

Daftar Pustaka

1. Undang-undang Republik Indonesia Nomor 36 Tahun 2009. UU tentang Kesehatan. 2009.
2. Undang-undang Republik Indonesia Nomor 35 Tahun 2014. UU tentang Perlindungan Anak. 2014.
3. Wang CH, Finkel RS, Bertini ES, Schroth M, Simonds A, Wong B, et al. Consensus statement for standard of care in spinal muscular atrophy. *J Child Neurol.* 2007;22(8):1027–49.
4. Dejsuphong D, Taweewongsounon A, Khemthong P, Chitphuk S, Stitchantrakul W, Sritara P, et al. Carrier frequency of spinal muscular atrophy in Thailand. *Neurol Sci.* 2019;40(8):1729–32.
5. Murrell D V., Lotze TE, Farber HJ, Crawford CA, Wiemann CM. The Experience of Families With Children With Spinal Muscular Atrophy Type I Across Health Care Systems. *J Child Neurol.* 2017;32(11):917–23.
6. Mercuri E, Finkel RS, Muntoni F, Wirth B, Montes J, Main M, et al. Diagnosis and management of spinal muscular atrophy: part 1: recommendations for diagnosis, rehabilitation, orthopedic and nutritional care. *Neuromuscul Disord.* 2018;28:103–15.
7. Farrar MA, Park SB, Vucic S, Carey KA, Turner BJ, Gillingwater TH, et al. Emerging therapies and challenges in spinal muscular atrophy. *Ann Neurol.* 2017;81:355–68.
8. Russman BS. Spinal muscular atrophy: Clinical classification and disease heterogeneity. *J Child Neurol.* 2007;22(8):946–51.
9. von Gontard A, Zerres K, Backes M, Laufersweiler-Plass C, Wendland C, Melchers P, et al. Intelligence and cognitive function in children and adolescents with spinal muscular atrophy. *Neuromuscul Disord.* 2002;12:130–6.
10. Lin CW, Kalb SJ, Yeh WS. Delay in Diagnosis of Spinal Muscular Atrophy: A Systematic Literature Review. *Pediatr Neurol.* 2015;53:293–300.
11. Darras BT, Finkel RS. Natural History of Spinal Muscular Atrophy. *Natural*

- History of Spinal Muscular Atrophy. US: Elsevier Inc.; 2017. 399–421 p.
12. Kaufmann P, McDermott MP, Darras BT, Finkel RS. Prospective cohort study of spinal muscular atrophy types 2 and 3. *Neurology*. 2012;79(18):1889–97.
 13. Mercuri E, Bertini E, Iannaccone ST. Childhood spinal muscular atrophy: Controversies and challenges. *Lancet Neurol*. 2012;11:443–52.
 14. Finkel RS, Sejersen T, Mercuri E, Bertini E, Chen K, Crawford TO, et al. 218th ENMC International Workshop: Revisiting the consensus on standards of care in SMA Naarden, The Netherlands, 19–21 February 2016. *Neuromuscul Disord*. 2017;27(6):596–605.
 15. Hendrickson BC, Donohoe C, Akmaev VR, Sugarman EA, Labrousse P, Boguslavskiy L, et al. Differences in SMN1 allele frequencies among ethnic groups within North America. *J Med Genet*. 2009;46:641–4.
 16. Sugarman EA, Nagan N, Zhu H, Akmaev VR, Zhou Z, Rohlfes EM, et al. Pan-ethnic carrier screening and prenatal diagnosis for spinal muscular atrophy: clinical laboratory analysis of >72,400 specimens. *Eur J Hum Genet*. 2012;20:27–32.
 17. Yoon S, Lee CH, Lee KA. Determination of SMN1 and SMN2 copy numbers in a Korean population using multiplex ligation-dependent probe amplification. *Korean J Lab Med*. 2010;30:93–6.
 18. Chan V, Yip B, Yam I, Au P, Lin CK, Wong V, et al. Carrier incidence for spinal muscular atrophy in southern Chinese. *J Neurol*. 2004;251:1089–93.
 19. Su YN, Hung CC, Lin SY, Chen FY, Chern JPS, Tsai C, et al. Carrier screening for spinal muscular atrophy (SMA) in 107,611 pregnant women during the period 2005-2009: A prospective population-based cohort study. *PLoS One*. 2011;6(2):1–7.
 20. Fang P, Li L, Zeng J, Zhou WJ, Wu WQ, Zhong ZY, et al. Molecular characterization and copy number of SMN1, SMN2 and NAIP in Chinese patients with spinal muscular atrophy and unrelated healthy controls. *BMC Musculoskelet Disord*. 2015;16(1):1–8.
 21. Feng Y, Ge X, Meng L, Scull J, Li J, Tian X, et al. The next generation of

