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Karnataka Paediatric Journal

Editorial

Uniquely cerebral palsy – World cerebral palsy day

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World Cerebral Palsy (CP) day, celebrated annually on October 6, is a global initiative aimed at raising awareness about CP and advocating for the rights of those affected by the condition. Established to highlight the challenges faced by the estimated 1.8 crore people living with CP worldwide, the day serves as a platform for individuals, families, and organisations to share experiences and promote understanding. There are more than 17 million people across the world living with CP. Another 350 million people are closely connected to a child or adult with CP. Nearly 15–20% of physically disabled children are affected by CP. In India, the estimated incidence is around 3/1000 live births.

Every year, October 6 is an opportunity to come together and celebrate the resilience, diversity, and vibrancy of people with CP. It is also a moment to campaign for meaningful, positive changes to accessibility and inclusion in society and recognize the contributions of family, carers, and allies. In 2024, the global campaign theme for World CP Day is uniquely CP. This theme celebrates the uniqueness and passions of individuals with CP, highlighting their individuality and unique experiences. Every individual with CP is unique, with passions, pursuits, and identities beyond just that of a person with a disability. CP is often misunderstood in the wider community, and we should overcome these stereotypes by celebrating the individuality and personality of the community.

The 2024 campaign theme aims to empower people with CP, family members, and supporters to share stories about their achievements and passions – how CP makes all unique. World CP Day is a global movement that started in 2012. The CP Alliance in 2012 declared October 6 as World CP Day to bring together people with CP, their friends, families, and organisations. It also aims to ensure that children and adults with CP have the same rights, opportunities, and access as everyone else globally. Last year, it reached over 10 million people. It aims to bring together people living with CP, their families, supporters, and organisations from over 100 countries. All with the aim to ensure a future in which children and adults with CP have the same rights, access, and opportunities as anyone else in our society.

CP Day is a social movement. The first World CP Day campaign was called 'Change My World in 1 Min.' The project sought ideas from the global community of people with CP for technologies and products that needed inventing – that had the potential to 'change the world' for people living with CP. In 2012, more than 470 ideas were posted on the World CP Day website. Three ideas were shortlisted, and then the call went out to inventors. A research team from the University of Virginia (USA) won the major prize. They developed a prototype solar-powered wheelchair, which was an idea posted by Alper Sirvan, a man with CP in Turkey. The wheelchair prototype was presented to Alper on World CP Day 2013. In 2015, the campaign evolved into a social

movement that targets the six key issues that affect people with CP around the world, irrespective of geographical, cultural, and economic differences.

CP is a physical disability that affects movement and posture. Many people with CP have other related vision, hearing, communication, and mobility needs. Its impact can range from a weakness in one hand to almost a complete lack of voluntary movement. As per a 2022 research report, the birth prevalence for pre-/perinatal CP in regions from high-income countries was 1.5/1000 live births and 1.6/1000 live births when post-neonatal CP was included. A majority (82.9%) of the children with CP had spastic CP. More than half (58.9%) of the children with CP could walk independently.

It is a complex disability:

- 1 in 4 children with CP cannot talk
- 1 in 4 cannot walk
- 1 in 2 have an intellectual disability
- 1 in 4 have epilepsy.

Here are five important facts about CP:

1. CP is the most common childhood disability, affecting 1 in 345 children
2. Approximately 1 million people in America and 18 million worldwide have CP
3. Among those with CP, 75% experience chronic pain, and 25% are non-verbal
4. CP is not progressive, meaning it does not get worse over time
5. The five types of CP include spastic, dyskinetic, ataxic, hypotonic, and mixed.

CP is one of the least understood disabilities due to a lack of awareness regarding this condition. The world of CP creates a platform to emphasise awareness with respect to the disease, diagnosis, and treatment, in addition to addressing the challenges CP patients face.

PREVENTION OF CP DURING PREGNANCY

CP prevention during pregnancy is dependent on maintaining good habits and staying healthy. The following are the ways to prevent CP at the time of pregnancy that includes:

- Staying away from exposure to infections or viruses that are known to harm foetal health, such as German Measles, Cytomegalovirus, or Zika
- Getting the necessary vaccinations
- Managing underlying health conditions
- Avoiding alcohol and cigarettes
- Recognising any potential Rh incompatibility between mother and child.

In 2024, the focus will continue to be on inclusion and celebrating the diverse contributions of individuals with CP across various fields. Activities may include awareness campaigns, fundraising events, and educational workshops designed to improve support and services for those living with CP. By fostering community engagement and collaboration, World CP Day aims to create a more inclusive society where individuals with CP can thrive and have equal opportunities. Let us join hands in this noble action.

Dr. Bhaskar Shenoy,
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Karnataka Paediatric Journal

Review Article

Pulmonary graphics – An insight into newborn lung on ventilator

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ABSTRACT

Neonatal care with technology has brought down mortality and morbidity in the world. More and more pre-term neonates are seeing the world with a quality life. Advanced software is used in the ventilators to check the breath-to-breath variability in the care of sick neonates. The anatomical and physiological aspects of a neonatal lung are essential before understanding and analysing the waves on the monitor. Constant monitoring of both the baby and the waves of lung mechanics is essential in clinical decision-making. Having a sophisticated machine with the graphics of lung mechanics is of no use if the clinician is incapable of correctly interpreting the waves. The pulmonary graphics is the graphical representation of the lung similar to the electrocardiogram and electroencephalogram routinely used to know the physiology of the heart and brain. With this background, the pulmonary graphics are dealt with simply, and therefore, the reader can understand the pulmonary graph well and use it efficiently in the care of sick neonates. The incorporation of microprocessors and sensors into the ventilators has made the clinician monitor lung function continuously in real time. The three basic parameters of the respiratory system – the pressure, the flow, and the volume – are visualised online. This can be utilised along with the clinical data, the blood gas, and the chest Roentgenogram.

Keywords: Neonate: Pulmonary graphics, Neonate, Lung, Pulmonary graphics

INTRODUCTION

Neonatal care with advanced technology has brought down mortality and morbidity in the world. More and more pre-term neonates are seeing the world with a quality life. Advanced software is used in the ventilators to check the breath-to-breath variability in the care of sick neonates.[1] The anatomical and physiological aspects of a neonatal lung are essential before

Figure 1: (a)The flow sensor-heated wire anemometer is a data link between the patient and the machine (arrow). (b)The monitor shows the pressure-time curve only when the sensor is not attached. (c)With an intact sensor, the flow and volume curves are also seen.

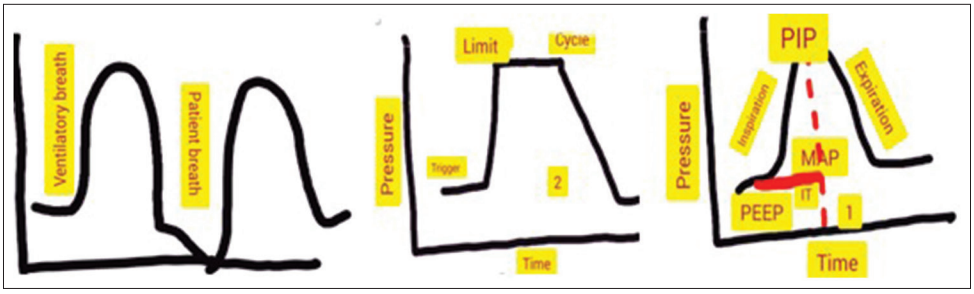

Figure 2: Characteristics of breath (ventilator and patient breath). (2) Trigger (cause for breath to begin), limit (cause for regulation of breath), cycle (cause for breath to end), and (1) the mean alveolar pressure. PEEP: Peak end-expiratory pressure, PIP: Peak inspiratory pressure, MAP: Mean alveolar pressure. The red- line represents IT. IT is inspiratory time. Until the dotted line is the inspiratory phase.

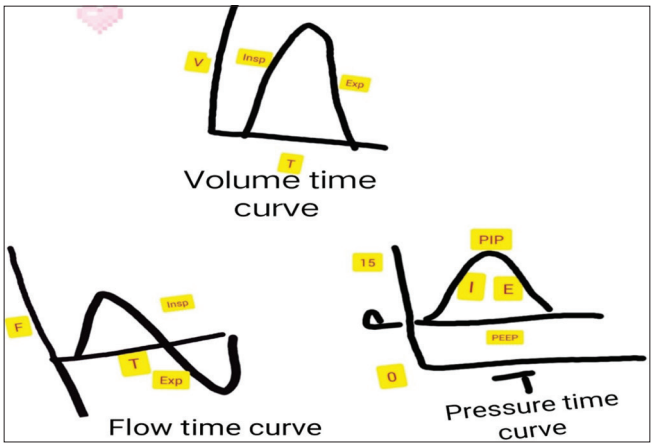

Figure 3: The scalar graphics demonstrate pressure, flow, and volume versus time curves. The flow time curve is biphasic, and the pressure time and volume time curves are monophasic. The pressure curve against the time shows peak end-expiratory pressure and peak inspiratory pressure (0-15), I (inspiration), E (expiration), the volume-time curve demonstrating inspiration and expiration, and the flow time curve demonstrating the inspiratory (above the baseline) and expiratory flow (below the baseline). V: Volume, T: Time, F: Flow, Insp: Inspiration, Exp: Expiration, PEEP: Peak end-expiratory pressure, PIP: Peak inspiratory pressure,

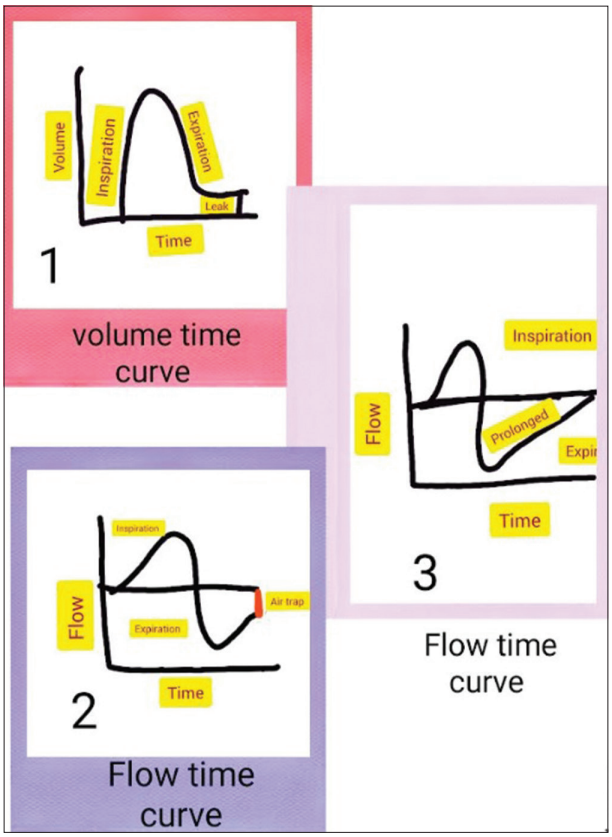

Figure 4: (1) Volume time curve demonstrating air leak (expiratory trace descends smoothly and then plateaus and does not reach the baseline), (2) flow time curve demonstrating prolonged expiration meaning increased expiratory resistance, (3) flow time curve demonstrating air trap where in the expiratory phase not reaching the baseline.

understanding and analysing the waves on the monitor. Constant monitoring of both the baby and the waves of lung mechanics is essential in clinical decision-making. Having a sophisticated machine with the graphics of lung mechanics is of no use if the clinician is incapable of correctly interpreting the waves. The pulmonary graphics are the graphical representation of the lung similar to the electrocardiogram and electroencephalogram routinely used to know the physiology of the heart and brain, respectively. With this background, the pulmonary graphics are dealt with simply, and therefore, the reader can understand the pulmonary graph well and use it efficiently in the care of sick neonates. The incorporation of microprocessors and sensors into the ventilators has made the clinician monitor lung function continuously in real time. The three basic parameters of the respiratory system – the pressure, the flow, and the volume – are visualised online. This can be utilised along with the clinical data, the blood gas, and the chest roentgenogram.

THE MICROPROCESSOR AND SENSORS

The technology helps the clinician to understand the breath-to-breathe interaction between the patient and the

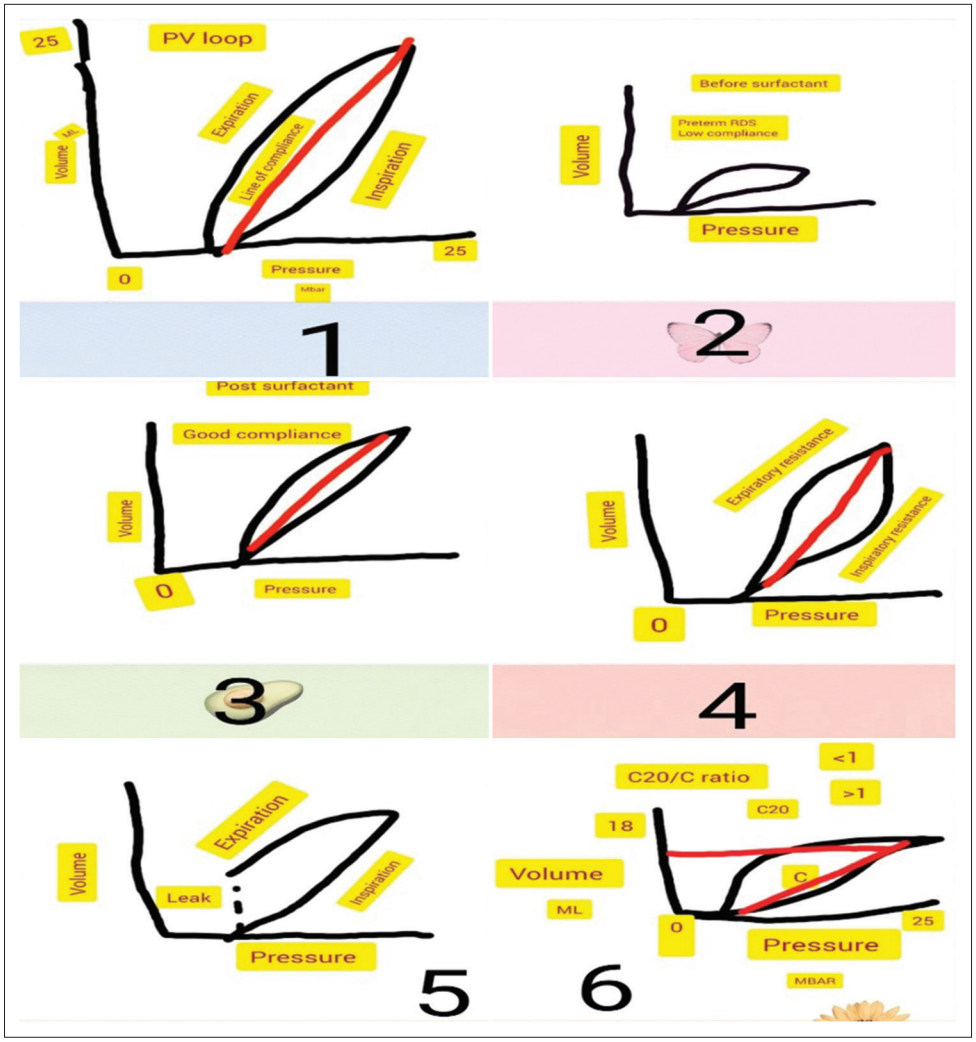

Figure 5: (1) Normal pressure-volume loop, (2) Low compliance loop as seen in pre-term respiratory distress syndrome with low compliance, (3) Good compliance loop post-surfactant administration, (4) Loop showing inspiratory and expiratory resistance, (5) Loop showing Endotracheal tube (ET) leak, (6) Loop showing beaking phenomena indicating hyperinflation of alveoli. The C20/C ratio is the ratio between the last 20% of the compliance to the total compliance. A ratio of more than one is normal, and <1 indicates hyperinflation. Pressure 0-25, C is compliance MBAR is the millibar.

ventilator. The flow sensor is the backbone of pulmonary graphics. The sensor measures the amount of cooling air that passes over the heated wire, and the cooling effect indicates more flow of gas. The pressure that is generated in a spontaneously breathing neonate by the respiratory muscle or by the ventilator in a mechanically ventilated initiates the flow to overcome the resistance of the compliance and the inertial properties of the respiratory system. This, in turn, creates a change in volume in the lung. The pressure and time are measured back at the machine, and the flow is measured at the patient. The volumes – the inspiratory and expiratory tidal volumes – are derived from flows and read at patient wye through a flow sensor [Figure 1].

