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Pediatric Beriberi Clinical Presentation

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Author: Simon S Rabinowitz, MD, PhD, FAAP; Chief Editor: Jatinder Bhatia, MBBS, FAAP more...

PRESENTATION

History

Thiamine deficiency has a wide range of clinical presentations. Although clinical overlap is common, the basic phenotypes are as follows:

- Wet beriberi
  - This phenotype affects the cardiovascular system and is divided into acute and chronic forms.
  - In acute wet beriberi or Shoshin beriberi, the predominant injury is to the heart, and rapid deterioration occurs because of the heart's inability to maintain function.
  - Presenting symptoms include tachycardia, low diastolic pressure, cardiomegaly, pulmonary edema, and cyanosis. Wet beriberi is characterized by elevated lactic acid levels. This condition can be readily reversed with thiamine infusion, if administered early.
  - Chronic wet beriberi with high-output cardiac failure has 3 stages.
    - Initially, peripheral vasodilatation occurs, yielding high-output cardiac failure.
    - Then, the progression of vasodilatation is perceived by the kidney as a relative loss of volume. The ensuing activation of the renin angiotensin system produces greater salt and water retention.
    - Consequently, further fluid overload results in peripheral edema and pulmonary effusions.
  - Cardiac overuse injury that occurs in the above setting results in tachycardia, hypertension, and chest pain.
  - Thyrotoxicosis, a more common cause of high-output cardiac failure, is always among the differential diagnoses.
- Dry beriberi
  - This phenotype affects the neuromuscular system.
  - Polyneuritis and symmetric, ascending paralysis of the peripheral nerve systems predominate.
  - The sensory system is affected first, followed by the motor and autonomic systems.
  - Typically, tactile sensation is the first to be lost, followed by pain, and, finally, temperature.
  - Paresthesia and hyperesthesia usually begin with the lower extremities and gradually involve the upper extremities and perioral area.
  - Deep tendon reflexes are lost, calf muscles become painful, and foot drop and, eventually, wrist drop occur.

- If untreated, progressive weakness, wasting of muscles, and, ultimately, complete paralysis occur.
- Encephalopathy is an alternative mode of presentation, with vomiting, disorientation, horizontal nystagmus, palsies of the eye movements (ophthalmoplegia), ataxia, and progressive mental impairment.
- Korsakoff syndrome is a more ominous condition that usually precludes complete recovery. Confusion is followed by the loss of recent memory and confabulation, which is the creation of accounts of events to cover up the loss of memory.

- Infantile thiamine deficiency
  - This occurs in various forms and typically affects breastfed infants whose mothers had beriberi.
  - Early on, the infant is constipated, crying, restless, and has emesis.
  - Three forms are recognized:
    - The pure cardiologic or pernicious form is common in infants aged 1-3 months. They present with cyanosis and features of acute cardiac failure. Infants usually die within 2-4 hours, but this type of deficiency responds very rapidly to thiamine.
    - The aphonic form is seen in infants aged 4-6 months. This milder form causes loss of voice due to paralysis of the vocal cords.
    - The pseudomeningitic form is encountered in infants aged 7-9 months. It presents with clinical signs of meningitis, but cerebrospinal fluid findings exclude infection. Vomiting, sweating, and seizures may be present.
- Wernicke-Korsakoff syndrome
  - This is an autosomal recessive genetic disease seen most often in individuals of European descent.
  - Affected patients have transketolases that bind thiamine pyrophosphate 10 times less tightly than normal; thus, the serum levels required to yield maximum enzyme activity are higher. Patients with Wernicke-Korsakoff are thus symptomatic with much less severe thiamine depletion.
  - This syndrome occurs most often in individuals with alcoholism who are malnourished. It is often precipitated by administration of glucose because excessive carbohydrate metabolism exacerbates a subclinical thiamine deficiency.
  - Clinically, it combines features of both Wernicke encephalopathy and Korsakoff psychosis.
- Subclinical thiamine deficiency
  - This deficiency is seen in people with high carbohydrate intake and low thiamine intake.
  - Other at-risk groups include those with increased thiamine requirements because of raised physiological or metabolic demands. Clinical scenarios include pregnancy and lactation, heavy physical exertion, intercurrent illness (eg, cancer, liver diseases, infections, hyperthyroidism), and surgery.
  - This deficiency is rarely encountered in patients with increased losses, such as those seen with dialysis, chronic diuretic use, and malabsorption.
  - Symptoms are usually mild, with anorexia often the presenting symptom. Anorexia is regarded to be a protective phenomenon because continued intake of a high-carbohydrate diet could be detrimental.
  - Other early symptoms include weakness, aching, burning sensation in the hands and feet, indigestion, irritability, and depression. After 6-8 weeks, the only objective signs at rest may be a slight fall in blood pressure and moderate weight loss. After 2-3 months, apathy and weakness become extreme and calf muscle tenderness develops. Also, loss of recent memory, confusion, ataxia, and, sometimes, persistent vomiting occur.
  - Pediatric associations include growth restriction and sudden infant death syndrome (SIDS).

