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AADARSH®

CONTENTS

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Aims and Scope of Indian Journal of Developmental and Behavioural Pediatrics (IJDBP)

IJDBP is a specialty journal in Developmental and Behavioural pediatrics published by Indian Academy of Pediatrics Chapter of Neurodevelopmental Paediatrics

The Journal welcomes Original papers, Review articles, Case reports and other articles relevant to child development & Behaviour including :

- Neuro developmental disorders,
- Developmental delays,
- Behavioural issues,
- Autism,
- Attention deficit hyperactivity disorder,
- Learning difficulties,
- Intellectual disabilities,
- Evidence based role of early intervention,
- Family centred multidisciplinary intervention,
- Neurogenetic disorders affecting child development,
- Neuroimaging & Neurological issues affecting child development,
- Corrective and assistive surgeries
- Home environmental and environmental issues affecting child development,
- Medical conditions
- Low birth weight and High-risk neonate requiring neonatal intensive care & its outcome,
- Preventive aspects in adolescents and pregnancy.
- Management of conditions covered in Rights of Persons with Disability Act,2016 of GOI.

It aim to promote advances in research in the field of child development and Behavioural issues so that latest evidenced based information is shared to enhance the quality of care and improve lives of children with special needs and their families.

The journal will be National Double Blind Peer review Open access journal published Quarterly. We will accept for publication manuscripts that were not published earlier in any form. The journal is devoted to publishing quality papers based on original innovative and most advance research in the field of developmental behavioural pediatrics.

The Journal aims to have the highest possible ethical and publication standards by scrutinizing the papers, through peer review assisted by eminent experts from prestigious teaching institutes from the country. For all Manuscripts submitted the journal will employ a plagiarism detection system for detecting plagiarism against previously published work.

INVITED GUEST EDITOR

GENERATION BETA: WHAT DO WE NEED TO KNOW?

Generation Beta, born between 2025 and 2039, will face unprecedented challenges stemming from pervasive digitalization. The dominance of screens and artificial intelligence may impede emotional development, social skills, and mental resilience. To counter this, parents, educators, and society must reintroduce the joy of direct human interaction, unstructured play, and physical activity into daily life.

Children born between 2025 and 2039 will belong to Generation Beta—a cohort that will inherit a world unlike anything before, shaped overwhelmingly by technology. As children of millennial and Gen Z parents, they will be raised amidst profound digital saturation. We already observe early signs of this with Gen Z and Generation Alpha, who are facing shifts in social cognition and behavior due to diminishing human engagement and rising screen exposure.

The root issue lies in the disconnect from reality. Our species evolved over millions of years through direct interactions with people and environments—learning, adapting, surviving. Now, in a matter of decades, we are inundated with a digital deluge our genes haven't evolved to process. The resulting internal world is often misaligned with actual reality.

In that sense, I liken AI to a genetic mutation—a sudden, radical shift for which we are not biologically prepared. What evolution would have gradually introduced over millennia is now being squeezed into a few decades. This mismatch may challenge Generation Beta's ability to interpret reality, navigate emotions, and engage socially.

The Impact on Emotional and Social Development

One major concern is the blurring of boundaries between reality and the virtual world. Generation Beta may grow up in a culture of instant gratification, where emotional regulation and resilience are underdeveloped. The ability to cope with disappointment, wait, adapt, or navigate complex social situations may diminish.

Marketing terms like "digital natives" or "AI natives" only serve to normalise this shift and promote the sale of gadgets. But I believe it's far too early to declare any generation as digitally native. Humans are, by design, socially native. Our evolution is deeply intertwined with social behavior—eye contact, facial expressions, turn-taking, cooperation—all essential for survival and thriving. Digital interactions, which often bypass these essential cues, are not just different; they are disruptive when consumed in excess.

Moreover, this constant bombardment of fragmented, non-contextual information creates cognitive noise—cluttering the brain with content that doesn't connect meaningfully with one's real-life experiences. Over time, this may hinder the development of authentic social behaviors and create distorted perceptions of the world.

Parenting in a Digitally Dominated World

So, what can we do?

First, let us stop glorifying digital immersion. Instead of dubbing children "digital natives," we must anchor them in what they truly need—human connections. Parents must model and teach children the value of bonding, playing, laughing, and simply "being" with others—family, friends, neighbors. Urbanization, smaller families, and delayed parenthood—particularly among millennials and Generation Alpha—are leading to reduced engagement between parents and children. Many parents, exhausted

by long work hours, struggle to offer the child time, energy, or nature exposure. The path of least resistance then becomes screen time.

In such scenarios, schools must become the surrogate social environment—encouraging interpersonal interaction, dialogue, group activities, and play. At the same time, the role of city planning cannot be ignored. Open spaces, parks, and child-friendly infrastructure are not luxuries—they are developmental necessities.

Structured and Unstructured Play

Much has been said about the value of organized sports. Indeed, they aid physical, psychological, and social development. But I want to stress the indispensable value of free, unstructured play.

Children need the freedom to explore, imagine, take small risks, solve minor conflicts, and create their own games. This builds independence, creativity, problem-solving, and self-regulation. Whether it's climbing a tree or playing hide and seek, unstructured play fosters brain development in ways digital stimulation never can.

Health Implications: Beyond the Physical

We already worry about childhood obesity, vision issues, and diabetes as offshoots of sedentary, screen-heavy lifestyles. But the deeper crisis is a social and emotional disconnect—children who are uncomfortable with eye contact, who lack empathy, and who struggle to navigate disappointment or boredom without reaching for a device.

As a developmental pediatrician, I fear that Generation Beta may struggle to distinguish perception from reality. This is not a trivial concern. It affects identity, social integration, learning, and mental health.

Yes, digital awareness is increasing, and yes, Generation Beta may have better access to mental health resources. But that's not enough. Mental health apps and AI-driven therapy can assist—but only in a symptomatic way. They cannot substitute human warmth, empathetic listening, or the subtle cues that form the bedrock of emotional healing.

What We Must Do Now

We must create environments—at home, school, and in communities—that support:

Reduced screen time, particularly in early childhood

Strong family and peer relationships

Free play and real-world exploration

Reduced reliance on digital tools for emotional support

Policies that favor open spaces and social schooling

Let us reintroduce the joy of simple human engagement. Smiles, stories, shared meals, family routines, games, and conversations—these are not old-fashioned. They are vital.

In the years ahead, we may have to rethink not only how we parent and teach, but also how we define intelligence, success, and connection. We cannot afford to be passive. Generation Beta deserves a world where digital tools assist but do not overwhelm. They deserve to grow up in real relationships, with real people, in the real world.

With Regards & Best Wishes,

Dr. Samir H Dalwai,
Developmental Behavioural Pediatrician
Founder Director, New Horizons Child Development Centre,

EDITORIAL

With the inputs from learned experts and marathon activity for last Two years the most comprehensive Guidelines for IAPCNDP High Risk Infant Developmentally supportive follow up were published in Indian Pediatrics May2025 issue. Feel Honoured as lead author to share the Link for the Consensus Guidelines of the Indian Academy of Pediatrics (IAP)—Neurodevelopmental Pediatrics Chapter on Developmentally Supportive Follow-Up for High-Risk Infants. https://rdcu.be/eqq7H.

Essential Reading for all dealing with Infant Care:

- Very practical Consensus Guideline on Developmentally Supportive Follow-Up of High-Risk Infants
- Clear definition of high-risk infants with color-coded risk stratification for easy identification.
- Habilitation right of every paediatrician
- Precise neonatal and follow-up care strategies with key monitoring and intervention methods.
- Screening, diagnostics, and genetic disorder insights with structured test recommendations.
- Growth monitoring, feeding, drug administration, and early stimulation guidelines for attainment of maximum potential
- Timeline for investigations like ROP, X-ray hip, BERA, MRI, and cerebral cooling assessments clearly outlined.
- Concise background information and boxed summaries for quick reference and better understanding.
- Clear simple holistic early intervention strategies which the care giver can explain to parents.
- It is an indispensable tool for improving infant care. Read, share, and implement these recommendations to make a meaningful impact!

Best Regards

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Knowledge, Attitudes, and Practices (KAP) Study on Autism Spectrum Disorder (ASD) awareness among pediatric residents

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Abstract

Introduction

ASD is a neurodevelopmental condition characterized by deficits in social communication and restrictive, repetitive behaviors. Early identification and intervention are critical for improving outcomes. Despite increasing awareness, gaps in ASD-related knowledge among pediatric residents persist, affecting early diagnosis and management.

Aims and Objectives

1. Assessing the residents' understanding of ASD, including its diagnostic criteria, prevalence, and associated factors.2.Evaluating their attitudes toward ASD, including perceptions, confidence in managing cases, and potential biases. 3 .Analyzing current practices in screening and management. 4.To identify obstacles pediatric residents face in ASD diagnosis and care.

Materials and Methods:

A questionnaire-based survey was conducted across multiple medical institutions in India. Pediatric residents pursuing MD, DNB, and DCH were invited to participate by disseminating the questionnaire through Google Forms. Responses were analyzed statistically using Microsoft Excel, with appropriate tests applied to determine significant variations. Ethical approval was obtained, and informed consent was secured from all participants.

Results : A total of 53 pediatric residents participated. The mean knowledge score was 79.23%, indicating a strong theoretical understanding of ASD. However, attitude scores were moderate (49.81%), suggesting gaps in confidence and perceptions. Practice scores were high (91.04%), demonstrating adherence to clinical guidelines. No strong correlations were found between knowledge, attitudes, and practices, indicating that higher knowledge did not necessarily translate into improved attitudes or clinical confidence.

