



UNIVERSITAS  
GADJAH MADA

PERBANDINGAN PROGRESIVITAS KLINIS MOTORIK DUCHENNE MUSCULAR DYSTROPHY DAN  
BECKER MUSCULAR DYSTROPHY

YANG DIDIAGNOSIS BERDASARKAN HASIL BIOPSI OTOT PADA PASIEN ANAK DI RSUP DR

SARDJITO

ZAKIAH NUR ISTIANAH, Prof. dr. Sunartini, PhD, SpAK; dr. Sasmito Nugroho, SpAK

Universitas Gadjah Mada, 2018 | Diunduh dari <http://etd.repository.ugm.ac.id/>

## DAFTAR PUSTAKA

- Aartsma-Rus, A., Ginjaar, I.B., Bushby, K. 2016. The importance of genetic diagnosis for Duchenne muscular dystrophy. *J Med Genet* 1–7.
- Ansved, T. 2001. Muscle training in muscular dystrophies. *Acta Physiol. Scand.* 171, 359–366.
- Barja, S., Pérez, R. 2016. Clinical assessment underestimates fat mass and overestimates resting energy expenditure in children with neuromuscular diseases. *Clin. Nutr. ESPEN* 15, 11–15.
- Bello, L., Campadello, P., Barp, A., Fanin, M., Semplicini, C., Sorarù, G., et al. 2016. Functional changes in Becker muscular dystrophy : implications for clinical trials in dystrophinopathies. *Nat. Publ. Gr.* 6 : 32439, 1–12.
- Bérard, C., Payan, C., Hodgkinson, I., Fermanian, J. 2005. A motor function measure scale for neuromuscular diseases. Construction and validation study. *Neuromuscul. Disord.* 15, 463–470.
- Bushby, K., Finkel, R., Birnkrant, D.J., Case, L.E., Clemens, P.R., Cripe, L., et al. 2009. *Lancet Neurology. Lancet Neurol.* 54–62.
- Connolly, A.M., Florence, J.M., Cradock, M.M., Malkus, E.C., Schierbecker, J.R., Siener, C.A., et al. 2013. Motor and cognitive assessment of infants and young boys with Duchenne Muscular Dystrophy: Results from the Muscular Dystrophy Association DMD Clinical Research Network. *Neuromuscul. Disord.* 23, 529–539.



Connolly, A.M., Pt, J.M.F., Cradock, M.M., Eagle, M., Flanigan, K.M., McDonald, C.M., Karachunski, P.I., et al. 2014. One Year Outcome of Boys With Duchenne Muscular Dystrophy Using the Bayley-III Scales of Infant and Toddler Development. *Pediatr. Neurol.* 50, 557–563.

Cruz Guzmán, O.D.R., Chávez García, A.L., Rodríguez-Cruz, M. 2012. Muscular dystrophies at different ages: Metabolic and endocrine alterations. *Int. J. Endocrinol.* 2012.

Daniels, M.J., Xu, D., Tennekoon, G.I., Erika, L., Russman, B.S., Finkel, R.S., Triplett, W.T., et al. 2018. Longitudinal timed function tests in duchenne muscular dystrophy: Imaging DMD cohort natural history. *Muscle and Nerve* 5, 1–29.

Davis, J., Samuels, E., Mullins, L. 2015. Nutrition Considerations in Duchenne Muscular Dystrophy. *Nutr. Clin. Pract.* XX, 511–521.

De Latre, C., Payan, C., Vuillerot, C., Rippert, P., De Castro, D., Bérard, C., et al. 2013. Motor function measure: Validation of a short form for young children with neuromuscular diseases. *Arch. Phys. Med. Rehabil.* 94, 2218–2226.

De Valle, K.L., Davidson, Z.E., Kennedy, R.A., Ryan, M.M., Carroll, K.M. 2016. Physical activity and the use of standard and complementary therapies in Duchenne and Becker muscular dystrophies. *J. Pediatr. Rehabil. Med.* 9, 55–63.

Eagle, M., Baudouin, S. V., Chandler, C., Giddings, D.R., Bullock, R., Bushby, K. 2002. Survival in Duchenne muscular dystrophy: improvements in life expectancy since 1967 and the impact of home nocturnal ventilation. *Neuromuscul. Disord.* 12, 926–929.

Fu, X.-N., Xiong, H. 2017. Genetic and Clinical Advances of Congenital Muscular Dystrophy. *Chin. Med. J. (Engl).* 130, 2624.