GRAPHICS

The pulmonary graphics help the clinician to understand the real-time data – scalar waveforms, loops, and trends from the proximal airway pressure displayed on the ventilator screen. The pressure, the flow, and the tidal volume waveform are the scalar display, and volume versus pressure, flow versus pressure, and the flow versus volume are the loops. The trends include compliance, resistance, minute volume maximum pressure, minimum pressure and mean pressure, oxygen %, and carbon dioxide diffusion coefficient against time (in High-frequency oscillation [HFO]). The respiratory parameters are plotted on a two-dimensional X-Y graph as pressure against time, flow against time and volume against

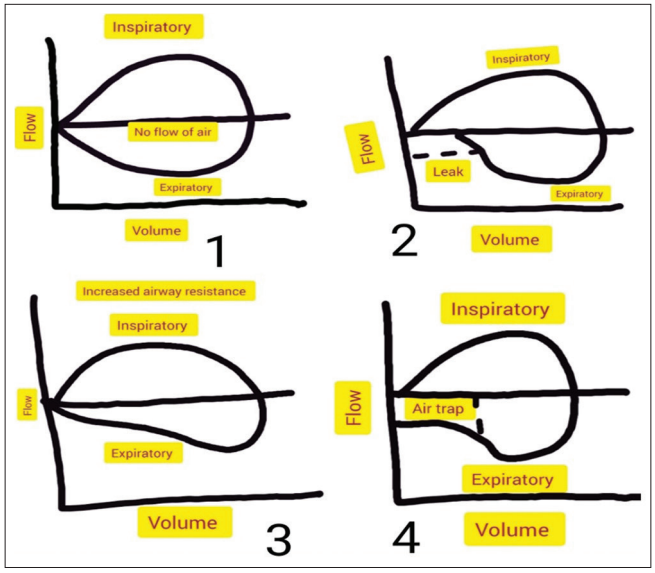

Figure 6: (1) Flow-volume curve showing normal (the inspiratory and expiratory flow come and join the same point where no airflow happens), (2) expiratory leak (the expiratory volume does not return to the zero volume level and the volume of leak indicates the magnitude), (3) increased expiratory resistance (scooped out expiratory tracing), (4) air trap (the expiratory trace does not return to baseline).

time and provide valuable information about the pattern of breathing and status of the lung. The interpretation of graphics is required when a baby is mechanically ventilated to understand the trend, to help the baby from the untoward effect of the ventilator, to study the impact of clinical morbidity on the graph, and to observe the improvement upon the drug administration. The scalar graphics and the loops are the two graphical representations of the respiratory mechanics in mechanically ventilated neonates. In the scalar graphics, the pressure, the flow, and the volume are measured against time. The flow and the pressure are depicted against the volume in the loop graphics. The pressure-time curve demonstrates two pressures, including the peak end-expiratory pressure (PEEP) and peak inspiratory pressure (PIP), and the mean alveolar pressure (MAP), the area under the curve. The PIP, the inspiratory time, and the MAP determine the oxygenation. The trigger, the limit, and the cycling are the breath characteristics in completing effective respiration.[2-5]

BREATH ON PULMONARY GRAPHICS

In spontaneous breathing, the inspiration is below the baseline and started by the patient's effort, and expiration is above the baseline and is by the machine's effort. The three characteristics of a breath include trigger, limit, and the cycle. The MAP is a function of PIP, PEEP, inspiratory time, and the rate [Figure 2].

CURVES

The pulmonary graphics are broadly classified into a scalar (three opened curves), loops (two closed curves), and trend. They are depicted in the images.

THE SCALAR GRAPHICS

In scalar graphics the pressure, the flow, and the volume are depicted against the time [Figures 3 and 4]. The flow volume and the pressure-volume curves are depicted in the loop graphics.[6,7] The normal and abnormal graphics are depicted with the explanation in the respective images.[8-15]

LOOPS

Graphical representation of the inspiratory and expiratory flow, including both the patient and ventilatory-triggered breath. Pressure volume and flow volume loops are described in normal and abnormal conditions [Figures 5 and 6].

THE TREND OF VENTILATOR PARAMETERS

The trend data are very important to understand the impact of different clinical events that happen in a ventilated baby over time and it helps in deciding the clinical improvement. The FIo2 requirement, the compliance, and the resistance can be recorded as a trend over time.

CONCLUSION

Pulmonary graphics are an innovation in the respiratory care of newborns. Having a basic knowledge of neonatal respiratory physiology with good physical examination and adequate knowledge of the respirator, the pulmonary graphics would be a great adjuvant as a bedside monitoring tool while ventilating a newborn.

Ethical approval

Institutional Review Board approval is not required.

Declaration of patient consent

Patient consent is not required as there are no patients in this study.

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Conflicts of interest

There are no conflicts of interest.

Use of artificial intelligence (AI)-assisted technology for manuscript preparation

The authors confirm that there was no use of artificial intelligence (AI)-assisted technology for assisting in the writing or editing of the manuscript and no images were manipulated using AI.

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Karnataka Paediatric Journal

Original Article

Demographic of deep vein thrombosis at a Tertiary Institution in Nigeria

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ABSTRACT

Objective: This study aims to determine the prevalence of deep vein thrombosis (DVT) in Calabar, Cross River State, Nigeria. Deep vein thrombosis is a spectrum of venous thrombo-embolism, which is due to a complex interplay between genetic and acquired risk factors and the second most common cause of morbidity and mortality. There is an increasing trend in the number of patients with DVT

Materials and methods: A retrospective study with 5 years of data (2018-2022) obtained from the medical register of the department of haematology, University of Calabar Teaching Hospital, Calabar. The data collected were analysed using Microsoft Excel 2016 and IBM Statistical Package for Social Sciences version 26.

Results: A total of 13 cases were seen during the 5-years period with male to female ratio of 1:2.2 and a mean age of 45.31 ± 12.45. Most patients were in their 30s and 40s and from and from the Efik tribe.

Conclusion: This study aims to awaken our consciousness of the increasing epidemiological burden of DVT in our environment and helps to enhance further investigation into the relationship between DVT, genetics, and tribe. Furthermore, establishing a venous thromboembolism multidisciplinary management team is sacrosanct for records and quality management.

Keywords: Deep vein thrombosis, Prevalence, Calabar

INTRODUCTION

Thrombosis is a silent killer and the second most common cause of morbidity and mortality worldwide.[1] The World Health Organization estimates that 17.5 million people die from cardiovascular disease (CVD) attributed to thrombosis, with over three-quarters of these cases occurring in low- and middle-income countries, such as Nigeria.[2-4] Venous thromboembolism (VTE) comprises deep vein thrombosis (DVT) and pulmonary embolism, which is the third most common cause of CVD globally.[5] Furthermore, it is associated with increased mortality rate, health costs, and recurrence.[6] The pathogenesis of DVT is a complex interplay between genetic predisposition and acquired risk factors. There are several risk factors for DVT, which includes; age, gender, race, surgery, use of oral contraceptives, cancer, paripartum period and prolonged immobilisation.[6-8] Several studies have, for example hospitalised patient reported a higher incidence of VTE in Africa, American than in Asian and Native American.[9-15] The implication of these studies is that the incidence of DVT might be higher in African population compared to other races.[9-15] Despite above information, there is paucity of data on the demographics and prevalence of DVT in our environment. Therefore, this study aims to estimate the demographic and prevalence adult DVT in our environment.

MATERIAL AND METHODS

Study design

This study is a retrospective study on DVT patient seen at the University of Calabar Teaching Hospital (UCTH) from January 2018 to October 2022.

Study area

The hospital is a 600 bed space tertiary health institution that renders specialist care to its host and neighbouring communities.

Subject

This includes 13 patients that were diagnosed and managed of DVT at the UCTH, out of the 2947 patient seen during the period of review. The diagnosis was made using the validated clinical wells scoring system with confirmation by a coloured compression Doppler-ultrasound scan.

Selection criteria

Those whose information were retrieved from the medical records with evidence of proper documented, well scored and Doppler findings of DVT where included, while those with any form of omission were excluded from the study. These data and results collected were analysed using Microsoft Excel 2016 and IBM Statistical Package for the Social Sciences version 26. The data were analysed using simple inferential statistics (frequency and percentage).

RESULTS

Deep Vein Thrombosis (DVT) at the University of Calabar Teaching Hospital involved 13 individuals, comprising 31% males and 69% females. The age distribution is presented in Table 1. The tribal distribution revealed that most cases were from the Efik tribe (5 individuals), followed by the Igbo tribe (2 individuals). Other tribes represented by one case each included Biase, Boki, Hausa, Obudu, Oron, and Ugep. The gender and tribe distributions are presented in [Figures 1 and 2] respectively.

The annual distribution of DVT cases varied over a five-year period: 2 cases in 2018, 5 in 2019, 3 in 2020, 1 in 2021, and 2 in 2022. This demographic overview highlights both the tribal diversity and the annual fluctuations in DVT incidence at the hospital.

The result is presented in [Figure 3].

DISCUSSION

This study reviews the prevalence of diagnosed adult DVT at UCTH but does not include individuals with asymptomatic DVT or those with symptoms not investigated. A total of 2947 patients were seen during the period of review. DVT constitutes 0.44% of all patients seen during the period review. This is at variance with a similar study conducted in the United States of America, which reported a higher prevalence of 5.04%.[16] Another study conducted by Danwang *et al.* in Africa reported a prevalence between 2.6% and 9.6% of

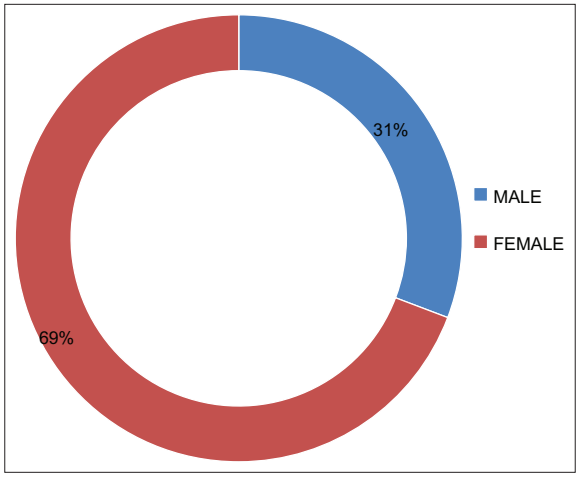

Figure 1: Sex distribution.

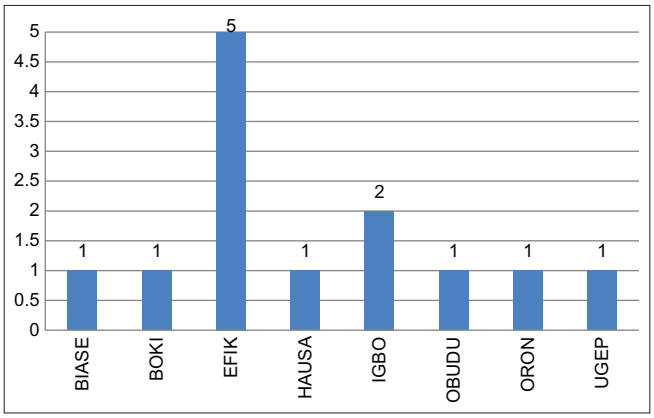

Figure 2: Tribe distribution.

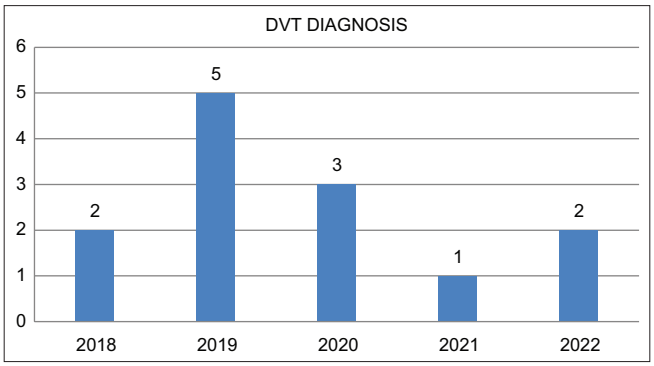

Figure 3: Yearly distribution. Total patients seen = 2947, and prevalence of deep vein thrombosis = 0.44%.

Table 1: Age range distribution.