Conclusions: While pediatric residents exhibit strong knowledge and clinical practices regarding ASD, their attitudes remain moderate, highlighting the need for improved training programs. Enhancing autism-specific education and increasing clinical exposure through structured workshops and interactive sessions can solve this issue.

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Keywords:

- Autism Spectrum Disorder (ASD)
- Pediatric Residents
- Developmental Screening
- ASD Prevalence

Introduction

ASD is a group of complex neurodevelopmental disorders marked by difficulties in social communication and interaction, as well as repetitive behaviors, restricted interests, and structured activity patterns [1,2,3]. Previously, ASD was classified into subtypes like autistic disorder, Asperger syndrome, and pervasive developmental disorder-not otherwise specified (PDD-NOS), but these are now considered under the broader ASD diagnosis in DSM-5[4].

According to the World Health Organization (WHO), approximately 1 in 160 children worldwide has ASD. Studies suggest variability in ASD prevalence based on geographic factors, with some findings indicating higher rates in rural areas[5]. In India, a study conducted in Chandigarh found that 2.25 per 1000 children are diagnosed with ASD[6]. Males are more prone to ASD than females, with a male-to-female ratio of approximately 3:1[7].

ASD arises from a combination of genetic predisposition and environmental influences. A meta-analysis reported that if one identical twin has ASD, the chances of the other twin being affected range from 64% to 91%[8]. The Interactive Autism Network also reported that identical twins often share similar levels of autistic traits, reinforcing the genetic influence on ASD[8]. Mutations in genes such as CHD8, DYRK1A, and SHANK3 are among those identified in individuals with autism[9]. Genetic research distinguishes between syndromic autism, which is associated with other genetic syndromes like Rett syndrome, Fragile X syndrome, and Tuberous Sclerosis Complex, and non-syndromic autism, which occurs without other identifiable syndromic conditions[10].

Prenatal and perinatal factors such as older parental age at the time of conception, maternal illnesses during pregnancy, extreme prematurity, low birth weight, and complications during birth such as hypoxia have been linked to an increased risk of autism[11]. Exposure to hazardous environmental substances, such as lead and mercury, pesticides, and air pollutants, during critical periods of brain development may contribute to the risk[12].

ASD is characterized by two primary domains of core symptoms:[4]

1. Social Communication Challenges
2. Restrictive and Repetitive Behaviors

The various diagnostic methods available are:[13]

1. Developmental Screening
2. Comprehensive Diagnostic Evaluation
3. Additional Assessments
   - Hearing and Vision Tests
   - Genetic Testing
   - Speech and Language Evaluation

Effective ASD management relies on multidisciplinary interventions, including:

1. Applied Behavior Analysis: Involves detailed behavioral assessments and individualized intervention plans.
2. Speech Therapy: Targets both verbal and non-verbal communication deficits.
3. Occupational Therapy: Helps individuals with autism improve skills needed for daily living.

Some special educational Strategies for children suffering from ASD include:

1. Individualized Education Plans
2. Visual Supports
3. Structured Teaching

Awareness of ASD among medical practitioners, and the general public varies significantly across regions and professions. Awareness among healthcare professionals can lead to timely and accurate diagnosis, which is essential for accessing early support measures that can significantly improve a child's developmental trajectory[11]. Educated communities are better equipped to support individuals with autism, from recognizing early signs to advocating for necessary resources and accommodations in educational and social settings[14].

The relevance of this study is rooted in its potential to enhance pediatric practice and patient outcomes. Increased understanding and acceptance of autism can lead to better support systems, greater advocacy for resources, and enhanced quality of life for autistic individuals and their loved ones.

Aim and Objectives

Aim

To assess the knowledge, attitudes, and practices

(KAP) of pediatric residents regarding Autism Spectrum Disorder (ASD).

Objectives

1. To evaluate knowledge of ASD, including its diagnostic criteria, clinical presentation, risk factors, and available interventions among residents of Pediatrics.
2. To assess their attitudes towards ASD, including perceptions of children with autism, confidence in managing ASD cases, and any existing misconceptions or biases.
3. To analyze current clinical practices among pediatric residents related to ASD.
4. To identify challenges and barriers faced by pediatric residents in diagnosing and managing ASD, such as inadequate training, lack of resources, and systemic limitations.

Materials and Methods:

Study Setting:

This is a study conducted among pediatric residents across various medical institutions in India.

Study Design:

A cross-sectional, questionnaire-based study designed to assess the knowledge, attitudes, and practices (KAP) regarding ASD.

Study Period:

This study took place over three months, from January 2025 to March 2025.

Study Population:

The study population consists of pediatric residents from medical institutions across India who are currently pursuing:

- MD (Doctor of Medicine)
- DNB (Diplomate of National Board)
- DCH (Diploma in Child Health)

Inclusion Criteria:

- Pediatric residents currently enrolled in medical colleges across India.
- Residents who voluntarily agree to participate and provide informed consent.

Exclusion Criteria:

- Pediatric residents who have completed their final examination and are no longer in training.
- Residents who do not provide consent for participation.

Sampling Technique:

A convenience sampling method will be used to recruit participants. The questionnaire will be disseminated electronically.

Data Collection Tool:

A Google Forms-based questionnaire will be used to collect responses from participants. The questionnaire is designed to evaluate knowledge, attitudes, and practices related to ASD.

Methodology:

Study participants will receive a structured, self-administered questionnaire via digital platforms. The responses will be collected electronically and compiled for further analysis.

Data Analysis:

- The data will be meticulously collected, expertly compiled, and rigorously analyzed using Microsoft Excel.
- Additionally, appropriate statistical tests will be applied to find any variations.

Ethical Considerations:

In adherence to ethical principles, this research study will ensure voluntary participation through an informed consent.

Results:

A total of 53 pediatric residents from various institutions in India participated in this study. The following observations were recorded (Table 1–4).

The mean scores for each domain of the KAP study were as follows:

Among the 53 pediatric residents in the study, 72.9%, 18.8% and 8.3% were pursuing MD, DCh and DNB degrees (Table 1).

Table 1. Distribution of Pediatric Residents by Course Pursued:

Course Pursuing	Number	Percentage
MD	39	72.9%
DCh	10	18.8%
DNB	4	8.3%

79.23% of the knowledge based questions were answered correctly (Table 2).

Table 2. Responses to Knowledge-Based Questions on ASD:

Knowledge about ASD	Number	Percentage
1.What kind of a disease is ASD?		
• Autoimmune	0	0
• Metabolic	0	0
• Neurodevelopmental	53	100
• Degenerative	0	0
2.ASD always has a genetic cause.		
• True	4	7.54
• False	49	92.45
3.What is the prevalence of ASD in India?		
• 1 in 1000 children	15	28.30
• 1 in 50 children	14	26.41
• 1 in 500 children	14	26.41
• 1 in 250 children	10	18.87
4.Which among the following is seen to be associated with ASD?		
• Mental retardation	22	41.50
• Reduced danger awareness	27	50.94
• Seizures	3	5.66
• Increased sexual drive	1	1.89
5.Name three symptoms or signs of ASD. (Multiple answers)		
• Repetitive behavior	26	NA
• No eye contact	14	NA
• Social isolation	34	NA
• Language delay	24	NA
• Sensory problems	3	NA
• Attention to detail	1	NA
• Self harm	2	NA
• Hyperactive	3	NA
• Poor school performance	2	NA
• Poor attention	4	NA
• Sensitivity to sound	1	NA
6.What is the best way to manage ASD?		
• Behavioral therapy	52	98.11
• Medications	1	1.89
• Hospitalisation	0	0
• Isolation from other children	0	0

7.What are the major reasons for the increasing prevalence of ASD? (Multiple answers)		
• Increased awareness	7	NA
• Increased screentime	24	NA
• Nuclear families	13	NA
• Genetic factors	5	NA
• Environmental factors	4	NA
• Poor parenting	7	NA
• Single child	3	NA
• Better diagnostics	2	NA
• Widespread screening	3	NA
8.What is your source of information about ASD?		
• Newspapers and Journals	2	3.77
• Medical textbooks	29	54.71
• Physicians and health care workers	17	32.07
• Internet	3	5.66
• Social media	1	1.89
• Parent of autistic child	1	1.89
9.ASD is more common in boys than girls		
• True	46	86.79
• False	7	13.20
10.Onset of ASD is usually in		
• Childhood	34	64.15
• Infantile period	18	33.96
• Neonatal period	1	1.89
11.Blood investigations are required for the confirmation of diagnosis of ASD		
• False	51	96.22
• True	2	3.77
12.ASD is not just a disease of children but could also progress into adulthood		
• True	50	94.34
• False	3	5.66
13.Which investigation is least useful in the routine workup of ASD?		
Thyroid function tests	2	3.77
Serum lead levels	7	13.20
Genetic testing	2	3.77
Complete blood count	42	79.23

Although the correct prevalence of ASD in India is 1 in 50 children, only 26.41% of respondents identified this correctly, indicating a gap in awareness.

Although mental retardation is the most accurately recognized association with ASD (41.50%), more than half of the respondents selected other options, highlighting gaps in understanding.

While ASD typically begins in the infantile period, only 33.96% of respondents identified this correctly, with the majority (64.15%) selecting childhood and a small minority (1.89%) incorrectly choosing the neonatal period.

49.81% of the attitude based questions were answered correctly (Table 3).

