

Case 1 参考文献

1. Lumb AB, Thomas CR. Nunn and Lumb's applied respiratory physiology. 9th ed. London : Elsevier, 2021.
2. 星野あつみ, 野村岳志. 二酸化炭素異常の生理学—二酸化炭素はなぜ溜まるのか?. INTENSIVIST 2022 ; 14 : 305-15.
3. UpToDate. The evaluation and management of the nonventilated adult with acute hypercapnic respiratory failure.
< <https://www.uptodate.com/contents/the-evaluation-and-management-of-the-nonventilated-adult-with-acute-hypercapnic-respiratory-failure> > Accessed Aug. 27, 2025.
4. 安藤哲朗. 脳神経内科医はハンマーで何を診ているのか?—神経学的診察—. 神経治療 2022 ; 39 : 229-31.
5. 平山恵造. 神経症候学II 改訂第二版. 東京 : 文光堂, 2010.
6. Inskip JA, Ramer LM, Ramer MS, et al. Autonomic assessment of animals with spinal cord injury : tools, techniques and translation. Spinal Cord 2009 ; 47 : 2-35. **PMID** : 18542091
7. 日本神経学会 (監修), 筋萎縮性側索硬化症診療ガイドライン作成委員会 (編集). 筋萎縮性側索硬化症 (ALS) 診療ガイドライン 2023. 東京 : 南江堂, 2023.
8. de Carvalho M, Matias T, Coelho F, et al. Motor neuron disease presenting with respiratory failure. J Neurol Sci 1996 ; 139 Suppl : 117-22. **PMID** : 8899670
9. Oki R, Izumi Y, Fujita K, et al. Efficacy and safety of ultrahigh-dose methylcobalamin in early-stage amyotrophic lateral sclerosis : a randomized clinical trial. JAMA Neurol 2022 ; 79 : 575-83. **PMID** : 35532908
10. Miller TM, Cudkowicz ME, Genge A, et al. Trial of antisense oligonucleotide tofersen for *SOD1* ALS. N Engl J Med 2022 ; 387 : 1099-110. **PMID** : 36129998
11. Korobeynikov VA, Lyashchenko AK, Blanco-Redondo B. Antisense oligonucleotide silencing of FUS expression as a therapeutic approach in amyotrophic lateral sclerosis. Nat Med 2022 ; 28 : 104-16. **PMID** : 35075293
12. 園生雅弘. MMT・針筋電図ガイドブック. 東京 : 中外医学社, 2018.
13. Kuwabara S, Sonoo M, Komori T, et al. Dissociated small hand muscle atrophy in amyotrophic lateral sclerosis : frequency, extent, and specificity. Muscle Nerve 2008 ; 37 : 426-30. **PMID** : 18236469