Age range	Frequency	Percentage
30–39	5	38.46
40–49	4	30.77
50–59	2	15.38
60–69	1	7.69
≥70	1	7.69
Total	13	100.00

Mean age – 45.31±12.45, Median age – 41, Minimum age – 33, Maximum age – 70

DVT in post-operation patients.[17] Moreso, another study by Muleledhu *et al.*[18] reported a prevalence of 2.4% in Nigeria. This variation can be attributed to differences in study design, poor awareness among Physicians, underdiagnosis due to lack of facilities and also, and DVT managed by other specialists such as the Vascular Surgeon, Pulmonologist, Cardiologist, and Anaesthiologist. This study also shows female preponderance, with a male-to-female ratio of 1:2.2. This is similar to the findings by Mugeni *et al.* on proximal DVT among hospitalised medical and obstetric patients in Rwanda.[19] This can be attributed to the fact that males at this age perform a lot of activities compared to their female counterparts. Furthermore, females at this age ingest more of oral contraceptives to prevent unwanted pregnancies apart from reduced activities. This study also reviews a mean age of 45.31 ± 12.45 and the median age of 41 years, with 15.3% above 60 years. This is similar to the finding by Mugeni *et al.*[19] who also reported that the bulk of the patients were below 65 years. This was in contrast with Western studies, which reported that DVT is predominantly a disease of middle-aged and elderly with markedly increasing incidence with age.[20] This study also showed that the prevalence of DVT was more with the Efik tribe, followed by the Igbos residing in Calabar. This may be attributed to either the location of the hospital within the Efik settlement, sedentary lifestyle due to the cultural practice of fattening females within the age of marriage, which falls within the age bracket of prevalence of DVT; also, it may be attributed to diet (Afang) which is rich in crude fat and carbohydrate. DVT was said to be highest in 2019 due to the increased number of Specialist Haematologist and facilities, which has improved diagnosis and management of DVT. This was followed by 2020, which may be attributed to the COVID-19 pandemic and it is associated with lockdown measures. The lowest value was recorded in 2021 due to increased awareness of the risk and prevention of DVT among physicians and the populace.

CONCLUSION

This study aims to awaken our consciousness on the increasing epidemiological burden of DVT in our environment and helps to enhance further investigation into the relationship between DVT, genetics, and tribe. Furthermore, the establishment of a VTE multidisciplinary management team is sacrosanct for records and quality management.

Limitation

Poor documentation, poor record keeping, and lack of VTE safety zone.

Authors' Contributions

Akaba Kingsley; Conceptualise, review. Edakabasi Akaba; Literature search. Omini Godwin; Methodology and discussions.

Ethical approval

Since this study is a retrospective study, the Institutional Review Board approval is not required.

Declaration of patient consent

Patient consent is not required as the patient's identity is not disclosed or compromised.

Financial support and sponsorship

Nil.

Conflicts of interest

There are no conflicts of interest.

Use of artificial intelligence (AI)-assisted technology for manuscript preparation

The authors confirm that there was no use of artificial intelligence (AI)-assisted technology for assisting in the writing or editing of the manuscript and no images were manipulated using AI.

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Karnataka Paediatric Journal

Case Report

Prostaglandin E1-induced urticaria with harlequin change in a neonate with transposition of great vessels: A case report

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ABSTRACT

Prostaglandin E1 (PGE1) is an emergency drug used in neonates with critical congenital heart diseases (CHDs) to maintain the ductal patency. There are many adverse effects of this drug explained – cutaneous side effects being one of them, of which limited literature is available. In this case report, we describe a term neonate with critical CHD, started on PGE1 infusion to maintain ductal patency. Baby developed erythematous migratory annular rashes to harlequin colour change in a dose-dependent manner. The severity of rashes decreased, and there was a complete resolution with tapering down of drug doses to a minimum level to maintain a ductal patency. No antihistamines or steroids were administered for the treatment. The PGE1 should be tapered to a minimum dose to maintain the required saturation in case of severe cutaneous reaction and should not be stopped abruptly as it is a lifesaving drug in critical CHD.

Keywords: Neonate, Cyanotic congenital heart disease, Prostaglandin E, Urticaria, Harlequin change

INTRODUCTION

Prostaglandin E1 (PGE1) is an emergency drug used in neonates with critical congenital heart diseases (CHDs) to maintain the ductal patency.[1] There are many adverse effects of this drug explained in the literature. The most common side effect is apnoea. The limited literature is available describing the adverse cutaneous manifestations of prostaglandin E1 infusion. We present a neonate on PGE1 developing an extensive erythematous rash with a harlequin color change.

CASE REPORT

An early-term male neonate was born to a non-consanguineous couple by lower-section *cesarean section*. He required intubation at birth, requiring high oxygen therapy to maintain saturation of 50–60%. The echocardiography demonstrated the transposition of great vessels with shunts, atrial septal defect and patent ductus arteriosus. He was started on PGE1 infusion @ 100 ng/kg/min. Atrial septostomy was performed on 2nd day of life. He developed an erythematous blanchable rash on day 3 of life involving the scalp, the face, and the upper trunk with sharp demarcation between the upper and lower abdomen, suggesting harlequin [Image 1]. The rash gradually progressed to erythematous annular migratory blanchable rash by day 4 of life when prostaglandin was gradually tapered to 60 mcg/kg/min [Image 2]. He was also

started on antibiotics for suspected sepsis. Gradually, the prostaglandin was weaned to 20 ng/kg/min; the cutaneous lesions also disappeared [Image 3]. He continued to be stable hemodynamically on the ventilator, and his septic markers were normal. On day 7 of life, he was taken for corrective surgery for transposition of the great arteries and discharged home on direct breastfeeding.

Management and outcome

In view of the erythematous rashes, the prostaglandin dose was tapered to a minimum therapeutic dose to maintain the target saturation. At the dose of 20 ng/kg/min, the rashes completely disappeared, and there was no further recurrence. No antihistamines or steroids were administered in our case for the skin lesions.

DISCUSSION

PGE1-related dermatological complications are not much reported in the neonatal population. After an extensive search, we could find only six cases characterized by migratory polycyclic erythema to harlequin changes.[2-4] To the best of our knowledge, this was the first case being reported from India. Similar case reports were published by Young *et al*. and Carter and Garzon.[2-4] Both the case reports described dose-dependent cutaneous reactions as seen in our index case.

PGE1 acts by activating adenylate cyclase and adenosine triphosphate sensitive potassium channel and, causing vasodilation and keeping the ductus arteriosus patent. The drug can induce vasodilatation and cause cutaneous inflammation through a non-allergic pathway of histamine release. Hypotension, rhythm disturbance, seizures, congestive cardiac failure, hypo- or hyperkalaemia, cortical proliferation, and hyperostosis of long bones[5] are the other adverse side effects of the drug, with apnoea being the most common seen in clinical practice. The harlequin color change generally resolves spontaneously without any residual skin reaction.[6,7] The non-allergic mechanism of histamine release causes the urticarial rash, and the severity is dose dependent. Similar skin reaction is noted secondary to vancomycin causing red man syndrome[8] and severity being dependant on dose and rate of infusion. Histopathological examination demonstrated sparse mononuclear cells. None of the medications, including the antihistamines and steroids, neither local nor systemic, will help in the management of such rash. The PGE1 should be tapered to a minimum dose to maintain the required saturation and should not be stopped abruptly as it is a lifesaving drug in critical CHD. The prompt recognition of cutaneous side effects of prostaglandin allows for suitable diagnosis and avoidance of unnecessary interventions and treatments, including antihistamines and topical or systemic steroids. As no evidence of systemic sequelae has been reported till now, the continuation of the lifesaving PGE1 treatment is warranted for critical CHD until the palliative or definitive surgery is performed.

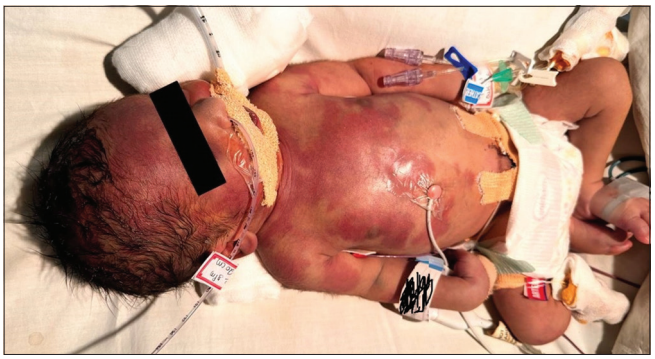

Image 1: Diffuse erythematous rash at prostaglandin E1 of 100 ng/kg/min.

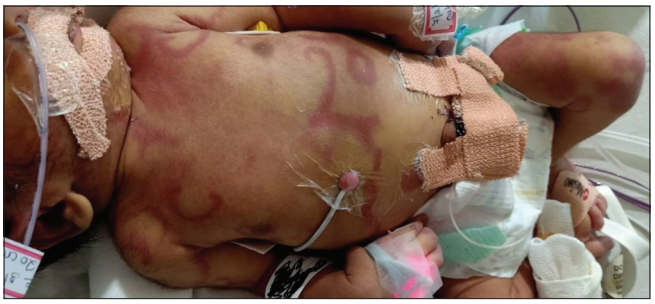

Image 2: Annular migratory rash at prostaglandin E1 of 40 ng/kg/min.

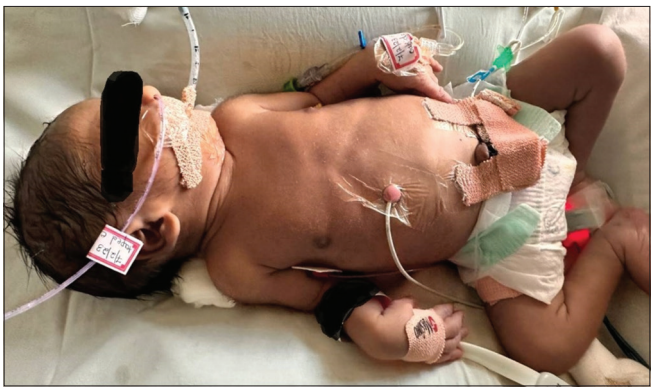

Image 3: Complete resolution of rash at prostaglandin E1 of 20 ng/kg/min.

Lessons learnt

1. Prostaglandin-related cutaneous effects present in varied morphology in a dose-dependent manner
2. Minimum effective dose of PGE1 to be continued to maintain ductal patency in critical CHD
3. Prompt recognition and diagnosis avoids unnecessary interventions.

CONCLUSION

Prostaglandin induced urticaria can be polymorphic in presentation, hence prompt diagnosis and cautious management in necessary in a neonate where it is a life saving drug.

Ethical approval

The Institutional Review Board approval is not required.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent.

Financial support and sponsorship

Nil.

Conflicts of interest

There are no conflicts of interest.

Use of artificial intelligence (AI)-assisted technology for manuscript preparation

The authors confirm that there was no use of artificial intelligence (AI)-assisted technology for assisting in the writing or editing of the manuscript and no images were manipulated using AI.

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Karnataka Paediatric Journal

Case Report

A case report of juvenile ocular myasthenia gravis in a toddler

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ABSTRACT

Myasthenia gravis (MG) is an acquired autoimmune disorder leading to abnormal fatiguability of muscles due to a deficiency of acetylcholine receptors (AChRs) caused by circulating antibodies directed against them. MG presents as ocular MG and generalised MG. We present a 1-year 9-month-old female child presenting with bilateral ptosis, worsening by the end of the day and improving with rest. On examination, pupillary reflexes were normal. Other cranial nerve examinations were normal. The child was one of the dizygotic twins. There was no similar history in the family members as well as the other twin. AChR antibodies assay was strongly positive. Chest X-ray revealed no thymus enlargement. The child was treated with neostigmine and prednisolone. Symptoms improved drastically; the child was discharged and is on regular follow-up. She did not have a relapse of symptoms on follow-up. We plan to taper steroids over the next 6 months and add on steroid-sparing agents.

Keywords: Myasthenia gravis, Acetylcholine receptors, Ptosis

INTRODUCTION

Myasthenia gravis (MG) is defined as an acquired autoimmune disorder where there is abnormal fatiguability of muscles due to a deficiency of acetylcholine receptors (AChRs) caused by circulating antibodies directed against AChR.[1] The most common antibodies are antibodies to nicotinic AChR. Antibodies affect the function by complement-mediated destruction of the motor end plate and internalisation of AChR antibodies.[2] Childhood MG has three types: Congenital myasthenic syndrome (CMS), transient MG, and Juvenile MG (JMG). JMG and CMS can be differentiated by the seropositivity of AChR antibodies. MG presents as ocular MG and generalised MG. Ocular MG is a form of MG clinically involving only the levator palpebrae superioris, the orbicularis oculi and the extraocular muscles. Ptosis and ophthalmoplegia, both unilateral and bilateral, constitute the only signs in about 20% of cases, whereas, in 80% of cases, ocular symptoms mark the onset of generalised MG.[1] Antibodies can also be directed against muscle-specific kinase (MuSK) and receptor-related low-density lipoprotein 4 (LRP4). MuSK antibodies act presynaptically by disrupting LRP4 function.[3] Cholinesterase inhibitors are used first-line as symptomatic treatment in JMG patients.

CASE REPORT

The one-year nine-month-old female child presented to our outpatient department with chief complaints of bilateral ptosis. Ptosis initially started on the left eyelid 4 weeks back, followed

by the right eyelid from the past 1 week, and was gradually progressive, better in the morning, and slowly worsening by the end of the day. At admission, the child had complete ptosis of the left eye associated with compensatory head tilt to the left side and partial ptosis of the right eye. There is no history suggestive of diplopia in this child. There is no history of similar illness in the family members as well as in the other twin. The child was one of the dizygotic twins of non-consanguineous marriage, first in birth order, born preterm (30 weeks), and delivered by emergency lower-segment caesarean section. The child had a stormy neonatal course with respiratory distress syndrome requiring surfactant, ventilator support, and other issues related to prematurity and had a neonatal intensive care unit stay for 2 months.

At admission, the child was conscious, alert, and oriented to time, place, and person. The child was hemodynamically stable with a heart rate of 102 bpm, respiratory rate of 20/min, blood pressure of 96/54 mmHg, and oxygen saturation of 96% in room air. Ocular examination revealed complete ptosis of the left eye and partial ptosis of the right eye with a 4 mm palpebral opening. Pupillary reflexes were intact. Other cranial nerves were normal. On motor system examination, power was normal in all four limbs, deep tendon reflexes were normal, abnormal cerebellar signs were absent, no bladder or bowel involvement. Other systems were within normal limits. Clinically, ocular MG was considered. Anti-AChR antibodies assay was done by enzyme-linked immunosorbent assay method and came to be strongly positive. Magnetic resonance imaging brain was normal. There is no evidence of thymoma on chest X-ray.

The child was started on neostigmine 1 mg/kg/day in three divided doses and prednisolone 1 mg/kg/day in two divided doses. The ptosis resolved completely, and the child was discharged after 2 days on neostigmine and prednisolone. The child was on regular follow-up, and prednisolone was tapered to 0.5 mg/kg/day on the first visit. The last hospital visit was 1 month back; no relapse of symptoms was noted.

Muscarinic side effects were not present during follow-up.

We plan to follow up with the child regularly and start on steroid-sparing agents (azathioprine) on the next visit and taper the steroids over the next 6 months. The child will be continued on neostigmine and azathioprine till remission.