Table 3. Responses to Attitude-Based Statements About ASD:

Practice about ASD	Number	Percentage
1.Routine screening and social awareness programmes are needed for ASD at schools and kindergartens.		
• Strongly agree	36	67.92
• Agree	15	28.30
• Disagree	1	1.89
• Strongly disagree	1	1.89
2. Mainstream schools should provide children with ASD the necessary accommodations and support.		
• Strongly agree	14	26.41
• Agree	26	49.05
• Disagree	13	24.53
• Strongly disagree	0	0
3. Allowing parents to be present in the classroom can enhance the support that kindergartens provide for children with ASD.		
• Strongly agree	8	15.09
• Agree	30	56.60
• Disagree	15	28.30
• Strongly disagree	0	0
4. Preschools should employ specialized educators and therapists to assist children with ASD in their learning environment.		
• Strongly agree	28	52.83
• Agree	21	39.62
• Disagree	1	1.89
• Strongly disagree	3	5.66
5. Health insurance policies need modifications to ensure they provide coverage for ASD?		
• Strongly agree	21	39.62
• Agree	28	52.83
• Disagree	4	7.55
• Strongly disagree	0	0

The vast majority of respondents (96.22%) supported the need for routine screening and social awareness programs for ASD in schools and kindergartens, with 67.92% strongly agreeing and 28.30% agreeing, while only 3.78% disagreed.

A significant majority (92.45%) favored the inclusion of special education teachers and therapists in preschools for children with ASD, with 52.83% strongly endorsing the idea and 39.62% agreeing, while only a small fraction (7.55%) opposed it.

91.04% of the practice based questions were answered correctly (Table 4).

Table 4. Responses to Practice-Based Questions Regarding ASD:

Practices regarding ASD	Number	Percentage
1.Do you know anyone who is possibly suffering from ASD?		
• Yes	23	43.40
• No	30	56.60
2.How would you feel if you were to spend some time with someone who is having ASD?		
• I would spend time to understand more about ASD	51	96.23
• Comfortable, because I feel I myself too have ASD	2	3.77
• I would prefer not to spend time because I am worried I might also develop some traits of ASD from him/her	0	0
• I would absolutely not spend time because they might harm me	0	0
3.If you were to come across a person suffering from ASD, what are the ways in which you would help him/her to have a better social life?		
• Teach them to greet people	4	7.56
• Simple story method to explain life situations	2	3.77
• Advise behavioral therapy	13	24.53
• Reduce screen time	7	13.21
• Neurodevelopmental screening	3	5.66
• Parental counselling	3	5.66
• Structured classroom program advisory	1	1.89
• Give comfort to express himself/herself	7	13.21
• Help with daily needs	3	5.66
• Improve family time	4	7.56
• Communication exercises	6	11.32
4.Would you attend a workshop or training on ASD if you were given an opportunity?		
• Yes	52	98.11
• No	1	1.89

A notable 43.40% of respondents reported knowing someone who may have ASD, while the majority (56.60%) did not.

An overwhelming majority (96.23%) expressed a willingness to spend time with an autistic individual to better understand autism.

a. **Knowledge (K) Score** (out of 13): Mean = 10.30, Median = 10, SD = 1.88
b. **Attitude (A) Score** (out of 5): Mean = 2.49, Median = 3, SD = 1.09
c. **Practice (P) Score** (out of 4): Mean = 3.64, Median = 4, SD = 0.48

These findings indicate that while knowledge about ASD is high (79.23%) and practices are well-adopted (91.04%), attitudes remain moderate (49.81%), suggesting areas for improvement in perceptions and confidence in managing ASD. (As illustrated in Figure 1)

Pearson correlation tests revealed:

- Knowledge vs. Attitude: 0.058 (very weak correlation)
- Knowledge vs. Practice: -0.090 (very weak correlation)
- Attitude vs. Practice: 0.048 (very weak correlation)

These results indicate no strong correlation between K, A, and P scores, meaning better knowledge does not necessarily lead to improved attitudes or practices.

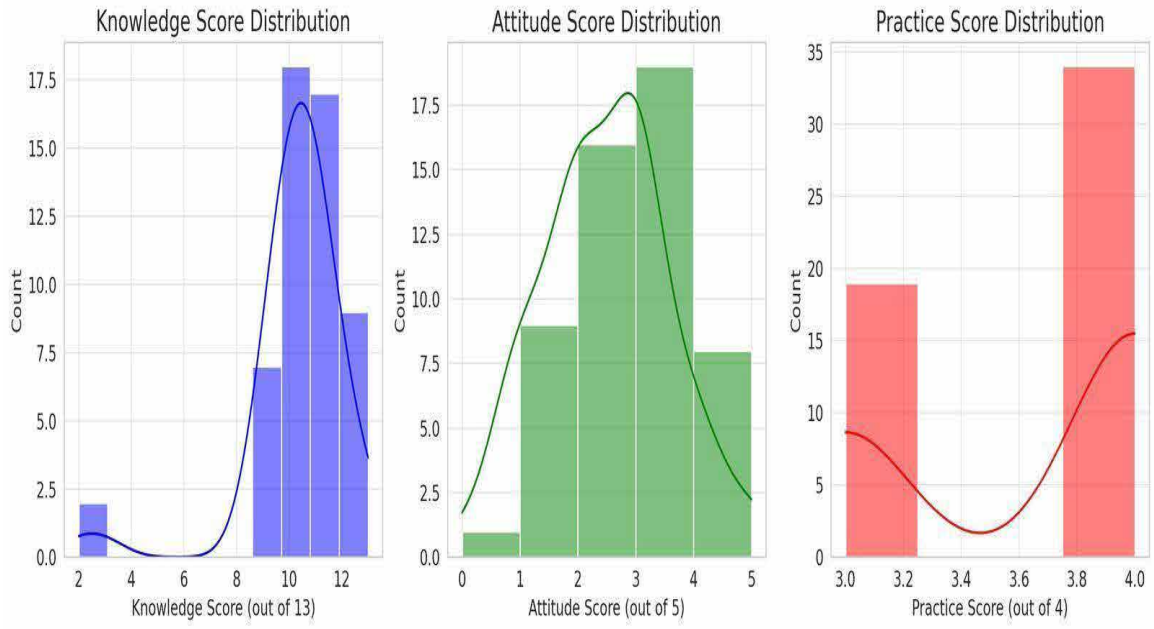

Figure 1. Distribution of Knowledge, Attitude, and Practice Scores Among Pediatric Residents

Discussion:

The study highlights that ASD is widely recognized as a neurodevelopmental disorder, though misconceptions about its cause persist. While 92.45% correctly identified that ASD does not always have a genetic cause, awareness regarding its prevalence in India was varied, with only 26.41% selecting the correct estimate (1 in 50 children).

41.50% identified mental retardation as a common comorbidity of ASD, though other factors such as reduced danger awareness (50.94%) were also mentioned. The most commonly recognized symptoms included social isolation (34%) and repetitive behavior (26%) reflecting general awareness of autistic traits.

A vast majority (98.11%) correctly identified behavioral therapy as the best management approach. Similarly, physicians express mixed views on alternative methods in ASD, with some approaches lacking scientific validation[15]. Misconceptions regarding the diagnostic process exist, as 3.77% incorrectly believed blood investigations were required. Encouragingly, 94.34% acknowledged that ASD persists into adulthood, reflecting an understanding of its lifelong nature.

The study also explored perceptions of ASD in educational settings. While 96.22% supported routine screening and awareness programs in schools, and 92.45% advocated for special educators in preschools, fewer respondents (75.46%) supported full inclusion of autistic children in regular schools. 71.69% agreed that parents should be allowed in classrooms.

When examining perceived factors behind the

increasing prevalence of ASD, the most cited reasons were increased screen time (24), nuclear families (13), and increased awareness (7).

In terms of social interactions and attitudes, 43.40% reported knowing someone with ASD, and 96.23% expressed willingness to engage with autistic individuals to understand them better. When asked about supporting autistic individuals, behavioral therapy (24.53%) was the most recommended approach, followed by reducing screen time (13.21%). Encouragingly, 98.11% were open to attending ASD workshops, reflecting strong interest in learning more.

Overall, the findings suggest good general awareness of ASD but reveal gaps in knowledge. Similar patterns have been reported in Indian studies exploring beliefs among healthcare disciplines[16]. While awareness of ASD among healthcare professionals is widespread, knowledge gaps persist in key areas such as early diagnosis and intervention strategies[17,18]. A systematic review found that healthcare providers' knowledge of ASD varied significantly, with correct response rates ranging from 47.37% to 71.05% on ASD knowledge assessments, indicating a need for enhanced training[19]. In some regions, such as Australia, about 62% of general practitioners achieved a high score in ASD knowledge tests[20], while in the USA, a substantial proportion of both pre-qualified medical students and practicing clinicians rated their knowledge as merely 'somewhat informed' or lower[19]. The lack of specialized healthcare professionals, such as developmental pediatricians and child psychologists, can hinder timely and accurate diagnosis and effective treatment[21]. Cultural perceptions and linguistic barriers can hinder ASD awareness, leading to underreporting and delayed interventions[22].

The results align with previous studies showing that healthcare professionals often have sufficient knowledge about ASD but may lack confidence in its management[23]. Despite high knowledge scores, moderate attitudes indicate possible misconceptions or lack of hands-on exposure. Factors like firsthand engagement with individuals diagnosed with ASD and demographic elements influence attitudes and knowledge levels. A study found that direct interaction with autistic individuals is a significant predictor of positive attitudes, although knowledge alone, without such contact, did not consistently predict positive attitudes across different ethnic groups[14].

