATION | EARLY BIRD (Till April 15th) | AFTER APRIL 15TH | SPOT |
|---|---|---|---|
| PG STUDENTS | Rs 3000 | Rs 3500 | Rs 4000 |
| PARAMEDICAL STAFF | Rs 3000 | Rs 3500 | Rs 4000 |
| DELEGATES | Rs 4000 | Rs 4500 | Rs 6000 |
| SENIOR DELEGATES (Age ⩾70years) | Rs 4000 | Rs 4000 | Rs 4000 |

▸ Registration amount includes the Workshop

SCAN THE QR CODE TO REGISTER

CLICK TO REGISTER

# ABSTRACTS OF SCIENTIFIC PAPERS ARE INVITED FOR THE NCDP 2024

## SUBMIT YOUR ABSTRACT

Poster Presentation | Oral Paper

**Last Date of Abstract Submission** 15th May 11:59pm

**All abstracts must follow the instructions listed below and be submitted within the time frame mentioned. Please note that all presenters must be central IAP members and must be a registered delegate for the conference.

NCDP 2024 reserves the right to remove any abstract that does not comply with the following guidelines.

## TEMPLATE TO FOLLOW

**Title**

ARIAL | 12 PT FONT | BOLD | LEFT ALIGNED | UPPERCASE

**Author/s**

Arial | 10pt font | first name initial | followed by surname list the name of the presenting author first and bold all presenters, separated by a comma, with superscript 1, 2 for organisation/affiliation - Example - J. Jones, M. Clark[2]

**Organisation or Affiliation**

Organisations numbered in superscript to indicate association with author or presenter: Arial | 8pt font | Organisation | City | Country Example: University of New South Wales, Sydney, Australia, Royal Brisbane Hospital, Brisbane, Australia

**Abstract Body**

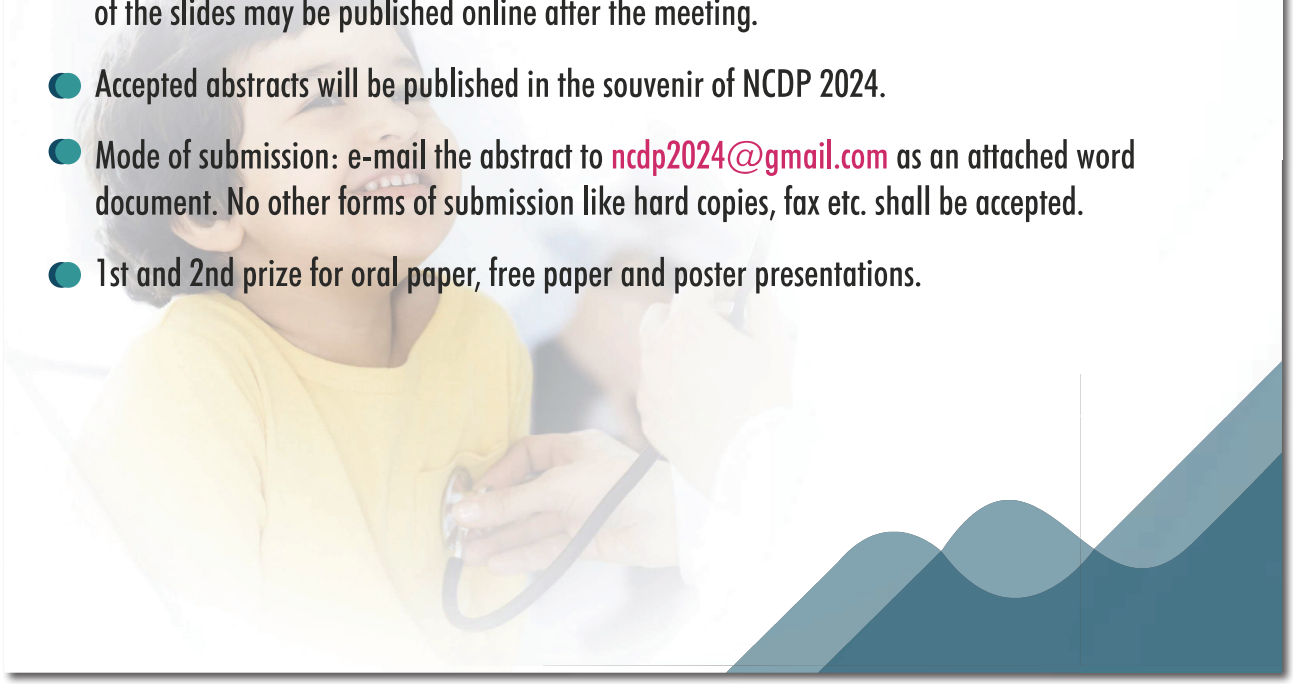
Arial, 10pt font, single spaced, justified, maximum of 300 words under the following headings

Background | AIMS | Methods | Results & Conclusions

References (optional)