DISCUSSION

We describe a case of JMG with ocular symptoms in a female child.

Ashraf *et Al.,* in their longitudinal study, described that JMG has less prevalence, higher incidence of ocular MG, equal sex ratio, and increased familial occurrence compared to adult MG. This study also revealed that JMG has a lower rate of seropositivity, lack of association of thymoma and other autoimmune disorders and more chances of complete remission.[4]

Peeler *et al*. in his, in their observational cohort, described an association between high AChR antibodies and progression from ocular myasthenia gravis (OMG) to generalised disease.[5] In this case report, the child had strongly positive antibodies at a younger age. There could have been a high incidence of generalised disease, thus resulting in higher mortality and morbidity if not diagnosed early.

Namba *et al*. reveals increased familial incidence of JMG and occurrence of disease in both members of monozygotic twins, but none of the dizygotic twins are affected. This study highlights the genetic role in the pathogenesis of MG probably autosomal recessive inheritance.[6] According to this study, the other twin had no risk of MG and was not evaluated.

A study by Monsul *et al*. suggested that early use of steroids may decrease the progression of OMG to generalised MG.[7] A randomised, double-blind trial by Palace *et al*.[8] suggested that a combination of prednisolone on alternate days with azathioprine in AChR-positive generalised MG patients reduces the maintenance dose of prednisolone and associated with reduced incidence of treatment failures, longer remission and fewer side effects.

Skeie *et al*.,[9] recommend mycophenolate mofetil, tacrolimus (FK506) may be used as second-line immunosuppressive drugs. Plasma exchange and intravenous immunoglobulin are equally effective and are recommended as a short-term treatment to induce remission in MG exacerbations. Thymectomy (TE) should be done irrespective of severity of MG if a thymoma is detected. TE increases the chances of remission and clinical improvement in non-thymomatous autoimmune MG.[9]

The latest therapeutic approaches in MG include (a) Rituximab-monoclonal antibody to CD 20 useful in refractory MG. Experimental treatments tested in animal model of MG experimental autoimmune MG includes (a) tumour necrosis factor-α inhibitors like etanercept, (b) complement inhibitors, (c) modulation of AChE expression by the use of oral antisense oligonucleotides which causes inhibition of targeted gene transcription, (d) cellular therapy, (e) specific removal of anti-AChR antibodies and (f) reduction of AChR modulation at neuromuscular junction.[10]

CONCLUSION

Juvenile Ocular Myasthenia Gravis is a rare and often overlooked condition. Pediatric patients presenting with ptosis or strabismus should prompt suspicion of childhood MG. Notably, familial incidence has been reported, emphasizing

the importance of vigilant monitoring of siblings, particularly in twin cases. Early recognition and diagnosis are crucial, as timely intervention with appropriate treatment significantly improves prognosis and decreases the risk of ambylopia.

Ethical approval

The Institutional Review Board approval is not required.

Declaration of patient consent

Patient consent not required as the patient's identity is not disclosed or compromised.

Financial support and sponsorship

Nil.

Conflicts of interest

There are no conflicts of interest.

Use of artificial intelligence (AI)-assisted technology for manuscript preparation

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Karnataka Paediatric Journal

Case Report

A nuanced presentation of congenital pleural effusion: The chylothorax

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ABSTRACT

This article details a rare instance of congenital pleural effusion, the cause of which was chylothorax. Congenital pleural effusions have several potential causes. To determine the cause, a variety of prenatal and postnatal tests can be performed. The range of presentations and the seriousness of the case's complications determine the management strategy. A case of chylothorax is presented in this article, which was treated conservatively after postnatal confirmation through pleural fluid analysis.

Keywords: Congenital chylothorax, Pleural effusion, Hydrops

INTRODUCTION

Neonates seldom experience pleural effusions. Both prenatal and postnatal acquisitions are possible. Prenatal pleural effusions have been linked to several conditions, including congenital heart defects, pulmonary malformations, hydrops fetalis, infections contracted in utero, congenital chylothorax and other uncommon infections. Among the most common postnatal causes are iatrogenic ones, such as peritoneal dialysis, central catheter-related complications, infections and newborn tachypnoea that are momentary and traumatic chylothorax.[1]

Prenatal pleural fusions are primarily caused by chylothorax and congenital heart defects. Prenatal pleural effusion most commonly results from congenital chylothorax. It frequently occurs in conjunction with hydrops foetalis and is most likely caused by abnormal lymphatic system development or obstruction.[1]

Antenatally, hydrops foetalis frequently include pleural effusions, which are identified by ultrasonogram (USG). Pulmonary hypoplasia, which can also manifest as severe respiratory distress at birth, can be caused by long-standing effusions that occur before 20 weeks of gestation. Physical findings include diminished breath sounds and dullness to percussion.[2,3]

Neonates who show no symptoms can be carefully treated and frequently observed. Neonates exhibiting symptoms ought to be managed symptomatically, utilising pleural fluid drainage, respiratory supports and empirical antibiotics if required. Pleural fluid should be analysed for all suspected aetiologies. Although serial needle aspirations may not always be successful in removing some of the effusions, tube thoracostomy is usually necessary.[2,3]

In this case study, we present a baby who had an antenatal pleural effusion and, following a thorough evaluation, was found to have chylothorax.

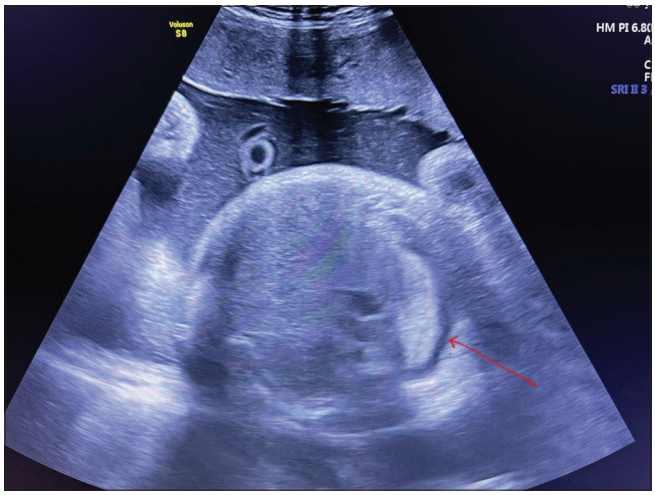

Figure 1: Antenatal ultrasound scan at 37 weeks showing right-sided pleural effusion (10–15 mL) of pleural fluid (red arrow).

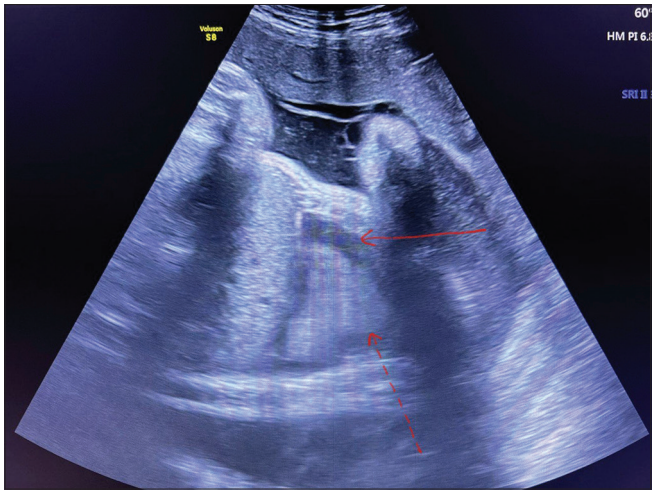

Figure 2: Antenatal ultrasound scan at 37 weeks showing right-sided pleural effusion (10–15 mL) of pleural fluid (red arrow) and collapsed lung (red dotted arrow).

CASE REPORT

A 25-year-old primigravida mother of a non-consanguineous marriage gave birth to a baby boy at 37 weeks and two days through elective caesarean section.

The mother experienced spontaneous conception, took folic acid supplements regularly and underwent routine prenatal care. The first trimester was uneventful. The nuchal translucency scan showed an increased nuchal fold thickness of 6.7 mm. The mother was treated with diet control after being diagnosed with gestational diabetes mellitus at 22 weeks of pregnancy. At 20 weeks, a level 2 scan revealed a bilateral pleural effusion, right more than left, with cardia displaced to the right side with sufficient foetal growth, and recommended additional testing. No noteworthy results were obtained from the quantitative

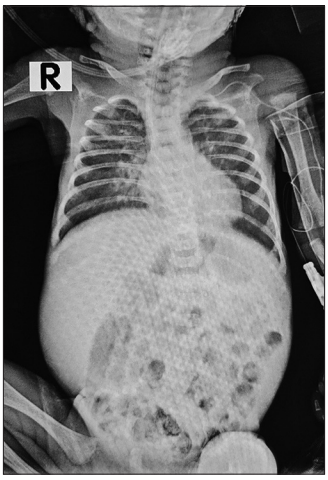

Figure 3: Frontal chest X-ray, supine film showing mild homogeneous haziness in bilateral lung fields at 2 hours of life.

fluorescent polymerase chain reaction, chromosomal microarray, karyotyping or TORCH panel (toxoplasma gondii, rubella, cytomegalovirus and herpes simplex virus 1 and 2), which was performed through amniocentesis. Antenatal 2 D echo was normal. The term scan also showed minimal bilateral pleural effusion with the right side more than the left side, polyhydramnios and adequate foetal growth [Figures 1 and 2]. Family history was insignificant. The maternal serology (human immunodeficiency virus, hepatitis B surface antigen, hepatitis C virus and venereal disease research laboratory) was non-reactive.

The baby was delivered through elective caesarean section in view of non-progressive labour. The baby cried immediately after birth. The newborn weighed 3.22 kg, measured 51 cm in length, had a head circumference of 31 cm and a chest circumference of 33 cm. A postnatal scan performed shortly after delivery revealed bilateral pleural effusion. Nothing remarkable was found after a head-to-toe examination. The infant began to exhibit tachypnoea, nasal flaring and mild chest retractions after 30 min of birth. A high-flow nasal cannula (HFNC) ventilation was given for the baby. Arterial blood gas results indicated adequate oxygenation. The results of the USG abdomen and neurosonogram were normal and did not reveal any hydrops-related characteristics. Culture revealed no growth, and the postnatal septic screen was negative. A bilateral pleural effusion was seen on the chest X-ray, and there were no additional contributing findings [Figure 3]. On day 4, HFNC settings were gradually weaned, and in view of unsettling tachypnoea, diagnostic pleural tapping was performed. An examination of the pleural fluid revealed characteristics that pointed to chylothorax–glucose – 53 mg/dL, total protein – 3 g/dL, lactate

dehydrogenase – 96 IU/L, total cholesterol 17 mg/dL and triglycerides – 10 mg/dL (before feeding). Over the 8th day, the baby's tachypnoea improved and was weaned off to room air. Vitamin K was administered along with a birth immunisation. The baby was put on IV fluids at first and then switched to trophic Ryle's tube feeds, and by day 8, he was being breastfed directly. No signs and symptoms of feed intolerance were noted. Very little pleural effusion and sufficient lower lobe expansion were seen on the pre-discharge USG chest.

The prognosis, recurrence, potential complications and consequent requirement for routine follow-up were explained to the patient's parents.

The baby was followed up after an interval of one month. Repeat chest X-ray and USG of the thorax showed no evidence of fluid collection or recurrence. The baby was healthy, active and feeding well and has received complete vaccination as per schedule, without any evidence of complications. Further, the baby is on a regular 3-month follow-up.

DISCUSSION

Congenital chylothorax is a rare disorder. It is characterised as a non-traumatic pleural effusion that is found 28 days after birth or before birth with a lymphocyte fraction of at least 80%. The term 'pleural lymph fluid accumulation' refers to the build-up of lymph that may result from direct lymphatic duct leakage, excessive production or drainage blockage. The incidence of congenital chylothorax is 1/10000–1/24000, with an overall survival rate of 30–70%. Congenital chylothorax may manifest independently or in conjunction with other genetic disorders or syndromes. The genetic syndromes associated with it are Turner syndrome, Noonan syndrome and Down syndrome. The identified congenital abnormalities include pulmonary lymphangiectasia, pulmonary hypoplasia and extrapulmonary lymphatic dysplasia or malformation.[4]

It is possible to detect congenital chylothorax in a foetus or during an early neonatal period. Accumulated pleural fluid in the foetal stage raises intrathoracic pressure, which, in turn, causes the foetal to swallow less amniotic fluid, leading to polyhydramnios and frequently premature labour. It also exacerbates issues that lead to impaired lung development. Prematurity, pulmonary dysplasia and hydrops foetalis are associated with unfavourable outcomes. In addition, pulmonary dysplasia is the most frequent cause of death.[4,5]

An analysis of pleural fluid reveals increased triglycerides, proteins and lymphocyte fractions (>80%). Thoracocentesis, thoracoamniotic shunting and pleurodesis are the available prenatal interventional options. Surgical intervention, nutritional support, drainage of effusions and respiratory support are all part of postpartum care.[6] When treating patients with refractory chylothorax, octreotide administration is recommended. Octreotide can be administered as a continuous intravenous infusion or given twice daily as an intravenous bolus or subcutaneous. The effective daily doses were from 7.2 μg/kg to 240 μg/kg for intravenous infusion. In 3–6 weeks, the majority of newborns experience total remission. Surgical interventions can be attempted, if not within six weeks. The last resort is surgical procedures that involve chemical pleurodesis. There may be a hereditary component to congenital chylothorax, as it is linked to recurrence in later siblings.[7]

CONCLUSION

A full-term baby boy diagnosed with mild antenatal pleural effusion was born. Nothing noteworthy was found during the prenatal workup. Chylothorax was diagnosed based on characteristics found in postnatal pleural fluid analysis that suggested chylous collection. The baby significantly improved after pleural fluid drainage. Any potential complications could be overlooked because there is no long-term follow-up included.

Ethical approval

The research/study complied with the Helsinki declaration of 1964.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent.

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Nil.

Conflicts of interest

There are no conflicts of interest.

Use of artificial intelligence (AI)-assisted technology for manuscript preparation

The authors confirm that there was no use of AI-assisted technology for assisting in the writing or editing of the manuscript and no images were manipulated using AI.

