

Table S2. Primer Sequences for qPCR Assays

Proband	Chromosome Abnormality	Primer Set	Forward Primer (5'-3')	Reverse Primer (5'-3')
8293	Dup (1)(p36.11p35.3)	qNUDC_1	GTAAGAACCGTCGCAAATATCGTG	TCCCCTACCACACACATTCCG
		qNUDC_2	TGATAGGAGGGAAGGGGTAAC	CTGGCATTGTTTTCTGTAGGT
		qNUDC_3	GATAGCTGTTTGCCTTTTCAGATG	CCTCTGCACTGCATATTTTCTCAT
		qSLC9A1_1	AGCCAGGGGTTTTCAAATGTT	CCAGAATGTGCTGCCTTTTTAAGT
		qEYA3_1	CAAGGGGAAGGCTCCTCATT	CACCAGGCTTTAGAGCTTGATTT
		qEYA3-EX8	GTTTGAGATGCTGGCTACTGC	CACAAATGCCAGCCTGATATCT
		qEYA3-EX7	ATAATGTGCTGGGCTTGGC	TCCATCTCCAAGTCAACACAGT
9148	Dup (2)(p22.1p16.1)	q9148_1	ATGCACCCAGCCACTGCTAG	TTATCTGCTCCCAGCCAAGTG
		q9148_2	TGAAGCTCTGCTGCTTGTATTATG	GACTTCCCAGAGTTCCTGTCTTAT
		q9148_3	ATTTTCCCAGGGACTTAGATTTT	CTGTCAAGTGGTAAAAGGGTTCAG
		q9148_4	CAGTGGGCAAATGGAGAAA	TGCTGAACAATATTCCATTGTGA
		q9148_5	GATTCCCCTCTTCCAAGTAACAT	AGGGTCCAAAGGGCTCAATAT
		q9148_6	CCATTCTCTGAATCCACAGAAA	AAGTTTCCAGGGGTGTTCATAC
8772	Mosaic Del (2)(p11.2p11.2)	q8772_LCR1	ACCCCACTACATATCCCCAAGAT	AAATGACTTCCTAGGGAAGGAGC
		q8772_LCR3	TTTAATGTTGGTGTGAACAGTGG	CGTAGGGGCTACAAGATGCTATTT
		q8772_LCR5	CAGGGCCTCGAGCCACTT	ACCGTGCCTGTGTGCTGG
		q8772_LCR7	CCCCTGCCTGTGGCACCT	CCATGGAGAGGGCAGCTGG
		qCD8A_1	TGCGCACAACAGTATTGTGAC	TGACGGGAAACAAAAGTAGGTAA
		qCD8B_1	GGTCTTTAGGATTTTGGGCAAATT	CCCTTGTGGAACCAAATGTATC
		qMGC4677_1	CGTAGGAAGGCTTGGGGTTAA	ACTTACATAAGGGCGGCATCC
		q8772telo_1	AGCTCCAGGGTTGCTTCTCAC	ATGCCTCTGCTTGGGTTGTC
		qKRCC1_1	GCAAAGGTCATTAATCCTCCAAT	AAATTGCTTTGATGGTGGAAATA
		qRPIA_2	AATTTTTAGTCACAAGCCTAGCTG	GACATCTGAAGTTGAAGCTATTGA
		qRPIA_3	AAAAGGCTTTAGATTTGTCAATCG	CCCAAAGCAAAAAGAGACTCAA
		qIGKC_3	ATCAGCTGTGTTACTACGGAGTGC	CCATTTCCCCTCCCTCATCT
		qIGKC_1	TTGCCACACTCCCCTGAG	CTGCCCTCTCCAGTGTGTCATG
		qIGKC_2	TGAGCAACTTTTCATAATTTCTCTT	AAGAGTTCCCTTGTGGCTAATAAA
9101	Del (5)(p15.2p14.3)	q9101-5p15_16	GCAAAAATATAATGACCCCAAAT	TGAATTGATCACACTGCTTAGAGA
		q9101-5p15_18	TCACCAAGCCTACGTAATGGA	TCTGGTATTTGGGATTTTCTATCT
		q9101-5p15_20	GTGTGGACTCATTTCCTGGAGATA	CTGAGACTTTAGCAGGCAACAGG
		q9101-5p15_11	AGAATTTTTCCCTGTTGAAACTCA	GCCAACTGTTTGAGCTGCTAAT
		q9101-5p15_10	GCTATGAAAGAAAAAGGCAGAGG	TGCATGTTTGTACCTTTGTTGTC
Del (7)(q11.23q11.23)	q9101-7q11_1	TTTGCACCAAATGTAACGATAAA	CAAGTCATGCAAGAATCCTGTTA	
	q9101-7q11_2	GCATACCTATAACAAAGGCCAAGA	TTAATGGACAACAAACCAGTGAA	
	q9101-7q11_3	ATCAAATGCTATAAAAAATCGTCAT	CTTTTGATCTATTCCCCTAAGGTT	
	q9101-7q11_4	TAAAAGGAGGACAGAAACACATGG	TGTGGGTGTGTGAAAATGGAGTA	