Future training programs should focus on improving attitudes by incorporating real-world case discussions, role-playing, and patient interactions. Surveys indicate that many residents feel inadequately prepared to address ASD, with deficiencies in their ability to conduct developmental screenings and manage ASD-related care[24,25]. Enhancing training programs to include comprehensive, evidence-based education on ASD can improve early detection rates and the overall quality of care provided to autistic children[26].

Conclusion

The study reveals a complex landscape of strengths and areas for growth among pediatric residents. Overall, these residents demonstrate solid knowledge of ASD, suggesting they are well-versed in its core characteristics, diagnosis, and general management. Their clinical practices, how they apply this knowledge in real-world settings, are also commendable, indicating effective patient handling and adherence to established medical protocols.

However, the study identified only a moderate level of attitude towards individuals with ASD. This suggests a potential emotional or empathetic disconnect that could influence patient-family interactions or long-term commitment to ASD care. Interestingly, there was no significant correlation found between knowledge, attitude, and practice. In essence, possessing more knowledge didn't necessarily lead to better attitudes or improved clinical behavior, and vice versa.

Key Findings:

- Pediatric residents have good knowledge but only moderate attitudes towards ASD
- Clinical practices related to ASD management are good
- No significant correlation was found between knowledge, attitude, and practice meaning that one does not affect the other

Recommendations:

1. Incorporate mandatory autism training into Pediatric residency programs which is holistic in nature that not only builds knowledge and skills

but also fosters empathy, cultural competence, and reflective practice.

2. Increase clinical exposure through ASD-specific rotations.
3. Develop interactive workshops and case-based discussions.

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Early Identification of Sensorineural Hearing Loss in a Neonate via Universal Screening : A Case Report as a Catalyst for State-Level Programmatic Change

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Abstract

Introduction:

Hearing impairment is one of the most common congenital conditions, yet early identification remains a challenge in low-resource settings.

Case summary:

We present a case from January 2003, during an early institutional attempt at newborn hearing screening, in which a neonate with moderate-to-severe bilateral sensorineural hearing loss was successfully diagnosed by Otoocoustic emission(OAE) screening and subsequently confirmed by Auditory Brainstem response (ABR) and rehabilitated by 6 months of age. The outcome—age-appropriate speech development and successful mainstream schooling—provided compelling momentum for scaling up district-wide screening initiatives.

Conclusion:

This report highlights the utility of OAE and ABR-based protocols in influencing health system planning and emphasizes the need for robust public health strategies to implement universal newborn screening in resource-constrained contexts. It documents how that clinical milestone inspired the Indian Academy of Pediatrics (IAP) Cochin Branch to spearhead a scalable model that evolved into a state-wide initiative by 2020. The Kerala experience offers a replicable pathway for other regions aiming to implement UNHS aligned with SDG 3.2.

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Keywords:

- Newborn Hearing Screening
- Public Health Implementation
- Kerala Model
- Universal Screening
- Pediatric Audiology

Background :

At the time of this case, newborn hearing screening was virtually non-existent in many Indian healthcare settings, with limited awareness among professionals and policy makers. There was minimal published evidence locally, and structured protocols for early detection and intervention were not in place.

Case Presentation:

Between January and March 2003, during a pilot phase of institutional newborn hearing screening in a tertiary care setting with low baseline awareness and no prior implementation protocol, 504 neonates were screened using universal otoacoustic emission (OAE).

Out of the screened cohort, one neonate failed the initial OAE test in both ears. This child also presented with significant neonatal hyperbilirubinemia, a known risk factor for auditory dysfunction.

Investigations:

The infant underwent initial otoacoustic emission (OAE) screening, which failed in both ears, prompting further evaluation. Auditory Brainstem Response (ABR) testing subsequently confirmed the presence of bilateral sensorineural hearing loss. Comprehensive assessments were conducted, including consultations with ENT specialists and pediatric neurology. A CT scan of the brain yielded normal results. However, genetic testing was not available at the time and could not be performed.

Treatment

- Early amplification with bilateral hearing aids
- Enrolment in a structured auditory-verbal therapy program
- Family counselling and speech-language follow-up

Differential Diagnosis

The following differentials were considered:

- Auditory Neuropathy Spectrum Disorder (ANSD) Given the association with hyperbilirubinemia, ANSD was a plausible consideration. However, consistent ABR findings and good response to amplification reduced its likelihood.
- Syndromic Hearing Loss Syndromes such as Jervell-Lange-Nielsen or Usher syndrome were considered, but there were no dysmorphic features or systemic abnormalities identified.
- Hyperbilirubinemia-Induced Auditory Toxicity Direct bilirubin toxicity affecting the auditory nerve or brainstem was considered the most probable etiology in the context of elevated bilirubin levels and absence of other systemic involvement.
- Genetic Non-Syndromic Sensorineural Hearing Loss Could not be ruled out due to the lack of genetic testing; however, the timing and severity aligned more closely with the clinical picture of bilirubin-induced damage.

Outcome and Follow-Up

The infant demonstrated:

- Age-appropriate auditory and speech-language development within expected milestones
- Effective use of hearing aids with regular audiological follow-up
- Normal cognitive and social development, leading to integration in mainstream schooling
- No signs of regression or additional disability through early childhood and school entry
- Parental support and early intervention services were pivotal in ensuring optimal rehabilitation and long-term functional outcomes.

This experience set the foundation to ponder implementation enablers—like responsive leadership, clinical vigilance, and the power of single-case advocacy—to build systemic newborn screening programs.

Discussion

This case underscores how one early success in newborn hearing screening within a resource-limited setting can serve as a policy lever. It served as a compelling "proof of concept," prompting clinicians and policymakers to ask: What if we could extend this benefit to every child born in Kerala?

a) **Building the Statewide NBHSP**

Phased Implementation

Phase	Timeframe	Coverage	Highlights
I: City Pilot	2003–2014	31 → 91 hospitals in Ernakulam	Coordinated by IAP Cochin Branch
II: State Rollout	2014–2020	516 hospitals (84 govt, 432 private)	Kerala declared "Hearing Friendly"

Screening Protocol

1. **OAE Screening**: For all newborns before discharge
2. **ABR Audiometry**: For NICU babies and those failing OAE twice (4-week interval)

b) **Stakeholder Engagement and Resource Optimization**

- *Partners:* IAP district branches, private audiology firms, hospital administrators
- *Strategies:*
  - Sharing portable OAE devices among facilities
  - Audiology services outsourced or pooled through regional hubs

- IAP-led procurement and training

- *Incentive:* Districts declared "Hearing Friendly" upon full compliance

c) **Outcomes**

Expansion timeline

- 2014: Ernakulam became first "Hearing Friendly" district
- 2020: All 14 districts onboarded; Wayanad was the final district to comply
- 516 facilities: Implemented screening before newborn discharge
- Universal ABR for NICU neonates: Enhanced detection of auditory neuropathy

Impact Snapshot:

Table 2: Screening coverage

Indicator	Outcome
Coverage	100% facility-level implementation statewide
Detection	High-risk infants diagnosed before 3 months
Intervention	Hearing aids fitted within therapeutic window
Systemic change	Proof-of-concept case → Full-state policy shift

d) **Key Insights**

The implementation of Kerala's statewide newborn hearing screening program was notably influenced by a single sentinel case that demonstrated the transformative impact of early identification and intervention. This case served as a powerful catalyst for broader systems change, illustrating how targeted clinical evidence can drive policy decisions when strategically leveraged.

Centralized coordination emerged as a vital enabler, allowing efficient resource allocation, streamlined training, and scalable implementation across public and private health facilities. By minimizing redundancy and maximizing cost-sharing, this approach ensured sustainability and uniform service delivery.

Collaboration between government entities, professional associations like the Indian Academy of Pediatrics, and private audiology providers proved essential. These partnerships extended service reach, particularly in underserved and resource-constrained areas, reinforcing the equity goals of the program.

Furthermore, the adoption of district-wise milestones created a sense of accountability and momentum. Declaring districts as "Hearing Friendly" only after achieving full implementation galvanized local health systems and encouraged structured, phased adoption throughout the state.

Comparison to Global Practices

Region	UNHS Implementation	Kerala's Approach
High-Income Countries	Since 1999	Tech-intensive, insurance-funded
Kerala	Since 2003 (full by 2020)	Cost-shared, centrally coordinated, policy-integrated

e) **Policy Implications and Future Directions**

- National Scaling Potential: Through IAP regional chapters and RBSK integration
- SDG 3.2 Alignment: Advances early diagnosis, inclusive education, and equity in child health
- Replicability Framework:
  - Case-based advocacy → Clinical piloting
  - Multi-stakeholder platform → District accountability
  - Monitoring & IEC → Community ownership

Conclusion

Kerala's newborn hearing screening journey—from a single success story in 2003 to a statewide movement by 2020—demonstrates how clinical data can be transformed into public health action. With clear stakeholder coordination, context-sensitive design, and policy anchoring, universal newborn hearing screening is not only feasible but scalable—even in resource-limited settings

Key points:

Proof of concept: Identification of hearing loss before 2 months of age, with intervention by 6 months, mirrored outcomes seen in high-income settings—normal communication and learning outcomes.

Role of hyperbilirubinemia: Reinforces its

significance as a red flag for auditory follow-up, aligning with global Joint Committee on Infant Hearing (JCIH) guidelines.

Low yield but high impact: Despite identifying only one case in 504, the life-changing impact on that child served as a powerful advocacy example.