14. 日本神経学会（監修），重症筋無力症/ランバート・イートン筋無力症候群診療ガイドライン作成委員会（編集）．重症筋無力症/ランバート・イートン筋無力症候群診療ガイドライン 2022. 東京：南江堂, 2022.
15. Nicolle MW, Stewart DJ, Remtulla H, et al. Lambert-Eaton myasthenic syndrome presenting with severe respiratory failure. *Muscle Nerve* 1996 ; 19 : 1328-33. **PMID** : 8808659
16. 埜中征哉, 西野一三. 臨床のための筋病理 第 5 版. 東京：日本医事新報社, 2021.
17. Sateia MJ. International classification of sleep disorders-third edition : highlights and modifications. *Chest* 2014 ; 146 : 1387-94. **PMID** : 25367475
18. van Doorn PA, Van den Bergh PYK, Hadden RDM, et al. European Academy of Neurology/Peripheral Nerve Society guideline on diagnosis and treatment of Guillain-Barré syndrome. *Eur J Neurol* 2023 ; 30 : 3646-74. **PMID** : 37814552
19. 日本神経学会（監修），ギラン・バレー症候群, フィッシャー症候群診療ガイドライン作成委員会（編集）．ギラン・バレー症候群, フィッシャー症候群診療ガイドライン 2024. 東京：南江堂, 2024.
20. 日本先天代謝異常学会（編集）．ポンペ病診療ガイドライン 2018. 東京：診断と治療社, 2018.
21. Kishnani PS, Steiner RD, Bali D, et al. Pompe disease diagnosis and management guideline. *Genet Med* 2006 ; 8 : 267-88. **PMID** : 16702877
22. Barba-Romero MA, Barrot E, Bautista-Lorite J, et al. Clinical guidelines for late-onset Pompe disease. *Rev Neurol* 2012 ; 54 : 497-507. **PMID** : 22492103
23. Cupler EJ, Berger KI, Leshner RT, et al. Consensus treatment recommendations for late-onset Pompe disease. *Muscle Nerve* 2012 ; 45 : 319-33. **PMID** : 22173792
24. North KN, Laing NG, Wallgren-Pettersson C. Nemaline myopathy : current concepts. The ENMC international consortium and nemaline myopathy. *J Med Genet* 1997 ; 34 : 705-13. **PMID** : 9321754
25. Romero NB, Sandaradura SA, Clarke NF. Recent advances in nemaline myopathy. *Curr Opin Neurol* 2013 ; 26 : 519-26. **PMID** : 23995272
26. Eymard B, Brouet JC, Collin H, et al. Late-onset rod myopathy associated with monoclonal gammopathy. *Neuromuscul Disord* 1993 ; 3 : 557-60. **PMID** : 8186711
27. Dalakas MC, Pezeshkpour GH, Flaherty M. Progressive nemaline (rod) myopathy associated with HIV infection. *N Engl J Med* 1987 ; 317 : 1602-3. **PMID** : 3683496
28. Dimitri D, Dubourg O. Sporadic late-onset nemaline myopathy in a patient with systemic lupus erythematosus. *J Neurol* 2013 ; 260 : 3171-3. **PMID** : 24178707
29. Ando T, Sato T, Kurahashi S, et al. A case of sporadic late-onset nemaline myopathy with monoclonal gammopathy of undetermined significance : long-term observation

of neurological symptoms after autologous stem-cell transplantation. Nagoya J Med Sci 2021 ; 83 : 641-7. **PMID** : 34552296

30. Utsugisawa K, Nagane Y, Akaishi T, et al. Early fast-acting treatment strategy against generalized myasthenia gravis. Muscle Nerve 2017 ; 55 : 794-801. **PMID** : 27603432
31. Lee SJ, Hur J, Lee TW, et al. Myasthenia gravis presenting initially as acute respiratory failure. Respir Care 2015 ; 60 : e14-6. **PMID** : 25185150
32. Mittal MK, Barohn RJ, Pasnoor M, et al. Ocular myasthenia gravis in an academic neuro-ophthalmology clinic : clinical features and therapeutic response. J Clin Neuromuscul Dis 2011 ; 13 : 46-52. **PMID** : 22361625
33. Fakiri MO, Tavy DL, Hama-Amin AD, et al. Accuracy of the ice test in the diagnosis of myasthenia gravis in patients with ptosis. Muscle Nerve 2013 ; 48 : 902-4. **PMID** : 23536427
34. Scherer K, Bedlack RS, Simel DL. Does this patient have myasthenia gravis? JAMA 2005 ; 293 : 1906-14. **PMID** : 15840866
35. Beekman R, Kuks JB, Oosterhuis HJ. Myasthenia gravis : diagnosis and follow-up of 100 consecutive patients. J Neurol 1997 ; 244 : 112-8. **PMID** : 9120493
36. 山寄博輝, 野寺裕之. 神経伝導検査・針筋電図検査—ICU での適応と注意点. INTENSIVIST 2016 ; 8 : 891-7.
37. Murai H, Utsugisawa K, Nagane Y, et al. Rationale for the clinical guidelines for myasthenia gravis in Japan. Ann N Y Acad Sci 2018 ; 1413 : 35-40. **PMID** : 29377151
38. Sanders DB, Wolfe GI, Benatar M, et al. International consensus guidance for management of myasthenia gravis : executive summary. Neurology 2016 ; 87 : 419-25. **PMID** : 27358333
39. Iorio R. Myasthenia gravis : the changing treatment landscape in the era of molecular therapies. Nat Rev Neurol 2024 ; 20 : 84-98. **PMID** : 38191918