

Study (first author, year)	Experimental type	Array platform	Number of targets/array density	Sample type	Patients/sites	Controls	Number of dysregulated genes with determined gene symbol / single nucleotide polymorphism (SNP)	Analysis
Pan, 2003	mRNA expression	Human Atlas 1.2 array kit	1,176 genes	Tissue biopsy	6 disease fascia	2 external control fascia (pooled into one sample)	22 genes	Consistent over- or under-expression in all disease tissues compared with control tissues
Qian, 2004	mRNA expression	Human Atlas 1.2 array kit	1,176 genes	Tissue biopsy	9 disease fascia	9 internal control fascia	17 genes	Fold change
Lee, 2006	mRNA expression	cDNA Chips (Stanford Functional Genomics Facility)	42,000 elements (30,000 unique genes)	Tissue biopsy	4 cord	4 internal control fascia 3 external control fascia	16 genes	Online software from Stanford Microarray Database.
Reham, 2008	mRNA expression	Affymetrix U133A GeneChip	approx. 22,000 probe (approx. 18,400 transcripts)	Tissue biopsy	4 cord 4 nodule	4 internal control fascia 4 external control fascia	72 genes (pairwise analysis study); 67 genes (network diagram)	GeneSifter microarray analysis tool. Stanford Microarray Database software.
Satish, 2008	mRNA expression	GE Code Link Human Whole Genome Bioarray System	approx. 57,000 transcripts	Fibroblast culture	6 cord	6 control fascia external	40 genes	Significance Analysis of Microarrays (SAM) software package.
		Illumina Sentrix Human-6 Expression Beadchip	approx. 48,000 transcript				40 genes	Significance Analysis of Microarrays (SAM) software package.
Zhang, 2008	mRNA expression	cDNA chips (Stanford Functional Genomics Facility)	42,000 elements (30,000 unique genes)	Tissue biopsy	4 cord	4 internal control fascia 3 external control fascia	29 genes	Online software from Stanford Microarray Database.
Forsman, 2008	mRNA expression	Affymetrix HGU133A array	over 22,000 probe sets (over 14,500 genes)	Tissue biopsy	Two randomized pools (each pool contained 6 disease tissue)	1 pool (contain three external controls)	2 genes	Analysis performed according to the Affymetrix GeneChip Expression Analysis Technical Manual.
Vi , 2009	mRNA expression	Affymetrix Human Genome U133 Plus 2.0 Array	approx. 54,000 probe (approx. 47,000 transcripts and variants)	Tissue biopsy	2 cord 1 nodule	2 internal control fascia 1 external control fascia	7 genes	Significance Analysis of Microarrays (SAM) software package.
Mosakhani, 2010	microRNA expression	Agilent miRNA Microarray system (V2)	723 human and 76 human viral miRNA	Tissue biopsy and fibroblast culture	29 cord	4 external control fascia (biopsy) 1 external (fibroblast culture)	8 genes	GeneSpring GX Analysis Software v 10
Kraljevic Pavelic, 2009	Protein expression	Two-dimensional gel electrophoresis and mass	n/a	Tissue biopsy	Two pools (each pool contained	2 pools (each pool contained 6 internal control)	44 genes	Five-fold changes in all gels from diseased tissue relative

		spectrometry analysis.			6 disease tissue)			to unaffected tissue
Hu, 2005	Genome-wide association	Microsatellite-based genome-wide scan (ABI apel)	Not specified	Blood	12	13 external	1 gene loci	MLINK (two-point linkage analysis)
Ojwang, 2010	Genome-wide association	Illumina HumanCytoSNP-12 array	Approximately 300,000 SNPs	Saliva	40	40	14 SNP	PLINK and ADMIXMAP program/software (MALD method)
Dolmans, 2011	Genome-wide association	Illumina HumanCytoSNP-12 (initial test for association); Affymetrix 6.0, Illumina 1.2M, Immunochip, GenomeLab SNP stream, and KASP by Design assays were used in tests for replication	301,232 SNPs (initial test for association)	Blood or saliva	856 (inintial genome-wide association); additional 1298 cases were included in test for replication.	2836 (initial typing); additional 15410 cases were included in test for replication.	11 SNP	Genome Studio
Kaur, 2008	Copy number variation	Agilent 44K oligonucleotide-based array comparative genomic hybridization	Approximately 44,000 probes	Tissue biopsy or blood	18	Pooled external control (blood)	No copy number changes found	Agilent CGH Analytics software
Shih, 2010	Copy number variation	Agilent 1M oligonucleotide-based array comparative genomic hybridization	Approximately 1,000,000 probes	Tissue biopsy or blood	4 nodule	Pooled external control (blood)	8 loci (none confirmed by downstream analysis)	Agilent CGH Analytics software
Shih (in press)	Copy number variation	Agilent 1M oligonucleotide-based array comparative genomic hybridization and NimbleGen 2.1M comparative genomic hybridization	Approximately 1,000,000 probes (Agilent) and 2,100,000 (NimbleGen)	Tissue biopsy or blood	8 nodule (4 from previously published data) vs. external control; 4 nodule vs. internal control	Pooled external control (blood) and 4 blood (internal control)	5 loci (2 confirmed by downstream analysis)	Nexus Copy Number