Programmatic momentum: This case was pivotal in initiating advocacy for district-level adoption of Universal Newborn Hearing Screening (UNHS), supported by Health system stakeholders, Pediatric and audiology communities, and Community sensitization efforts

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Conflict of interest statement

"None declared."

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Criterion validation of 'learning difficulty related items' in INCLEN- NDST against NIMHANS battery among children with suspected learning problems

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Abstract

Background:

The prevalence of learning difficulties is increasing in recent years as a result of better awareness, support systems and better neonatal care. Many tools available in this domain are western and effective screening tools are rare in the Indian context.

Objective:

To evaluate the criterion validity of the Neurodevelopmental Screening Tool (NDST-Research form) as a screening tool for learning difficulties in children aged 7-13 years, using the NIMHANS Battery as the gold standard diagnostic tool.

Methods:

This hospital-based criterion validation study was conducted, over eight months. Forty consecutive children aged 7-13 years with suspected learning problems were screened using the NDST-Research form, followed by assessment with the NIMHANS Battery for Specific Learning Disability (SLD). The NDST-Research form was administered by a Developmental Nurse Counsellor, while the NIMHANS Battery was conducted by a consultant clinical psychologist, blinded to the screening results. Statistical analysis with SPSS version 20, calculated sensitivity, specificity, predictive values, diagnostic accuracy, and likelihood ratios.

Results:

NDST-Research form: 35 children (87.5%) screened positive for SLD, while 5 children (12.5%) did not. NIMHANS Battery results: 34 children (85%) diagnosed with SLD, while 6 children (15%) were not. NDST-Research form had a sensitivity: 97.06%; specificity: 66.7%; Positive Predictive Value (PPV): 94.29%; Negative Predictive Value (NPV): 80%; Positive Likelihood Ratio: 1.48; Negative Likelihood Ratio: 0.46, and Diagnostic Accuracy of 92.5%

Conclusion:

The acceptable psychometric properties suggest that NDST can be used in pediatric outpatient clinics for early identification, enabling timely intervention given the

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Keywords:

- Specific Learning Disorder
- NDST-Research Form
- NIMHANS Battery
- Pediatric Screening
- Learning Difficulties
- Criterion Validation

high prevalence of attention deficits, arithmetic difficulties, and reading/writing impairments to improve early detection and intervention strategies.

Introduction

According to DSM-5, Specific Learning Disorder is defined as persistent difficulties in reading, writing, arithmetic or mathematical reasoning, with academic skills well below average scores [1]. The NIMHANS battery for Specific Learning Disability (SLD) characterizes it as a performance level three grades below the child's current academic standard [2]. The INCLEN study, conducted across five regions in India, reported a community prevalence of SLD at 1.6% (95% CI: 1.0–2.5) among children aged 6 to 9 years, utilizing the Grade Level Assessment Device (GLAD) [3]. In a cross-sectional, school-based study in Ernakulum, the prevalence of SLD was found to be 16.49% (95% CI: 14.59–18.37), with impairments in reading, written expression, and mathematics being 12.57%, 15.6%, and 9.93%, respectively. Binary logistic regression analysis identified male gender, low birth weight, developmental delay, family history of poor scholastic performance, and syllabus as independent factors associated with SLD[4].

The administration of the NIMHANS battery in a school setting is impractical without a trained psychologist, necessitating an alternative approach. According to the Australian Disability Clearinghouse on Education and Training, learning difficulty is defined non-categorically, encompassing individuals who experience challenges in acquiring one or more basic academic skills[5] This definition emphasizes functional educational difficulties, which may be amenable to interventions, rather than the primary etiology. The Learning Disabilities Association of America has proposed broad-spectrum intervention strategies that may benefit children with learning difficulties[6]. Consequently, the objective of a screening tool should be to identify children at risk for learning difficulties, enabling the implementation of appropriate interventions.

The Neurodevelopmental Screening Tool (NDST-Research form), developed by the INCLEN-NDD study team under the leadership of Arora NK, is a 39-item screening instrument that assesses 10 neurodevelopmental disorders, including four specific questions related to SLD. A failure in any one item is considered a failure of the test. According to a gazette notification by the Ministry of Social Justice and Empowerment, the NIMHANS battery for SLD is designated as a diagnostic assessment tool for certifying benchmark disability in India[7].

Rationale: Diagnostic tools for identifying Specific Learning Disability require extended administration time, highlighting the need for screening to identify 'at-risk' children. Early identification during the initial school years (from age 7 onwards) is crucial for facilitating timely intervention. Therefore, this study aims to determine the criterion validity of the 'LD-related items' in the NDST-Research form as a screening tool, with the NIMHANS battery serving as the diagnostic tool administered by a trained psychologist.

Objectives

1. To administer neurodevelopmental screening tool (NDST- Research form) screening test among children in the age group of 7-13 years with suspected learning problems attending NIMS spectrum CDRC.
2. To administer NIMHANS battery for SLD tool among the same children.
3. Criterion validation of LD related items in neurodevelopmental screening tool (NDST-Research form) against (NIMHANS battery) as gold standard for specific learning disabilities and to calculate sensitivity, specificity, predictive values, diagnostic accuracy and likelihood ratios.

Methods

The present study is a hospital based criterion validation study which was carried out from January to August 2022 over a period of 8 months, at Thiruvananthapuram NIMS-Spectrum-Child Development Research Centre (CDRC), Noorul Islam Centre for Higher Education (NICHE), Deemed-to-be University. Forty consecutive children 7-13 years of age with suspected learning problems, coming to NIMS-Spectrum-CDRC, a tertiary care centre for children with neurodevelopmental problems, were included. Ethical clearance was obtained from Institutional Ethical Committee clearance (Regn. No. ECR/218/Inst/Ker/2013/RR-16 and Approval No. NIMS/IEC/2022/01/04, dtd. 10/01/2022) and due consent from each individual parent was obtained. Consecutive 40 children aged 7-13 years who attended the clinic with suspected learning

problems, with at least average intelligence, were included in the study. Children without a primary care giver and non-consenting parents were excluded. LD related items in NDST R/F were used as screening tool and NIMHANS battery for SLD Level 2 as the gold standard test.

Data was collected by interview method. Evaluation using NDST-Research form was done by Developmental Nurse Counsellor and evaluation using NIMHANS battery by consultant clinical psychologist trained in NIMHANS Battery, blind to the screening results. The analysis was performed using Statistical Package for Social Science (SPSS version 20).

Results

Out of the study population of 40 children,

- Age: 7-10 years – 15; 11-13years – 25.
- Gender: Male 25 (62.5%); Female 15 (37.5%).

Using NDST-Research form 35 (87.5%) children had at least one question positive suggestive of LD and 5 (12.5%) children did not have any one question positive suggestive of LD (Table.1).

Table 1: Distribution of 'SLD related items' in NDST-Research form (n= 40)

NDST No.	NDST Research form Items			
	School change	No	Required	Dropped out
55a.	Did your child change school/dropped out of school due to poor school performance?	32	8	0
	School performance	No	Some-times	Most times
56a.	Do the teachers complained about your child's poor performance in studies?	7	28	5
58a.	Does your child find it difficult to read or write or do simple calculations?	5	29	6
	Subject specific	No	One subject	Two or more
57a.	Does your child have significant difficulty in any subject?	9	17	14
NDST-Research form Impression = Not suggestive of LD: 5(12.5%); Suggestive of LD: 35(87.5%)				

This study showed that using NIMHANS battery, maximum number of children had issues with attention followed by poor arithmetic skills, reading skills, writing skills, comprehension, spelling, perceptual motor ability and auditory memory in descending order. (Table.2)

Table 2: LD in the NIMHANS Battery Domains (n= 40)

Domains	Adequate No. (%)	Not adequate/ need to improve No. (%)
Attention	7 (17.5)	33 (82.5)
Arithmetic	13 (32.5)	27 (67.5)
Reading	19 (47.5)	21 (52.5)
Writing	21 (52.5)	19 (47.5)
Comprehension	22 (55.0)	18 (45.0)
Spelling	25 (62.5)	15 (37.5)
Perceptual motor ability	28 (70.0)	12 (30.0)
Auditory memory	31 (77.5)	9 (22.5)
NIMHANS Battery Impression= No LD: 6 (15); LD=34 (85)		

On doing criterion validation of 'LD Related items' in NDST-Research form against NIMHANS Battery, the psychometric properties were as follows: sensitivity 97.06%; specificity 66.7%; positive predictive value (PPV) 94.29%; negative predictive value (NPV) 80%; positive likelihood ratio 1.48; negative likelihood ratio 0.46 and diagnostic accuracy 92.5%. (Table 3)

Table 3: 'LD Related items' in NDST-Research form vs NIMHANS Battery

NDST-Research form Impression	NIMHANS Battery Impression		Total
	LD	No LD	
LD	33 (TP)	2 (FP)	35
No LD	1 (FN)	4 (TN)	5
Total	34	6	40

Discussion

Specific Learning Disorder (SLD) is a neurodevelopmental condition that affects academic skills such as reading, writing, and arithmetic, often leading to long-term educational challenges if not identified early [8]. In India, the pooled prevalence of SLD in community and school settings has been estimated at 8% (95% CI: 4–11%), highlighting the need for early identification and intervention [9,10]. However, rather than confirming SLD at a young age, the priority should be identifying learning difficulties early to facilitate timely intervention [11]. The benefit for the participants of this study was the group administration of an intervention module in local language (Malayalam) that focussed on improving; (i) phonemic awareness; (ii) reading; (iii) writing; (iv) copying; (v) number concepts; (vi) mathematical reasoning; (vii) mathematical operations and (viii) visuospatial training.

