

## Chapter 87

### REFERENCES

1. Zhou J, Pessah IN, Allen PD, Naguib M. Neuromuscular disorders and malignant hyperthermia. In: Miller R, ed. *Miller's Anesthesia*. Vol 1. Philadelphia, PA: Churchill Livingstone; 2010:1171-1195.
2. Monnier N, Procaccio V, Stieglitz P, Lunardi J. Malignant-hyperthermia susceptibility is associated with a mutation of the alpha 1-subunit of the human dihydropyridine-sensitive L-type voltage-dependent calcium-channel receptor in skeletal muscle. *Am J Hum Genet*. 1997;60(6):1316-1325.
3. Hopkins PM. Malignant hyperthermia: advances in clinical management and diagnosis. *Br J Anaesth*. 2000;85(1):118-128.
4. Nelson TE. Malignant hyperthermia: a pharmacogenetic disease of Ca<sup>++</sup> regulating proteins. *Curr Mol Med*. 2002;2(4):347-369.
5. Denborough MA, Lovell RR. Anesthesia deaths in a family. *Lancet*. 1960;(7140):45.
6. Denborough MA, Forster JF, Lovell RR, Maplestone PA, Villiers JD. Anaesthetic deaths in a family. *Br J Anaesth*. 1962;34:395-396.
7. Britt BA, Locher WG, Kalow W. Hereditary aspects of malignant hyperthermia. *Can Anaesth Soc J*. 1968;16:89-98.
8. Kalow W, Britt BA, Terreau ME, Haist C. Metabolic error of muscle metabolism after recovery from malignant hyperthermia. *Lancet*. 1970;2(7679):895-898.
9. Ellis FR, Harriman DG, Keaney NP, Kyei-Mensah K, Tyrrell JH. Halothane-induced muscle contracture as a cause of hyperpyrexia. *Br J Anaesth*. 1971;43(7):721-722.
10. Mickelson JR, Louis CF. Malignant hyperthermia: excitation-contraction coupling, Ca<sup>2+</sup> release channel, and cell Ca<sup>2+</sup> regulation defects. *Physiol Rev*. 1996;76(2):537-592.
11. MacLennan DH, Duff C, Zorzato F, et al. Ryanodine receptor gene is a candidate for predisposition to malignant hyperthermia. *Nature*. 1990;343(6258):559-561.
12. McCarthy TV, Healy JM, Heffron JJ, et al. Localization of the malignant hyperthermia susceptibility locus to human chromosome 19q12-13.2. *Nature*. 1990;343(6258):562-564.
13. Harrison GG. Control of the malignant hyperpyrexia syndrome in MHS swine by dantrolene sodium. *Br J Anaesth*. 1975;47:62-65.
14. Kolb ME, Horne ML, Martz R. Dantrolene in human malignant hyperthermia. *Anesthesiology*. 1982;56(4):254-262.
15. Chelu MG, Goonasekera SA, Durham WJ, et al. Heat- and anesthesia-induced malignant hyperthermia in an RyR1 knock-in mouse. *Faseb J*. 2006;20(2):329-330.
16. Ørding H. Incidence of malignant hyperthermia in Denmark. *Anesth Analg*. 1985;64(7):700-704.
17. Brady JE, Sun LS, Rosenberg H, Li G. Prevalence of malignant hyperthermia due to anesthesia in New York State, 2001-2005. *Anesth Analg*. 2009;109(4):1162-1166.
18. Li G, Warner M, Lang BH, Huang L, Sun LS. Epidemiology of anesthesia-related mortality in the United States, 1999-2005. *Anesthesiology*. 2009;110(4):759-765.
19. Larach MG, Localio AR, Allen GC, et al. A clinical grading scale to predict malignant hyperthermia susceptibility. *Anesthesiology*. 1994;80(4):771-779.
20. Britt B, Kwong FHF, Endrenyi L. The clinical and laboratory features of malignant hyperthermia management—a review. In: EO Henschel, ed. *Malignant Hyperthermia: Current Concepts*. New York, NY: Appleton-Century-Crofts; 1977:9-45.
21. Larach MG, Gronert GA, Allen GC, Brandom BW, Lehman EB. Clinical presentation, treatment, and complications of malignant hyperthermia in North America from 1987 to 2006. *Anesth Analg*. 2010;110(2):498-507.