Online supplementary Table S1. Summary of experimental parameters in gene expression microarray platforms

Gene Symbol	Meta-analysis			Published gene-expression microarray studies				Whole proteome profiling [21]	microRNA profiling [13]*	Fibromatosis (regulation from Gene Expression Atlas)
	Percentage of false positive predictions	Regulation	Fold change	Reference	P-value or false positive rate	Regulation	Fold change			
ACTA2	0.015	Up	2.3	[6, 12]	>0.05	Up	1.4 to 1.8	-		Up
ADAM12	0.000	Up	2.9	[6, 9]	0.001 to 0.002, n/a	Up	4, n/a	-	miR-301a, miR-26a, miR-204, miR-1, miR-101, miR-498	Up
ADH1B	0.000	Down	-4.1	[9, 10]	0.003 to 0.052	Down	-3 to -5	-		-
AKR1C2	0.003	Down	-1.9	[7, 10]	n/a, <0.05	Down	-2.3 to -11.8	-		-
ALDH1A1	0.003	Down	-2.2	[9]	0.002 to 0.59	Down	-2 to -4	-		-
ALDH2	0.015	Down	-1.7	[7, 10]	n/a, <0.05	Down	-1.7 to -22.5	-		-
ANGPTL7	0.000	Down	-1.9	[9, 10]	0.033, <0.05	Down	-2.5 to -4	-		-
APOD	0.039	Down	-1.3	[9]	0.009	Down	-4	-		-
AR	0.000	Down	-2.1	[10]	<0.05	Down	-2.9	-		-
BASP1	0.013	Up	1.9	[12]	n/a	Up	n/a	-	miR-539	Up
CD79B	0.000	Down	-4.2	[10]	<0.05	Down	-5.6	-		-
CDO1	0.000	Down	-2.2	[9-11]	0.023, <0.05,, <0.1	Down	-2 to -4.8	-		Up
CHI3L2	0.000	Down	-2.3	[10]	<0.05	Down	-4.5	-		-
CLU	0.000	Down	-4.6	[9, 10]	0.015 to 0.040, <0.05	Down	-3 to -5	-	miR-1236	-
COL15A1	0.000	Down	-2.5	[10]	<0.05	Down	-3	-	miR-505	Up
COL1A1	0.042	Up	1.9	[9, 12]	0.002 to 0.052,, <0.05	Up	2 to 4.4 , n/a	-	miR-133a, miR-338-3p, miR-382, miR-498	Up
COL4A2	0.009	Up	2.4	[8, 9]	0.001, n/a	Up	3 to 10.4	-		Up
COL5A1	0.000	Up	2.7	[6, 9]	0.002 to 0.052, n/a	Up	2 to 4, n/a	-	miR-135a, miR-143	Up
COL5A2	0.000	Up	2.9	[6, 9, 12]	0.002 to 0.049,	Up	2 to 4, n/a	-		Up

