

**TABLE S1.** The target sequences of each pair of probes in CNVplex technique.

Probe	Genome region (GRCh37/hg19)	Binding region	Product length	Binding sequences
DMDE01A	33229573	EXON01	155	CTGGGAGGCAATTACCTTCGAGAAAAACGAATAGGAAAAACT GAAGTGTTACTTTTT
DMDE01B	33229475	EXON01	160	GCCTACTGGAGCAATAAAGTTTGAAGAAGTTTACCAGGTTTTT TTTATCGCTGCCTTG
DMDE02A	33038465	INTRON01	145	TTTAATTTGGATGCCCCAAACCAGCATCACTCATGTTTAATTCC ATTTATCAATG
DMDE02B	33038296	EXON02	167	GCATTTTAGATGAAAGAGAAGATGTTCAAAGAAAACATTCAC AAAATGGGTAAATGCACA
DMDE02C	33038116	INTRON02	169	CATTTTATTTAGCTGTGCAAATCTGGAAATATTATGTTCACTCT TATTTAAGGAGTAAGCAGCAGA
DMDE03A	32867916	EXON03	110	TTCAGTTTGGGAAGCAGCATATTGAGAACCTCTTCAGTGACCTA CAGGATGGG
DMDE03B	32867865	EXON03	130	CGCCTCCTAGACCTCCTCGAAGGCTGACAGGGCAAAAAGT
DMDE04A	32862943	EXON04	101	AAGGATCCACAAGAGTTCATGCCCTGAACAATGTCAACAAGGC ACTGC
DMDE04B	32862899	EXON04	148	CACTGCGGGTTTTGCAGAACAATAATGTAAGTAGTACCCTGGA CAAGGTCTGG
DMDE05A	32841495	EXON05	164	TGTTTTACCCTTTCTTTAACAGGTTGATTTAGTGAATATTGGAA GTAAGTACATCGTAGATGG
DMDE05B	32841410	EXON05	116	GATTTGGAATATAATCCTCCACTGGCAGGTAAGAATCCTGATGA ATGGTTTCCTTTTG
DMDE06A	32834689	EXON06	148	ACCAACAGTGAAAAGATTCTCCTGAGCTGGGTCCGACAATCAA CTCGTAATT
DMDE06B	32834627	EXON06	136	TGTAATCAACTTCACCACCAGCTGGTCTGATGGCTGGCTTTGA ATG
DMDE07A	32827691	EXON07	139	GACTGGAATAGTGTGGTTTGCCAGCAGTCAGCCACACAACGAC TGGA
DMDE07B	32827628	EXON07	122	AACATCGCCAGATATCAATTAGGCATAGAGAACTACTCGATC CTGAAGGTTGGTAAATT
DMDE08A	32717403	EXON08	163	GATGTGTAGTGTTAATGTGCTTACAGATGTTGATACCACCTATC CAGATAAGAAGTCCATCTTAATG
DMDE08B	32717319	EXON08	101	GCCTCAACAAGTGAGCATTGAAGCCATCCAGGAAGTGGAATG TTGCC
DMDE09A	32716073	EXON09	151	GCACAGGGATATGAGAGAAGTCTTCCCTAAGCCTCGATTCAA GAGCTATGCC
DMDE09B	32716031	EXON09	142	GAGCTATGCCTACACACAGGCTGCTTATGTCACCACCTCTGACC CTACACG
DMDE10A	32663239	EXON10	113	TTGGAAGCTCCTGAAGACAAGTCATTTGGCAGTTCATTGATGGA GAGTGAAGTAA

DMDE10B	32663170	EXON10	116	TCAAACAGCTTTAGAAAGAAGTATTATCGTGGCTTCTTTCTGCTG AGGACACATTGCA
DMDE11A	32662410	EXON11	136	AGGGGTACATGATGGATTTGACAGCCCATCAGGGCCGGTTGG T
DMDE11B	32662303	EXON11	154	GAAACTGAAGTACAAGAGCAGATGAATCTCCTAAATTCAAGAT GGGAATGCCTCAGG