- population-based spinal muscular atrophy carrier screening: Comprehensive pan-ethnic SMN1 copy-number and sequence variant analysis by massively parallel sequencing. *Genet Med.* 2017;19(8):936–44.
22. Shafie AA, Chaiyakunapruk N, Supian A, Lim J, Zafra M, Hassali MAA. State of rare disease management in Southeast Asia. *Orphanet J Rare Dis.* 2016;11:107.
 23. Augustine EF, Dorsey ER, Saltonstall PL. The care continuum: An evolving model for care and research in rare diseases. *Pediatrics.* 2017;140(3):e20170108.
 24. Qian Y, McGraw S, Henne J, Jarecki J, Hobby K, Yeh WS. Understanding the experiences and needs of individuals with Spinal Muscular Atrophy and their parents: a qualitative study. *BMC Neurol.* 2015;15:217.
 25. Lawton S, Hickerton C, Archibald AD, McClaren BJ, Metcalfe SA. A mixed methods exploration of families' experiences of the diagnosis of childhood spinal muscular atrophy. *Eur J Hum Genet.* 2015;23:575–80.
 26. Yang BH, Mu PF, Wang WS. The experiences of families living with the anticipatory loss of a school-age child with spinal muscular atrophy – the parents' perspectives. *J Clin Nurs.* 2016;25(17–18):2648–57.
 27. Finkel RS, Mercuri E, Meyer OH, Simonds AK, Schroth MK, Graham RJ, et al. Diagnosis and management of spinal muscular atrophy: part 2: pulmonary and acute care. *Neuromuscul Disord.* 2018;28:197–207.
 28. Tizzano EF. Treating neonatal spinal muscular atrophy: A 21st century success story? *Early Hum Dev.* 2019;138.
 29. Dangouloff T, Servais L. Clinical Evidence Supporting Early Treatment Of Patients With Spinal Muscular Atrophy: Current Perspectives. *Ther Clin Risk Manag.* 2019;15:1153–61.
 30. the Centers for Disease Control and Prevention (CDC). Measuring Healthy Days: Population Assessment of Health-Related Quality of Life. CDC. 2000.
 31. Cho MH. Clinical approach to quality of life in children with end-stage renal disease. *Korean J Pediatr.* 2013;56(8):323–6.
 32. Iannaccone ST, Hynan LS, Morton A, Buchanan R, Limbers CA, Varni JW.

- The PedsQL™ in Pediatric Patients with Spinal Muscular Atrophy: Feasibility, Reliability, and Validity of the Pediatric Quality of Life Inventory™ Generic Core Scales and Neuromuscular Module. *Neuromuscul Disord.* 2009;19(12):805–12.
33. Jaglal SB, Guilcher SJT, Bereket T, Kwan M, Munce S, Conklin J, et al. Development of a Chronic Care Model for Neurological Conditions (CCM-NC). *BMC Health Serv Res.* 2014;14:409.
 34. Epping-Jordan JE, Pruitt SD, Bengoa R, Wagner EH. Improving the quality of health care for chronic conditions. *Qual Saf Heal Care.* 2004;13:299–305.
 35. Quinlivan R, Matthews E, Hanna MG. Innovative care model for patients with complex muscle diseases. *Curr Opin Neurol.* 2014;27:607–13.
 36. Arisanti N, Sasongko EPS, Pandia V, Hilmanto D. Implementation of palliative care for patients with terminal diseases from the viewpoint of healthcare personnel. *BMC Res Notes.* 2019;12:217.
 37. Arnold WD, Kassar D, Kissel JT. Spinal Muscular Atrophy: Diagnosis and Management in a New Therapeutic Era. *Muscle Nerve.* 2015;51(2):157–67.
 38. Kolb SJ, Kissel JT. Spinal muscular atrophy: a timely review. *Arch Neurol.* 2011;68(8):979–84.
 39. Sutarjo. Apakah Spinal Muscular Atrophy(SMA)? [Internet]. 2018. Available from: <https://smaindonesia.org/blog/2018/01/02/apakah-spinal-muscular-atrophy-sma/>
 40. D'Amico A, Mercuri E, Tiziano FD, Bertini E. Spinal muscular atrophy. *Orphanet J Rare Dis.* 2011;6:71.
 41. Baioni MTC, Ambiel CR. Spinal muscular atrophy: Diagnosis, treatment and future prospects. *J Pediatr (Rio J).* 2010;86(4):261–70.
 42. Lorson CL, Rindt H, Shababi M. Spinal muscular atrophy: Mechanisms and therapeutic strategies. *Hum Mol Genet.* 2010;19(R1):111–8.
 43. Lunn MR, Wong CH. Spinal muscular atrophy. *Lancet.* 2008;371:2120–33.
 44. Magnani L, Bertolotti T. *Springer Handbook of Model-Based Science.* Vol. 80. Springer; 2017.
 45. Gall MD, Gall JP, R W, Borg. *Educational Research: An Introduction.* 7th