					n/a					
CRTAC1	0.000	Down	-2.8	[9]	0.050	Down	-2	-		-
CSF1	0.028	Down	-1.9	[8]	n/a	Up	6.4	-	miR-205	-
CSR2P	0.000	Up	2.5	[9]	0.002 to 0.033	Up	2 to 4	-	miR-101	Up
CYP4B1	0.000	Down	-3.6	[9]	0.032 to 0.052	Down	-2 to -3	-		-
DDX3Y	0.000	Up	7.1	[10]	<0.05	Up	6.3	-	miR-101, miR-135a	-
FAP	0.025	Up	2	[12]	n/a	Up	n/a	-		Up
FBLN1	0.002	Down	-2.2	[10]	<0.05	Down	-4 to -4.2	-		Up
FBXO2	0.000	Down	-2	[10]	<0.05	Down	-2.6 to -4.8	-		-
FRZB	0.003	Down	-1.5	[9]	0.001 to 0.023	Down	-2 to -3	-		-
GLUL	0.044	Down	-1.5	[9]	0.059	Down	-2	-	miR-140-5p	Down
HPD	0.047	Down	-1.5	[10]	<0.05	Down	-3	-	miR-654-5p	-
HSD11B1	0.004	Down	-2	[10]	<0.05	Down	-3 to -3.6	-		-
KDM5D	0.000	Up	7.4	[10]	<0.05	Up	8.13 to 9.4	-		Up
LRRC17	0.000	Up	4.4	[6, 9, 12]	0.001 to 0.003, n/a	Up	3 to 4, n/a	-		Up
MAFB	0.022	Up	1.6	[6, 12]	n/a	Up	n/a	-	miR-223, miR-338-3p, miR-365, miR-631	Up
MARCKS	0.000	Up	2.3	[9]	0.011	Up	3	-	miR-1237, miR-140-3p, miR-143, miR-30b, miR-563, miR-634, miR-766, miR-205, miR-23a, miR-30b, miR-34c-3p, miR-584, miR-648	Up
MAASP1	0.000	Down	-2.7	[10]	<0.05	Down	-4.5	-	miR-648	-
MMP2	0.012	Up	1.8	[8, 9, 12]	0.042, n/a	Up	2 to 29, n/a	-		Up
MXRA5	0.010	Up	1.8	[6, 12]	n/a	Up	n/a	-		Up
MYOC	0.000	Down	-2.1	[9]	0.001 to 0.052	Down	-2 to -5	-		-
NCAM1	0.000	Up	2.7	[12]	n/a	Up	n/a	-	miR-30b	-
NT5DC2	0.007	Up	2	[9]	0.001 to 0.006	Up	3 to 4	-		Up

PCOLCE2	0.000	Down	-2.3	[9]	0.002 to 0.023	Down	-3 to -6	-		-
PDPN	0.007	Up	2	[12]	n/a	Up	n/a	-	<i>miR-934</i>	Up
POSTN	0.000	Up	5	[8, 9, 11]	0.002 to 0.055, <0.1	Up	3 to 62.79	-		Up
PPAP2B	0.000	Down	-2.3	[9]	0.052 to 0.054	Down	-2	-	<i>miR-140-3p, miR-149, miR-363, miR-936</i>	Up
PPL	0.000	Down	-2.6	[9]	0.005 to 0.026	Down	-2 to -4	-		-
PRC1	0.006	Up	2	[12]	n/a	Up	n/a	-		Down
PTGER2	0.020	Down	-1.8	[10]	<0.05	Down	-3.8	-	<i>miR-519e</i>	-
PTPRD	0.000	Up	3.2	[10]	<0.05	Up	2.94	-	<i>miR-1, miR-1228, miR-1237, miR-133a, miR-135a, miR-204</i>	Up
PTPRN2	0.000	Down	-4.5	[10]	<0.05	Down	-6.3	-		-
RAB31	0.022	Up	2	[12]	n/a	Up	n/a	-	<i>miR-129-3p</i>	Up
RGS16	0.046	Down	-1.3	[10]	<0.05	Down	-2	-	<i>miR-1236, miR-204</i>	-
RGS3	0.001	Up	2.5	[9, 12]	0.003, n/a	Up	3, n/a	-	<i>miR-1237, miR-126, miR-133a, miR-22, miR-296-5p, miR-382</i>	Up
RPS4Y1	0.000	Up	15.3	[10]	<0.05	Up	6.1	-		-
SEPP1	0.026	Down	-1.4	[10]	<0.05	Down	-2.9 to -4.3	-		Up
SGK1	0.017	Down	-1.5	[10]	<0.05	Down	-2 to -2.8	-	<i>miR-101, miR-133a, miR-365, miR-23a, miR-382</i>	-
SOD2	0.003	Down	-2	[10]	<0.05	Down	-1.8 to -2.2	-		Down
TGFB2	0.009	Up	2.6	[12]	n/a	Up	n/a	-	<i>miR-133a, miR-23a, miR-584</i>	Up
THBS2	0.007	Up	2.2	[12]	n/a	Up	n/a	-		Up
THY1	0.023	Up	2	[9]	0.001	Up	3	-		Up
TMEM15	0.007	Up	2	[12]	n/a	Up	n/a	-		-
TNC	0.007	Up	1.9	[6, 9, 12]	0.001 to 0.053, n/a	Up	2 to 5.2, n/a	-		Up