Fujii, K., Minami, N., Hayashi, Y., Nishino, I., Nonaka, I., Tanabe, Y., et al. 2009.

Homozygous female becker muscular dystrophy. Am. J. Med. Genet. 149, 1052–1055.

Gao, Q., McNally, E.M. 2016. The Dystrophin Complex: structure, function and implications for therapy. Compr Physiol 5, 1223–1239.

Gianola, S., Pecoraro, V., Lambiase, S., Gatti, R., Banfi, G., Moja, L. 2013. Efficacy of Muscle Exercise in Patients with Muscular Dystrophy: A Systematic Review Showing a Missed Opportunity to Improve Outcomes. PLoS One 8, 1–9.

Goldstein, J.A., McNally, E.M. 2010. Mechanisms of muscle weakness in muscular dystrophy. J. Gen. Physiol. 136, 29–34.

Helderman-Van Den Enden, A.T.J.M., Van Den Bergen, J.C., Breuning, M.H., Verschueren, J.J.G.M., Tibben, A., Bakker, E., et al. 2011. Duchenne/Becker muscular dystrophy in the family: Have potential carriers been tested at a molecular level? Clin Genet 79, 236–242.

Jeronimo, G., Nozoe, K.T., Polesel, D.N., Moreira, G.A., Tufik, S., Andersen, M.L. 2016. Impact of corticotherapy, nutrition, and sleep disorder on quality of life of patients with Duchenne muscular dystrophy. Nutrition 32, 391–393.

Kaczorowska, E., Zimowski, J., Cichoń-Kotek, M., Mrozińska, A., Purzycka, J., Wierzba, J., et al. 2016. Coincidence of Turner Syndrome and Duchenne Muscular Dystrophy – an Important Problem for the Clinician. Dev. Period Med. XX, 273–278.

Koeks, Z., Bladen, C.L., Salgado, D., van Zwet, E., Pogoryelova, O., McMacken, G., et al. 2017. Clinical Outcomes in Duchenne Muscular Dystrophy: A Study of 5345 Patients from the TREAT-NMD DMD Global Database. J. Neuromuscul. Dis. 4, 1–14.



Kohler, M., Clarenbach, C.F., Bahler, C., Brack, T., Russi, E.W., Bloch, K.E. 2009.

Disability and survival in Duchenne muscular dystrophy Disability and survival in Duchenne muscular dystrophy 80.

Kostek, M.C., Gordon, B. 2018. Exercise is an adjuvant to contemporary dystrophy treatments, Exercise and Sport Sciences Reviews.

Li, X., Zhao, L., Zhou, S., Hu, C., Shi, Y., Shi, W., et al. 2015. A comprehensive database of Duchenne and Becker muscular dystrophy patients ( 0 – 18 years old ) in East China. Orphanet J. Rare Dis. 10, 1–10.

Maciel Pizzato, T., Jesus Alves de Baptista, C.R. de, Martinez, E.Z., Sobreira, C.F. da R., Mattiello-Sverzut, A.C. 2016. Prediction of Loss of Gait in Duchenne Muscular Dystrophy Using the Ten Meter Walking Test Rates. J. Genet. Syndr. Gene Ther. 7, 1–6.

Mah, J. 2016. Current and emerging treatment strategies for Duchenne muscular dystrophy. Neuropsychiatr. Dis. Treat. Volume 12, 1795–1807.

Nozoe, K.T., Akamine, R.T., Mazzotti, D.R., Polesel, D.N., Grossklauss, L.F., Tufik, S., et al. 2016. Phenotypic contrasts of Duchenne Muscular Dystrophy in women: Two case reports. Sleep Sci. 9, 129–133.

Omonova, U.T., 2013. INTERNATIONAL OF BIOMEDICINE Clinical-Diagnostic Features of Duchenne Muscular Dystrophy in Children 3, 266–268.

Passamano, L., Taglia, A., Palladino, A., Viggiano, E., D'Ambrosio, P., Scutifero, M, et al. 2012. Improvement of survival in Duchenne Muscular Dystrophy: Retrospective analysis of 835 patients. Acta Myol. 31, 121–125.



Politano, L., Scutifero, M., Patalano, M., Sagliocchi, A., Zaccaro, A., Civati, F., et al. 2017. Integrated care of muscular dystrophies in Italy. Part 1. Pharmacological treatment and rehabilitative interventions. *Acta Myol.* XXXVI, 19–24.

