

REFERENCES

Homepage:

1. Auron A, Brophy PD. Hyperammonemia in review: Pathophysiology, diagnosis, and treatment. *Pediatr Nephrol* 2012;27:207-222.

Overview:

1. Haberle J. Clinical practice: The management of hyperammonemia. *Eur J Pediatr* 2011;170:21-34.

2. Orton DJ, Gifford JL, Seiden-Long I, Khan A, de Koning L. Critically high plasma ammonia in an adolescent girl. *Clin Chem* 2016;62(12):1565-1569.

3. Broomfield A, Grunewald S. How to use serum ammonia. *Arch Dis Child Educ Pract Ed* 2012;97:72-77.

4. Burton BK. Inborn errors of metabolism in infancy: A guide to diagnosis. *Pediatrics* 1998;102(6):E69.

5. Summar ML, Barr F, Dawling S, Smith W, Lee B, Singh RH, Rhead WJ, Sniderman King L, Christman BW. Unmasked adult-onset urea cycle disorders in the critical care setting. *Crit Care Clin* 2005;21:S1-S8.

6. Haberle J, Boddaert N, Burlina A, Chakrapani A, Dixon M, Huemer M, Karall D, Martinelli D, Sanjurjo Crespo P, Santer R, Servais A, Valayannopoulos V, Lindner M, Rubio V, Dionisi-Vici C. Suggested guidelines for the diagnosis and management of urea cycle disorders. *Orphanet J Rare Dis* 2012;7:32.

7. LaBuzetta JN, Yao JZ, Bourque DL, Zivin J. *Am J Med* 2010;123(10):885-891.

8. Cesar Machado MC, da Silva FP. Hyperammonemia due to urea cycle disorders: A potentially fatal condition in the intensive care setting. *J Intensive Care* 2014;2:22.

9. Smith W, Kishnani PS, Lee B, Singh RH, Rhead WJ, Sniderman King L, Smith M, Summar M. Urea cycle disorders: Clinical presentation outside the newborn period. *Crit Care Clin* 2005;21:S9-S17.

REFERENCES

10. Ah Mew N, Lanpher BC, Gropman A, Chapman KA, Simpson KL, Urea Cycle Disorders Consortium, Summar ML. Urea cycle disorders overview. In Pagon RA, Adam MP, Ardinger HH et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington, Seattle; 1993-2017. <http://www.ncbi.nlm.nih.gov/books/NBK1217>. Updated June 22, 2017. Accessed September 22, 2017.
11. N-acetylglutamate synthetase deficiency. National Organization for Rare Disorders. 2016. Available at: <https://rarediseases.org/rare-diseases/n-acetylglutamate-synthetase-deficiency>. Accessed September 11, 2017.
12. Panlaqui OM, Tran K, Johns A, McGill J, White H. Acute hyperammonemic encephalopathy in adult onset ornithine transcarbamylase deficiency. *Intensive Care Med* 2008;34:1922-1924.

Neonatal:

1. Haberle J, Boddaert N, Burlina A, Chakrapani A, Dixon M, Huemer M, Karall D, Martinelli D, Sanjurjo Crespo P, Santer R, Servais A, Valayannopoulos V, Lindner M, Rubio V, Dionisi-Vici C. Suggested guidelines for the diagnosis and management of urea cycle disorders. *Orphanet J Rare Dis* 2012;7:32.
2. Haberle J. Clinical practice: The management of hyperammonemia. *Eur J Pediatr* 2011;170:21-34.
3. Al Kaabi EH, El-Hattab AW. N-acetylglutamate synthase deficiency: Novel mutation associated with neonatal presentation and literature review of molecular and phenotypic spectra. *Mol Genet Metab Rep* 2016;8:94-98.
4. Ah Mew N, Caldovic L. N-acetylglutamate synthase deficiency: an insight into the genetics, epidemiology, pathophysiology, and treatment. *Appl Clin Gen* 2011;4:127-135.
5. Cartagena A, Prasad AN, Rupar CA, Strong M, Tuchman M, Ah Mew N, Prasad C. Recurrent encephalopathy: NAGS (N-acetylglutamate synthase) deficiency in adults. *Can J Neurol Sci* 2013;40:3-9.