TXNIP	0.012	Down	-1.8	[9]	0.053	Down	-3	-	<i>miR-135a</i>	Down
USP9Y	0.000	Up	6.3	[10]	<0.05	Up	5.76	-		-
ZFY	0.000	Up	5.3	[10]	<0.05	Up	8.9	-	<i>miR-498</i>	-
COL6A3	0.008	Up	2.7	-	-	-	-	Up	<i>miR-130b</i>	Up
MFAP4	0.000	Down	-2.5	-	-	-	-	Up		Up
MYL3	0.015	Down	-1.5	-	-	-	-	Up		-
FABP4	0.019	Down	-1.2	-	-	-	-	Up		-
ACTG1	-	-	-	-	-	-	-	Up		-
ARHGDI1A	-	-	-	[8]	n/a	Up	3.5	Up	<i>hsa-miR-133a</i>	Down
HSPA8	-	-	-	-	-	-	-	Up	<i>miR-365, miR-4295, miR-4465, miR-519d</i>	Down
IDH1	-	-	-	-	-	-	-	Up	<i>miR-133a, miR-30a, miR-367, miR-23a, miR-30a</i>	-
LGALS1	-	-	-	-	-	-	-	Up	<i>miR-22</i>	Up
PCBP1	-	-	-	-	-	-	-	Up	<i>miR-338-3p</i>	-
PGK1	-	-	-	-	-	-	-	Up	<i>miR-1229, miR-143, miR-365</i>	Down
TAGLN	-	-	-	-	-	-	-	Up	<i>miR-22, miR-539</i>	Up
TGFBI	-	-	-	-	-	-	-	Up	<i>miR-766</i>	-
TPM4	-	-	-	[11]	n/a	Up	3.24 to 4.13	Up	<i>miR-10a, miR-133a, miR-143, miR-30b, miR-32, miR-338-3p, miR-613, miR-648, miR-617, miR-30b</i>	Up
CALR	-	-	-	-	-	-	-	Up	<i>miR-181a</i>	-
VCP	-	-	-	-	-	-	-	Up	<i>miR-23a</i>	Down

Online supplementary Table S2. Comparisons made between genes reported to be dysregulated in the meta-analysis of this study, whole-proteome study, microRNA study and fibromatosis regulation determined in European Bioinformatics Institute Gene Expression Atlas

Italic font indicate the regulation is inconsistent with those reported by other studies, using the first column in the left as the basis for comparison.

* The column indicates that the gene is a predicted target of a microRNA suggested to be dysregulated in DD. Red indicates the microRNA is preferentially expressed in DD, blue indicates the microRNA is down-regulated in DD. MicroRNA negatively regulates its target mRNA. Italic indicates the microRNA up-/down-regulation is inconsistent with the mRNA (or protein, if there has not been mRNA report of the gene) regulation reported in DD.

Signalling pathway	Gene Ontology term	Accession	Genes dysregulated in DD from this term
Beta-catenin and Wnt signalling pathway	Wnt receptor signalling pathway	GO: 0016055	COL1A1, COL18A1, CXXC4, DKK1, DKK2, EDIL3, FRZB, GNB2L1, GPC3, LEF1, NDRG2, PPAP2B, PTK7, RGS3, ROR2, RSPO1, RSPO2, SFRP1, SFRP4, SMO, SOX4, SULF1, WNT2, WNT4, WNT5A, WNT7B, ZIC1
	Regulation of Wnt receptor signalling pathway	GO:0030111	COL1A1, CXXC4, DKK1, DKK2, EDIL3, FRZB, GNB2L1, GPC3, LEF1, PPAP2B, RGS3, ROR2, RSPO1, RSPO2, SFRP1, SFRP4, SOX4, SULF1, WNT2, WNT4, WNT5A, ZIC1
	Beta-catenin binding	GO:0008013	AR, CTNNA1, LEF1
Akt signalling pathway	Protein kinase B signalling cascade	GO:0043491	AKR1C2, AKR1C3, ARRB2, C1QTNF1, CCL19, CCL5, IGF2
	Regulation of protein kinase B signalling cascade	GO:0051896	AKR1C2, AKR1C3, ARRB2, C1QTNF1, CCL19, IGF2
	Protein kinase B binding	GO:0043422	ARRB2
TGF-beta signalling pathway	Cellular response to transforming growth factor beta stimulus	GO:0071560	CCL5, CLEC3B, COL1A1, COL4A2, PDE3A, SFRP1, WNT2, WNT4, WNT5A
	Transforming growth factor beta binding	GO:0050431	CD36, TGFB3
	Regulation of transforming growth factor beta production	GO:0071634	PTGS2, TGFB2
	Regulation of transforming growth factor beta receptor signalling pathway	GO:0017015	ASPN, C5orf13, SULF1, TGFB3
	Transforming growth factor beta receptor signalling pathway	GO:007179	ARRB2, ASPN, C5orf13, COL1A2, FOS, GDF10, SULF1, TGFB2, TGFB3, USP9Y

Online supplementary Table S3. Genes reported to be dysregulated in global gene expression studies and involved in signalling pathways suggested to be involved in Dupuytren's disease