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Karnataka Paediatric Journal

Case Report

Lumps and bumps in a young child: Hyaline fibromatosis syndrome

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ABSTRACT

Hyaline fibromatosis syndrome (HFS) is a rare deposition disorder, wherein the skin and internal organs have unusual amounts of amorphous hyaline material. HFS can present as a milder form called Juvenile Hyaline Fibromatosis or severe Infantile Systemic Hyalinosis (ISH). Skin changes common to both the entities are skin thickening, perianal nodules, facial papules, gingival hyperplasia, hyperpigmented plaques over flexural joints and massive subcutaneous scalp tumours. Conspicuous involvement of the joints in the form of joint contractures affects patient morbidity. Furthermore, ISH involves the gastrointestinal tract characterised by protein-losing enteropathy and malabsorption. This contributes to the high mortality seen in ISH. We describe a 2-year-old male patient with grade 2 HFS who succumbed to a lower respiratory tract infection, a manifestation not typically observed in moderate HFS cases.

Keywords: Juvenile hyaline fibromatosis, Infantile hyaline fibromatosis, Hyaline fibromatosis syndrome, Joint contractures, Subcutaneous scalp tumours

INTRODUCTION

Hyaline Fibromatosis Syndrome (HFS) is a rare deposition disorder of autosomal recessive inheritance. Juvenile hyaline fibromatosis (JHF), the milder form, presents with cutaneous papules and nodules, gingival hyperplasia and variable degrees of bone involvement. However, Infantile Systemic Hyalinosis (ISH), the severe form, is distinguished by widespread hyaline deposition in various organs, particularly the gastrointestinal tract. It presents with protein-losing enteropathy and malabsorption, which, in turn, leads to recurrent infections. Most affected individuals succumb to the illness within the first 2 years of life.[1]

CASE REPORT

A 2-year-old male child, born of a second-degree consanguineous marriage and normal full-term vaginal delivery, presented with inability to extend the knees and wrist joints since 6 months of age, and asymptomatic skin lesions involving the scalp, neck and back for 1 year. There was no history of diarrhoea or recurrent respiratory tract infections. He had short stature and was severely underweight, that is <−3 Standard deviation (SD) in the World Health Organization growth chart.

Cutaneous examination showed multiple, skin-coloured tumours over the scalp, chest and back [Figures 1 and 2]. Innumerable, grouped, pink to skin-coloured, smooth, flat to dome shaped papules were noted over the face, scalp, ears, neck, chest and back [Figure 3]. Gingival tumours were also seen. Movements of the wrist and knee joints were limited, with fixed contractures over

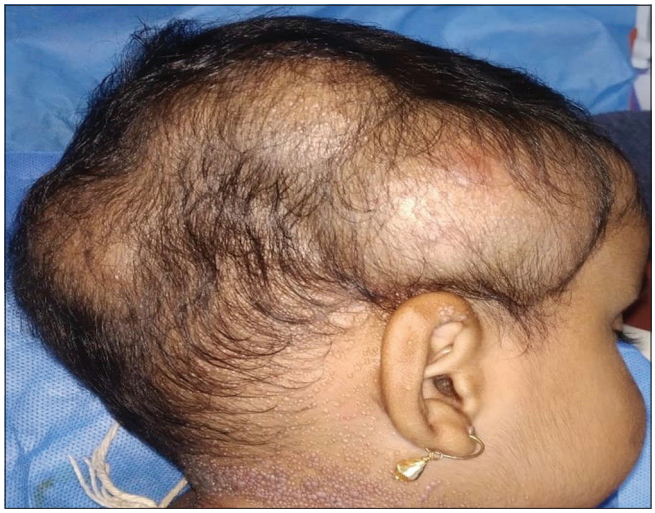

Figure 1: Multiple, large, firm, skin-coloured and non-tender tumours involving the scalp.

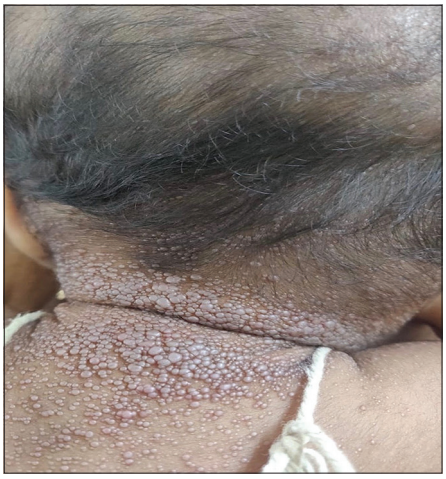

Figure 3: Innumerable, grouped, pink to skin-coloured, smooth, flat to dome-shaped papules over the neck, scalp and ears.

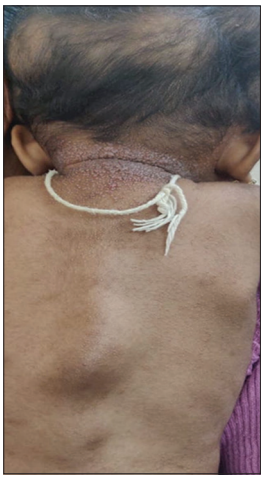

Figure 2: Tumours over the spine.

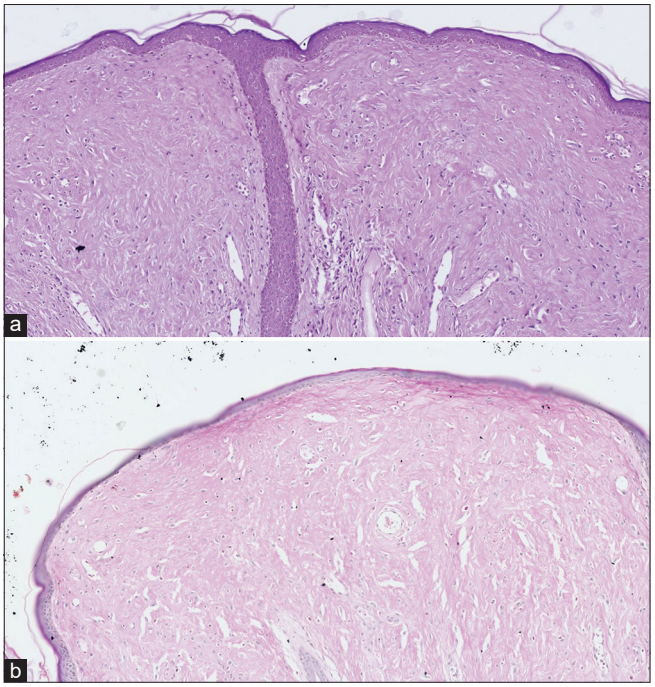

Figure 4: (a) Homogenous, eosinophilic deposits in the papillary dermis, with epidermal atrophy (Hematoxylin and eosin (H&E), ×10). (b) Periodic acid-Schiff positivity in the papillary dermis (×10).

forearm and thighs. Developmental milestones were adequate for age. Systemic examination was normal. Laboratory investigations including complete blood count, liver function tests and renal function tests were within normal limits. Radiographic examination of the long bones revealed osteopenia and soft-tissue contractures. Ultrasonography of scalp showed lobulated soft-tissue intensity lesions without the involvement of underlying bone. Magnetic resonance imaging brain and fundoscopy were normal. Histopathological examination of skin lesions showed Periodic acid-Schiff (PAS)-positive, diastase resistant homogenous, eosinophilic deposits in the papillary dermis and with epidermal atrophy [Figure 4a and b]. The above features were diagnostic of HFS. The patient was referred to plastic surgery for palliative removal of the tumours and to orthopaedics for release of soft-tissue contractures. The parents were offered genetic counselling. However, genetic testing was not done due to financial constraints. Unfortunately, the child passed away 6 months later due to a lower respiratory tract infection.

DISCUSSION

HFS is a spectrum disease that includes JHF and infantile hyaline fibromatosis (IHF). It is marked by intracellular hyaline deposits.

The incidence of the disease is not well-known. However, medical literature shows fewer than 70 documented cases of JHF and 20 cases of ISH.[2-5]

HFS is an autosomal recessive disorder caused by loss-of-function mutations in the anthrax toxin receptor 2 (ANTXR2) gene located on chromosome 4q21.21. ANTXR2 encodes for a transmembrane protein, which plays a critical role in endothelial cell morphogenesis and is essential for maintaining skin integrity. Mutations in ANTXR2, which can be homozygous or compound heterozygous, disrupt its function, leading to the abnormal deposition of hyaline material observed in HFS. According to genotype-phenotype tests, ISH is typically caused by truncating mutations and mutations affecting the extracellular domain, whereas JHF is caused by mutations in the cytoplasmic domain.[2,3]

HFS presents with a spectrum of cutaneous manifestations. Pink to pearly-white papules, plaques are seen affecting the chin, forehead, ears, posterior neck, perianal region and nasolabial folds. The scalp is characterised by large subcutaneous nodules. In addition, hyperpigmented macules and patches may be observed over bony prominences, particularly the metacarpophalangeal joints, wrists and ankles. Oral cavity involvement is seen as gingival hyperplasia, curved dental roots and tooth misalignment.[3] Radiographic investigations reveal osteopenia and periosteal reactions. Bony involvement in the form of flexion contractures affecting the proximal and distal interphalangeal joints of the hands and elbows is seen. Early adulthood is when progressive joint contractures are first noticed. In early adulthood, progressive joint contractures, often involving major joints, restrict patients to either bed or wheelchair.[3]

IHF is comparable to JHF, but the former affects the joints much more severely, resulting in joint contractures. In contrast to JHF, ISH presents with significant visceral involvement within the first few weeks or months of life. The most common feature is hyaline infiltration of the small intestine and colon, disrupting their absorptive function. This manifests clinically as malabsorption and protein-losing enteropathy, leading to diarrhoea, failure to thrive, growth failure, osteopenia and an increased susceptibility to infection. Severe joint pain and restriction cause respiratory insufficiency and immobility. By the time, a child is 2 years old, sepsis with associated renal, pulmonary and cardiac failure frequently results in death.[6]

Histologically, the epidermis appears normal, while the dermis shows characteristic hyaline deposition. This eosinophilic, PAS-positive material accumulates in both papillary and reticular dermis, within the extracellular and perivascular spaces. Spindle cell proliferation without atypia and minimal inflammation are also observed.[3]

Proposed grading system for HFS adapted from Denadai *et al*.[7]

Our child had skin, gingival, joint and bone involvement but did not present with protein-losing enteropathy, recurrent infections, diarrhoea and failure to thrive and was thus graded as moderate HFS (Grade 2) [Table 1]. Diagnosis in our case was based on presence of typical clinical manifestations and subsequently confirmed by histopathological analysis. Molecular testing was not done due to financial constraints.

The differential diagnosis of HFS includes several disorders, such as congenital generalised fibromatosis, lipoid proteinosis and Farber disease. Farber disease can be distinguished from HFS by its neurologic involvement and lack of hyperpigmented patches. Lipoid proteinosis presents with hoarseness as the initial symptom followed by tongue enlargement, dental hypoplasia, the formation of papules around the eyelids and on the face and specific skin lesions (vesicles and crusted bullae that develop into waxy plaques) which are not seen in HFS. Multiple, or generalised, nodules made up of smooth muscle and differentiated fibroblast cells without the typical HFS skin abnormalities are the hallmarks of congenital generalised fibromatosis.[8]

At the moment, HFS lacks any definitive treatment. The sole goal of therapy is palliative. Early surgical intervention may be attempted to halt disease progression in localised lesions; however, recurrences may occur after excision.[9]

Intralesional corticosteroids may offer some benefit in managing early-stage HFS lesions. However, their effectiveness is limited in advanced lesions, which can become quite large and ulcerate, causing significant patient discomfort.[9]

Table 1: Grading system for Hyaline fibromatosis syndrome (HFS).

Feature	Grade 1 (Mild)	Grade 2 (Moderate)	Grade 3 (Severe)	Grade 4 (Lethal)
Skin and/or Gingival Involvement	Present (+)	Present (+)	Present (+)	Present (+)
Joint and/or Bone Involvement	Absent (–)	Present (+)	Possible (±)	Possible (±)
Internal Organ Involvement	Absent (–)	Absent (–)	Possible (±)	Present (+)
Severe Clinical Decompensation	Absent (–)	Absent (–)	Absent (–)	Present (+)

Physiotherapy has little impact, although capsulotomy of the joints may have some short-term, positive effects like increased joint mobility.[10] Partial or radical gingivectomy is done for gingival overgrowth.[9]

CONCLUSION

Our patient had grade 2 HFS. Unfortunately, the patient succumbed to a lower respiratory tract infection, a manifestation not typically observed in moderate HFS cases. Given this atypical outcome, genetic testing is essential for further management and a deeper understanding of the pathogenesis of HFS.

Ethical approval

The Institutional Review Board approval is not required.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent.

Financial support and sponsorship

Nil.

Conflicts of interest

There are no conflicts of interest.

Use of artificial intelligence (AI)-assisted technology for manuscript preparation

The authors confirm that there was no use of artificial intelligence (AI)-assisted technology for assisting in the writing or editing of the manuscript and no images were manipulated using AI.

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Karnataka Paediatric Journal

Case Report

Kartagener's syndrome in an adolescent male: A case report and review of literature

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ABSTRACT

Kartagener's syndrome (KS) is a rare autosomal recessive genetic condition causing disruption to ciliary movement, leading to the triad of sinusitis, situs inversus, and bronchiectasis. Mutations in genes such as *DNAI1* and *DNAH5* increase susceptibility to recurrent sinopulmonary infections, infertility and errors with the left-right body orientation. A teenage boy with a decade-long history of sinusitis, chronic cough and ear infections showed bronchiectasis and situs inversus in clinical and imaging examinations. He had a neonatal intensive care unit stay for 1 month due to respiratory distress at birth, where dextrocardia was noted. Treatment with antibiotics, mucolytics, chest therapy and vaccination improved his symptoms. KS should be considered in newborns with dextrocardia and breathing problems. Genetic counselling and fertility issues should be addressed once KS is diagnosed.

Keywords: Kartagener's syndrome, Chronic sinusitis, Bronchiectasis, Situs inversus

INTRODUCTION

Kartagener's syndrome (KS) is a rare autosomal recessive genetic disorder affecting ciliary movement, characterised by sinusitis, situs inversus and bronchiectasis. It occurs in approximately 1 in 30,000 live births and was first described by Siewert in 1904. The clinical syndrome, including chronic sinusitis, bronchiectasis and situs inversus, was recognised by Manes Kartagener in 1933.[1] Ciliary dyskinesia was suggested as its cause by Camner *et al.*, in 1975, while the term 'immotile cilia syndrome' was coined by Eliasson *et al.*, in 1977 to highlight infertility alongside sinopulmonary infections.[2,3] KS results from gene mutations of more than 50 genes, including *DNAI1* and *DNAH5*, impairing ciliary motility crucial for respiratory defence, sperm motility and proper embryonic visceral orientation during embryogenesis.[4] This case report describes an adolescent boy with KS, reported due to its rarity and aims to contribute toward a greater understanding of KS.