Physical

See the list below:

- Wet beriberi (cardiac)
  - Edema
  - Waxy skin
  - Vomiting
  - Widened pulse pressure
  - Systolic flow murmur
  - Gallop rhythm (third heart sound); best heard in left-lateral position during inspiration
  - Jugular venous distension
  - Tachycardia
  - Cardiomegaly
  - Pallor
  - ECG changes (prolonged QT, T-wave inversion, low voltage)
- Acute fulminant cardiovascular beriberi (Shoshin beriberi or occidental beriberi)
  - Breathlessness, cyanosis
  - Wet crackles in lower lung fields
  - Classic heart failure
- Dry beriberi (paralytic or nervous)
  - Pallor
  - Wasting
  - Listlessness
  - Tachycardia
  - Hepatomegaly
  - Symmetric peripheral neuropathy
  - Symmetric paresthesias, especially of the distal extremities, with diminished touch sensation
  - Weakness (starting with feet)
  - Loss of ankle and knee reflexes
  - Normal vibration and pain sensation
- Wernicke disease
  - Irritability, forgetfulness
  - Ataxia
  - Mental confusion
  - Nystagmus (horizontal more common than vertical)
  - Ptosis
  - Ophthalmoplegia
  - Delirium
  - Coma
- Korsakoff psychosis
  - Retrograde amnesia
  - Inability to learn
  - Confabulation
- Wernicke-Korsakoff syndrome - Combined features of both Wernicke disease and Korsakoff psychosis
- Infantile beriberi
  - Physical findings depend on the form but usually begin with nonspecific early findings, including the following:
    - Fatigue
    - Anorexia
    - Headaches
    - Irritability, fatigue
  - The physical findings in acute cardiac infantile beriberi are similar to those found in Shoshin beriberi, often with cyanosis.

- The physical findings in aphonic infantile beriberi include hoarseness that progresses to a complete loss of crying.
- The physical findings in pseudomeningeal infantile beriberi include nystagmus, muscle twitching, bulging fontanelle, convulsions and coma.

Causes

See the list below:

- Beriberi is caused by thiamine deficiency, which can be the result of different physiologic, dietary, and environmental factors.
- Frequently, more than one risk factor is present.

Differential Diagnoses

References

1. Aasheim ET. Wernicke encephalopathy after bariatric surgery: a systematic review. *Ann Surg*. 2008 Nov. 248(5):714-20. [Medline].

2. Institute of Medicine of the National Academies. *Dietary Reference Intakes for Thiamin, Riboflavin, Niacin, Vitamin B6, Folate, Vitamin B12, Pantothenic Acid, Biotin and Choline*. Washington DC: National Academy Press; 1998. [Full Text].

3. World Health Organization. Thiamine Deficiency and its prevention and control in major emergencies. 1999. Available at http://whqlibdoc.who.int/hq/1999/WHO_NHD_99.13.pdf.

4. McCandless DW, Schenker S, Cook M. Encephalopathy of thiamine deficieny: studies of intracerebral mechanisms. *J Clin Invest*. 1968 Oct. 47(10):2268-80. [Medline].

5. Martin PR, Singleton CK, Hiller-Sturmhofel S. The role of thiamine deficiency in alcoholic brain disease. *Alcohol Res Health*. 2003. 27(2):134-42. [Medline].

6. Greenspon J, Perrone EE, Alaish SM. Shoshin beriberi mimicking central line sepsis in a child with short bowel syndrome. *World J Pediatr*. 2010 Nov. 6(4):366-8. [Medline].

7. Muri RM, Von Overbeck J, Furrer J, Ballmer PE. Thiamin deficiency in HIV-positive patients: evaluation by erythrocyte transketolase activity and thiamin pyrophosphate effect. *Clin Nutr*. 1999 Dec. 18(6):375-8. [Medline].

