

FSHD related bibliography

Wijmenga C, Padberg GW, Moerer P, Wiegant J, Liem L, Brouwer OF, Milner EC, Weber JL, van Ommen GB, Sandkuyt LA, et al. Mapping of facioscapulohumeral muscular dystrophy gene to chromosome 4q35-qter by multipoint linkage analysis and in situ hybridization. *Genomics*. 1991 Apr;9(4):570-5.

Wijmenga C, Hewitt JE, Sandkuijl LA, Clark LN, Wright TJ, Dauwerse HG, Gruter AM, Hofker MH, Moerer P, Williamson R, Van Ommen GJB, Padberg GW, Frants RR. Chromosome 4q DNA rearrangements associated with facioscapulohumeral muscular dystrophy. *Nat Genet* 1992 Sep;2(1):26-30.

Hewitt JE, Lyle R, Clark LN, Valleley EM, Wright TJ, Wijmenga C, van Deutekom JC, Francis F, Sharpe PT, Hofker M, et al. Analysis of the tandem repeat locus D4Z4 associated with facioscapulohumeral muscular dystrophy. *Hum Mol Genet*. 1994 Aug;3(8):1287-95.

Bakker E, Wijmenga C, Vossen RH, Padberg GW, Hewitt J, van der Wielen M, Rasmussen K, Frants RR. The FSHD-linked locus D4F104S1 (p13E-11) on 4q35 has a homologue on 10qter. *Muscle Nerve*. 1995;2:S39-44.

Bakker E, Van der Wielen MJ, Voorhoeve E, Ippel PF, Padberg GW, Frants RR, Wijmenga C. Diagnostic, predictive, and prenatal testing for facioscapulohumeral muscular dystrophy: diagnostic approach for sporadic and familial cases. *J Med Genet*. 1996 Jan;33(1):29-35.

van Deutekom JC, Bakker E, Lemmers RJ, van der Wielen MJ, Bik E, Hofker MH et al. Evidence for subtelomeric exchange of 3.3 kb tandemly repeated units between chromosomes 4q35 and 10q26: implications for genetic counselling and etiology of FSHD1. *Hum Mol Genet*. 1996 Dec;5(12):1997-2003.

Deidda G, Cacurri S, Piazza N, Felicetti L. Direct detection of 4q35 rearrangements implicated in facioscapulohumeral muscular dystrophy (FSHD). *J Med Genet* 1996;33:361-365.

Upadhyaya M, Maynard J, Rogers MT, et al. Improved molecular diagnosis of facioscapulohumeral muscular dystrophy (FSHD): validation of the differential double digestion for FSHD. *J Med Genet* 1997;34:476-479.

Lunt PW. 44th ENMC International Workshop on facioscapulohumeral muscular dystrophy: molecular studies, 19-21 July 1996, Naarden, The Netherlands, 1998:126-30.

Lemmers RJ, van der Maarel SM, van Deutekom JC, van der Wielen MJ, Deidda G, Dauwerse HG et al. Inter- and intrachromosomal sub-telomeric rearrangements on 4q35: implications for facioscapulohumeral muscular dystrophy (FSHD) aetiology and diagnosis. *Hum Mol Genet*. 1998 Aug;7(8):1207-1214.

van Overveld PG, Lemmers RJ, Deidda G, Sandkuijl L, Padberg GW, Frants RR et al. Interchromosomal repeat array interactions between chromosomes 4 and 10: a model for subtelomeric plasticity. *Hum Mol Genet.* 2000 Nov 22;9(19):2879-2884.

van der Maarel SM, Deidda G, Lemmers RJ, van Overveld PG, van der Wielen M, Hewitt JE et al. De novo facioscapulohumeral muscular dystrophy: frequent somatic mosaicism, sex-dependent phenotype, and the role of mitotic transchromosomal repeat interaction between chromosomes 4 and 10. *Am J Hum Genet.* 2000 Jan;66(1):26-35.

Lemmers RJL, de Kievit P, van Geel M, van der Wielen MJ, Bakker E, Padberg GW, Frants RR, van der Maarel SM. Complete allele information in the diagnosis of facioscapulohumeral muscular dystrophy by triple DNA analysis. *Ann Neurol.* 2001 Dec;50(6):816-9.

van Geel M, Dickson MC, Beck AF, Bolland DJ, Frants RR, van der Maarel SM, de Jong PJ, Hewitt JE. Genomic analysis of human chromosome 10q and 4q telomeres suggests a common origin. *Genomics.* 2002 Feb;79(2):210-7.

Lemmers RJ, de Kievit P, Sandkuijl L, Padberg GW, van Ommen GJ, Frants RR et al. Facioscapulohumeral muscular dystrophy is uniquely associated with one of the two variants of the 4q subtelomere. *Nat Genet.* 2002 Oct;32(2):235-236.

Butz M, Koch MC, Müller-Felber W, Lemmers RJ, van der Maarel SM, Schreiber H. Facioscapulohumeral muscular dystrophy. Phenotype-genotype correlation in patients with borderline D4Z4 repeat numbers. *J Neurol.* 2003 Aug;250(8):932-7.

Lemmers RJ, Osborn M, Haaf T, Rogers M, Frants RR, Padberg GW, Cooper DN, van der Maarel SM, Upadhyaya M. D4F104S1 deletion in facioscapulohumeral muscular dystrophy: phenotype, size, and detection. *Neurology.* 2003 Jul 22;61(2):178-83.

van Overveld PG, Lemmers RJ, Sandkuijl LA, Enthoven L, Winokur ST, Bakels F et al. Hypomethylation of D4Z4 in 4q-linked and non-4q-linked facioscapulohumeral muscular dystrophy. *Nat Genet.* 2003 Dec;35(4):315-317.