CASE REPORT

A 13-year-old male developmentally normal adolescent, born out of a non-consanguineous marriage, presented to the paediatrics outpatient department complaining of recurrent episodes of productive cough since 3 years of age. There was a history of increased coughing when exposed to dust and shortness of breath on exertion. The child had daily symptoms of cough with shortness of breath for which he was being treated as poorly controlled bronchial asthma; however, it was

mildly relieved on metered dose inhaler (Salbutamol and Formoterol). The child also had a history of suppurative otitis media of the left ear and chronic sinusitis since 3 years of age. There is no history of chest pain, haemoptysis, fever or weight loss. There was no similar history or history of atopy in the family. There was no history of tuberculosis contact. The child was born at full term by vaginal delivery with birth weight of 3 kg. The baby was admitted to the neonatal intensive care unit immediately after birth in view of respiratory distress for about a month, suspected to be meconium aspiration syndrome. A chest X-ray was done, which suggested the heart to be on the right side. General examination revealed no remarkable findings. Vitals were stable. The child was well-nourished, with anthropometry within normal limits. Respiratory examination revealed bilateral coarse crepitations and extensive wheeze all over the lung fields. Cardiovascular examination revealed heart sounds being heard clearer on the right side of the chest with no murmur.

A complete hemogram was done, which showed haemoglobin – 12.1 g/dL, total leucocyte count – 14.15 × 10*9/L, red blood cell count – 5.55 × 10*12/L, platelets – 554 × 10*6/L, mean corpuscular volume – 66.4 fl, mean corpuscular haemoglobin – 21.8 pg and mean corpuscular haemoglobin concentration – 32.8 g/dL. Differential leukocyte count showed N – 62, L – 26, M – 8 and E – 4. Serum immunoglobulin E levels showed 150.5 IU/mL (Normal range: 0 – 160 IU/mL). Sputum for acid-fast bacilli was negative. C-reactive protein and erythrocyte sedimentation rates were within normal limits. The Mantoux test was negative.

Chest X-ray showed apex and aortic arch of the heart on the right side, gastric bubble on the right side and prominent bilateral bronchovascular marking in lower lobes [Figure 1]. An electrocardiogram (ECG) was done, which suggested right axis deviation, inversion of all complexes in the lead I (inverted P wave, negative QRS, inverted T wave), upright p wave in aVL lead and an absent R wave progression and prominence of S wave in the anterior leads and low voltage in leads V4–V6 [Figure 2]. The ECG findings were normal for the right-sided chest leads. Ultrasonography abdomen revealed a liver on the left side and spleen on the right side of the abdomen, suggesting situs inversus. His pulmonary function tests suggested decreased forced vital capacity (FVC) (1.59 L or 64% predicted), decreased forced expiratory volume in one second (FEV1) (1.37 L or 65.0% predicted) and Tiffeneau-Pinelli index (FEV1/FVC) (86.1% with 98% predicted) suggestive of restriction. Echocardiography was done, which confirmed situs inversus, with no other structural heart defect. A High-resolution computed tomography scan of the lungs was performed, which showed bilateral, central, cylindrical, and varicose bronchiectatic changes with extensive mucosal plugging along with centrilobular benching nodules in bilateral lungs suggestive of infective bronchiolitis [Figures 3 and 4]. With the above-mentioned history, the primary ciliary dyskinesia clinical prediction rule (PICADAR) score[5] came to 11. Along with clinical signs and imaging studies, the diagnosis of KS was made.

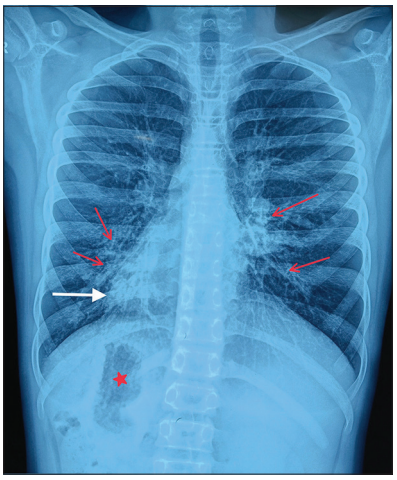

Figure 1: Chest X-ray showing apex of heart on the right side (shown as white arrow), gastric bubble on the right side (shown as red star mark), bilateral bronchovascular marking prominent in lower lobes, consistent with bronchiectasis (red arrow).

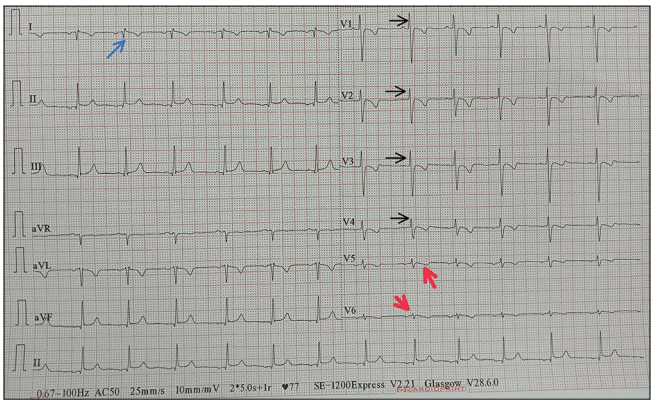

Figure 2: Electrocardiogram showing right axis deviation, inversion of all complexes in lead I (inverted P wave, negative QRS, inverted T wave) shown as blue arrow, an absent R wave progression and prominence of S wave in the anterior leads (shown as black arrow) and low voltage in leads V4–V6 (shown as red arrow).

Antibiotics, mucolytics, inhaled bronchodilators and chest physiotherapy were advised. The child was vaccinated with influenza and pneumococcal vaccine. He was advised to avoid cough suppressants. Otorhinolaryngology consultation was taken in view of chronic suppurative otitis media of the left ear and chronic sinusitis. Pus culture revealed *Staphylococcus aureus* bacterial growth and the child was advised of medical management and close follow-up.

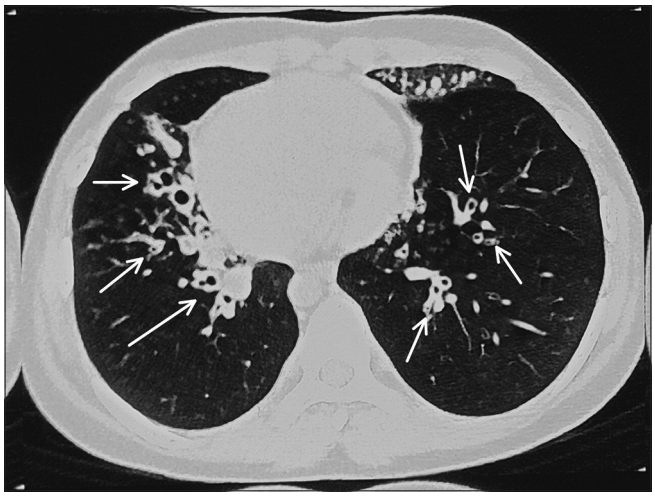

Figure 3: High-resolution computed tomography showing bilateral central cylindrical and varicose bronchiectatic changes with extensive mucosal plugging (shown as white arrow) with centrilobular benching nodules in bilateral lungs suggestive of infective bronchiolitis.

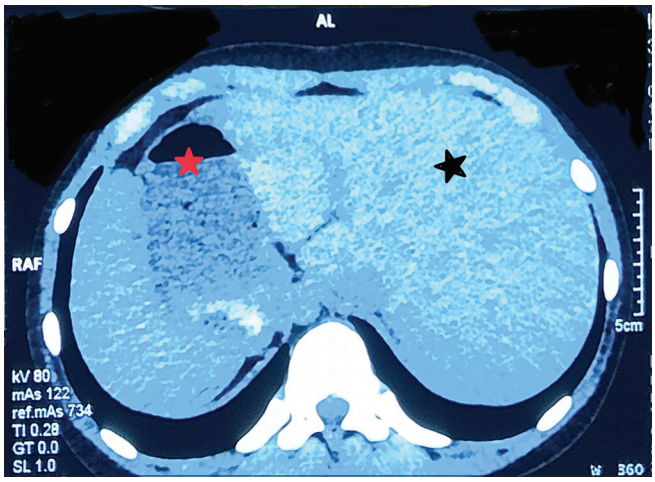

Figure 4: High-resolution computed tomography showing liver on the left (black star) and stomach bubble on the right side (red star).

The nature, outcome and prognosis of the condition with a risk of future infertility and the importance of regular follow-up have been explained to the child's parents.

DISCUSSION

KS is a rare disorder occurring in about 1 in 30,000 live births.[6] It falls under the category of primary ciliary dyskinesia (PCD), a congenital condition characterised by abnormalities in ciliary structure or function. This condition, which follows an autosomal recessive inheritance pattern, involves faulty coding for dynein protein, with DNAH5 and DNAI1 being the most commonly mutated genes.[7] These genetic mutations result in cilia that are misshapen, incorrect in size, or moving improperly, leading to impaired ciliary motility.[4] Normal ciliary function is crucial for proper embryonic development, respiratory health and sperm movement. Malfunctioning cilia can cause defects in the left-right body orientation, such as situs solitus or situs inversus totalis, as well as recurrent sinus and lung infections and infertility.

KS manifests as a combination of chronic sinusitis, bronchiectasis and situs inversus, though symptoms can vary among individuals. Neonates may initially experience respiratory distress, progressing to chronic cough due to bronchiectasis, unresponsive asthma, recurrent sinus infections, otitis and fertility issues such as ectopic pregnancy in females or infertility in males.[8] Diagnosis should be considered in term neonates with dextrocardia and unexplained respiratory distress or pneumonia.[9] In our case, the patient displayed the classic triad of sinopulmonary infections, situs inversus and central bronchiectasis. While initially presenting with neonatal respiratory distress and situs inversus, the diagnosis was missed. Subsequent treatment for poorly responsive asthma should have prompted consideration of ciliopathy. PICADAR score is a diagnostic predictive tool used for identifying patients who may have PCD. A full score of 14 points corresponds to a 99.80% probability of having PCD, while a score of ≥10 indicates a 92.6% probability, and a score of ≥5 indicates an 11.10% probability.[5]

Additional diagnostic tests may involve assessing nasal epithelial mucociliary function, typically measured by decreased nasal nitric oxide levels. Electron microscopy can reveal reduced ciliary beat frequency (<11 Hz/s) and absent ciliary dynein arms. Genetic studies may also be conducted to identify mutations in the DNAI1 and DNAH5 genes.[10,11] These tests have not been performed in our case due to financial limitations.

Infertility primarily results from compromised ciliary motility affecting sperm tail function and fallopian tube movement, alongside significant lung implications. In rare instances, cardiac anomalies beyond dextrocardia, such as ventricular septal defects, transposition of great arteries and pulmonary outflow defects, may also occur.[12] In addition, conditions such as pancreatic malformations, polycystic kidney disease, polysplenia and vascular malformations, although uncommon, have been documented in some cases. As a result, PCD is increasingly acknowledged as a multi-system disorder.[13]

Timely identification and consistent medical monitoring are imperative for patients with these conditions to mitigate potential complications, given the absence of a definitive cure. Supportive care for sinopulmonary issues in KS typically involves chest physiotherapy, inhaled mucolytics and antibiotic therapy. Since ciliary function is compromised, cough serves as a vital mechanism for mucus clearance; therefore, cough suppressants should be avoided. Those experiencing frequent bronchiectasis exacerbations (≥3 times/year) may require long-term and low-dose

prophylactic antibiotics. Due to heightened susceptibility to upper and lower respiratory infections, vaccinations such as influenza and pneumococcal are administered. Late diagnosis of bronchiectasis is associated with a poor prognosis, even with optimal management strategies.[14]

CONCLUSION

Recognizing and considering rare conditions like Kartagener syndrome and primary ciliary dyskinesia (PCD) in the differential diagnosis of chronic respiratory infections is crucial. Early identification leads to timely treatment and improved outcomes, preventing complications such as pneumonia and bronchiectasis. Awareness among healthcare providers is key, especially in specific patient groups like neonates with dextrocardia and respiratory distress, or children with persistent wheezing unresponsive to typical asthma therapies. In these cases, considering these syndromes is essential for early diagnosis, optimal management, and enhanced quality of life.

Ethical approval

The Institutional Review Board approval is not required.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent.

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Nil.

Conflicts of interest

There are no conflicts of interest.

Use of artificial intelligence (AI)-assisted technology for manuscript preparation

The authors confirm that there was no use of artificial intelligence (AI)-assisted technology for assisting in the writing or editing of the manuscript, and no images were manipulated using AI.

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Karnataka Paediatric Journal

Case Report

Emergent challenges in paediatric tracheostomy care: Insights from a case report

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ABSTRACT

Tracheostomy is a common surgical procedure in paediatric patients that can lead to life-threatening complications such as tube breakage. This case report highlights the management of tracheostomy tube breakage in a 3-year-old male with Down's syndrome and subglottic stenosis. On presentation with cough and cyanosis, the patient was found with a missing tracheostomy tube, leading to prompt intervention. Imaging revealed a broken tube in the left main bronchus, prompting successful extraction under general anaesthesia. The case underscores the importance of timely action and multidisciplinary care in such emergencies. The literature review reveals sparse discussion on paediatric tracheostomy tube breakage, emphasising the need for increased awareness and preventative measures. This report contributes to understanding and managing this uncommon yet critical complication, emphasising the necessity of thorough inspection and adherence to quality standards to ensure patient safety.

Keywords: Tracheostomy, Stenosis, Extubation, Emergencies, X-ray

INTRODUCTION

Tracheostomy is a surgical procedure in which an opening is created in the anterior trachea to facilitate respiration. Indications of tracheostomy include subglottic stenosis, ventilator dependence for a prolonged period, refractory obstructive sleep apnoea, and chronic aspiration, to mention a few. Complications such as accidental decannulation, obstruction, and haemorrhage are life-threatening, while tracheal stenosis, infections, and tracheocutaneous fistula formation are the minor ones, with a total occurrence rate of >50%.[1] Tracheostomy-related emergencies, if not managed properly, lead to significant morbidity and mortality, with the 30-day mortality being high, emphasising the need for multidisciplinary evaluation and a well-prepared emergency care provider.[2]

Paediatric tracheostomy rates have decreased over the years, but the complications have become more severe. Early wound identification protocols, reducing prolonged pressure, and skin care methods have helped significantly reduce the incidence and impact of these complications.[3] Tracheostomy tube breakage, though rare, is a dangerous complication, especially in the paediatric age group, as the broken tube needs to be taken out while maintaining the airway patency adequate for ventilation. Fragmented and migrated tracheostomy tubes have been reported as a life-threatening complication by Al-Momani *et al.* in the literature.[4] Corrosion and breakage of metal tracheostomy tubes have also been reported, imposing strict regulations over production control and quality.[5]

We present to you a case report with tracheostomy tube breakage in the paediatric age group, with the primary objective being the fact that such an emergency requires prompt action, along with highlighting ways to manage them. We hope to enhance our understanding of this emergency and its treatment by presenting the case and reviewing prior studies. Through this rare case study, we aim to provide readers with a better knowledge of tracheostomy tube breakage, which will make it easier for medical practitioners to identify and treat patients presenting with such a complication.

