ORIGINAL RESEARCH PAPER
Volume - 13 | Issue - 11 | November - 2024 | PRINT ISSN No. 2277 - 8179 | DOI : 10.36106/ijsr
INTERNATIONAL JOURNAL OF SCIENTIFIC RESEARCH

NUTRITIONAL DEFICIENCY AS A CAUSE OF ACUTE SEVERE SUPRA-SYSTEMIC PULMONARY HYPERTENSION IN AN INFANT: A CASE REPORT OF REVERSIBLE CRISIS

Cardiology

Uma Devi Karuru*	ESIC Medical College and Super Speciality Hospital, Assistant Professor, Department of Cardiology. *Corresponding Author
Ashirbad Parhi	ESIC Medical College and Super Speciality Hospital, Assistant Professor, Department of Cardiology.
Naveen T	ESIC Medical College and Super Speciality Hospital, Assistant Professor, Department of Cardiology.
Sai Kumar Mysore	ESIC Medical College and Super Speciality Hospital, Assistant Professor, Department of Cardiology.
Sadanand Reddy Tummala	ESIC Medical College and Super Speciality Hospital, Assistant Professor, Department of Cardiology.
Kiran Kumar Kanjerla	ESIC Medical College and Super Speciality Hospital, Assistant Professor, Department of Cardiology.

ABSTRACT

Acute supra-systemic pulmonary hypertension (PH) in infants is a rare but life-threatening condition that can lead to right heart failure if untreated. This case report describes a 2-month-old male infant admitted with severe respiratory distress, tachycardia, and tachypnea. Born at term and exclusively breastfed, he exhibited cardiomegaly and significant PH, indicated by echocardiography. Investigation revealed the mother's inadequate diet, consisting mainly of polished white rice, resulting in thiamine (Vitamin B1) deficiency in both mother and infant. The infant received thiamine supplementation and sildenafil, leading to a rapid reduction in pulmonary pressures within a week and complete recovery by the six-month follow-up. Thiamine deficiency, while uncommon, can cause severe cardiovascular complications, especially in populations with poor maternal nutrition. As thiamine is essential for carbohydrate metabolism and myocardial energy production, its deficiency can deplete myocardial energy and precipitate high-output heart failure and severe PH. This case, alongside reports of other infants with PH and right heart failure linked to thiamine deficiency, underscores the need for clinicians to consider nutritional deficiencies as reversible causes of infantile PH. Early diagnosis and intervention are crucial in managing infants with severe pulmonary hypertension, particularly in low-income families facing nutritional inadequacies.

KEYWORDS

Nutritional deficiency, Pulmonary hypertension, Infant, Reversible crisis

BACKGROUND:

Acute Supra-Systemic pulmonary hypertension (PH) in infants is a rare, yet life-threatening condition characterized by pulmonary arterial pressures exceeding systemic pressures. This elevation in pulmonary pressure induces significant right heart strain, which can culminate in right heart failure if left untreated. Rapid identification and intervention are critical to avoid irreversible cardiovascular damage. The etiology of pulmonary hypertension in infancy can be multifactorial, stemming from congenital heart defects, persistent pulmonary hypertension of the new born (PPHN), and acquired conditions, such as infections or nutritional deficiencies. Among these causes, thiamine (Vitamin B1) deficiency although uncommon, can manifest with cardiovascular complications, especially in settings where maternal and infant nutritional intake is inadequate (1).

This report presents a case of acute Supra-Systemic pulmonary hypertension in an infant who exhibited a striking response to thiamine supplementation in conjunction with pulmonary vasodilator therapy. The case underscores the importance of considering nutritional deficiencies, such as thiamine deficiency, in the differential diagnosis of pulmonary hypertension in infants.

Case Presentation:

A 2-month-old male infant presented to the emergency department with severe respiratory distress, minimal peripheral cyanosis though without abdominal distension, loss of consciousness, or seizures. The infant was delivered at term via vaginal delivery, with a birth weight of 2.5 kg, to non-consanguineous parents belonging to lower socioeconomic status. The neonatal period was uneventful, with no history of perinatal asphyxia. The baby was on exclusive breast feeding.

On clinical examination, the infant exhibited tachycardia with a heart rate of 170 beats per minute, tachypnoea with a respiratory rate of 70 breaths per minute, blood pressure of 80/50 mm Hg and oxygen saturation of 80%, accompanied by minimal peripheral cyanosis. The infant was promptly started on nasal oxygen at 2 litres per minute. Initial laboratory investigations revealed normal haemoglobin levels and unremarkable renal and liver function tests. Viral markers, tuberculosis, bacterial and fungal cultures, and screening for primary and secondary immunodeficiencies returned negative results. A chest radiograph revealed cardiomegaly, with a cardiothoracic ratio of 70% and evidence of right ventricular (RV) enlargement. Electrocardiography (ECG) indicated normal sinus rhythm, with signs of right atrial (RA) enlargement and right ventricular (RV) hypertrophy.

