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A 2-Month-Old Girl from Laos With Dyspnoea, Cyanosis and Irritability

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Clinical Presentation

History

You are working in the paediatric intensive care unit (PICU) of a tertiary hospital in Vientiane, Laos. A 2-month-old baby girl is presented with 3 days of irritability, dyspnoea and grunting. Her mother is a 24-year-old rice farmer who describes that the baby suddenly became unwell but was neither feverish nor coughing. The infant was born at term and had been very well until 3 days previously.

Clinical Findings

Irritable infant, crying and grunting, with a temperature of 37.0°C (98.6°F), pulse 140 bpm (normal range 100–160), respiratory rate 40 breath cycles per minute (normal range 30–60). The blood pressure is not taken. The child is dyspnoeic and has central cyanosis, hepatomegaly and oedematous extremities. The rest of the physical examination appears normal, with a clear chest and no heart murmurs.

Questions

1. What are your most important differential diagnoses?
2. What additional information do you need to obtain from the mother and what would be your immediate management?

Discussion

A young Lao mother presents with her 2-month-old baby girl, who has been acutely unwell for the past 3 days. The child has been breastfeeding poorly and was noted to have been grunting, which is a non-specific sign of severe systemic illness in infants. On examination, the child is irritable, cyanosed and shows signs of heart failure.

Answer to Question 1

What Are Your Most Important Differential Diagnoses?

The most important differential diagnoses to consider are congenital heart disease, respiratory diseases (e.g. bronchopneumonia, bronchiolitis and laryngitis), meningitis and infantile beriberi (thiamine or vitamin B_1 deficiency).

However, the absence of cough, wheeze and fever, along with a normal chest examination, suggest that respiratory diseases are unlikely. The fact that the cyanosis has started well after birth along with the sudden onset of symptoms and the absence of a heart murmur suggests that congenital heart disease is unlikely. Absence of fever or bulging fontanels makes meningitis unlikely. The combination of dyspnoea, poor breastfeeding, abnormal cry, grunting and swollen extremities in a suitable endemic setting (South-east Asia) suggest infantile beriberi as the most likely diagnosis.

Answer to Question 2

What Additional Information Do You Need to Obtain from the Mother and What Would be Your Immediate Management?

The most important information to be obtained from the mother is whether the child has been exclusively breastfed, whether the mother had practiced food avoidance during pregnancy and/or post-partum, and whether the mother herself has had any symptoms and signs suggestive of beriberi, such as paresthesias and difficulty rising from a squatting position.

Prolonged food avoidance post-partum is common in lowland Lao culture. It is based upon the traditional belief that certain food items may harm the newborn. Most lowland mothers eat milled glutinous rice but avoid eating fruits and vegetables, which results in low diet diversity for some months before and after delivery.

Thiamine deficiency may be one result of such dietary restriction, and affected mothers secrete insufficient levels of thiamine in their breastmilk. The cardiac form of thiamine deficiency usually manifests during the second or third month of life. Infants present with dyspnoea, cyanosis, vomiting and irritability.

Children with infantile beriberi respond rapidly (i.e. within 30–60 min) to intravenous or intramuscular thiamine (50 mg), which should urgently be administered. The child should be closely monitored.

The Case Continued…

The mother reported that her child had been exclusively breastfed and that she had practiced food avoidance since delivery: She had avoided eating beef, pork, vegetables and fruits. The mother said that she herself had anorexia, weakness, a husky voice, and paresthesias affecting her limbs – symptoms of thiamine deficiency.

Intravenous thiamine (50 mg) was given immediately to the child after admission and she quickly and dramatically responded. Six hours later the baby was able to breastfeed normally and was discharged the following day. Blood samples were taken from the child and the mother. Analysis showed that both were thiamine-deficient.

The mother was given oral thiamine supplementation and advised to return to a well-balanced diet including thiamine-rich foods.

SUMMARY BOX

Infantile Beriberi

Infantile beriberi, or clinical thiamine (vitamin B1) deficiency in infants, is a largely forgotten, fatal but inexpensively treated disease. It mainly occurs in South and South-east Asia, where 50 to 100 years ago it was recognized as a major public health problem. It remains relatively common in Laos, probably because of prolonged maternal food avoidance during pregnancy and postpartum. There is also evidence that it is still of importance in Cambodia, India and Myanmar, and cases have been reported from refugee populations in Thailand.

Infantile beriberi occurs in exclusively breastfed babies of approximately 2 to 3 months of age, whose mothers have thiamine deficiency resulting from inadequate thiamine intake.

It commonly manifests mainly as the "wet" form of beriberi, characterized by heart failure with hepatomegaly and marked peripheral oedema. The disease typically presents as shock, often preceded by a hoarse cry, grunting, poor breastfeeding, irritability, dyspnoea and cyanosis. Clinically unapparent thiamine deficiency was also found to be common among sick infants admitted to the hospitals in Laos.

A thiamine-deficient diet is largely made up of milled, sticky rice that has had most of its thiamine removed as a result of the milling process. In the late nineteenth century, the advent of mechanical rice milling, which removes the main dietary source of thiamine in the rice husk, is thought to be the precipitant for beriberi becoming a major public health problem in Asia, responsible for considerable mortality. However, there has been very little recent epidemiological research, despite evidence that it remains focally important.

The pathophysiology of infantile beriberi remains unclear, but the cardinal problem is usually myocardial dysfunction. Thiamine deficiency may also present with a variety of other clinical syndromes, including encephalopathy, hypoglycemia and lactic acidosis.

Infantile beriberi is usually diagnosed clinically because of the need to act before biochemical results are available and the lack of laboratories in endemic areas that can perform thiamine biochemical assays. Treatment of infantile beriberi with thiamine is simple, inexpensive and highly effective. It should be administered parenterally to rapidly increase tissue thiamine levels. Thiamine supplementation should also be given to the mothers of infants with beriberi. Education about thiamine-rich foods (e.g. pulses, groundnuts, whole wheat, fruits and vegetables) and on the danger of food avoidance should be provided before discharge. Prevention is crucial: better understanding of post-partum diets, and on how to change the practice of food avoidance is very important and thiamine supplementation for mothers is urgently needed.

Further Reading

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