CASE REPORT

The patient is a 3-year-old male child known case of Down's syndrome and subglottic stenosis (tracheostomy done at day 65 of life), who presented to the paediatric casualty with increased cough and bluish discoloration of the face. The child had decreased alertness, and the tracheostomy tube was missing, with only the flange intact. The child's vitals were a pulse rate of 128/min, blood pressure of 90/52 mmHg, respiratory rate of 48/min, and the child was afebrile. There was no history of seizures, fever, or any other complaints. Physical examination revealed reduced air entry to the left lung. The rest of the systemic examination was normal.

Past history was significant for transient myeloproliferative disorder for 2 years, which had been managed effectively with multiple platelet transfusions, leading to resolution. Karyotyping revealed the Trisomy of chromosome 21. In addition, the patient had severe tricuspid regurgitation suggestive of pulmonary arterial hypertension, for which sildenafil and inotropes were administered. Subsequent echocardiography indicated normalisation at 2 months. Furthermore, the patient experienced a year back, right upper lobe pneumonia, which was successfully treated with sulbactam/cefoperazone and amikacin over a 10-day period. Due to repeated extubation failures, the child underwent a tracheostomy at 65 days of life. On-going ENT follow-ups advised tube changes every 15 days, with the last change performed at home 10 days prior.

On admission, the child was supported by bag and tube ventilation while maintaining satisfactory saturation with 5L O2 through T piece. Subsequently, subcutaneous emphysema was observed on the left chest wall along with pneumothorax. A chest X-ray revealed a broken tracheostomy tube in the left main bronchus area [Figure 1], prompting an ENT consultation for foreign body removal. Under general anaesthesia, the skin stoma was revised, enabling visualisation of the tracheostomy tube. Using optical forceps and a telescope introduced through the tracheostomy, the broken tube in the left bronchus was successfully extracted and replaced with a size 4 cuffed tracheostomy tube [Figure 2]. Haemostasis was achieved, and the child

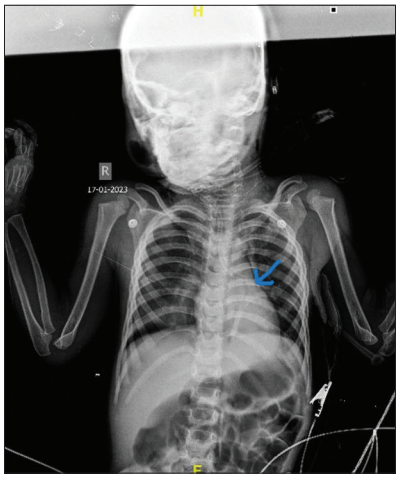

Figure 1: X-ray showing broken tracheostomy tube in the left main bronchus (Blue arrow).

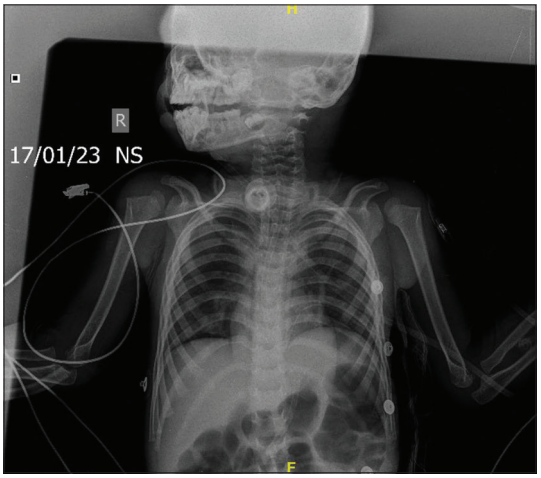

Figure 2: X-ray taken after removal of the broken tracheostomy tube.

was then transferred back to the paediatric intensive care unit. Post-procedure, the child's condition remained stable, with resolved pneumothorax and persistent subcutaneous emphysema. Due to borderline saturation levels (89–91%), the child was initiated on 2 L/min O2 through T piece. Suture removal was done on the 7th day, and ENT follow-up was advised.

DISCUSSION

In critically ill patients who require extended mechanical ventilation for acute respiratory failure and airway problems, tracheostomy is a routinely done intervention. With an average of over 100,000 tracheostomies being performed every year,[6] the complications that arise out of the procedure require prompt management to avert life-threatening crises

or significant morbidity. While tracheostomy is considered a life-saving treatment in certain clinical scenarios – the paediatric population is still at a higher risk than adults. The increased survival rates among premature newborns and those born with severe congenital disabilities, however, have led to a dramatic change in the rationale for tracheostomy in paediatric patients over the past few decades.

Being a life-saving intervention often employed in pediatric patients with complex medical conditions, for essential respiratory support and management, tracheostomy-related emergencies can be very difficult to handle unless recognised and managed urgently. Tracheostomy tube breakage has been reported in the literature[4,5,7] in which a broken tube in a 4.5-year-old child was removed after imaging using rigid bronchoscopy in the first case. Kock *et al*.[5] focused more on the need for strict regulation practices during the production of tracheostomy tubes, while Erdozáin Rodríguez *et al*.[7] reported the aspiration of the inner cannula into the left tracheobronchial tree and is one of the earliest reported cases of tracheostomy tube breakage.[4] Krishnamurthy and Vijayalakshmi reported right main bronchus and trachea are the most common sites for the dislodged components of a broken tracheostomy tube being trapped.[8] The common approach of removal of the broken parts through rigid bronchoscopy was done by Atwood *et al*. in a case where the tube broke at the junction of the cannula and neck plate.[9]

Tracheal polyps causing rupture of the tracheostomy tube cuff have also been reported in a rare case by Feifei *et al*.[10] Tracheobronchopathia osteochondroplastica, a rare benign disorder of the bronchi and trachea, was also reported to cause cuff rupture. Wilson *et al*., in a case similar to the one reported by Erdozáin Rodríguez *et al*., reported that the aspirated foreign body was removed by flexible bronchoscopy.[7,11] Studies have well addressed, how to prevent tracheostomy tube breakage. In one case, Loh *et al*. recommended regular follow-up and tube replacement.[12] In another case involving a 14-year-old child, Piromchai *et al*. suggested that proper cleaning and following a schedule for tube changes can help prevent breakage and associated complications.[13]

Breakage is caused by a number of causes, including manufacturing flaws, repeated handling, and exposure to specific cleaning agents – which can cause the tracheostomy tube to fragment and migrate. The available literature highlights the importance of early detection and treatment in general, as patients are immediately at risk from foreign body aspiration.

Paediatric tracheostomy tube breakage and its management have not been discussed much before in literature.

Prompt and effective treatment is crucial to avoid life-threatening situations from rare but dangerous tracheostomy tube breakage in young patients. This case, involving a child with Down's syndrome and subglottic stenosis, exemplifies the unique challenges of managing such a break. Through this article, we have explored the strategies and difficulties associated with the management of this paediatric emergency.

Reviews of the literature highlight the importance of careful inspection and quality checks prior to the initial use of tracheostomy tubes, particularly in situations where inner cannulas may be prone to fracture. These incidents highlight the necessity of thorough preventative measures to avoid tracheostomy tube breaking, which include careful examination for manufacturing flaws, adherence to correct handling procedures, and routine inspection for potential corrosion in metal tubes. Such preventative measures are necessary to guarantee the security and welfare of patients who depend on tracheostomies for breathing support.

CONCLUSION

Paediatric tracheostomy tube breakage is a serious though not a well-reported area in scientific literature. Through this case report, we aim to provide insightful information about the efficient management and preventative measures essential in ensuring the well-being of paediatric patients with tracheostomies by critically analysing the cases and situating them within the larger framework of paediatric tracheostomy care.

Ethical approval

The Institutional Review Board approval is not required.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent.

Financial support and sponsorship

Nil.

Conflicts of interest

There are no conflicts of interest.

Use of artificial intelligence (AI)-assisted technology for manuscript preparation

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Karnataka Paediatric Journal

Case Report

Expanding the phenotypic spectrum of Rauch–Steindl syndrome: A case study of a novel NSD2 mutation in an Indian patient

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ABSTRACT

Rauch–Steindl syndrome (RAUST) is a rare genetic disorder characterised by poor growth, distinctive facial dysmorphisms and variable developmental delays. It is a milder variant of Wolf–Hirschhorn syndrome and is associated with mutations in the *NSD2* gene. This study presents a case of RAUST with a novel NSD2 mutation, expanding the phenotypic spectrum of the syndrome. An 18-month-old Indian boy was referred to Basaveshwar Teaching and General Hospital for failure to thrive. Born preterm at 1.7 kg with intrauterine growth restriction, he presented with severe growth retardation, microcephaly and facial dysmorphism including dolichocephaly, triangular facies and cleft palate. Developmental assessment revealed significant delays in motor and speech milestones. Genetic analysis identified a heterozygous *NSD2* gene variant (chr 4: g.1978839del; p.Pro1343GlnfsTer49), leading to a frameshift and premature truncation of the protein. Whole exome sequencing was performed on peripheral blood DNA, revealing the pathogenic NSD2 mutation. The analysis also highlighted the variability in phenotypic expression of RAUST, including additional anomalies such as undescended testes. The proband's clinical features align with RAUST, with the identified NSD2 mutation contributing to the observed dysmorphisms and developmental delays. Despite normal neuroimaging, the wide spectrum of symptoms, including additional anomalies, underscores the complex phenotypic variability of RAUST. The mother's similar phenotypic features suggest possible autosomal dominant inheritance with variable expressivity. This case highlights the diverse presentation of RAUST and the critical role of NSD2 mutations in its pathogenesis. It emphasises the need for genetic analysis in diagnosing RAUST and understanding its broad phenotypic spectrum. Further studies are necessary to explore the full range of clinical manifestations and inheritance patterns associated with RAUST.

Keywords: Rauch–Steindl syndrome, NSD2 gene mutation, Developmental delay, Dysmorphic features, Whole exome sequencing

INTRODUCTION

Rauch–Steindl syndrome (RAUST) is characterised by poor pre- and postnatal growth, sometimes with short stature and small head circumference, characteristic dysmorphic facial features and variable developmental delay with delayed motor and speech acquisition and impaired intellectual function that can be mild.[1] Other features may include hypotonia and behavioural abnormalities.[1] The phenotype represents a mild form of Wolf–Hirschhorn syndrome (WHS; 194190), which is a contiguous gene deletion syndrome caused by heterozygous deletion of several genes on chromosome 4p16.[2] The clinical features of RAUST are similar to but milder than those of WHS, with less severe dysmorphic facial features, less severe developmental

disabilities in general and absence of a seizure disorder.[2] The phenotype and expressivity of RAUST are highly variable.[3]

The heterozygous mutations in the *NSD2* gene that were identified in patients with RAUST by Lozier *et al.* (2018), Derar *et al.* (2019) and Barrie *et al.* (2019) occurred *de novo*.[3-5] The transmission pattern of RAUST in the Chinese family reported by Hu *et al.* (2020) was consistent with autosomal dominant inheritance with incomplete penetrance and variable expressivity.[6] Most of the heterozygous mutations in the NSD2 gene identified in RAUST patients by Zanoni *et al.* (2021) occurred *de novo*.[7] However, there were 2 families (families 6 and 7) in which the transmission pattern was consistent with autosomal dominant inheritance.[7]

Patients with RAUST exhibit a wide range of mild phenotypic features, with core manifestations of microcephaly, intrauterine growth restriction, facial dysmorphisms, autism, intellectual disability, low birth weight, feeding difficulties, failure to thrive, short stature, speech delay and muscular hypotonia.[8] In this study, we identified a heterozygous *NSD2* gene variant (chr 4: g.1978839del; Depth:86x) that results in a frameshift and premature truncation of the protein 49 amino acids downstream to codon 1343 (p.Pro1343GlnfsTer49; ENST00000508803.6) [Figure 1] in an Indian boy diagnosed with RAUST. In addition, our study further expands the phenotypic spectrum of RAUST with genetic mutations and associated maternal phenotype.[9]

CASE REPORT

Patient

An 18-month-old was referred to Basaveshwar Teaching and General Hospital Kalaburagi for failure to thrive on 29th April 2024. The proband's parents provided written informed consent for the publication of photographs as well as clinical and genetic data. The present study was approved by Department of Paediatrics, Basaveshwar Teaching and General Hospital attached to M R Medical College, Kalaburagi.

Genetic analysis

Whole exome analysis

Genomic DNA was extracted from 2-mL peripheral blood samples from the proband. Whole exome sequencing was performed at MedGenome Labs Ltd., Bangalore. DNA extracted from blood was used to perform targeted gene capture using a custom capture kit. The libraries were sequenced to mean depth of >80-100X on Illumina sequencing platform. The sequences obtained are aligned to human reference genome (GRCh38) using BWA aligner [Sentieon] and analysed using Sentieon for removing duplicates, recalibration and re-alignment of indels [Sentieon]. Sentieon haplotype caller is then used to identify variants in the sample. The germline variants identified in the sample is deeply annotated using VariMAT pipeline. In addition to single nucleotide variant (SNV) and small Indels, copy number variants are detected from targeted sequence data using the ExomeDepth method.

RESULTS

Clinical description

The proband [Figure 2] is an 18-month-old boy born to non-consanguineous parents, admitted for delayed development and growth. He was born late preterm with intrauterine growth restriction (birth weight: 1.7 kg), accompanied by cleft palate and respiratory distress, requiring a 6-day neonatal intensive care unit stay. Initial concerns included low birth weight and feeding difficulties due to the cleft palate.

Developmentally, at 7 months, he had a partial neck hold, social smile and alertness to sounds. By 8 months, he could roll

RESULTS

PATHOGENIC VARIANT CAUSATIVE OF THE REPORTED PHENOTYPE WAS DETECTED

SNV(s)/INDELS

Gene# (Transcript)	Location	Variant	Zygosity	Disease (OMIM)	Inheritance	Classification$
NSD2 **(+)** (ENST00000508803.6)	Exon 22	c.4028del **(p. Pro1343GlnfsTer49)**	Heterozygous	Rauch-Steindl syndrome (OMIM#619695)	Autosomal dominant	Pathogenic (PVS1, PM2, PPS)

Figure 1: Genetic report, Whole Exome Sequencing showing pathogenic variant of Rauch-Steindl syndrome (red color), SNV: Single Nucleotide Variant, INDEL: Insertion or Deletion, OMIM: Online Mendelian Inheritance in Man, NSD2: NSD2 nuclear receptor binding SET domain protein 2, PVS1: Pathogenic very strong, PM2: Pathogenic moderate, PPS: Pathogenic supporting. #: The in-silico prediction# of the variant, $: Due to inadequate literature evidence, this NSD2 variation is classified as a pathogenic variant and has to be carefully correlated with the clinical symptoms.