Echocardiography showed gross dilation of the RA, RV (Video 01, https://youtu.be/f62bEm1t9y8), main pulmonary artery, alongside moderate tricuspid regurgitation and significant PH, with an estimated right ventricular systolic pressure (RVSP) of 144 mm Hg + right atrial pressure (RAP) (Figure 01).

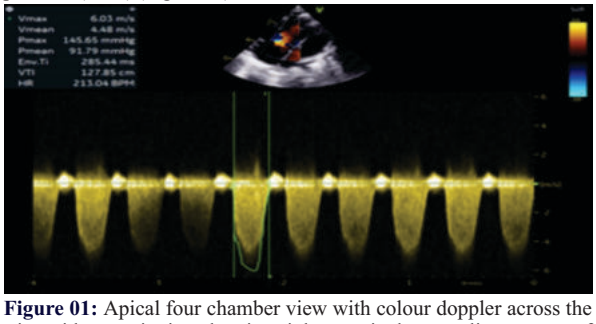

Figure 01: Apical four chamber view with colour doppler across the tricuspid regurgitation showing right ventricular systolic pressure of 144 mm Hg + right atrial pressure.

No intracardiac shunt was observed. Computed tomography (CT) of the chest ruled out pulmonary arteriovenous malformations, pulmonary veno-occlusive disease, pulmonary alveolar proteinosis, pulmonary hemosiderosis and extracardiac shunts.

Further history revealed that the infant's mother had been adhering to food taboos, consuming an almost exclusive diet of polished, washed white rice during the postnatal period without appropriate nutritional supplementation and baby was exclusively on breast feeding. Given the clinical context, thiamine deficiency was suspected, and subsequent tests confirmed low thiamine levels in both mother and infant. Both mother and infant were started on thiamine supplementation. After initiating thiamine supplementation and pulmonary vasodilator therapy with sildenafil at 1 mg/kg/day, the infant demonstrated a significant reduction in pulmonary pressures, with the RVSP decreasing to 20 mm Hg +RAP (Figure 02) within a span of 7days with drastic improvement in respiratory distress.

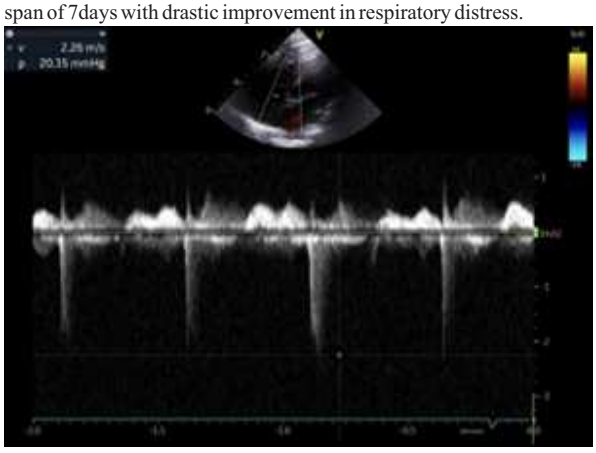

Figure 02: Apical four chamber view with colour doppler across the tricuspid regurgitation showing right ventricular systolic pressure of 20 mm Hg + right atrial pressure

At the 6-month follow-up, the infant's pulmonary pressures and right heart chambers had normalized (Video 02, https://youtu.be/fB5W2IUv8Qg), and there was marked clinical improvement, with adequate weight gain and normal developmental milestones.

DISCUSSION:

Thiamine, a water-soluble vitamin, has a short half-life and requires continuous dietary intake since the human body has limited storage capacity and cannot synthesize it endogenously. In the absence of adequate intake, thiamine levels can deplete within two weeks, especially in individuals with high metabolic demands, such as infants. Thiamine deficiency can lead to severe cardiovascular and neurological complications, particularly in cases of pulmonary hypertension. This is due to thiamine's essential role in myocardial energy metabolism and the regulation of vascular resistance.

Diagnosing thiamine deficiency can be challenging because serum thiamine levels often do not reflect the body's functional thiamine status. Functional thiamine deficiency is typically suggested by low erythrocyte transketolase activity (ETKA) and an elevated thiamine pyrophosphate effect (TPPE), though these tests are expensive and not widely accessible (2-4). A confirmed diagnosis can be made if there is a 25% increase in ETKA activity after thiamine supplementation. However, in many clinical settings, empirical treatment with thiamine is both diagnostic and therapeutic, as evidenced by the rapid resolution of symptoms (1,5) and normalization of pulmonary pressures in this infant.

