

**Supplementary Table II MLPA probes.**

Probe name	Cytoband	Size (bp)	5' half-probe <sup>a</sup>	3' half-probe <sup>b</sup>	Comment	SD
GABRA	4p12	84	CAGCCTGTTGCATAACCATCG	AGCAAAGTTCAGGATGCG	Pilot	
RELN2	7q22.1	87	CAGCATTACGGAATGAAGGTCA	CCACAAGAAGTGGCTTACAACC	Control	0.04
KRTAP2-1	17q21.2	93	CCCGGAACGCGTAACTCACCTT	CTGAAAAAGTCACACTCCCATTCTCTAC	A	0.05
UQCRQ	5q31.1	93	CACAGTCACCGATCCGGTTGACTCGTT	CGTCATTTCCGGCAAGGTTGGAAG	GDF9	0.08
C5IGS1	5q31.1	96	GCTTTTCCACATGCCTTAGAAGTTTCAGC	AAGCACCTAGCGAACATCTTAATCC	GDF9	0.04
SH3GL3	15q25.2	96	GAACCTGAGGCCAGCTCTAACAGTCTT	TCATGGATGCATACACCTGCCTGCAG	A	0
C2ORF67	2q34	99	CAAATCGTCATCAGAGCTGCTATCAGTTGCA	TCGGAATCCAAACCCTCTTCAACATG	A	0.03
C5IGS2	5q35.1	99	GTAGACAAAGGAGGTGGTGCAAGAGCGCAA	ACTACTTCATGGGGAGGACGTCTCAG	GDF9	0.05
CXIGS1	Xp22.33	99	CAAAATTAAGTGTAGGATTCATTTTCTGC	ATCTACACATCACTGGCATAGGCTCTG		
DAX1	Xp21.2	102	GCAGCCTCAGCGGCCTGTTGAAGACGCTG	CGCTTCGTCAGTACTTGCCCTGCTTCCAG	Control	nd
KRTAP2-3	17q21.2	102	GATCCTCAAGCACGAATCAACAATAACAT	CCTATCTATTTCCCCACATAATTATGCAGC	A	0.03
PCLN1 <sup>c</sup>	3q28	105	GACACAAGGSGTGTAAATGCACG	TTTCAGGGTGTGTTGCATATGATTTAATCAATCAGTATG	Control	0.04
CACNA1C_3	12p13.33	108	CTCCTTCAGGCTTGATTTGAGAAGGACTGGGGC	AGTTTGCTCAAGATCCCAAGTTCATCGAGGTC		
DNAH6	2p11.2	108	GTTTCCTGAGTGACTTCCACCAGCTGTAAGC	AAACTGCATCAAGCATTGTAGAAGCCTCAGTTG	A	0.03
GDF9_exon2-1	5q31.1	108	CCTTCCATCAGTGGAACTGCTATTTAACCTGGAT	CGCATTACTACCGTTGAACACTTACTCAAGTC	GDF9	0.09
GDF9_exon1-1	5q31.1	111	GCAGCTGATAACCTTATTTAGCCAATTT	GTTAATTAGATACAGATTTACTTGGTTATTTAGCTTTCC	GDF9	0.04
KRTAP2-4	17q21.2	111	GTGGACTTTCGAAACACATTTACAACCTGATGG	TTTTTACTGCTCAAAGTGCACCATCATCTTTTGC	A	0.04
SH3GL3_2	15q25.2	111	CTTTGCTGGGATAATATGCAATCAAGCTAATAGGTGCC	TTACCTGGACCTCCCAACAGGTAATAAATGG		
SMARCC1	3p21.31	111	CCTGCCAAAGGTGATCAGAGTCGATCAGTTGA	CCTTGGGGAAGATAATGTGACAGAGCAGACCAATCAC		
C5IGS5	5q31.1	114	GTATACTAATTCTCCTTGTAACTAGCATATATAAATCAATCG	AGTCACACAGAACAATATAACATTTGTCC	GDF9	0.08
TJP3	19p13.3	114	CCCGTGGCCGACATTGCTATGCAGAAGTTGACT	GCTGAGATGCCTGACCAGTTTGAATCGCAGGTGAGAAG	A	0.02
C2IGS12	2q34	117	GAGTGGAGACCACCTGGACAGTGACAGAGATCACTCATT	CCCCAGAAAGGTAGCATTATGACTTCAATCCCATC		
C2IGS13	2p16.2	117	CTCTATTGCTATCCTCCATGGCCTGGAGAACGAATGCA	GGCCTCTTTCTCTTCTACCTATGCAAGATAAGAATG		
GDF9_exon1-2	5q31.1	117	GCCTCCTTCCCGCCTTTTCAAAG	TTCTATCTGTTGGGCGAGGTGGGTACCTAGGCTGCAGC CAGACTCCAGAG	GDF9	0.03
P2RY8	Xp22.33	117	GCTACTACCACGTGTACAAGCTCACGCTGTGTCTCA	GCTGCCTCAACAAGTGTCTGGACCCGTTTGTATTAC	A	0.03
KRTAP2-2	17q21.2	120	CATACTAGCTCACACTATGCATTGAAGACACCTTTT	CAGACCAACCACAGATGAGAAATCTTTTCTAGGACTCCAG	A	0.02
SRY	Yp11.31	120	GAATGCGAAAAGTCAAGATCAGCAAGCAGCTGGGA	TACCAGTGGAAAATGCTTACTGAAGCCGAAAAATGGCCATTCC	Control	nd
C15IGS2	15q26.2	123	GGAATATCCCATCGTTACTTAGGTGTCAATATGTGTAT	CAGATGACTGGATTAGTATTTTAAACAGCAATGGCAAAGAAG		
CSPG5	3p21.31	123	GCCTGCTTACTTATACGATGATTTACCCCCCTTCGATGAATCT	GATTTCACCCACCACATCCTTTTATGATGACTTGG	A	0.03
D.NAH6_2	2p11.2	123	GTCACACCTACACCCTGCAGGAATTTAA	GGCCGCACAAGTCATACGGCTAGCAGAGGTAACAAATTTTGTCT TATAAGAATC		
GDF9_exon2-2	5q31.1	123	CTTCCCATCACCGTCACCGCAGAGGTGAGGAACTGTCAGTT	CTGAATTGAAGAAGCCCTTGGGCCAGCTTCTTCAATC	GDF9	0.02
C5IGS4	5q31.1	126	CCCATTGCTCAAGGTCTTACATACAAAGCAAGAGATGGCTT	CTAAGGGTTCTAACCACTCTGATGTTTTAAGCATGTGTC	GDF9	0.04
KRTAP2-3_2	17q21.2	126	GTGGAGTCTCATCTGATCCAGAAGGGGTAGAAGAGTCTGC	ACAAGCTTCTGTGCACCTGGGAATGTTTCGTGCGTTGAGAGG		
TSPYL6	2p16.2	126	GTGGTTGAGGGGACATGTCAGACCCTATCCTCAT	GGCAAGGTTAATATGTTGGTGTCTTGACAAGTACCAGGCTTT CTCAGCC	A	0.03