DMDE12A	32632494	EXON12	114	GAATGACTGGCTAACAAAAACAGAAGAAAGAACAAGGAAAAT GGAGGAAGAGCCTC
DMDE12B	32632442	EXON12	160	CCTACCTTATGTTGTTGACTTGGCGTTTTAGGTCTTCAAGATCA GGTCCAAGAGG
DMDE13A	32613957	EXON13	154	AAGAAGATCTAGAACAAGAACAAGTCAGGGTCAATTCTCTCAC TCACATGGTGGTGGT
DMDE13B	32613894	EXON13	113	GAATCTAGTGGAGATCACGCAACTGCTGCTTTGGAAGAACAAC TTAAGGTCAGA
DMDE14A	32592049	INTRON13	125	TCCAACCTTAGCGTACATAGGAGACTGAGATACTTTGGCAAAT TATTCATGCCATTTTTAA
DMDE14B	32591965	INTRON13	139	GCTGTGCTTGATTGTCTCTTCTCCAGGTATTGGGAGATCGATGG GCAA
DMDE15A	32591710	EXON15	119	TGGCTTTCAGAAAAAGAAGATGCAGTGAACAAGATTCACACAA CTGGCTTTAAAGATC
DMDE15B	32591643	INTRON15	125	GAAATGTTATCAAGTCTTCAAAAAGTGGCCGTATGTACTTTCTA GCTTTCAATGGTCTTATAAAAA
DMDE16A	32584028	INTRON15	162	ACTATAGTGGTGTATGGAATGCAACCCAGGCTTATCTGTGATC TTTCTTGTTTTAACAGG
DMDE16B	32583978	EXON16	148	TTTAACAGGTTTTAAAAGCGGATCTAGAAAAAGAAAAGCAATC CATGGGCAAAGT
DMDE16C	32583853	EXON16	154	GCTGGATAACTTTGCCCGTGTGGGATAATTTAGTCCAAAAAC TTGAAAAGAGTACA
DMDE17A	32563526	INTRON16	113	CAACAAGCAAGAACAGTTTCTCATTATTTCTTTGCCACTCCA AGCAGTCTTTACTG
DMDE17B	32563367	EXON17	139	AGTAACTACGGTGACCACAAGGGAACAGATCCTGGTAAAGCAT GCTCAAGA
DMDE17C	32563327	EXON17	99	TGCTCAAGAGGAACTTCCACCACCACCTCCCCAAAAGAAGAGG CAGA
DMDE18A	32536210	EXON18	122	TGATATAACTGAACTTCACAGCTGGATTACTCGCTCAGAAGCTG TGTTGCAGAGTCCTG
DMDE18B	32536162	EXON18	151	CAGAGTCTGAATTTGCAATCTTTCGGAAGGAAGGCAACTTCTC AGACTTAAAAGA
DMDE19A	32519959	INTRON18	142	ACTCATCTTTGCTCTCATGCTGCAGGCCATAGAGCGAGAAAAA GCTGAGA
DMDE19B	32519866	INTRON19	157	CCCTGGTGAACAGATGGTGAATGGTAATTACACGAGTTGATT AGATAATCTTCTTA

DMDE20A	32509585	EXON20	139	CAAGCCTCAGAACAACCTGAACAGCCGGTGGATCGAATTCTGCC AGTTG
DMDE20B	32509467	EXON20	107	TTGGAGCAGATGACAACTACTGCTGAAAACCTGGTTGAAAATCC AACCCACC
DMDE21A	32503128	EXON21	98	CCCTGAAAGAGAAAGGACAAGGACCCATGTTCTGGATGCAGA CTTG
DMDE21B	32503087	EXON21	141	CAGACTTTGTGGCCTTTACAAATCATTTTTAAGCAAGTCTTTTCTG ATGTGCA
DMDE21C	32502923	INTRON21	125	TGAAACAATAATTTCTGTAATGGAACCATTCTCCCTACAACCT GTATTAACAAGGAACGAAATACTG
DMDE22A	32490275	INTRON22	104	GCAGAGACTCGGGGAATTGCAGGTCTGTGAATATTTGAATGTC AAAACAATAAA