Bhat et al.(5) conducted a study involving 29 infants with a mean postnatal age of 78.45 days, all of whom presented with acute onset PH. These infants, exclusively breastfed and from lower and middle socioeconomic backgrounds, exhibited signs of right heart failure and acute metabolic acidosis. Similarly, a case series by Narsimha et al. reported 55 infants with an average age of 3.9 months, all of whom exhibited high-output cardiac failure and pulmonary hypertension. These infants, like their mothers, were thiamine-deficient, as evidenced by low ETKA levels, and showed rapid improvement with thiamine supplementation. Among these infants, 19 were followed for 2–3 weeks, and both clinical and echocardiographic improvements were observed.

Another study by Uppalapati T et al. (6) focused on 10 cases of infants aged 1-5 months, with 90% of cases occurring after 2 months. Additionally, a case series by Panigrahy et al. (7) highlighted that the most common age for infantile thiamine deficiency-related symptoms is between 1 and 7 months. Mothers who primarily consume polished white rice that has been repeatedly washed are at the highest risk of thiamine deficiency. The infants most vulnerable to developing thiamine deficiency are those aged 1-3 months who are exclusively breastfed.

In comparison with other documented cases, none demonstrated such an acute exacerbation of pulmonary pressures as seen in this case. This case emphasizes the critical importance of early recognition of thiamine deficiency as a reversible cause of PH. Untreated thiamine deficiency can lead to severe cardiovascular morbidity, including wet beriberi, which is characterized by high-output cardiac failure and elevated pulmonary vascular resistance. The rapid clinical and hemodynamic improvements following thiamine supplementation in this case confirm the diagnosis.

In addition to thiamine supplementation, pulmonary vasodilator therapy, specifically sildenafil, played a crucial role in stabilizing the infant's condition during the acute phase by significantly reducing pulmonary pressures. The combination of thiamine supplementation and sildenafil resulted in sustained clinical and hemodynamic improvements, as evidenced by the normalization of pulmonary pressures at the 6-month follow-up and the infant's overall recovery.

This case underscores the need for clinicians to remain vigilant for reversible causes of pulmonary hypertension, such as nutritional deficiencies, especially in populations with limited access to adequate maternal and infant nutrition. While rare, thiamine deficiency can cause life-threatening pulmonary hypertension, but it is highly treatable if recognized early. The successful outcome in this case highlights the importance of timely intervention with thiamine supplementation and pulmonary vasodilator therapy.

CONCLUSION:

Pulmonary hypertension in infants is frequently attributed to congenital heart or pulmonary anomalies. However, clinicians must remain vigilant for reversible causes such as nutritional deficiencies, particularly in populations with inadequate maternal nutrition. Thiamine deficiency, although uncommon, can precipitate life-threatening PH but is highly treatable if recognized early. This case underscores the critical importance of timely diagnosis and intervention, highlighting the synergistic role of thiamine supplementation and pulmonary vasodilator therapy in managing acute suprasystemic pulmonary hypertension.

REFERENCES:

1. Qureshi UA, Sami A, Altaf U, et al.: Thiamine responsive acute life threatening metabolic acidosis in exclusively breast-fed infants. Nutrition. 2016, 32:213-216. . https://doi.org/10.1016/j.nut.2015.08.007
2. Whitfield KC, Bourassa MW, Adamolekun B, et al.: Thiamine deficiency disorders: diagnosis, prevalence, and a roadmap for global control programs. Ann N Y Acad Sci. 2018:3-43. . https://doi.org/10.1111/nyas.13919
3. Khounnorath S, Chamberlain K, Taylor AM, et al.: Clinically unapparent infantile thiamin deficiency in Vientiane, Laos. PLoS Negl Trop Dis. 2011, 5:969. . https://doi.org/10.1371/journal.pntd.0000969
4. Bhat JI, Rather HA, Ahangar AA, et al.: Shoshin beriberi-thiamine responsive pulmonary hypertension in exclusively breastfed infants: A study from northern India. Indian Hear J. 2017, 69:24-7. . https://doi.org/10.1016/j.ihj.2016.07.015
5. Uppalapati T, Krishna YS, Eregowda A: Clinical spectrum of infants with thiamine responsive pulmonary arterial hypertension. Int J Contemp Pediatr. 2023, 10:380-6. . https://doi.org/10.18203/2349-3291.ijcp20230441
6. Panigrahy N, Chirla DK, Shetty R, et al.: Thiamine-Responsive Acute Pulmonary Hypertension of Early Infancy (TRAPHEI)—A Case Series and Clinical Review. Children. 2020. https://doi.org/10.3390/children7110199.