REFERENCES

6. Ah Mew N, Lanpher BC, Gropman A, Chapman KA, Simpson KL, Urea Cycle Disorders Consortium, Summar ML. Urea cycle disorders overview. In Pagon RA, Adam MP, Ardinger HH et al, eds. GeneReviews® [Internet]. Seattle, WA: University of Washington, Seattle; 1993-2017. <http://www.ncbi.nlm.nih.gov/books/NBK1217>. Updated June 22, 2017. Accessed September 22, 2017.
7. Broomfield A, Grunewald S. How to use serum ammonia. Arch Dis Child Educ Pract Ed 2012;97:72-77.
8. Burton BK. Inborn errors of metabolism in infancy: A guide to diagnosis. Pediatrics 1998;102(6):E69.
9. Maggio PM. Sepsis and septic shock. Merck Manual, Professional Version, 2017. Revised April 2016. Available at: <http://www.merckmanuals.com/professional/critical-care-medicine/sepsis-and-septic-shock/sepsis-and-septic-shock#v928482>. Accessed October 2, 2017.
10. Zea-Vera A, Ochoa TJ. Challenges in the diagnosis and management of neonatal sepsis. J Trop Pediatr 2015;61:1-13.
11. Broomfield AA, Walter JH. Treatment of hyperammonemia in the newborn. A report. Willink Biochemical Genetics Unit, Royal Manchester Children's Hospital. Eur Paediatr 2008;36-39.

REFERENCES

Infants/Children/Adults:

1. Haberle J, Boddaert N, Burlina A, Chakrapani A, Dixon M, Huemer M, Karall D, Martinelli D, Sanjurjo Crespo P, Santer R, Servais A, Valayannopoulos V, Lindner M, Rubio V, Dionisi-Vici C. Suggested guidelines for the diagnosis and management of urea cycle disorders. *Orphanet J Rare Dis* 2012;7:32.
2. Haberle J. Clinical practice: The management of hyperammonemia. *Eur J Pediatr* 2011;170:21-34.
3. Batshaw ML, Tuchman M, Summar M, Seminara J. A longitudinal study of urea cycle disorders. *Mol Genet Metab* 2014;113:127-130.
4. Al Kaabi EH, El-Hattab AW. N-acetylglutamate synthase deficiency: Novel mutation associated with neonatal presentation and literature review of molecular and phenotypic spectra. *Mol Genet Metab Rep* 2016;8:94-98
5. Kolker S, Cazorla AG, Valayannopoulos V, Lund AM, Burlina AB, Sykut-Cegielska J, Wijburg W, Teles EL et al. The phenotypic spectrum of organic acidemias and urea cycle disorders. Part 1: the initial presentation. *J Inherit Metab Dis* 2015;38:1041-1057.
6. Ah Mew N, Caldovic L. N-acetylglutamate synthase deficiency: an insight into the genetics, epidemiology, pathophysiology, and treatment. *Appl Clin Gen* 2011;4:127-135.
7. Cartagena A, Prasad AN, Rupar CA, Strong M, Tuchman M, Ah Mew N, Prasad C. Recurrent encephalopathy: NAGS (N-acetylglutamate synthase) deficiency in adults. *Can J Neurol Sci* 2013;40:3-9.
8. Broomfield A, Grunewald S. How to use serum ammonia. *Arch Dis Child Educ Pract Ed* 2012;97:72-77.
9. Caldovic L, Morizono H, Tuchman M. Mutations and polymorphisms in the human N-acetylglutamate synthase (NAGS) gene. *Hum Mutat* 2007;28:754-759.
10. Summar ML, Tuchman M. Proceedings of a consensus conference for the management of patients with urea cycle disorders. *J Pediatr* 2001;138(1 Suppl):S6-S10.

REFERENCES

Infants/Children/Adults:

1. Haberle J, Boddaert N, Burlina A, Chakrapani A, Dixon M, Huemer M, Karall D, Martinelli D, Sanjurjo Crespo P, Santer R, Servais A, Valayannopoulos V, Lindner M, Rubio V, Dionisi-Vici C. Suggested guidelines for the diagnosis and management of urea cycle disorders. *Orphanet J Rare Dis* 2012;7:32.
2. Haberle J. Clinical practice: The management of hyperammonemia. *Eur J Pediatr* 2011;170:21-34.
3. Batshaw ML, Tuchman M, Summar M, Seminara J. A longitudinal study of urea cycle disorders. *Mol Genet Metab* 2014;113:127-130.
4. Al Kaabi EH, El-Hattab AW. N-acetylglutamate synthase deficiency: Novel mutation associated with neonatal presentation and literature review of molecular and phenotypic spectra. *Mol Genet Metab Rep* 2016;8:94-98
5. Kolker S, Cazorla AG, Valayannopoulos V, Lund AM, Burlina AB, Sykut-Cegielska J, Wijburg W, Teles EL et al. The phenotypic spectrum of organic acidemias and urea cycle disorders. Part 1: the initial presentation. *J Inherit Metab Dis* 2015;38:1041-1057.
6. Ah Mew N, Caldovic L. N-acetylglutamate synthase deficiency: an insight into the genetics, epidemiology, pathophysiology, and treatment. *Appl Clin Gen* 2011;4:127-135.
7. Cartagena A, Prasad AN, Rupar CA, Strong M, Tuchman M, Ah Mew N, Prasad C. Recurrent encephalopathy: NAGS (N-acetylglutamate synthase) deficiency in adults. *Can J Neurol Sci* 2013;40:3-9.
8. Broomfield A, Grunewald S. How to use serum ammonia. *Arch Dis Child Educ Pract Ed* 2012;97:72-77.
9. Caldovic L, Morizono H, Tuchman M. Mutations and polymorphisms in the human N-acetylglutamate synthase (NAGS) gene. *Hum Mutat* 2007;28:754-759.
10. Summar ML, Tuchman M. Proceedings of a consensus conference for the management of patients with urea cycle disorders. *J Pediatr* 2001;138(1 Suppl):S6-S10.