DMDE23A	32486682	EXON23	95	TTGAGGGACGCTGGAAGAAGCTCTCTCCAGCTGGTTGAGC
DMDE24A	32482721	EXON24	107	CCCTTGGGGATTTCAGAAATTTAAAAAGCAGCTGAAACAGTG CAGAGTAAGATTT
DMDE25A	32481552	INTRON25	139	GTGGGATCACATGTGCCAACAGGTATAGACAATCTCTTTCAGTG TGGCTTG
DMDE26A	32472852	EXON26	110	GAGATGCACGAATGGATGACACAAGCTGAAGAAGAGTATCTTG AGAGAGATTTTGAA
DMDE27A	32466571	INTRON27	101	AATGGGAAATGCAAGACTTTGGAAGTCAGTTGCTTTTCTGGTC TTTGTC
DMDE28A	32459298	EXON28	113	GCGGAGCTGAGGAAATCTCTGAGGTGCTAGATGTAAGTTGTAA ATTAAGCCAAATGATGA
DMDE29A	32456363	EXON29	125	GAGACATTTAATTCTCGTTGGAGGGAACACATGAAGAGGTAT GAAGATAAGTGAAAAATCTCTTT
DMDE30A	32429866	INTRON30	136	CAATGCCTCAGGAAGCCCAGGCAAGTACATCTGGGAATCAGC TTCC
DMDE31A	32408200	EXON31	125	GGCTGCCCAAAGAGTCCTGTCTCAGATTGATGTTGCACAGGTAT ATGTTATTCA
DMDE32A	32407620	EXON32	163	GGAAGTAGTACAGTCACAGCTAAATCATTTGTGTGGTATGTATTT CTGGTGGCAAATACGCAG
DMDE33A	32404428	EXON33	142	TGCATTATAATGAGCTGGGAGCAAAGGTGTGTGCATGCTGAGA CCACA
DMDE34A	32398622	INTRON34	160	GGATTCTGAAGTTGCCTGGGGAAAGGTAACCTATATCACTG AAGGTTATTTGAACA
DMDE35A	32383215	EXON35	148	CAGTTTGGGCAAGAAGGAGACGTTGGTGGAAAGATAAACTCAG TCTTCTGAATAGT
DMDE36A	32382707	EXON36	121	GAGAAAAAGAAACCCAGCAAAAAGAAGACGTGCTTAAGGTA GCAAATAAAATATGAAAAG
DMDE37A	32380904	INTRON37	163	GCCATTTACACAGAATTAAGACTGGAAAGGTAGGAAGATCTA CTCCAAGGTGGAACTTG
DMDE38A	32366539	EXON38	151	CAGCAGGGGGTGAATCTGAAAGAGGAAGACTTCAATAAAGATA TGGTAAATTGGTTG

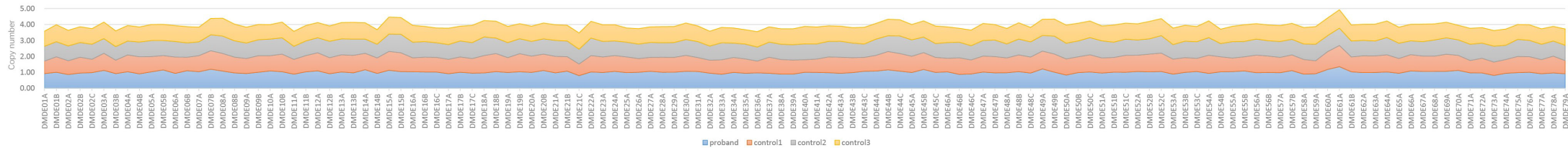
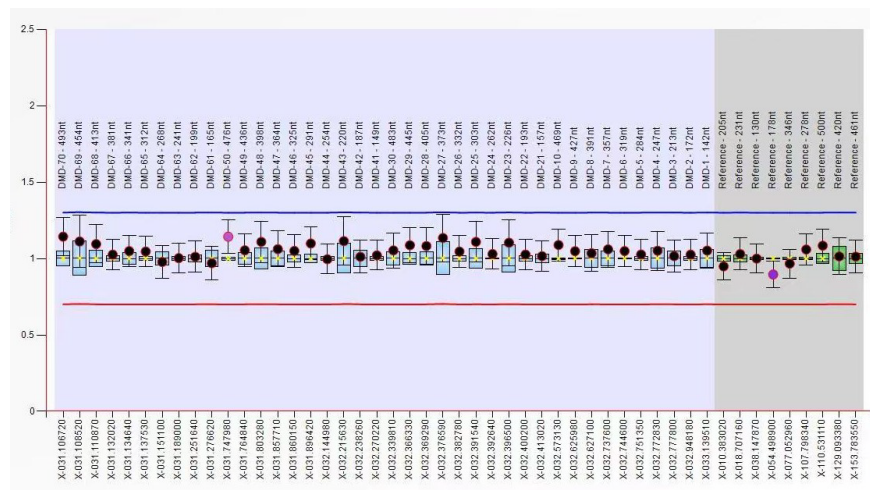
DMDE39A	32364150	EXON39	166	GGTACTGTAAAAGAATTGTTGCAAAGAGGAGACAACCTACAAC AAAGAATCACAGATGAGAGAAA
DMDE40A	32361268	EXON40	122	AAAAAATTAGCCAGCTACCTGAGCCCAGAGATGAAAGGAAAA TAAAGGTAATGTTGTTTT
DMDE41A	32360229	EXON41	110	GCAAATTTGCTCAGTTTCGAAGACTCAACTTTGCACAAATTGTG AGTTGTTACTGG
DMDE42A	32328241	EXON42	116	CAACTTCTCAATGCTCCTGACCTCTGTGCTAAGGACTTTGAAGA TCTCTTTAAGCAAGA
DMDE43A	32306005	INTRON42	160	GAGCGATCCACTCTCTCAGGATGAGGTCTGTGATTACCTATTG AATTTGAACATGTCA
DMDE43B	32305708	EXON43	98	TGTGGAAAGGGTGAAGCTACAGGAAGCTCTCTCCAGCTTGAT TTCCA
DMDE43C	32305664	EXON43	110	GATTTCCAATGGGAAAAAGTTAAACAAAATGTACAAGGACCGAC AAGGGTAGGTAACA