# GENERAL GUIDELINES

- Language to follow: English.
- Multiple abstract submissions permitted maximum of 3.
- Word Limit - not to exceed 300 words (Excluding the title, authors and affiliations)
- Format of Abstract submitted - Word doc.
- Only original scientific material which have not been previously published or presented will be accepted.
- All abbreviations must be spelt out on first use.
- Abstracts should be thoroughly spell checked.
- The presenting author has to obtain permission from all authors to submit the abstract & has to be available to present in-person.
- A submission as a particular presentation type - oral, poster or theme may be changed to a different type or theme following review in order to be accepted and included in the programme.
- Presenters may be recorded for live streaming. A recording of the presentation and a copy of the slides may be published online after the meeting.
- Accepted abstracts will be published in the souvenir of NCDP 2024.
- Mode of submission: e-mail the abstract to ncdp2024@gmail.com as an attached word document. No other forms of submission like hard copies, fax etc. shall be accepted.
- 1st and 2nd prize for oral paper, free paper and poster presentations.



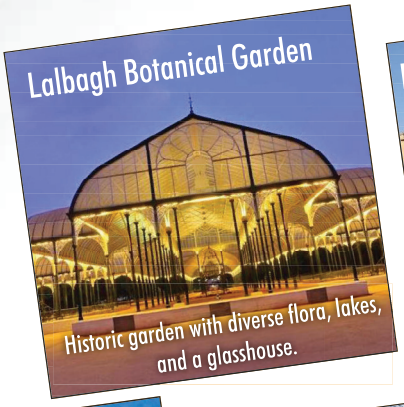
# PRE CONFERENCE WORKSHOP

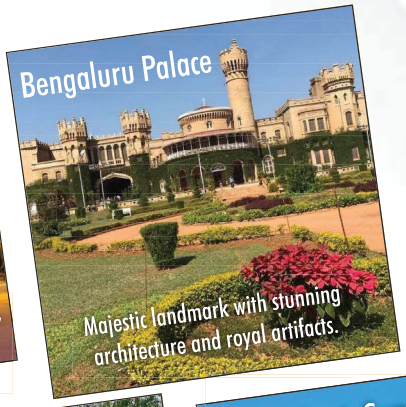
## AUGUST 30TH

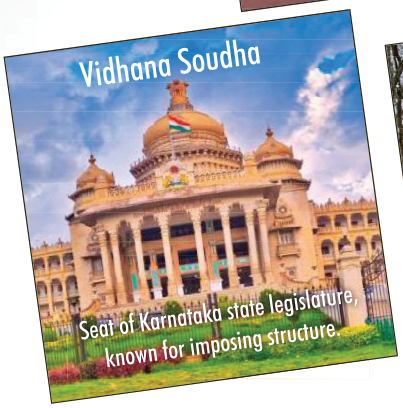
| TOPIC | VENUE | WORKSHOP COORDINATOR |
|---|---|---|
| Identification of Developmental Differences and Early Action ( IDDEA) | Banglore Medical College and Research Institute | Dr Gayathri Devi |
| Applied Clinical Neurology in Developmental Paediatrics | Raja Rajeswari Medical College (RRMC) | Dr Kishore Kotha |
| RACE Module for High Risk Newborn follow up | Indira Gandhi Institute of Child Health | Dr Uddhav Kinhal |
| Vision in Neurodevelopmental Disorders - Seeing through the CVI Lens | The Unit of Hope St. John's Centre for Children with Special Needs | Dr Maria Lewin |

Note : Conference registration is compulsory to attend the workshop

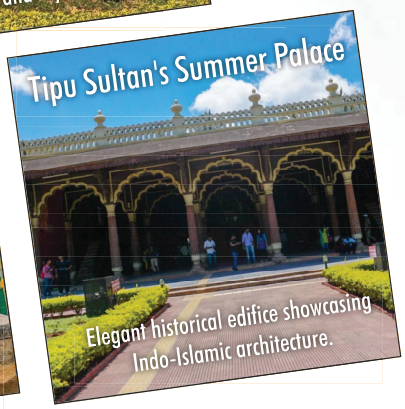
# TOP 5 HIGHLIGHTS FOR TOURISTS IN BENGALURU








## HIGHLIGHTS OF JN TATA AUDITORIUM AT IISC, BENGALURU

### Modern Facility
Equipped with cutting-edge technology for various events.

### Architectural Brilliance
Features striking design elements and spacious layout.

### Impeccable Acoustics
Provides optimal sound quality for diverse activities.

### Versatile Venue
Ideal for conferences, concerts, and cultural events.

### Prestigious Setting
Located within the esteemed IISC campus amidst greenery.

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| ORGANISING JOINT SECRETARY | DR. GAYATRI DEVI | 9845929945 |
| ORGANISING JOINT SECRETARY | DR. KISHORE KOTHA | 7795922388 |
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