

References for *ACTA1*

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- Agrawal, P. B., C. D. Strickland, et al. (2004). "Heterogeneity of nemaline myopathy cases with skeletal muscle alpha-actin gene mutations." *Ann Neurol* **56**(1): 86-96.
- Brooke, M. (1973). Congenital fiber type disproportion. in Kakulas, BA (ed): *Clinical Studies in Myology*. Proc. of the 2nd Int. Cong. on Muscle Diseases, 1971, Perth, Australia, Experta Medica (pub): 147-59.
- Ilkovski, B., S. T. Cooper, et al. (2001). "Nemaline myopathy caused by mutations in the muscle alpha-skeletal-actin gene." *Am J Hum Genet* **68**(6): 1333-43.
- Ilkovski, B., K. J. Nowak, et al. (2004). "Evidence for a dominant-negative effect in ACTA1 nemaline myopathy caused by abnormal folding, aggregation and altered polymerization of mutant actin isoforms." *Hum Mol Genet* **13**(16): 1727-43.
- Kaindl, A. M., F. Ruschendorf, et al. (2004). "Missense mutations of ACTA1 cause dominant congenital myopathy with cores." *J Med Genet* **41**(11): 842-8.
- Laing, N. G., N. F. Clarke, et al. (2004). "Actin mutations are one cause of congenital fibre type disproportion." *Ann Neurol* **56**(5): 689-94.
- Laing, N. G., D. E. Dye, et al. (2009). "Mutations and polymorphisms of the skeletal muscle alpha-actin gene (ACTA1)." *Hum Mutat* **30**(9): 1267-77.
- McKusick, V. A. (2008). "102610 ACTIN, ALPHA, SKELETAL MUSCLE 1; ACTA1." OMIM, created 1986.
- McKusick, V. A. (2008). "161800 NEMALINE MYOPATHY 3; NEM3." OMIM, created 1986.
- McKusick, V. A. (2008). "255310 MYOPATHY, CONGENITAL, WITH FIBER-TYPE DISPROPORTION; CFTD." OMIM, created 1986.
- North, K. and M. M. Ryan (2006). "Nemaline Myopathy." GeneReviews, created 2002. <http://www.ncbi.nlm.nih.gov/bookshelf/br.fcgi?book=gene&part=nem>.
- Nowak, K. J., D. Wattanasirichaigoon, et al. (1999). "Mutations in the skeletal muscle alpha-actin gene in patients with actin myopathy and nemaline myopathy." *Nat Genet* **23**(2): 208-12.
- Ryan, M. M., C. Schnell, et al. (2001). "Nemaline myopathy: a clinical study of 143 cases." *Ann Neurol* **50**(3): 312-20.
- Ryan, M. M., B. Ilkovski, et al. (2003). "Clinical course correlates poorly with muscle pathology in nemaline myopathy." *Neurology* **60**(4): 665-73.
- Wallgren-Pettersson, C., K. Pelin, et al. (1999). "Clinical and genetic heterogeneity in autosomal recessive nemaline myopathy." *Neuromuscul Disord* **9**(8): 564-72.