This study assessed the NDST-Research form as a screening tool for learning difficulties, comparing its results with the NIMHANS Battery, a standardized diagnostic tool. However, the present study did not analyse the grade-level performance of the child while using NIMHANS battery because the intention was to use NDST as a validated screening tool. The findings indicate that 87.5% of children screened positive for learning difficulties using NDST, while 85% were diagnosed with SLD using the NIMHANS Battery, demonstrating strong agreement between the two tools. The criterion validation of NDST against the NIMHANS Battery showed high sensitivity (97.06%) and positive predictive value (94.29%), suggesting that NDST is effective in identifying children at risk for SLD. In this context, the observed sensitivity, Positive Predictive Value, Positive Likelihood Ratio and diagnostic accuracy of LD related items in NDST against NIMHANS battery suggests utility of this simple tool at clinic level. The acceptable psychometric properties of "LD related items" in NDST-research form make it a quick screening tool that could be used in the paediatric OPD setting as well. Among the domains assessed using the NIMHANS Battery, attention deficits were the most prevalent (82.5%), followed by difficulties in arithmetic (67.5%), reading (52.5%), writing (47.5%), comprehension (45%), spelling (37.5%), perceptual motor ability (30%), and auditory memory (22.5%). These findings align with previous studies indicating that attention deficits and arithmetic difficulties are common in children with learning disorders [8,11].

The NDST-Research form demonstrated acceptable psychometric properties, making it a quick and effective screening tool for learning difficulties in pediatric outpatient settings. Given its high sensitivity and diagnostic accuracy (92.5%), NDST can serve as a first-line screening tool, allowing clinicians to identify children who may require further assessment and intervention.

Conclusion:

This study highlights the utility of NDST-Research form as a validated screening tool for learning difficulties in children. The high sensitivity, positive predictive value, and diagnostic accuracy suggest that NDST can be effectively used in pediatric outpatient clinics to identify children at risk for SLD. The findings also emphasize the importance of early intervention, as children with

learning difficulties benefit from targeted support in phonemic awareness, reading, writing, number concepts, mathematical reasoning, and visuospatial training.

Given the high prevalence of learning difficulties, integrating NDST screening into routine pediatric assessments could facilitate early identification and intervention, ultimately improving academic outcomes for children with learning challenges. Future research should explore longitudinal outcomes of children identified through NDST and assess the impact of early intervention strategies on their academic performance.

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Limiting Cognitive Impairment in Genetic Disorders : Kabuki Syndrome – A Case Report

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Abstract

Kabuki Syndrome (KS) is a rare genetic disorder presenting in children with unique facial features, health issues, and developmental delays that are often mistaken for other syndromic diagnoses. Children with KS, in addition, develop short stature and growth delays later in life. Being a multisystem disorder, all findings may not be present at birth and may evolve, making it difficult for clinicians to diagnose clinically during the initial days of life. Growth retardation happens in the childhood for a child born with normal weight at birth. They go on to develop intellectual disability also later in life. It is known that delays in the language domain contribute to intellectual deficiency. Literature detailing the features of this disorder, especially in the socio-emotional and the language domain, and approaches to the same are deficient. The progress achieved, especially in the cognitive and language domain, is not sufficiently documented, probably due to the paucity of reported cases. In this article, we present a case description and language-based approach to development for limiting possible intellectual disability in a child with KS. A brief review of KS is also provided, highlighting its signs and symptoms.

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Keywords:
- Kabuki syndrome
- Cognitive impairment
- Polydactyl
- Everted nares
- REELS

Introduction

Kabuki Syndrome (KS) is a rare genetic disorder in children with unique facial features, health issues, and developmental delays that are often mistaken for other syndromic diagnoses. In addition, KS children develop short stature and growth delays [1]. It is a rare genetic disorder presenting in children, with unique facial features, health issues, and developmental delays that are often mistaken for other syndromic diagnoses. In addition, children with KS develop short stature and growth delays later in life. Being a multisystem disorder, not all findings may be present at birth and may evolve, making it difficult for clinicians to diagnose clinically during the initial days of life. Growth retardation occurs in childhood in children born with normal weight at birth. They continue to develop intellectual disabilities later in life. Delays in language are known to contribute to intellectual deficiency. Literature detailing the features of this disorder, especially in the socio-emotional and language domains, and approaches to the same are lacking. The possible progress that can be achieved, especially in the cognitive and language domains, has not been sufficiently documented, probably due to the paucity of reported cases. In this article, we present a case description and a language-based developmental approach to limit the possibility of intellectual disability in chil-

dren with KS. A brief review of KS has also been provided, highlighting its signs and symptoms.

Case Presentation

A 3.5-year-old girl was referred for speech and language evaluation following parental complaints of speech delay with a genetically proven diagnosis of KS. She had abnormal facies with wide eyes, bushy arched eyebrows, long eyelashes, depressed nasal bridge, broad nasal tip, cleft palate, large protruding ears, eversion of the lower lateral eyelids, sparse hair in the lateral one-third of the eyebrows, and Class I malocclusion. Many may also have a hairy body, and one-third may have polydactyly, as seen in Figure 1A-1D.

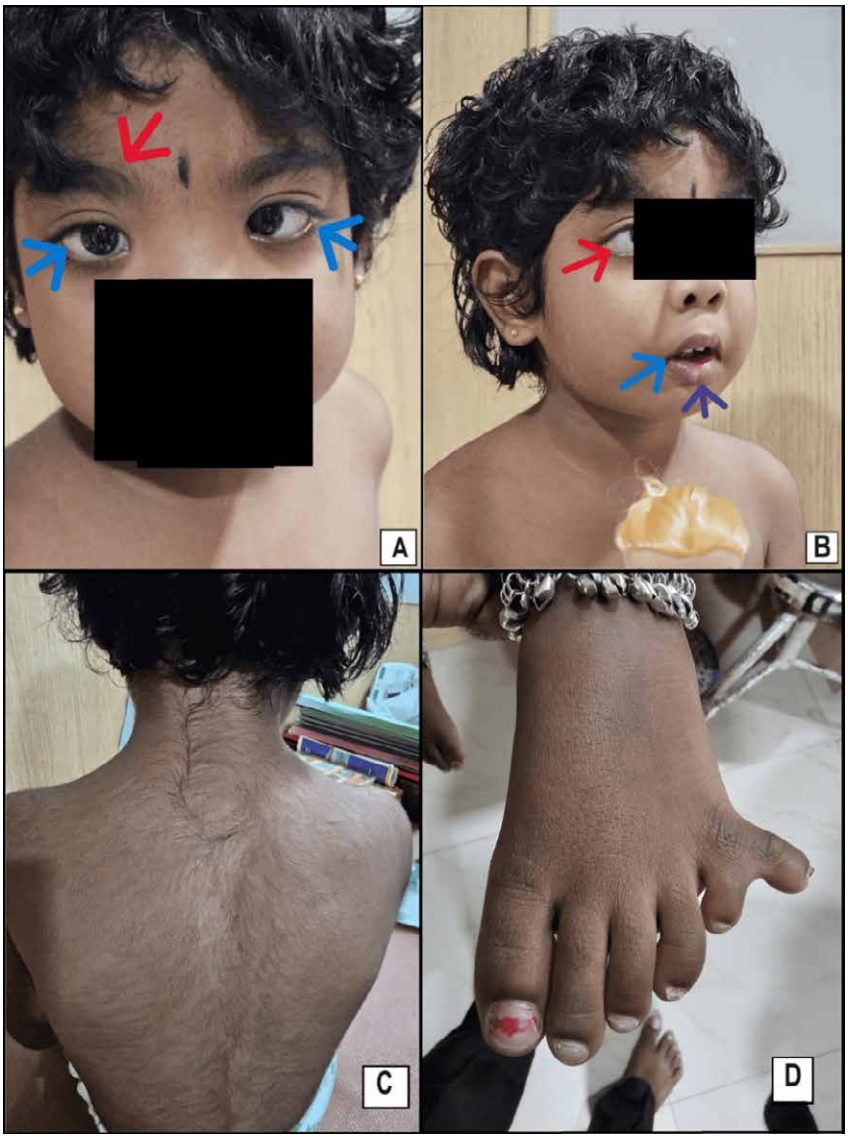

Figure 1: Dysmorphisms in Kabuki syndrome

(A) Long eyelashes with bushy arched eyebrows, depressed nose, and sparse hair on the lateral side of the eyebrow. (B) Wide eyes with everted eyelashes on the lateral side of the lower eyelids, lower lip pit, class one malocclusion, (C) hairy trunk, and (D) polydactyly. Red arrows- show the bushy eyebrows and lack of hair in the outer one-third of the eyebrows. Blue arrows- show the eversion of lateral one-third of eyelids, wide palpebral fissure, and long eyelashes. The purple arrow in Figure 1B shows the pit in the lower lip.