Supplemental Table 1. Primer Sequences for qPCR Assays (continued)

Del (7)(q11.23q11.23)	q9101-7q11_5	TTTATGATTTGGATTGCCAATTGA	AGGCAGATTTTTTTTTCTGGCAGTA	
	q9101-7q11_6	CAACAGTGCCATGTTCACTAAGA	ACGCCTACTCCATTTATCCAAA	
	q9101-7q11_7	AATACAAATGCCACAAAGATGC	TAGTGTTGAGAGTGCTTTAAAAAA	
	q9101-7q11_8	TGAGTACTACACAAGGGTTCATGC	ATGGGGCTTTCTACAAGGCATA	
	q9101-7q11_9	GAGGTCCGAAGATGAGAAAGCA	CCAACCCAGCTGGATCATTTA	
	q9101-7q11_10	GGAGCACAGGAAATCACTGTCA	TGGTGCTGTTGTTCTGATGA	
	q9101-7q11_11	GCCAAATCTTTGAGGGATTCT	TGTGGGAGATCCTTTATTCTCTG	
	q9101-7q11_12	GGGGAGGGAAATGGATTTGAGT	CCTGGAATATCCTTTCCTGTCT	
	q9101-7q11_13	ACCCACTGCCTGAATGTA AAA	TTAGGCCATGGATTTCTTTGAA	
	q9101-7q11_14	GGGGCCAGAGATGTAATGTGT	CTTGGCAGGCAATAGTTCACTT	
	q9101-7q11_15	TGGATAAATGGAGTAGGCGTACTT	AAAATGTCTGGGCAGTAAATAAA	
	9152 Del (7)(q11.23q11.23)	q9152_1	ACAGGAATCTCAGGCATGTGGT	AGGGTCTGGCTTTTGGAAAC
		q9152_2	CCAGTCTGTTGAGTCCATGAGAA	AGGCGGGAATGTGGCAAG
		q9152_3	CCTCCACATGGAGCAGAGTTAAAG	ACAGGCAGTGACCCAGTGTT
		q9152_4	GTGTGAGGGAGGGCAGAATGT	GGCCCGTGGTATCATTCTCT
q9152_5		TTGGTCATTATTTATGAAAGTGGT	AATAAGACCCTACCCTCCAAAAAT	
q9152_6		TGTGCCCTGATGCCTCTATT	CACCACAGGCAGACTGGA	
q9152_7		ACCTGGTTGCCCTTCAGGA	TACCCTCCATCAGACGCAGAGA	
q9152_8		ACGTGCCACTGCGGTTGT	TGTGATAAGGGGAGGCATTCTG	
q9152_9		GTAGCGGTCAGCATGGGTAT	CCCTCAACCCCCAGAGATT	
q9152_10		AGCCTTCCAACAGGTGAGGAG	TTGACAGATGTCAGCCGAGCC	
q9152_11		GGTTAAGCAGGAGGGGCTCT	TAGGATGACAGCAGATCTCCC	
qGTF2I_6		AGTTCGTTTATGATGAGTGGCAGTT	ACACAAGGTAAATAATTAGACAGTGATTGCA	
qGTF2I_7		TGGGAAGTTTGTATTTACCTGATAAGTTT	CCCAGCACATCCA ACTATTACCA	
qGTF2I_8	GGAACATATGGAAACATGCTTAATAATG,	CAGGGTCCTCACTTGTCTGAA		
8464 Dup (16)(p12.2p11.2)	qCDR2	GCCGGTTTCTCCTGGTCAC	GCCAAGTGGAGGAGCTGAAGT	
	qATXN2L	TACCCAGATGTTGAAGGGATTA AA	AAACATTTCAACAAGTCCAATCAC	
	qZP2	TCTCTTACCTGGGTAGCTTTGC	TCAGTGAAGTTGGGTCCATTT	
	qIGSF6	ACCTGTGTCCCCAGACGA	GCTGCTCATAGGCTCCATCA	
	qCLN3	GGGCCTCGGTTCTTATGA	GGAGGGGAAGAAGAAGCAGA	