

## Research Letter

### *Cardiac Beriberi: Often a Missed Diagnosis*

#### Summary

**Thiamine deficiency leads to various manifestations due to dysfunction of nervous or cardiovascular system, commonly known as dry and wet beriberi, respectively. The latter, also known as cardiac beriberi is usually missed in clinical practice because of the absence of classically described symptoms such as pedal edema/anasarca. We investigated 55 such infants and prospectively followed their clinical course. All the babies were exclusively breast-fed and their mothers belonged to low socio-economic status with their staple diet consisting of non-parboiled polished rice. Majority presented with tachypnea, chest indrawing and tachycardia and cardiomegaly with dilatation of right heart and pulmonary hypertension on 2D-echocardiography. Low levels of erythrocyte transketolase activity suggested thiamine deficiency that was confirmed by reversion of several clinical features including cardiologic abnormalities to normalcy on thiamine supplementation. We recommend thiamine therapy for infants with unexplained congestive cardiac failure or acute respiratory failure from precarious socio-economic background since it is life-saving in many instances.**

**Key words: thiamine deficiency, nutrition-infant, cardiac beriberi, right heart failure, public health.**

Thiamine is a water-soluble vitamin that functions as a co-enzyme in several key reactions in carbohydrate metabolism [1]. Its deficiency in young infants predisposes to various manifestations, commonly known as dry and wet beriberi [2, 3]. The latter is also known as cardiac beriberi and often goes unrecognized since it is easily confused with other illnesses [2]. Thiamine deficiency is reported to have been eliminated in India through active advocacy of consumption of under-milled rice [4]. Recently, we

cared for several infants at Niloufer Hospitals for Children in Hyderabad, India, who presented with features of acute lower respiratory tract infection, primary pulmonary hypertension with congestive cardiac failure (CCF) and had remarkable recovery on thiamine administration. We prospectively investigated 55 such infants to evaluate their clinical profile, thiamine status and therapeutic efficacy of thiamine supplementation.

All enrolled babies were exclusively breast-fed with an appropriate weight for their age. Mean age of presentation was 3.9 months and mean duration of illness was 7.5 days. All of them presented with tachypnea, chest indrawing and tachycardia. Other major manifestations included hepatomegaly ( $n = 44$ ; 80%), cough ( $n = 42$ ; 76.3%) and fever ( $n = 29$ ; 52.7%), while aphonia ( $n = 10$ ; 18.2%), external ophthalmoplegia with seizures and altered consciousness ( $n = 4$ , 7.3%) were present in few (Table 1). None of them had anemia or congenital heart disease. On examination, all the infants had rapid high-volume pulse, normal heart sounds and cardiomegaly with prominent pulmonary conus. This indicates high-output cardiac failure with pulmonary hypertension but none had anasarca or pedal edema [1, 5]. The 2D-echocardiography revealed right atrial and ventricular dilatation with tricuspid regurgitation, pulmonary hypertension and good myocardial contractility [5], while ECG changes were non-specific and non-contributory. Computed tomography scan of the brain revealed bilateral hypodensity of the basal ganglia in four cases with associated encephalitic beriberi [4]. All the babies and their mothers had low levels of erythrocyte transketolase activity (ETKA), suggesting a deficient thiamine status. The infants were administered 75 mg of intramuscular thiamine twice a day for 5 days in addition to routine supportive care. Diagnosis of cardiac beriberi was confirmed by rapid therapeutic response to thiamine administration [2, 3] and reversal of echocardiographic abnormalities in 19 babies, who reported for follow-up after 2–3 weeks.

We attempted to explore the cause of thiamine deficiency in these babies and their mothers. Mothers of these infants, who belonged to low socio-economic status were undernourished and calorie deficient. Their staple diet consisted of non-parboiled polished rice from which soup was discarded after cooking, and they hardly consumed meat, legumes and vegetables that are rich sources of thiamine [3, 4]. More than half of the probands had cough and fever, which is known to increase the demand for thiamine, thus precipitating the clinical presentation [2, 3]. However, despite having low ETKA levels, the mothers were asymptomatic since clinical

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TABLE 1  
Summary of clinical features in 55 infants recruited in the study

Clinical features	N (%)
Tachypnea	55 (100)
Chest indrawing	55 (100)
Tachycardia	55 (100)
Hepatomegaly	44 (80)
Cough	42 (76.3)
Fever (100–102°F)	29 (52.7)
Aphonia	10 (18.2)
Pulmonary crepitations	6 (10.9)
Bilateral ptosis, seizures and hypotonia	4 (7.3)

presentation of thiamine deficiency is known to depend on the ingestion of calories, especially carbohydrates [3].

Thus, we conclude that all the babies had thiamine deficiency and presented with features of cardiac beriberi with high-output cardiac failure and pulmonary hypertension. These observations emphasize that thiamine deficiency in India is far from controlled, further substantiated by our recent study which reported large number of infants with a diagnosis of infantile encephalitic beriberi having overlapping features of Leigh's disease [4]. The diagnosis is quite often missed because of the lack of awareness and non-availability of a confirmatory test, which is technically demanding and expensive [2]. Hence, we propose thiamine supplementation as a potential lifesaving therapeutic option in infants from low socio-economic background presenting with

unexplained refractory CCF or acute respiratory failure.

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