- ed. *Nursing Research*. Boston: Pearson Education Inc.; 2003.
46. Putra N. *Research dan development : Penelitian dan Pengembangan : Suatu Pengantar*. Jakarta: Rajawali; 2012. 214 p.
 47. Tham TY, Tran TL, Prueksaritanond S, Isidro JS, Setia S, Welluppillai V. Integrated health care systems in asia: An urgent necessity. *Clin Interv Aging*. 2018;13:2527–38.
 48. De Visser M, Oliver DJ. Palliative care in neuromuscular diseases. *Curr Opin Neurol*. 2017;30.
 49. Gotay CC, Korn EL, McCabe MS, Moore TD, Cheson BD. Quality-of-Life Assessment in Cancer Treatment Protocols: Research Issues in Protocol Development. *J Natl Cancer Inst*. 1992;84(8):575–9.
 50. Theofilou P. Quality of life: Definition and measurement. *Eur J Psychol*. 2013;9(1):150–62.
 51. Klug C, Schreiber-Katz O, Thiele S, Schorling E, Zowe J, Reilich P, et al. Disease burden of spinal muscular atrophy in Germany. *Orphanet J Rare Dis*. 2016;11:58.
 52. Wan HWY, Carey KA, D’Silva A, Kasparian NA, Farrar MA. “Getting ready for the adult world”: How adults with spinal muscular atrophy perceive and experience healthcare, transition and well-being. *Orphanet J Rare Dis*. 2019;14:74.
 53. Creswell J., Clark VLP. *Designing and Conducting Mixed Methods Research*. 3rd ed. SAGE Publications Inc.; 2017. 520 p.
 54. Tashakkori A, Johnson RB, Teddlle C. *Foundations of Mixed Methods Research*. SAGE Publications Inc.; 2020. 472 p.
 55. Miles MB, Huberman AM, Saldana J. *Qualitative Data Analysis A Methods Sourcebook*. SAGE Publications Inc.; 2019. 408 p.
 56. Hsu CC, Sandford BA. The Delphi technique: Making sense of consensus. *Pract Assessment, Res Eval*. 2007;12(10).
 57. Mosadeghrad AM, Ghazanfari F. Developing a hospital accreditation model: a Delphi study. *BMC Health Serv Res*. 2021;21:879.
 58. Porter J, Charlton K, Tapsell L, Truby H. Using the Delphi process to