Reddy, A., Elizabeth, C., Nicholas, H. 2013. Nutritional status and admission risk in Duchenne muscular dystrophy (DMD). *Eur. Respir. Soc. Annu. Congr.* 2013 36, 1.

Romitti, P.A., Zhu, Y., Puzhankara, S., James, K.A., Nabukera, S.K., Zamba, G.K.D., et al. 2015. Prevalence of Duchenne and Becker Muscular Dystrophies in the United States. *Pediatrics* 135, 1–12.

Ryder, S., Leadley, R.M., Armstrong, N., Westwood, M., De Kock, S., Butt, T., et al. 2017. The burden, epidemiology, costs and treatment for Duchenne muscular dystrophy: an evidence review. *Orphanet J. Rare Dis.* 12, 1–21.

Salera, S., Menni, F., Moggio, M., Guez, S., Sciacco, M., Esposito, S. 2017. Nutritional challenges in duchenne muscular dystrophy. *Nutrients* 9, 1–10.

Sardone, V., Ellis, M., Torelli, S., Feng, L., Chambers, D., Eastwood, D., et al. 2018. A novel high-throughput immunofluorescence analysis method for quantifying dystrophin intensity in entire transverse sections of Duchenne muscular dystrophy muscle biopsy samples. *PLoS One* 13, 1–21.

Schram, G., Fournier, A., Leduc, H., Dahdah, N., Therien, J., Vanasse, M., et al. 2013. All-cause mortality and cardiovascular outcomes with prophylactic steroid therapy in Duchenne muscular dystrophy. *J. Am. Coll. Cardiol.* 61, 948–954.



Silva, E.C. Da, Machado, D.L., Resende, M.B.D., Silva, R.F., Zanoteli, E., Reed, U.C. 2012. Motor function measure scale, steroid therapy and patients with Duchenne muscular dystrophy. *Arq. Neuropsiquiatr.* 70, 191–5.

Suriyonplengsaeng, C., Dejthevaporn, C., Khongkhatithum, C., Sanpantan, S., Tubthong, N., Pinpradap, K., et al. 2017. Immunohistochemistry of sarcolemmal membrane-associated proteins in formalin-fixed and paraffin-embedded skeletal muscle tissue: A promising tool for the diagnostic evaluation of common muscular dystrophies. *Diagn. Pathol.* 12, 1–10.

Syarif, I., Widiastuti, 2009. Distrofi muskular duchenne. *Maj. Kedokt. Andalas* 33, 196–206.

Taglia, A., Petillo, R., D'Ambrosio, P., Picillo, E., Torella, A., Orsini, C., et al. 2015. Clinical features of patients with dystrophinopathy sharing the 45-55 exon deletion of DMD gene. *Acta Myol.* 34, 9–13.

Tayeb, M.T. 2010. Deletion mutations in Duchenne muscular dystrophy (DMD) in Western Saudi children. *Saudi J. Biol. Sci.* 17, 237–240.

Taylor, P.J., Maroulis, S., Mullan, G.L., Pedersen, R.L., Baumli, A., Elakis, G., et al. 2007. Measurement of the clinical utility of a combined mutation detection protocol in carriers of Duchenne and Becker muscular dystrophy. *J Med Genet* 44, 368–372.

Theadom, A., Rodrigues, M., Roxburgh, R., Balalla, S., Higgins, C., Bhattacharjee, R., et al. 2014. Prevalence of muscular dystrophies: A systematic literature review. *Neuroepidemiology* 43, 259–268.

Van den Bergen, J.C., Ginjaar, H.B., van Essen, A.J., Pangalila, R., de Groot, I.J.M., Wijkstra, P.J., et al. 2014. Forty-Five Years of Duchenne Muscular Dystrophy in The Netherlands. *J. Neuromuscul. Dis.* 1, 99–109.



Wong, B.L., Rybalsky, I., Shellenbarger, K.C., Tian, C., Mcmahon, M.A., Rutter,

M.M., et al. 2016. Long-term outcome of interdisciplinary management of patients with duchenne muscular dystrophy receiving daily glucocorticoid treatment. *J. Pediatr.* 182, 296–303.e1.

Zhong, J., Xu, T., Chen, G., Lan, D. 2017. Genetic analysis of the dystrophin gene in children with duchenne and becker muscular dystrophies. *Muscle Nerve* 117, 1–20.