8. Fattal-Valevski A, Kesler A, Sela BA, et al. Outbreak of life-threatening thiamine deficiency in infants in Israel caused by a defective soy-based formula. *Pediatrics*. 2005 Feb. 115(2):e233-8. [Medline].

9. Fattal-Valevski A, Azouri-Fattal I, Greenstein YJ, Guindy M, Blau A, Zelnik N. Delayed language development due to infantile thiamine deficiency. *Dev Med Child Neurol*. 2009 Aug. 51(8):629-34. [Medline].

10. Hanninen SA, Darling PB, Sole MJ, Barr A, Keith ME. The prevalence of thiamin deficiency in hospitalized patients with congestive heart failure. *J Am Coll Cardiol*. 2006 Jan 17. 47(2):354-61. [Medline].

11. Khounnorath S, Chamberlain K, Taylor AM, Soukaloun D, Mayxay M, Lee SJ, et al. Clinically unapparent infantile thiamin deficiency in Vientiane, Laos. *PLoS Negl Trop Dis*. 2011 Feb 22.

5(2):e969. [Medline]. [Full Text].

12. Djoenaidi W, Notermans SL, Verbeek AL. Subclinical beriberi polyneuropathy in the low income group: an investigation with special tools on possible patients with suspected complaints. *Eur J Clin Nutr*. 1996 Aug. 50(8):549-55. [Medline].

13. McGready R, Simpson JA, Cho T, et al. Postpartum thiamine deficiency in a Karen displaced population. *Am J Clin Nutr*. 2001 Dec. 74(6):808-13. [Medline].

14. Ahoua L, Etienne W, Fermon F, et al. Outbreak of beriberi in a prison in Cote d'Ivoire. *Food Nutr Bull*. 2007 Sep. 28(3):283-90. [Medline].

15. Fozi K, Azmi H, Kamariah H, Azwa MS. Prevalence of thiamine deficiency at a drug rehabilitation centre in Malaysia. *Med J Malaysia*. 2006 Dec. 61(5):519-25. [Medline].

16. Morovvati S, Nakagawa M, Sato Y, Hamada K, Higuchi I, Osame M. Phenotypes and mitochondrial DNA substitutions in families with A3243G mutation. *Acta Neurol Scand*. 2002 Aug. 106(2):104-8. [Medline].

17. Lira PI, Andrade SL. [Beriberi epidemic in Maranhão State, Brazil]. *Cad Saude Publica*. 2008 Jun. 24(6):1202-3. [Medline].

18. Rao SN, Mani S, Madap K, Kumar MV, Singh L, Chandak GR. High prevalence of infantile encephalitic beriberi with overlapping features of Leigh's disease. *J Trop Pediatr*. 2008 Oct. 54(5):328-32. [Medline].

19. Shivalkar B, Engelmann I, Carp L. Shoshin syndrome: two case reports representing opposite ends of the same disease spectrum. *Acta Cardiol*. 1998. 53(4):195-9. [Medline].

20. Lu J, Frank EL. Rapid HPLC measurement of thiamine and its phosphate esters in whole blood. *Clin Chem*. 2008 May. 54(5):901-6. [Medline].

21. Wrenn KD, Murphy F, Slovis CM. A toxicity study of parenteral thiamine hydrochloride. *Ann Emerg Med*. 1989 Aug. 18(8):867-70. [Medline].

22. Kitamura K, Yamaguchi T, Tanaka H, et al. TPN-induced fulminant beriberi: a report on our experience and a review of the literature. *Surg Today*. 1996. 26(10):769-76. [Medline].

23. Angstadt JD, Bodziner RA. Peripheral polyneuropathy from thiamine deficiency following laparoscopic Roux-en-Y gastric bypass. *Obes Surg*. 2005 Jun-Jul. 15(6):890-2. [Medline].

24. Centers for Disease Control and Prevention. Lactic acidosis traced to thiamine deficiency related to nationwide shortage of multivitamins for total parenteral nutrition--United States, 1997. *JAMA*. 1997 Jul 9. 278(2):109, 111. [Medline].

25. Indraccolo U, Gentile G, Pomili G, et al. Thiamine deficiency and beriberi features in a patient with hyperemesis gravidarum. *Nutrition*. 2005 Sep. 21(9):967-8. [Medline].

26. Kornreich L, Bron-Harlev E, Hoffmann C, et al. Thiamine deficiency in infants: MR findings in the brain. *AJNR Am J Neuroradiol*. 2005 Aug. 26(7):1668-74. [Medline].

