Thiamine Deficiency: New Fads Old Problems
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Thiamine is a water-soluble vitamin involved in many cellular metabolic activities which result in the production of ATP.

The primary source of thiamine is from diet including whole grains, pork and legumes.

Thiamine stores can be depleted in as little as 18 days resulting in inadequate ATP production that disrupts organ systems with high metabolic demands such as the cardiovascular and nervous systems.

This depletion leads to the clinical presentations of wet and/or dry beriberi which rapidly progress without thiamine replacement.

In extreme cases, beriberi is associated with Wernicke-Korsakoff syndrome which damages the thalamic and hypothalamic regions of the brain leading to confusion and visual problems.

Thiamine levels were tested in all three patients due to clinical symptoms and history.

All patients presented with a history of almond milk consumption as the primary source of milk.

All patients presented with Thiamine levels < 7 nmol/L (norm 74-222).

Patient One: 15-month old male who presented with encephalopathy and seizures

Patient Two: 22-month old male who presented with seizure activity

Patient Three: 13-month old female who presented with acute respiratory failure

Biochemically, the measurement of erythrocyte transketolase activity (ETKA) and the thiamine pyrophosphate effect (TPPE), provide a sensitive thiamine deficiency test.

Under recognition of thiamine deficiency can lead to severe neurological manifestations.

Once thiamine replaced, full recovery is possible.

References:

Figure 1. Patient One: Head MRI revealed a symmetric abnormal signal with restricted diffusion within the putamen and caudate nucleus with increased T2 signal in the bilateral thalami and periaqueductal white matter.
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Abstract:

Thiamine is a water-soluble B1 vitamin involved in many cellular metabolic activities which result in the production of ATP. The primary source of thiamine is from the diet including whole grains, pork and legumes. Thiamine deficiency is uncommon in the United States and most western civilized cultures due to a high prevalence of thiamine-containing foods. Dietary stores can be depleted in as little as 18 days if not replaced, resulting in inadequate ATP production that significantly disrupts organ systems with high metabolic demands such as the cardiovascular and nervous systems, leading to the clinical presentations of wet and dry beriberi. In contrast, thiamine deficiency is more prevalent in areas of the world where the diet is comprised of polished rice products, vegetables and seafood which are poor sources of thiamine.

A case series is presented with a 15-month old male, 22-month old male, and 13-month old female. Each patient presented with severe metabolic or respiratory compromise. All patients presented with a history of almond milk consumption as the primary source of milk. Two presented with seizure activity while the third presented in respiratory failure. Thiamine levels were tested in all three patients due to clinical symptomology and history. Each patient saw a decreased thiamine level at <7 nmol/L (normal: 74-222 nmol/L). Thiamine administration was administered to two of the three patients with rapid reversal of symptoms. The third patient recovered prior to thiamine administration however the pediatrician was notified of the results.

Infantile thiamine deficiency exists in 3 forms: cardiologic, aphonic or pseudo-meningitic. Testing for this disease should be considered when there is a history of low thiamine diet or clinical manifestations. Recommended biochemical diagnosis include thiamine level (blood or urine), erythrocyte transketolase activity (ETKA) and thiamine pyrophosphate effect (TPPE). Due to the complex clinical presentation of thiamine deficiency and decreased prevalence in the developed world, this life-threatening disorder may be under recognized and easily missed. Once thiamine is replaced, symptoms rapidly improve, often leading to a full recovery. On the contrary, if thiamine deficiency is not recognized, patients can have severe neurologic damage while cardiac manifestations can lead to death.

The presentation of these patients reminds us that classical vitamin deficiencies can always make a comeback when food fads change rapidly even in developed economies. It is important for pediatric providers to always explore diet history during well-checks in order to anticipate adverse events. Finally, we wonder if thiamine deficiency can make common viral illnesses more intense resulting in PICU admissions.

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