Lemmers RJ, van der Wielen MJ, Bakker E, Padberg GW, Frants RR, van der Maarel SM. Somatic mosaicism in DFEH often goes undetected. *Ann Neurol.* 2004 Jun;55(6):845-850.

van Overveld PG, Enthoven L, Ricci E, Rossi M, Felicetti L, Jeanpierre M, Winokur ST, Frants RR, Padberg GW, van der Maarel SM. Variable hypomethylation of D4Z4 in facioscapulohumeral muscular dystrophy. *Ann Neurol.* 2005 Oct;58(4):569-76.

Lemmers RJ, Wohlgemuth M, van der Gaag KJ, van der Vliet PJ, van Teijlingen CM, de Knijff P, Padberg GW, Frants RR, van der Maarel SM. Specific sequence variations within the 4q35 region are associated with facioscapulohumeral muscular dystrophy. *Am J Hum Genet.* 2007 Nov;81(5):884-94.

Ehrlich M, Jackson K, Tsumagari K, Camano P, Lemmers RJ. Hybridization analysis of D4Z4 repeat arrays linked to FSHD. Chromosoma 2007 Apr;116(2):107-16.

de Greef JC, Lemmers RJ, van Engelen BG, Sacconi S, Venance SL, Frants RR et al. Common epigenetic changes of D4Z4 in contraction-dependent and contraction-independent FSHD. Hum Mutat. 2009 Oct;30(10):1449-1459.

Lemmers RJ, van der Vliet PJ, Klooster R, Sacconi S, Camañ P, Dauwerse JG, Snider L, Straasheijm KR, van Ommen GJ, Padberg GW, Miller DG, Tapscott SJ, Tawil R, Frants RR, van der Maarel SM. A unifying genetic model for facioscapulohumeral muscular dystrophy. Science. 2010 Sep 24;329(5999):1650-3.

de Greef JC, Lemmers RJ, Camañ P, Day JW, Sacconi S, Dunand M, van Engelen BG, Kiuru-Enari S, Padberg GW, Rosa AL, Desnuelle C, Spuler S, Tarnopolsky M, Venance SL, Frants RR, van der Maarel SM, Tawil R. The clinical features of facioscapulohumeral dystrophy 2. Neurology 2010 Oct 26;75(17):1548-54

Lemmers RJ, van der Vliet PJ, van der Gaag KJ, Zuniga S, Frants RR, de Knijff P, van der Maarel SM. Worldwide population analysis of the 4q and 10q subtelomeres identifies only four discrete interchromosomal sequence transfers in human evolution. Am J Hum Genet. 2010 Mar 12;86(3):364-77.

Snider L, Geng LN, Lemmers RJ, Kyba M, Ware CB, Nelson AM, Tawil R, Filippova GN, van der Maarel SM, Tapscott SJ, Miller DG. Facioscapulohumeral dystrophy: incomplete suppression of a retrotransposed gene. PLoS Genet. 2010 Oct 28;6(10):e1001181

Lemmers RJ, Tawil R, Petek LM, Balog J, Block GJ, Santen GW, Amell AM, van der Vliet PJ, Almomani R, Straasheijm KR, Krom YD, Klooster R, Sun Y, den Dunnen JT, Helmer Q, Donlin-Smith CM, Padberg GW, van Engelen BG, de Greef JC, Aartsma-Rus AM, Frants RR, de Visser M, Desnuelle C, Sacconi S, Filippova GN, Bakker B, Bamshad MJ, Tapscott SJ, Miller DG, van der Maarel SM. Digenic inheritance of an SMCHD1 mutation and an FSHD-permissive D4Z4 allele causes facioscapulohumeral muscular dystrophy type 2. Nat Genet. 2012 Dec;44(12):1370-4.

Sacconi S, Camañ P, de Greef JC, Lemmers RJ, Salviati L, Boileau P, Lopez de Munain Arregui A, van der Maarel SM, Desnuelle C. Patients with a phenotype consistent with facioscapulohumeral muscular dystrophy display genetic and epigenetic heterogeneity. J Med Genet. 2012 Jan;49(1):41-6.

Lemmers RJ, Goeman JJ, van der Vliet PJ, van Nieuwenhuizen MP, Balog J, Vos-Versteeg M, Camano P, Ramos Arroyo MA, Jerico I, Rogers MT, Miller DG, Upadhyaya M, Verschuren JJGM, Lopez de Munain A, BGM van Engelen BGM, Padberg GW, Sacconi S, Tawil R, Tapscott SJ, Bakker B, van der Maarel SM. Inter-individual Differences in CpG Methylation at D4Z4 Correlate with Clinical Variability in FSHD1 and FSHD2. Hum Mol Genet. 2015 Feb 1;24(3):659-69.

van den Boogaard M, Lemmers RJ, Camano P, van der Vliet PJ, Voermans N, van Engelen BG, López de Munain A, Tapscott SJ, van der Stoep N, Tawil R, van der Maarel SM. Double SMCHD1 variants in FSHD2: the synergistic effect of two SMCHD1 mutations on D4Z4 hypomethylation and disease penetrance in FSHD2. Eur J Hum Genet. 2016 Jan;24(1):78-85