27. Lehninger, Albert L. Vitamins and Coenzymes. *Biochemistry*. 2nd ed. New York, NY: Worth Publishers; 1977. 337-9.

28. Stryer, Lubert. Citric acid cycle. *Biochemistry*. 2nd ed. San Francisco, CA: Freeman; 1981. 290-5.

29. Suter PM, Vetter W. Diuretics and vitamin B1: are diuretics a risk factor for thiamin malnutrition?. *Nutr Rev*. 2000 Oct. 58(10):319-23. [Medline].

30. Weise Prinzo Z, de Benoist B. Meeting the challenges of micronutrient deficiencies in emergency- affected populations. *Proc Nutr Soc*. 2002 May. 61(2):251-7. [Medline].

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Contributor Information and Disclosures

Author

Simon S Rabinowitz, MD, PhD, FAAP Professor of Clinical Pediatrics, Vice Chairman, Clinical Practice Development, Pediatric Gastroenterology, Hepatology, and Nutrition, State University of New York Downstate College of Medicine, The Children's Hospital at Downstate

Simon S Rabinowitz, MD, PhD, FAAP is a member of the following medical societies: American Gastroenterological Association, American Academy of Pediatrics, Phi Beta Kappa, American Association for the Advancement of Science, American College of Gastroenterology, American Medical Association, New York Academy of Sciences, North American Society for Pediatric Gastroenterology, Hepatology and Nutrition, Sigma Xi

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Specialty Editor Board

Mary L Windle, PharmD Adjunct Associate Professor, University of Nebraska Medical Center College of Pharmacy; Editor-in-Chief, Medscape Drug Reference

Disclosure: Nothing to disclose.

Jatinder Bhatia, MBBS, FAAP Professor of Pediatrics, Medical College of Georgia, Georgia Regents University; Chief, Division of Neonatology, Director, Fellowship Program in Neonatal-Perinatal Medicine, Director, Transport/ECMO/Nutrition, Vice Chair, Clinical Research, Department of Pediatrics, Children's Hospital of Georgia

Jatinder Bhatia, MBBS, FAAP is a member of the following medical societies: Academy of Nutrition and Dietetics, American Academy of Pediatrics, American Association for the Advancement of Science, American Pediatric Society, American Society for Nutrition, American Society for Parenteral and Enteral Nutrition, Society for Pediatric Research, Southern Society for Pediatric Research

Disclosure: Serve(d) as a director, officer, partner, employee, advisor, consultant or trustee for: Nestle.

Chief Editor

Jatinder Bhatia, MBBS, FAAP Professor of Pediatrics, Medical College of Georgia, Georgia Regents University; Chief, Division of Neonatology, Director, Fellowship Program in Neonatal-Perinatal Medicine, Director, Transport/ECMO/Nutrition, Vice Chair, Clinical Research, Department of Pediatrics, Children's Hospital of Georgia

Jatinder Bhatia, MBBS, FAAP is a member of the following medical societies: Academy of Nutrition and Dietetics, American Academy of Pediatrics, American Association for the Advancement of Science, American Pediatric Society, American Society for Nutrition, American Society for Parenteral and Enteral Nutrition, Society for Pediatric Research, Southern Society for Pediatric Research

Disclosure: Serve(d) as a director, officer, partner, employee, advisor, consultant or trustee for: Nestle.

Additional Contributors

Maria Rebello Mascarenhas, MBBS Associate Professor of Pediatrics, University of Pennsylvania School of Medicine; Section Chief of Nutrition, Division of Gastroenterology and Nutrition, Director, Nutrition Support Service, Children's Hospital of Philadelphia

Maria Rebello Mascarenhas, MBBS is a member of the following medical societies: American Gastroenterological Association, American Society for Parenteral and Enteral Nutrition, North American Society for Pediatric Gastroenterology, Hepatology and Nutrition

Disclosure: Nothing to disclose.

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L Arturo Batres, MD Assistant Professor, Department of Pediatrics, Division of Gastroenterology and Nutrition, The Children's Hospital of the King's Daughters

L Arturo Batres, MD is a member of the following medical societies: American Academy of Pediatrics, American Association for the Study of Liver Diseases, American Gastroenterological Association, and North American Society for Pediatric Gastroenterology and Nutrition

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Sheela Moorthy, MD Staff Physician, Department of Pediatrics, Richmond University Medical Center

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