22. Monnier N, Kozak-Ribbens G, Krivosic-Horber R, et al. Correlations between genotype and pharmacological, histological, functional, and clinical phenotypes in malignant hyperthermia susceptibility. *Hum Mutat*. 2005;26(5):413-425.
23. Ibarra C, Wu S, Murayama K, et al. Malignant hyperthermia in Japan: mutation screening of the entire ryanodine receptor type 1 gene coding region by direct sequencing. *Anesthesiology*. 2006;104(6):1146-1154.
24. Dulhunty AF, Haarmann CS, Green D, Laver DR, Board PG, Casarotto MG. Interactions between dihydropyridine receptors and ryanodine receptors in striated muscle. *Prog Biophys Mol Biol*. 2002;79(1-3):45-75.
25. Franzini-Armstrong C. RYR-DHPR relationship in skeletal and cardiac muscles. In: Wehrens X, Marks A, eds. *Ryanodine Receptors Structure, Function, and Dysfunction in Clinical Disease*. Vol. 1. New York, NY: Springer; 2005:35-41.
26. Loke J, MacLennan DH. Malignant hyperthermia and central core disease: disorders of Ca<sup>2+</sup> release channels. *Am J Med*. 1998;104(5):470-486.
27. Jurkat-Rott K, McCarthy T, Lehmann-Horn F. Genetics and pathogenesis of malignant hyperthermia. *Muscle Nerve*. 2000;23(1):4-17.
28. Pessah IN, Lynch C 3rd, Gronert GA. Complex pharmacology of malignant hyperthermia. *Anesthesiology*. 1996;84(6):1275-1279.

29. MacLennan DH, Phillips MS. Malignant hyperthermia. *Science*. 1992;256(5058):789-794.
30. Durham WJ, Aracena-Parks P, Long C, et al. RyR1 S-nitrosylation underlies environmental heat stroke and sudden death in Y522S RyR1 knockin mice. *Cell*. 2008;133(1):53-65.
31. Yang T, Riehl J, Esteve E, et al. Pharmacologic and functional characterization of malignant hyperthermia in the R163C RyR1 knock-in mouse. *Anesthesiology*. 2006;105(6):1164-1175.
32. Zvaritch E, Kraeva N, Bombardier E, et al. Ca<sup>2+</sup> dysregulation in Ryr1(I4895T/wt) mice causes congenital myopathy with progressive formation of minicores, cores, and nemaline rods. *Proc Natl Acad Sci USA*. 2009;106(51):21813-21818.
33. Robinson R, Carpenter D, Shaw MA, Halsall J, Hopkins P. Mutations in RYR1 in malignant hyperthermia and central core disease. *Hum Mutat*. 2006;27(10):977-989.
34. Carpenter D, Ringrose C, Leo V, et al. The role of CACNA1S in predisposition to malignant hyperthermia. *BMC Med Genet*. 2009;10:104.
35. Wu S, Ibarra MC, Malicdan MC, et al. Central core disease is due to RYR1 mutations in more than 90% of patients. *Brain*. 2006;129(Pt 6):1470-1480.
36. Sambuughin N, Holley H, Muldoon S, et al. Screening of the entire ryanodine receptor type 1 coding region for sequence variants associated with malignant hyperthermia susceptibility in the north american population. *Anesthesiology*. 2005;102(3):515-521.
37. Sambuughin N, Sei Y, Gallagher KL, et al. North American malignant hyperthermia population: screening of the ryanodine receptor gene and identification of novel mutations. *Anesthesiology*. 2001;95(3):594-599.
38. Muldoon SM, Sambuughin N, Voelkel M, Bunger R, Grocott H, Sulzer C. Malignant hyperthermia, thermoregulation and peri-operative hypothermia. In: Longnecker D, ed. *Anesthesiology*. New York, NY: McGraw-Hill; 2008:1964-1997.
39. Monnier N, Krivosic-Horber R, Payen JF, et al. Presence of two different genetic traits in malignant hyperthermia families: implication for genetic analysis, diagnosis, and incidence of malignant hyperthermia susceptibility. *Anesthesiology*. 2002;97(5):1067-1074.