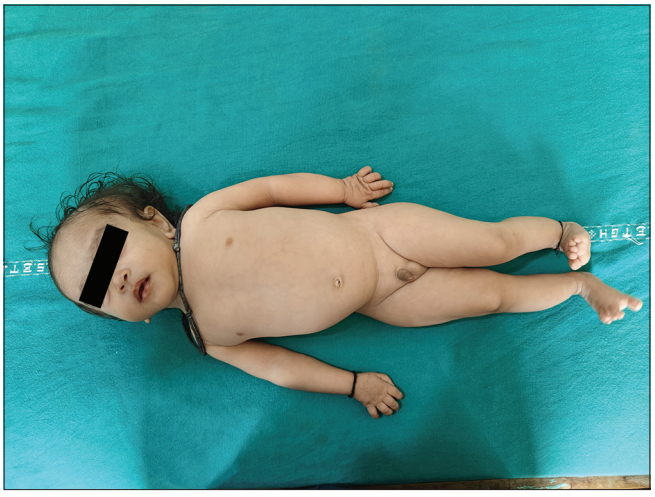

Figure 2: Proband.

Table 1: Comparison of Proband features with a reference case.

Clinical Features	Patient	Reference Case (Yang *et al*)
Variant	p.Pro1343GlnfsTer49	c.2721delT(p.Asn907Lysfs*5)
Gender	Male	Female
Age at examination	18 months	7 years
Gestation	36 weeks (Late preterm)	Full-term
Birth weight	1.7 kg	2.15 kg (<−2.5 SD)
Birth length	Not provided	45 cm (<−2 SD)
Feeding difficulty	Yes	Yes
Hypotonia	Yes	Yes
Weight	2.7 kg (<-3 SD)	15 kg (<−3 SD)
Length/Height	52 cm (<−3 SD)	118 cm (<−1 SD)
Head circumference (OFC)	35 cm (<−3 SD)	49 cm (<−2 SD)
Developmental delay	Yes, global developmental delay	Yes, cognitive impairment, delayed motor milestones
Age of walking	Not reached (18 months)	20 months
Age of first words	Not reached	25 months
Brain anomalies	MRI brain – Normal	MRI normal
Facial dysmorphism	Microcephaly, dolichocephaly, wide-open anterior fontanelle, broad forehead, triangular facies, eccentric pupils, prominent ears, deep-set eyes, periorbital fullness, wide/flat nasal bridge, cleft palate, micrognathia, delayed dentition	Triangular face, proptosis, hypertelorism, broad forehead, high anterior hairline, short/upslanted palpebral fissures, sparse eyebrows, short philtrum, micrognathia, abnormal teething
Other anomalies	Widely spaced nipples, bilateral undescended testes	Small hands/feet, mild clinodactyly of the right hand, loose skin on hands/feet

MRI: Magnetic resonance imaging, SD: Standard deviation, OFC: Occipito-Frontal Circumference

over and reach for toys. At 17 months, he sits with support, has an immature pincer grasp, coos and recognises his mother. His developmental quotient (DQ) is 13.8% for his age.

At present, he has severe acute malnutrition (weight: 2.7 kg, <−3 standard deviation [SD]) and severe short stature (length: 52 cm, <−3 SD). Table 1 depicts comparison of proband features with a reference case by Yang *et al*.[10] Dysmorphic features include microcephaly (head circumference: 35 cm, <−3 SD), dolichocephaly, wide-open anterior fontanelle, broad forehead, triangular facies, eccentric pupils, prominent ears, deep-set eyes, periorbital fullness, flat nasal bridge, micrognathia, delayed dentition, widely spaced nipples, delayed bone age and undescended testes.

Investigations (magnetic resonance imaging [MRI] brain, spine X-ray, chest X-ray [Figure 3], echocardiography, abdominal ultrasonography and brainstem evoked response audiometry (BERA)) were normal.

The mother [Figure 4] exhibits phenotypic traits of RAUST, including low birth weight, short stature, triangular facies, wide forehead, micrognathia, strabismus and refractive errors.

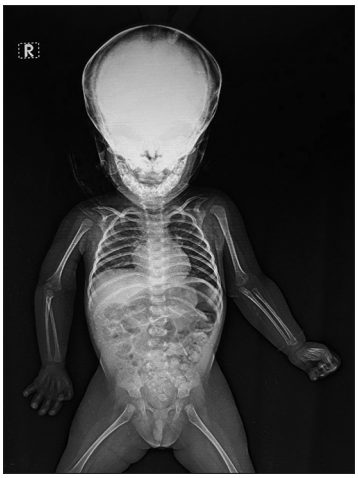

Figure 3: Skeletal survey.

DISCUSSION

Clinical presentation and dysmorphic features

The proband exhibits multiple dysmorphic features such as microcephaly, dolichocephaly, triangular facies, deep-set eyes and cleft palate. These are consistent with Rauch-Steindl syndrome (RSS), as described in the literature. The presence of less commonly discussed features, such as bilateral eccentric pupils and prominent ears might reflect the phenotypic variability within RSS or an additional genetic anomaly. The NSD2 variant identified in the proband has been linked to developmental phenotypes similar to those observed in RSS, suggesting that the genetic mutation directly contributes to craniofacial and developmental abnormalities.[1]

Figure 4: Proband's mother.

Growth parameters and development

The child's severe growth retardation and global developmental delay align with the typical RSS presentation. The proband's DQ of 13.8%, severe hypotonia and delayed motor milestones (e.g., sitting with support at 17 months) are characteristic of the developmental challenges associated with pathogenic NSD2 mutations. The identified NSD2 variant, known for causing frameshifts and truncation, likely disrupts normal protein function, leading to the observed growth and developmental delays. The literature supports that truncating mutations in NSD2 are a significant cause of the profound growth and developmental impairments seen in RSS.[2]

Genetic findings

The heterozygous 1 base pair deletion in the *NSD2* gene (p.Pro1343GlnfsTer49) is a critical finding that correlates with the proband's clinical presentation. This specific mutation has been reported in similar cases, further supporting its pathogenic role. NSD2 mutations are well-documented in RSS cases, particularly those involving frameshifts and premature truncations, which lead to the loss of essential protein functions. The proband's mutation is classified as pathogenic, consistent with other reported cases where NSD2 mutations result in the developmental and dysmorphic features characteristic of RSS.[11]

Brain and organ anomalies

Despite the significant developmental delays, the proband's brain MRI and other organ evaluations are normal. This is consistent with many RSS cases where neuroimaging does not reveal structural anomalies despite severe developmental issues. The identified NSD2 mutation, while leading to profound developmental delays, may not necessarily cause structural brain anomalies, as seen in the proband. The literature suggests that functional disruptions due to NSD2 mutations primarily manifest as neurodevelopmental delays rather than structural abnormalities.[12]

Additional anomalies

The proband exhibits bilateral undescended testes and widely spaced nipples, features that are less commonly associated with RSS but may represent an extended phenotypic spectrum due to the NSD2 mutation. The variability in phenotypic expression, including these additional anomalies, might be due to the specific truncating mutation in the *NSD2* gene, highlighting the broad spectrum of clinical manifestations that can arise from such mutations.[13]

CONCLUSION

This case of an 18-month-old boy with a heterozygous *NSD2* gene variant (p.Pro1343GlnfsTer49) provides valuable insights into RAUST. The patient's clinical presentation, including microcephaly, developmental delay and dysmorphic features, aligns with RAUST. The identified NSD2 mutation correlates with severe growth and developmental challenges, consistent with previously reported cases. Notably, the proband's mother also exhibits phenotypic features associated with RAUST, such as low birth weight, short stature and facial dysmorphisms, suggesting a possible autosomal dominant inheritance pattern with variable expressivity. Despite normal neuroimaging and additional anomalies like undescended testes, the findings highlight the broad phenotypic spectrum of RAUST and reinforce the role of NSD2 mutations in its pathogenesis. This case underscores the importance of genetic analysis in diagnosing RAUST and understanding its clinical variability.

Ethical approval

The Institutional review board approval is not required.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent.

Financial support and sponsorship

Nil.

Conflicts of interest

There are no conflicts of interest.

Use of artificial intelligence (AI)-assisted technology for manuscript preparation

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Karnataka Paediatric Journal

Letter to the Editor

Sialuria due to GNE pathogenic variant masquerading as cerebral palsy

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Quick Response Code:

Dear Editor,

Sialuria (OMIM#269921) is an inborn error of metabolism that leads to abnormally high free sialic acid and occurs due to dominant mutations in the *GNE* gene.[1] A total of ten case studies have been published to date. Here, we report the 11th case from the world and the first Indian case of Sialuria, which masqueraded as cerebral palsy.

A 7-year-old male child, without any significant family and birth history presented with developmental delay and seizures from 6 months of age. The child had a global developmental delay with a predominant delay in the motor domain. He currently walks with difficulty, needs help for activities of daily living, and speaks in phrases with dysarthria. Seizures were noted at 6 months of age, with frequency of 1–2 episodes per year, requiring 2 antiseizure medications. The child also had stiffness of limbs starting from 2 years of age. On examination, weight -15.5 kg, height -101 cm, and head circumference -48 cm all are <3. Standard deviation, spasticity in all four limbs, exaggerated deep tendon reflexes, and extensor planters. Hepatosplenomegaly and coarse facies were absent. On investigations, complete hemograms, liver and kidney function tests, ammonia, lactate, blood gases, and tandem mass spectrometry were normal. Magnetic resonance imaging (MRI) brain showed mild cerebral atrophy with dilated ventricles. Exome sequencing identified a likely pathogenic missense variant c.2086G>A, p.(Val696Met) in the *GNE* gene. Parents did not consent to testing. Urinary total N-Acetylneuraminic acid: 1.4 (0.41 ± 0.23 mmol/g creatinine) and Free N-Actylneuraminic acid: 1.0 (0.24 + 0.14 mmol/g creatinine) was elevated.

In summary, the child has a global developmental delay, seizures, and spastic quadriparesis with no organomegaly, which prompted us to think of cerebral palsy, hereditary spastic paraparesis, and arginase deficiency. Exome sequencing gave us a clue like the case identified by Champaigne *et al.*[2] On reverse phenotyping and biochemical investigation, the diagnosis of sialuria was confirmed. Mutations affecting the allosteric site Arg 263 and Arg 266 and other variants like p.Asp84 His have been described.[3] The mutation in the current case does not lie in the allosteric domain and awaits explanation by further functional studies. In conclusion, any child with cerebral palsy and no significant birth history or changes on an MRI brain scan should be investigated for genetic aetiology.

Ethical approval

The Institutional Review Board has waived the ethical approval for this study.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent.

Financial support and sponsorship

Nil.

Conflicts of interest

There are no conflicts of interest.

Use of artificial intelligence (AI)-assisted technology for manuscript preparation

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Karnataka Paediatric Journal

Letter to the Editor

Hyperpigmentation in a child as a clue to chikungunya

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Dear Editor,

A 2-month-old male child born of non-consanguineous marriage presented with high-grade, intermittent fever for 1 month along with history of reduced feeding and lethargy for 2 days. The birth weight of the child was 2.5 kg. The mother observed pigmentation initially appearing over the face, which progressively extended to involve the rest of the body for 20 days. All milestones were appropriate for age.

On examination, diffuse macular brownish-black pigmentation was observed over the nose, perioral area, periphery of the face, trunk, and extremities [Figures 1 and 2]. Differentials thought of were Vitamin B12 deficiency, Addison's disease, congenital adrenal hyperplasia (CAH), and chikungunya.

Routine investigations showed leucocytosis. In view of hyperpigmentation, Vitamin B12 deficiency and CAH were ruled out because Vitamin B12 and serum cortisol were normal. In view of fever, lethargy, and leucocytosis, cerebrospinal fluid (CSF) analysis was performed to rule out meningitis. CSF analysis was normal, with no evidence of meningitis. To rule out chikungunya-induced hyperpigmentation, CSF serology was performed, which tested positive

Figure 1: Two-month-old male, with chikungunya, with brown black macules and patches over nose, perioral area, forehead, and sides of the face.

Figure 2: Patchy brownish-black hyperpigmentation over the trunk and extremities.

for chikungunya immunoglobulin (IgM). The mother also tested positive for chikungunya IgM; however, she had no antenatal or postnatal history suggestive of chikungunya. This suggests that both the child and mother acquired the infection postnatally through vectors.

The fever and dehydration were attributed to be secondary to sepsis, and the diffuse type of pigmentation was due to chikungunya.

The child was managed conservatively with IV fluids and analgesics, with gradual resolution of hyperpigmentation spontaneously.

Chikungunya is a re-emerging infection caused by Arbovirus, transmitted by *Aedes aegypti* and *Aedes albopictus* mosquito. Other rare modes of transmission include blood-borne and vertical transmission in the first or second trimester of pregnancy. A higher risk of vertical transmission occurs when there is a maternal infection within 1 week of delivery, with greater chances of severe disease in the neonate.[1] Pigmentary alterations reported in chikungunya include localised, grey-black, macular hyperpigmentation over the ala of the nose known as chik sign, centrofacial freckle-like/melasma-like macules/patches, flagellate hyperpigmentation, diffuse hyperpigmentation of the face and extremities, mucosal hypermelanosis of the tongue and palate, periorbital hypermelanosis, Addisonian-type palmar pigmentation, and melanonychia.[2]

Generalised pigmentation is more commonly seen in infants compared to centrofacial and neck pigmentation seen in adults. Children more commonly have neurological and dermatological symptoms compared to infected adults.[1]

Pigmentary changes have been attributed to pigment dispersion by the chikungunya virus and post-inflammatory response to the virus.[3]

Other cutaneous manifestations of chikungunya include maculopapular eruption, aphthae like orogenital ulcers, palmar erythema, vesiculobullous lesions, haemorrhagic manifestations, exacerbating of existing dermatosis, telogen effluvium, erythema nodosum, erythema multiforme-like lesions, lichenoid lesions, photosensitivity, lobular panniculitis, lymphoedema, and urticaria.[2]

Chikungunya should be included in the differential diagnosis when evaluating patients presenting with fever and pigmentary changes, particularly in endemic countries. Clinicians should also recognise the diverse cutaneous manifestations associated with chikungunya.

Ethical approval

Institutional review board approval is not required.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent.

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Nil.

Conflicts of interest

There are no conflicts of interest.

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