She weighed 10 kg (less than the third centile), and her height was 85 cm (less than the third centile), short of her age. She had a developmental delay with mild delays in gross and fine motor domains,

as she sat at 11 months, walked at 19 months, could only scribble, spill when she fed herself, and was unable to tell a single word, although she could understand and follow commands. She did not mingle with peers and was deficient in her preverbal skills. She was born with a birth weight of 2.9 Kg, developed difficulty in feeding during the postnatal period, and was on nasogastric feeding for a few days. During the initial years, the patient was hypotonic and developed recurrent chest infections. She underwent grommet insertion for recurrent middle ear infection. Her speech delay was initially attributed to middle ear pathology, which led to a delay in workup. Although she had an initial motor delay, she recovered well, and at presentation, she only had significant speech, language, and socio-emotional delays. Her full blood count was normal; the absence seizure diagnosed with the electroencephalogram (EEG) was treated medically; MRI was normal; and the echocardiogram(ECHO) showed an ostium secundum type of atrial septal defect and a small patent ductus arteriosus. Multisystem involvement and developmental delay led to suspicion of a genetic disorder, and whole exome sequencing was performed, which was reported as a KMT2D mutation. Wechsler's Preschool and Primary School (WPPS-IV) assessment showed that the Full-Scale Intelligence Quotient (FSIQ) score was within the low average range for average verbal comprehension, below average visual processing, low average working memory, fluid reasoning, and speed of processing. The child had limitations in intelligence, as expected, which is bound to increase with age, considering the language and fine-motor limitations. Her formal speech assessment showed that the child communicated mainly through the gestural mode. She followed a two-step command. A few misarticulation (dental, fricative, and sibilant) errors were noted, which were attributed to dental malocclusion (class I malocclusion). Simplification of clusters was also observed in addition to prosody and articulation errors, which became pronounced following speech therapy since the spontaneous verbal utterances increased in length and complexity, negatively impacting intelligibility. Abnormal oral resonance was also observed owing to the high-arched palate and oral motor hypotonia. The Communication Deal Developmental Checklist (CDDC) was administered, and its oro-motor assessment subscale showed oro-motor deficiency. The Auditory Brainstem Response test (ABR test) showed the 5th wave at 31.1/s repetition rate using click stimuli and rarefaction polarity until 50 dBnHL for the right ear and 45 dBnHL for the left ear. This indicated mild hearing loss in the right ear, and minimal hearing loss in the left ear. These findings could not explain the initial significant language and speech delays she exhibited. Immittance audiometry showed a B-type tympanogram bilaterally, with an ear canal volume of 0.4 cm3, and an acoustic reflex test showed the absence of ipsilateral reflex (left ear, no reflex). An otolaryngology consultation was performed, corrective measures were taken, and the child was given speech therapy along with developmental therapy for the affected domains.

The child attended therapy sessions and formal school. The speech therapist worked on low and high vowels with visual feedback, syllable repetition, articulatory placements, auditory feedback, and oro-motor interventions aimed at the muscles involved in cheek and tongue movements. Once the child began experimenting with sounds, the phonological and developmental errors that crept in were managed successfully. A plastic surgery review of the corrected cleft palate was performed because of the velopharyngeal insufficiency. Mild hypernasality and audible nasal turbulence due to the presence of obligatory velopharyngeal insufficiency were corrected by reducing the loudness, which in turn was thought to contribute to the velopharyngeal insufficiency. Developmental therapists, who work on multiple domains simultaneously by selecting targeted activities based on the level of development of each domain, work in tandem with the speech therapist to produce better expression of speech and language by the child. The CDDC noted improvement in the developmental domains of gross motor (34 months), fine motor (30 months), activities of daily living (34 months), receptive language (30 months), expressive (22 months), cognition (30 months), social (32 months), and emotional (32 months). The WPPS-IV showed a significant improvement in intelligence following the intervention. The Receptive Emergent Expressive Language Scale 4 (REELS-4) score improved receptive and expressive language as in *Table 1*.

Test	Pre-intervention Findings	Post intervention findings
WPPS		
FSIQ	low average; SS=89; PR= 23.	SS=99, PR= 47, CI: 93-105
verbal comprehension	average range (SS= 92; PR= 30)	average range (SS= 92, PR= 30)
visual processing	below average range (SS= 80; PR= 9)	low average range (SS= 85, PR= 16)
working memory	low average range (SS= 85; PR=16)	low average range (SS= 87, PR= 19)
fluid reasoning	low average range (SS= 88; PR= 21)	average range (SS= 94, PR= 34)
speed of processing	low average range (SS= 87; PR=19)	average range (SS= 94, PR= 34)
REELS-4		
RLA	18-20 months	28-32 months
ELA	11-12 months	16-18 months

Table 1: Improvement in Intelligence and Language abilities following interventions

WPPS-4-Wechsler Preschool and Primary Scale of Intelligence Fourth Edition; SS- standard score; PR- percentile; REELS-Receptive Expressive Emergent Language-4th edition; RLA_ Receptive language age; ELA- Expressive language age

Discussion

The phenotypic features of the index child, including everted outer eyelids, a high-arched palate, and frontal bossing, align with those documented in previous Kabuki syndrome (KS) case series [1]. Approximately one-third of patients have been reported to exhibit cleft lip and palate, while a high-arched palate is observed in nearly two-thirds of cases. KS remains an underdiagnosed condition among patients with cleft lip and palate [2]. In addition to typical cleft palates, preauricular tags and Tessier type 7 lateral clefts have been infrequently reported in KS [3]. In such instances, the use of a 3D-printed obturator device has been attempted to facilitate early direct breastfeeding, thereby promoting better weight gain and subsequent surgical cleft repair [4]. A notable delay in expressive language, as observed in this child, is a common characteristic of KS, as highlighted in previous studies that reported delayed speech and language acquisition [5,6]. Interventions addressing hypernasality and turbulence due to velopharyngeal insufficiency have improved speech intelligibility. However, these issues were ameliorated through loudness modification, which justifies such interventions. If postnasal turbulence remains untreated, it may lead to the development of posterior nasal fricatives, potentially resulting in reduced hearing, as noted in earlier research [7]. Children with KS often exhibit mild cognitive delays [8]. However, the child in this study demonstrated significantly lower intelligence, likely attributable to deficiencies in language acquisition. Hearing interventions, combined with speech therapy, developmental therapy, and correction of velopharyngeal turbulence, may have contributed to improved speech clarity and intelligence. This underscores the necessity of assessing each factor contributing to developmental delays individually and targeting interventions accordingly. Furthermore, the WPPS-IV assessment indicated that improvements in visuospatial skills were less pronounced compared to other domains. This finding aligns with recent research attributing such deficits to challenges in visual construction due to visual perceptual processing and visuospatial skills [9]. In the absence of effective treatments for this genetic disorder, management strategies focus on correcting manifestations and enhancing functional skills and quality of life for affected children [6]. Following three months of intervention, the previously nonverbal child began to imitate, vocalize, and follow two-step commands. The REELS-4 assessment six months later recorded a receptive language age of 28-32 months and an expressive language age of 16-18 months. This underscores the importance of managing these syndromic children in an integrated manner rather than addressing issues in isolation. Correcting hearing deficits, addressing speech

and language challenges associated with cleft palate, and addressing the inherent language delay of the syndrome, alongside activities that enhance fluid reasoning, verbal comprehension, working memory, and processing speed in a developmentally appropriate, domain-specific manner, can improve socioemotional and speech domains as well as the intelligence of these children, even in the presence of genetic compromise.

Endocrinology evaluation though initially unremarkable, should be done on follow up as they may develop endocrine abnormalities resulting in short stature as well as immune deficits, increased susceptibility to infection, and auto immune disorders mandating a immunology consultation.

Conclusions

In rare syndromic children with developmental delays, the number needed to treat, produce, and document any significant progress is often limited. Hence, the true potential for the development of such children is rarely recorded. This is true in KS, where intelligence is mildly affected. Addressing the hearing issues with the management of cleft palate, with post-surgical training and multi-domain targeted developmental therapy, helps the child to improve not only in speech and language but also in intelligence. Therefore, a detailed assessment of each domain of development and speech mechanism is needed, and efforts should be made to get maximum output from these children. Surgical correction of the cleft lip or palate produces cosmetic differences, but follow-up actions aimed at optimizing the skill acquisition produce a significant difference in functional ability. Improvement in intelligence and socioemotional domains is possible in genetic conditions like KS if training on skill attainment is well managed.

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Navigating the rare : A case report of Angelman syndrome coexisting with oculocutaneous albinism

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Abstract

Abstract: An association of oculocutaneous albinism with Angelman syndrome is a rare entity. Here, we discuss about a four year old male child born with classic albino features. He presented to us at nine months of age with genetically proven OCA2, Global developmental delay and seizures. As the child had refractory seizures and very slow response to multidisciplinary therapy, he was suspected to have a dual pathology and hence specific genetic testing for Angelman syndrome was done which helped to have greater clinical insight into his special challenges.

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Keywords:
- Angelman syndrome
- Oculocutaneous albinism
- Global development delay
- Autism spectrum disorder

Introduction

Oculocutaneous albinism (OCA) refers to a rare group of genetically diverse disorders inherited in an autosomal recessive manner, caused by mutations in genes involved in melanin production. Individuals with OCA typically present with various eye-related issues, including nystagmus, reduced vision, light sensitivity, strabismus, underdevelopment of the fovea, light-colored irises, and difficulties with color perception. (1) The severity of OCA symptoms can differ widely—from partial reduction in skin and hair pigmentation to a complete absence of melanin, primarily linked to disruptions in tyrosine metabolism. So far, seven subtypes of OCA (OCA-7) have been identified, with OCA2 being the most prevalent. Notably, about 1% of individuals with Angelman or Prader-Willi syndromes also exhibit OCA2.