RBI ex23	13q14.2	129	GTCACCAATACCTCACATTCTCGAAGCCCTTACAAGTTTCCT	AGTTCACCCTTACGGATTCTGGAGGGAACATCTATATTTACC	Control	0.03
CREBBP	16p13.3	131	CCAGACCCACCCAGGCCCTCCTCAATAGTAACTCTGGCCATAGCTTAAT	TAATCAGGCTTCACAAGGGCAGGCGCAAGTCATGAATGGATC	Control	0.03
CACNA1C	12p13.3	132	CTGTGTGCATATGTCCTGCCCGTGTATATGCACCCACACCATGTGCCCG	TGCACACCAGTGACTACGCAGTCCCCCCTTTCTGGTTTAGC	A	0.03
C15IGS1	15q26.2	135	CCTACTTTTTCTGACACTGTGCTATGTTTTGAATTCTGTAGGAATCC	TTGGCGGAAGTTGCCTTCAGCATTTTAGGCCTGTCTAAACAGGTAG		
CACNA1C_2	12p13.33	135	GATGATTTGTAACACACAGTCTGTGCAGAAAGATCCCCTTCAGGAGG	TGTCTCCAGCATCCCAAAGCTGTGCGCACCTTCTCTTTTCCTGC		

Bp, basepair; SD, Standard deviation in probe validation; A, included in an MLPA kit for screening of array-CGH findings; GDF9, included in the MLPA kit for *GDF9* analysis; nd, not determined.

<sup>a</sup>The 5' half-probes are preceded by the universal tag sequence GGGTTCCTAAGGGTTGGA.

<sup>b</sup>The 3' half-probes are followed by the universal tag sequence TCTAGATTGGATCTTGTGGCAC.

<sup>c</sup>From Stern *et al.* (2004).