- identify priorities for Dietetic research in Australia 2020-2030. *Nutr Diet.* 2020;77:437–43.
59. Davies A, Teare L, Falder S, Dumville J, Shah M, Jenkins ATA, et al. Consensus demonstrates four indicators needed to standardize burn wound infection reporting across trials in a single-country study (ICon-B study). *J Hosp Infect.* 2020;106(2):217–25.
 60. Braddom RL. *Braddom's Physical Medicine and Rehabilitation*. 5th ed. Cifu DX, editor. Philadelphia: Elsevier; 2016.
 61. Zohrabi M. Mixed method research: Instruments, validity, reliability and reporting findings. *Theory Pract Lang Stud.* 2013;3(2):254–62.
 62. Jamieson S. Likert scales: How to (ab)use them? *Med Educ.* 2004;38(12):1217–8.
 63. Kyriazos TA. Applied Psychometrics: Sample Size and Sample Power Considerations in Factor Analysis (EFA, CFA) and SEM in General. *Psychology.* 2018;09:2207–30.
 64. MacCallum RC, Widaman KF, Preacher KJ, Hong S. Sample size in factor analysis: The role of model error. *Multivariate Behav Res.* 2001;36(4):611–37.
 65. Undang-undang Republik Indonesia Nomor 23 Tahun 1997. UU tentang Pengelolaan Lingkungan Hidup. 1997;
 66. Ferdinand A. *Structural equation modeling dalam penelitian manajemen : aplikasi model- model rumit dalam penelitian untuk tesis S-2 & disertasi S-3*. Semarang: Badan Penerbit Universitas Diponegoro; 2000.
 67. Ghozali I, Latan H. *Partial Least Squares Konsep, Teknik dan Aplikasi Menggunakan Program SmartPLS 3.0 Untuk Penelitian Empiris*. 2nd ed. Semarang: Badan Penerbit Universitas Diponegoro. Semarang: Badan Penerbit Universitas Diponegoro; 2015.
 68. Fouka G, Mantzourou M. What are the major ethical issues in conducting research? is there a conflict between the research ethics and the nature of nursing? *Heal Sci J.* 2011;5(1):3–14.
 69. Peraturan Menteri Pendidikan Nasional Republik Indonesia no. 70 tahun

2009. Permendiknas RI tentang Pendidik Inklusif bagi Peserta Didik yang Memiliki Kelainan dan Memiliki Potensi Kecerdasan dan/atau Bakat Istimewa. 2009.
70. Peraturan Menteri Pendidikan dan Kebudayaan Republik Indonesia no. 44 tahun 2019. Permendikbud RI tentang Penerimaan Peserta Didik Baru pada Taman Kanak-Kanak, Sekolah Dasar, Sekolah Menengah Pertama, Sekolah Menengah Atas, dan Sekolah Menengah Kejuruan. 2019.
 71. Undang-undang Republik Indonesia no. 8 tahun 2016. UU tentang Penyandang Disabilitas. 2016.
 72. Peraturan Pemerintah Republik Indonesia no. 13 tahun 2020. PP RI tentang Akomodasi yang Layak untuk Peserta Didik Penyandang Disabilitas. 2020.
 73. Peraturan Menteri Pendidikan dan Kebudayaan Republik Indonesia no. 6 tahun 2021. Permendikbud RI tentang Petunjuk Teknis Pengelolaan Dana Bantuan Operasional Sekolah Reguler. 2021.
 74. Peraturan Menteri Pendidikan dan Kebudayaan Republik Indonesia no.22 tahun 2016. Permendikbud RI tentang Standar Proses Pendidikan Dasar dan Menengah. 2016.
 75. Landfeldt E, Pechmann A, McMillan HJ, Lochmüller H, Sejersen T. Costs of Illness of Spinal Muscular Atrophy: A Systematic Review. *Appl Health Econ Health Policy*. 2021;19:501–20.
 76. Cohen JS, Biesecker BB. Quality of Life in Rare Genetic Conditions: A Systematic Review of the Literature. *Am J Med Genet*. 2010;152A(5):1136–56.
 77. Farrar MA, Carey KA, Paguinto S-G, Chambers G, Kasparian NA. Financial, opportunity and psychosocial costs of spinal muscular atrophy: An exploratory qualitative analysis of Australian carer perspectives. *BMJ Open*. 2018;8:e020907.
 78. Alvarez K, Suarez B, Palomino MA, Hervias C, Calcagno G, Martínez-Jalilie M, et al. Observations from a nationwide vigilance program in medical care for spinal muscular atrophy patients in Chile. *Arq Neuropsiquiatr*. 2019;77(7):470–7.

