

1. Dubowitz V, Fardeau M (1995) Proceedings of the 27th ENMC sponsored workshop on congenital muscular dystrophy, 22–24 April 1994, The Netherlands. Neuromuscul Disord 5:253–258.
2. Tome FM, Evangelista T, Leclerc A, Sunada Y, Manole E, Estournet B, Barois A, Campbell KP, Fardeau M (1994) Congenital muscular dystrophy with merosin deficiency. CR Acad Sci [III] 317:351–357.
3. Helbling-Leclerc A, Zhang X, Topaloglu H, Cruaud C, Tesson F, Weissenbach J, Tome FM, Schwartz K, Fardeau M, Tryggvason K, et al (1995) Mutations in the laminin alpha2-chain gene (*LAMA2*) cause merosin-deficient congenital muscular dystrophy. Nat Genet 11:216–218.
4. Allamand V, Guicheney P (2002) Merosin-deficient congenital muscular dystrophy, autosomal recessive (MDC1A, MIM#156225, *LAMA2* gene coding for alpha2 chain of laminin). Eur J Hum Genet 10:91–94.
5. Philpot J, Cowan F, Pennock J, Sewry C, Dubowitz V, Bydder G, Muntoni F (1999) Merosin-deficient congenital muscular dystrophy: the spectrum of brain involvement on magnetic resonance imaging. Neuromuscul Disord 9:81–85.
6. Pini A, Merlini L, Tome FM, Chevallay M, Gobbi G (1996) Merosin-negative congenital muscular dystrophy, occipital epilepsy with periodic spasms and focal cortical dysplasia. Report of three Italian cases in two families. Brain Dev 18:316–322.
7. Sunada Y, Edgar TS, Lotz BP, Rust RS, Campbell KP (1995) Merosin-negative congenital muscular dystrophy associated with extensive brain abnormalities. Neurology 45:2084–2089.
8. Yoshida A, Kobayashi K, Manya H, Taniguchi K, Kano H, Mizuno M, Inazu T, Mitsuhashi H, Takahashi S, Takeuchi M, Herrmann R, Straub V, Talim B, Voit T, Topaloglu H, Toda T, Endo T (2001) Muscular dystrophy and neuronal migration disorder caused by mutations in a glycosyltransferase, POMGnT1. Dev Cell 1:717–724.
9. Beltran-Valero de Bernabe D, Currier S, Steinbrecher A, Celli J, Beusekom E van, Zwaag B van der, Kayserili H, Merlini L, Chitayat D, Dobyns WB, Cormand B, Lehesjoki AE, Cruces J, Voit T, Walsh CA, Bokhoven H van, Brunner HG (2002) Mutations in O-mannosyltransferase gene *POMT1* give rise to the severe neuronal migration disorder Walker-Warburg syndrome. Am J Hum Genet 71:1033–1043.
10. Kobayashi K, Nakahori Y, Miyake M, Matsumura K, Kondo-Iida E, Nomura Y, Segawa M, Yoshioka M, Saito K, Osawa M, Hamano K, Sakakihara Y, Nonaka I, Nakagome Y, Kanazawa I, Nakamura Y, Tokunaga K, Toda T (1998) An ancient retrotransposon insertion causes Fukuyama-type congenital muscular dystrophy. Nature 394:388–392.
11. Brockington M, Blake DJ, Prandini P, Brown SC, Torelli S, Benson MA, Ponting CP, Estournet B, Romero NB, Mercuri E, Voit T, Sewry CA, Guicheney P, Muntoni F (2001) Mutations in the fukutin-related protein gene (*FKRP*) cause a form of congenital muscular dystrophy with secondary laminin alpha2 deficiency and abnormal glycosylation of alpha-dystroglycan. Am J Hum Genet 69:1198–1209.
12. Quijano-Roy S, Galan L, Ferreiro A, Cheliout-Heraut F, Gray F, Fardeau M, Barois A, Guicheney P, Romero NB, Estournet B (2002) Severe progressive form of congenital muscular dystrophy with calf pseudohypertrophy, macroglossia and respiratory insufficiency. Neuromuscul Disord 12:466–475.
13. Topaloglu H, Brockington M, Yuva Y, Talim B, Haliloglu G, Blake D, Torelli S, Brown SC, Muntoni F (2003) *FKRP* gene mutations cause congenital muscular dystrophy, mental retardation and cerebellar cysts. Neurology 60:988–992.