REFERENCES

11. Ah Mew N, Lanpher BC, Gropman A, Chapman KA, Simpson KL, Urea Cycle Disorders Consortium, Summar ML. Urea cycle disorders overview. In Pagon RA, Adam MP, Ardinger HH et al, eds. GeneReviews® [Internet]. Seattle,WA: University of Washington, Seattle; 1993-2017. <http://www.ncbi.nlm.nih.gov/books/NBK1217>. Updated June 22, 2017. Accessed September 22, 2017.
12. Roth K. Genetics of hyperammonemia. eMedicine/Medscape. Updated Mar 29, 2016. Available at: <http://emedicine.medscape.com/article/944996-overview>. Accessed September 7, 2017.
13. LaBuzetta JN, Yao JZ, Bourque DL, Zivin J. Adult nonhepatic hyperammonemia: A case report and differential diagnosis. *Am J Med* 2010;123(10):885-891. 13. Cesar Machado MC, da Silva FP. Hyperammonemia due to urea cycle disorders: A potentially fatal condition in the intensive care setting. *J Intensive Care* 2014;2:22.
14. Cesar Machado MC, da Silva FP. Hyperammonemia due to urea cycle disorders: A potentially fatal condition in the intensive care setting. *J Intensive Care* 2014;2:22.
15. Haberle J, Burlina A, Chakrapani A, Dixon M, Karall D, Lindner M, Mandel H, Martinelli D, Pintos-Morell G, Santer R, Skouma A, Servais A, Tal G, Rubio V, Huemer M, Dionisi-Vici C. Suggested guidelines for the diagnosis and management of urea cycle disorders: First revision. *J Inher Metab Dis*. 2019;1-39.

REFERENCES

Testing:

1. Haberle J. Clinical practice: The management of hyperammonemia. *Eur J Pediatr* 2011;170:21-34.
2. Broomfield A, Grunewald S. How to use serum ammonia. *Arch Dis Child Educ Pract Ed* 2012;97:72-77
3. Ammonia. Lab Tests Online. American Association for Clinical Chemistry (AACC). February 29, 2016. Available at: <https://labtestsonline.org/understanding/analytes/ammonia/tab/test>. Accessed September 11, 2017.
4. Hawke L. Ammonia (plasma, blood). The Association for Clinical Biochemistry and Laboratory Medicine. Available at: <http://www.acb.org.uk/whatwedo/science/amalc.aspx>. Published 2012. Accessed September 7, 2017.
5. Barsotti RJ. Measurement of ammonia in blood. *J Pediatr* 2001;138(1):S11-S20.
6. Orton DJ, Gifford JL, Seiden-Long I, Khan A, de Koning L. Critically high plasma ammonia in an adolescent girl. *Clin Chem* 2016;62(12):1565-1569.
7. Gupta A. To err is genetics: Diagnosis and management of inborn errors of metabolism (IEM). Chapter 32, pp. 415-423. In *Anthropology Today: Trends, Scope and Applications*. Eds. V Bhasin & MK Basin. New Delhi, India: Kamla-Raj Enterprises, 2007. *Anthropologist Special Volume No. 3* 2007:415-423.
8. Blair NF, Cremer PD, Tchan MC. Urea cycle disorders: a life-threatening yet treatable cause of metabolic encephalopathy in adults. *Pract Neurol* 2015;15:45-48.
9. Inherited metabolic disorders. Most recent review: October 20, 2016. Available at: <http://www.webmd.com/a-to-z-guides/inherited-metabolic-disorder-types-and-treatments#1-3>. Accessed September 11, 2017.