79. Lamb C, Peden A. Understanding the Experience of Living with Spinal Muscular Atrophy. *J Neurosci Nurs*. 2008 Aug;40(4):250–6.
80. Rokom. Transformasi Layanan Primer Kemenkes Fokus Pada Pencegahan di Puskesmas dan Posyandu [Internet]. 2023. Available from: <https://sehatnegeriku.kemkes.go.id/baca/rilis-media/20220727/4140743/transformasi-layanan-primer-kemenkes-fokus-pada-pencegahan-di-puskesmas-dan-posyandu/>
81. Indonesia ISMKM. Transformasi Layanan Kesehatan Nasional [Internet]. 2023. Available from: <https://lldikti5.kemdikbud.go.id/home/detailpost/transformasi-layanan-kesehatan-nasional>
82. Janssen M, Sagasser MH, Laro EAM, De Graaf J, Scherpbier-De Haan ND. Learning intraprofessional collaboration by participating in a consultation programme: What and how did primary and secondary care trainees learn? *BMC Med Educ*. 2017;17:25.
83. Soetomenggolo TS, Ismael S, Handryastuti S. *Buku Ajar Neurologi Anak*. 1st ed. Badan Penerbit Ikatan Dokter Anak Indonesia; 2022.
84. Peraturan Gubernur Jawa Barat no.92 tahun 2019. Pergub Jawa Barat tentang Pemenuhan Standar Pelayanan Minimal Penyelenggaraan Pendidikan Khusus di Daerah Provinsi Jawa Barat. 2019.
85. Peraturan Menteri Pendidikan, Kebudayaan, Riset, dan Teknologi Republik Indonesia no.31 tahun 2021. Permendikbudristek RI tentang Perubahan atas Permendikbudristek no16 tahun 2021 tentang Petunjuk Teknis Pengelolaan Dana Bantuan Operasional Sekolah Kinerja dan Bantuan Operasional Sekolah Afirmasi Tahun Anggaran 2021. 2021.
86. Cruz R, Belter L, Wasnock M, Nazarelli A, Jarecki J. Evaluating Benefit-risk Decision-making in Spinal Muscular Atrophy: A First-ever Study to Assess Risk Tolerance in the SMA Patient Community. *Clin Ther*. 2019;41(5):943–60.
87. Peraturan Menteri Pendidikan, Kebudayaan, Riset, dan Teknologi Republik Indonesia no.16 tahun 2021. Permendikbudristek RI tentang Petunjuk Teknis

- Pengelolaan Dana Bantuan Operasional Sekolah Kinerja dan Bantuan Operasional Sekolah Afirmasi Tahun Anggaran 2021. 2021.
88. Undang Undang Republik Indonesia no.20 tahun 2003. UU RI tentang Sistem Pendidikan Nasional. 2003.
 89. Peraturan Pemerintah Republik Indonesia no.19 tahun 2005. PP RI tentang Standar Nasional Pendidikan. 2005.
 90. Hair JF, Risher JJ, Sarstedt M, Ringle CM. When to use and how to report the results of PLS-SEM. *Eur Bus Rev.* 2019;31(1):2–24.
 91. Hair JF, Sarstedt M, Hopkins L, Kuppelwieser VG. Partial least squares structural equation modeling (PLS-SEM): An emerging tool in business research. *Eur Bus Rev.* 2014;26(2):106–21.
 92. Hair JF, Hult GTM, Ringle CM, Sarstedt M. A primer on partial least squares structural equation modeling (PLS-SEM). 2nd ed. Los Angeles: SAGE Publications Inc.; 2017.
 93. Mix L, Schreiber-Katz O, Wurster CD, Uzelac Z, Platen S, Gipperich C, et al. Executive function is inversely correlated with physical function: the cognitive profile of adult Spinal Muscular Atrophy (SMA). *Orphanet J Rare Dis.* 2021;16:10.
 94. Kolb SJ, Kissel JT. Spinal Muscular Atrophy. *Neurol Clin.* 2015;33(4):831–46.
 95. Sumintono B, Widhiarso W. Aplikasi Model Rasch untuk Penelitian Ilmu-Ilmu Sosial. Revisi. Cimahi: Trim Komunikata Publishing House; 2014.
 96. Ariffin SR, Omar B, Isa A, Sharif S. Validity and reliability Multiple Intelligent item using Rasch measurement model. *Procedia Soc Behav Sci.* 2010;9:729–33.
 97. Wibisono S. Aplikasi Model Rasch Untuk Validasi Instrumen Pengukuran Fundamentalisme Agama Bagi Responden Muslim. *J Pengukuran Psikol dan Pendidik Indones.* 2018;5(1).