40. Litman RS, Rosenberg H. Malignant hyperthermia: update on susceptibility testing. *JAMA*. 2005;293(23):2918-2924.
41. Karan SM, Crowl F, Muldoon SM. Malignant hyperthermia masked by capnographic monitoring. *Anesth Analg*. 1994;78(3):590-592.
42. Groom L, Muldoon SM, Tang ZZ, et al. Identical de novo mutation in the type 1 ryanodine receptor gene associated with fatal, stress-induced malignant hyperthermia in two unrelated families. *Anesthesiology*. 2011;115(5):938-945.
43. Bouchama A, Knochel JP. Heat stroke. *N Engl J Med*. 2002;346(25):1978-1988.
44. Burkman JM, Posner KL, Domino KB. Analysis of the clinical variables associated with recrudescence after malignant hyperthermia reactions. *Anesthesiology*. 2007;106(5):901-906.
45. Larach MG, Brandom BW, Allen GC, Gronert GA, Lehman EB. Cardiac arrests and deaths associated with malignant hyperthermia in North America from 1987 to 2006: a report from the North American Malignant Hyperthermia Registry of the Malignant hyperthermia Association of the United States. *Anesthesiology*. 2008;108(4):603-611.
46. Gronert GA, Thompson R, Onofrio B. Human malignant hyperthermia: awake episodes and correction by dantrolene. *Anesth Analg*. 1980;59:377-378.
47. Tobin JR, Jason DR, Challa VR, Nelson TE, Sambuughin N. Malignant hyperthermia and apparent heat stroke. *JAMA*. 2001;286(2):168-169.
48. Wappler F, Fiege M, Steinfath M, et al. Evidence for susceptibility to malignant hyperthermia in patients with exercise-induced rhabdomyolysis. *Anesthesiology*. 2001;94(1):95-100.
49. Kim T, Nemergut M. Preparation of modern anesthesia workstations for malignant hyperthermia-susceptible patients—a review of past and present practice. *Anesthesiology*. 2011;114(1):205-212.
50. Durham W, Wehrens X, Sood S, Hamilton SL. Diseases associated with altered ryanodine receptor activity. In: Carafoli E, Brini M, eds. *Calcium Signalling and Disease*. New York, NY: Springer; 2007:273-321.
51. Endo M. Calcium release from the sarcoplasmic reticulum. *Physiol Rev*. 1977;57(1):71-108.
52. Rosenberg H, Antognini JF, Muldoon S. Testing for malignant hyperthermia. *Anesthesiology*. 2002;96(1):232-237.
53. Allen GC, Larach MG, Kunselman AR. The sensitivity and specificity of the caffeine-halothane contracture test: a report from the North American Malignant Hyperthermia Registry. The North American Malignant Hyperthermia Registry of MHAUS. *Anesthesiology*. 1998;88(3):579-588.
54. Ørding H, Brancadoro V, Cozzolino S, et al. In vitro contracture test for diagnosis of malignant hyperthermia following the protocol of the European MH Group: results of testing patients surviving fulminant MH and unrelated low-risk subjects. The European Malignant Hyperthermia Group. *Acta Anaesthesiol Scand*. 1997;41(8):955-966.
55. Karan S, Lojeski EW, Muldoon SM. Malignant hyperthermia. In: Tremper K, ed. *Principles of Anesthetic Techniques and Anesthetic Emergencies*. Vol 4. Philadelphia, PA: Churchill-Livingstone; 1998:9.10-19.13.
56. Girard T, Treves S, Voronkov E, Siegemund M, Urwyler A. Molecular genetic testing for malignant hyperthermia susceptibility. *Anesthesiology*. 2004;100(5):1076-1080.
57. Sei Y, Sambuughin N, Muldoon S. Malignant hyperthermia genetic testing in North America Working Group Meeting. Bethesda, Maryland. September 4-5, 2002. *Anesthesiology*. 2004;100(2):464-465.
58. Sei Y, Brandom BW, Bina S, et al. Patients with malignant hyperthermia demonstrate an altered calcium control mechanism in B lymphocytes. *Anesthesiology*. 2002;97(5):1052-1058.
59. Girard T, Cavagna D, Padovan E, et al. B-lymphocytes from malignant hyperthermia-susceptible patients have an increased sensitivity to skeletal muscle ryanodine receptor activators. *J Biol Chem*. 2001;276(51):48077-48082.