Angelman syndrome (AS) is a complex neurodevelopmental disorder characterized by impaired motor coordination, significant speech delay, intellectual disability, and distinct dysmorphic features. (2) The condition arises from the loss of function of the *UBE3A* gene, which encodes a ubiquitin-protein ligase E3A, located on chromosome 15. This gene is typically maternally expressed in neurons, and its dysfunction leads to abnormalities in neuronal signaling, synaptic development, and neural plasticity. Clinically, AS is associated with microcephaly, absent or severely limited verbal communication, a characteristic happy demeanor with frequent smiling or laughter, stereotyped or autistic-like behaviors, ataxic gait, and epilepsy. (3) Seizures in AS are often refractory and usually require treatment with broad-spectrum antiepileptic medications. (4)

Case Report

A 9 months old boy born of spontaneous conception to a nonconsanguineous couple at maternal age of 30 and paternal age of 35 at time of conception, was brought with complaints of developmental delay and recurrent seizures. Mother had no history of earlier pregnancy losses. Double marker test, nuchal translucency and anomaly scan were normal. Mother had history of hypothyroidism and pregnancy induced hypertension managed with medications. Pregnancy was terminated at 35 weeks of gestation following preeclampsia. Baby had good apgar at birth and had albino features - blond hair with pale skin and eyelash. He was admitted in NICU on Day 01 of life in view of prematurity, low birth weight (1.6kg) and recurrent seizures requiring two anti-seizure medications- Phenobarbitone and levetiracetam. Neurosonogram and metabolic screening during neonatal period was normal. He was discharged on day 07 of life on anti-seizure medications and parents were counselled for high risk follow up and development surveillance.

At 09 months he had microcephaly, squint, horizontal nystagmus, generalised hypertonia with only partial neck control and not rolling over. Ophthalmologic evaluation showed iris heterochromia, hypopigmented retina and foveal hypoplasia. He was initiated on intense development therapy sessions, seizure medications and muscle relaxants. Meanwhile he continued to have multiple episodes of seizures requiring add on medications. Despite intensive continuation of multidisciplinary therapy his developmental age remained almost static.

At three years of age he continued to have no verbal output, dystonic movements with hand manipulation, mannerism and restricted socialisation. Hearing evaluation was normal. Modified Checklist for Autism in Toddlers assessment at 3 years of age was 11 showing high risk for Autism Spectrum Disorder (ASD). At four years of age his CARS score was 32 showing mild to moderate ASD. REELS assessment showed receptive and expressive language age less than six months. Development assessment with DDST II showed gross motor age of 15 months and fine motor, expressive language and social milestones corresponding to 09 months.

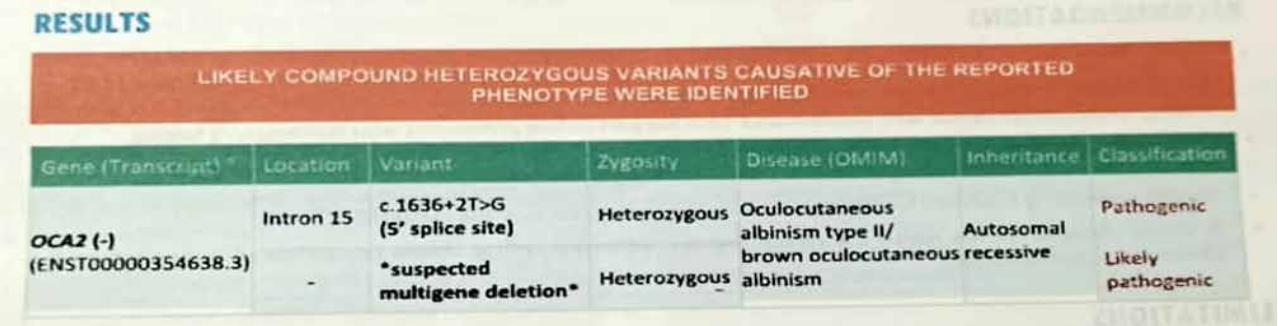
At four years he was diagnosed as Global developmental delay (Development quotient -22) and non verbal Autism Spectrum Disorder (Fig 3). Repeat EEG showed continuous burst of generalised epileptiform activity and suppression which was suggestive of epileptiform encephalopathy.

As the seizures were refractory with global development delay and a happy demeanour Angelman Syndrome was clinically suspected and genetic analysis was done (Fig 2, Fig 3). Results of PWS DNA methylation analysis was pathognomic for Angelman syndrome. Parents were counselled about the long term need of intensive autism appropriate therapy along with physical rehabilitation He is enrolled into special school. His seizures are controlled on two anti-seizure medications-sodium valproate and clobazam.

Figure 1: Phenotypic presentation in a case of Angelman syndrome with oculocutaneous albinism

Discussion

While case reports have established a connection between hypomelanotic disorders such as tuberous sclerosis and autism, cases involving albinism are rare. Raj G et al. (2022) reported a case involving a three-year-old female child presenting with low birth weight, a history of neonatal intensive care unit (NICU) admission, and developmental delays in fine motor skills, social cognition, and language. The child also exhibited hyperactivity, socially inappropriate behaviors, limited eye contact, and a preference for solitary play. [5] Ophthalmologic evaluation revealed foveal hypoplasia, leading to a diagnosis of oculocutaneous albinism. Another



RESULTS

LIKELY COMPOUND HETEROZYGOUS VARIANTS CAUSATIVE OF THE REPORTED PHENOTYPE WERE IDENTIFIED

Gene (Transcript)	Location	Variant	Zygosity	Disease (OMIM)	Inheritance	Classification
OCA2 (-) (ENST00000354638.3)	Intron 15	c.1636+2T>G (5' splice site)	Heterozygous	Oculocutaneous albinism type II/ brown oculocutaneous albinism	Autosomal recessive	Pathogenic
	-	*suspected multigene deletion*	Heterozygous			Likely pathogenic

Figure 2 :Genetic study revealing OCA2 pathogenic variant of oculocutaneous albinism type II

PATHOGENIC VARIANT CAUSATIVE OF THE SUSPECTED PHENOTYPE WAS IDENTIFIED

Sl. No.	Deletions /Duplications	No. of probes showing deletion/duplication/methylation	MLPA probe ratio (Dosage quotient)#	Disease (OMIM)	Inheritance	Classification
1.	Heterozygous deletion	35	0.45 – 0.55	Angelman syndrome	-	Pathogenic
2.	Aberrant methylation	7	0.00			

Figure 3 :Genetic study revealing Angelmann syndrome

study was reported by Chaitanya Varma et al (2023) describing association between OCA and ASD pointing out a genetic relationship between OCA and ASD.

The association between oculocutaneous albinism with autism has been rarely reported in literature. In a case report by Yurie Fukiyama et al (2018) at Osaka, a similar presentation was noted which aligns with our findings.[6] Another case report was reported by Chinese literature , Zhou QJ et al (2023) showing genetic relationship and complications associated with a combination of OCA 2 mutation with Angelman syndrome.[7] To the best of our literature reaserch there have been no reported case studies from India to date.

Since refractory seizures are uncommon with OCA , further evaluation helped us identify the coexisting Angelman syndrome. Most patients with epilepsy are diagnosed as AS following their diagnostic EEG patterns as reported by Ranasinghe JC et al. Patients with Angelman syndrome may experience both provoked and unprovoked seizures. Epileptic manifestations tend to be more severe in individuals with a deletion involving the 15q11–q13 chromosomal region.

CONCLUSION

Diagnosing AS early is a challenging due to nonspecific symptoms and increase chance of misdiagnosis with other common genetic condition.[8]AS should be taken into account in the differential diagnosis of infants with delayed developmental milestones, seizures, especially in children with happy facies and ocular abnormalities like strabismus. Though oculocutaneous albinism in a child with Angelman syndrome is only 1%, it must also be considered during evaluation process. Vision correction, proper stimulation, intensive speech and physiotherapy must be started early.

An earlier diagnosis will help clinicians to foresee complications, better compliance and coperation with treatment plan. Hence high index of suspicion and earlier genetic diagnosis will help decrease ambiguity and enable clearer decision making hence improving overall patient outcome.

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3. Key words (5 to 7).
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6. Material & Methods should follow the introduction & should provide enough information to permit repetition of experimental work/ project, giving how the study was designed, carried out and data was analyzed. State whether study was approved by ethics committee or not. Describe statistical analysis methods used with required details. Give actual p-values, t-values define statistical terms, symbols and abbreviations.
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3. The top and bottom margins are 1 inch.
4. The title is written using capital letters only at first word or special name (example: location name), 17 font size, center position.
5. Sub titles are written using capital letters only at first word or special name, 12 font size, starting from the left margin.
6. Sub of a sub of sub titles, if any, are written using capital letters only at the beginning of each word except for connecting words, all in italics. They should be started from the left margin.
7. All references should be preferably of the last ten years publication.

Tables and Figures : Tables and figures should be presented as follows:

1. The name of tables and figures should follow a numbering system (Arabic numbering system). The title of the tables and figures are placed at the top and the bottom respectively.
2. The tables and figures should provide the source of information, if any, at the bottom.
3. Each image should be less than 4096 kb(4MB).All the Images/Figures should be submitted in separate files. Do not Zip the file.

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: _____MiddleName: _____
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 _____VoluntaryAgency _____
9.

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

10. Areas of Interest of Work : _____
11. Membership Subscription:
    a) Life Membership for Central IAP Members – Rs1500
    b) Life Associate Membership for Doctors other than Pediatricians – Rs1500
    c) Life Affiliate Membership for All Other Professionals – Rs1500
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_____