14. Kawazaki ES (1990) Sample preparation from blood, cells, and other fluids. In: Innis M, Gelfand D, Sninsky G, White T (eds) PCR protocols. A guide to methods and application. Academic Press, San Diego, pp 146–52.

15. Triki C, Louhichi N, Meziou M, Choyakh F, Kechaou MS, Jlidi R, Mhiri C, Fakhfakh F, Ayadi H (2003) Merosin-deficient congenital muscular dystrophy with mental retardation and cerebellar cysts in three Tunisian patients unlinked to the LAMA2, FCMD, MEB and CMD1B. *Neuromuscul Disord* 13:4–12
16. Brockington M, Yuva Y, Prandini P, Brown SC, Torelli S, Benson MA, Herrmann R, Anderson LV, Bashir R, Burgunder JM, Fallet S, Romero N, Fardeau M, Straub V, Storey G, Pollitt C, Richard I, Sewry CA, Bushby K, Voit T, Blake DJ, Muntoni F (2001) Mutations in the ukutin related-protein gene (FKRP) identifies limb girdle muscular dystrophy 2I as a milder allelic variant of congenital muscular dystrophy MDC1C. *Hum Mol Genet* 10:2851–2859
17. Mercuri E, Brockington M, Straub V, Quijano-Roy S, Yuva Y, Herrmann R, Brown SC, Torelli S, Dubowitz V, Blake DJ, Romero NB, Estournet B, Sewry CA, Guicheney P, Voit T, Muntoni F (2003) Phenotypic spectrum associated with mutations in the fukutin-related protein gene. *Ann Neurol* 53:537–542.
18. Quijano-Roy S, Romero NB, Louhichi N, Brockington M, Many H, Yeliz Y, Richard P, Estournet B, Muntoni F, Fardeau F, Barois A, Guicheney P (2002) FKRP (fukutin related protein) gene mutations associated with intermediate phenotype between CMD type 1C and LGMD2I (abstract). *Neuromuscul Disord* 12:743.
19. Muntoni F, Valero de Bernabe B, Bittner R, Blake D, Bokhoven H van, Brockington M, Brown S, Bushby K Campbell KP, Fiszman M, Gruenewald S, Merlini L, Quijano-Roy S, Romero N, Sabatelli P, Sewry CA, Straub V, Talim H, Topaloglu H, Voit T, Yurchenco PD, Urtizberea JA, Wewer U, Guicheney P (2004) 114th ENMC International Workshop on Congenital Muscular Dystrophy (CMD) 17-19 January 2003, Naarden, The Netherlands: (8th Workshop of the International Consortium on CMD; 3rd Workshop of the MYO-CLUSTER project GENRE). *Neuromuscul Disord*. 2003, 13:579–88.
20. Osawa M, Sumida S, Suzuki N, Arai Y, Ikenaka H, Murasugi H, Shishikura K, Suzuki H, Saito K, Fukuyama Y (1997) Fukuyama type congenital progressive muscular dystrophy. In: Fukujama Y, Osawa M, Saito K (eds) *Congenital muscular dystrophies*. Elsevier Science, pp 31–68.
21. Esapa CT, Benson MA, Schroder JE, Martin-Rendon E, Brockington M, Brown SC, Muntoni F, Kroger S, Blake DJ (2002) Functional requirements for fukutin-related protein in the Golgi apparatus. *Hum Mol Genet* 11:3319–3331.
22. Ervasti JM, Campbell KP (1993) A role for the dystrophin-glycoprotein complex as a transmenbrane linker between laminin and actin. *J Cell Biol* 122:809–823.
23. Michele DE, Barresi R, Kanagawa M, Saito F, Cohn RD, Satz JS, Dollar J, Nishino I, Kelley RI, Somer H, Straub V, Mathews KD, Moore SA, Campbell KP (2002) Post-translational disruption of dystroglycan-ligand interactions in congenital muscular dystrophies. *Nature* 418:417–422.
24. Moore SA, Saito F, Chen J, Michele DE, Henry MD, Messing A, Cohn RD, Ross-Barta SE, Westra S, Williamson RA, Hoshi T, Campbell KP (2002) Deletion of brain dystroglycan recapitulates aspects of congenital muscular dystrophy. *Nature* 418:422–425.