DMDE44A	32235175	EXON44	104	CCATATGCTTTTACCTGCAGGCGATTTGACAGATCTGTTGAGAA ATGGCG
DMDE44B	32235123	EXON44	160	AACTGTTTCAGCTTCTGTTAGCCACTGATTAATATCTTTATATCA TAATGAAAACGCCGCC
DMDE44C	32235067	EXON44	163	CAGTTTCTCAGAAAGACACAAATTCCTGAGAATTGGGAACATG CTAAATACAAATGGTATCT
DMDE45A	31986742	INTRON44	122	TCATAGGGAAATTTTACATGGAGCTTTTGTATTTCTTTCTTTC CAGTACAACCTGCA
DMDE45B	31986576	EXON45	116	CAAACGTGTGTCAGAACATTGAATGCAACTGGGGAAGAAATAA TTCAGCAATCCTC
DMDE45C	31986495	EXON45	96	TGGGAAGCCTGAATCTGCGGTGGCAGGAGGTCTGCAAACAGC
DMDE46A	31950247	EXON46	150	GGAAGCAGATAACATTGCTAGTATCCCCTGAACTGGAAAA GAGCAGCACTAA
DMDE46B	31950197	EXON46	163	GCAACTAAAAGAAAAGCTTGAGCAAGTCAAGGTAATTTTATTT TCTCAAATCCCCAGGG
DMDE46C	31949769	EXON46	166	GAACAAAAGAATATCTTGTCAGAATTTCAAAGAGATTTAAATG AATTTGTTTTATGGTTGGAGGAAG
DMDE47A	31947846	EXON47	98	GTTTCAGTACTGGTGAAGAGTTGCCCTGCGCCAGGGAATTC TCA
DMDE47B	31947753	EXON47	159	TCCCATAAGCCCAGAAGAGCAAGATAAACTTGAAAATAAGCTC AAGCAGACAAATCTC
DMDE48A	31893457	EXON48	127	CAGAGCTTTACCTGAGAAACAAGGAGAAATTGAAGCTCAAATA AAAGACCTTGGGC
DMDE48B	31893400	EXON48	104	CTTGAAAAAAGCTTGAAGACCTTGAAGAGCAGTTAAATCATC TGCTGCTGTGG
DMDE48C	31893353	EXON48	118	GCTGCTGTGGTTATCTCCTATTAGGAATCAGTTGAAAATTTATA ACCAACCAAACCAAGA
DMDE49A	31854926	EXON49	113	GGGTTCTTTTCCCCAGGAAACTGAAATAGCAGTTCAAGCTAAAC AACCGGATG

DMDE49B	31854883	EXON49	110	CAACCGGATGTGGAAGAGATTTTGTCTAAAGGGCAGCATTGTG ACAAGGAAA
DMDE50A	31838389	INTRON49	157	GGAGAAAGGGTTTTGTATGGAGCAATTGATAAATATTTGTAG GGTGGTTGGCTAAAA
DMDE50B	31838129	EXON50	133	TTCAAGAGCTGAGGGCAAAGCAGCCTGACCTAGCTCCTGGACT GA
DMDE50C	31838088	INTRON50	148	CCTGGACTGACCACTATTGGAGCCTGTAAGTATACTGGATCCCA TTCTCTTTGGCT
DMDE51A	31792267	EXON51	125	TGTTACTCTGGTGACACAACCTGTGGTTACTAAGGAACTGCCA TCTCCAACTAGAA
DMDE51B	31792212	EXON51	101	GCCATCTTCCTTGATGTTGGAGGTACCTGCTCTGGCAGATTCA ACCG
DMDE51C	31792127	EXON51	154	GCTTGATCAAGTTATAAAATCACAGAGGGTGATGGTGGGTGAC CTTGAGGATATCA
DMDE52A	31747836	EXON52	142	CAATGCAGGATTTGGAACAGAGGCGTCCCCAGTTGGAAGAACT CATTACC
DMDE52B	31747794	EXON52	104	CATTACCCTGCCAAAAATTTGAAAAACAAGACCAGCAATCAA GAGGCT
DMDE52C	31747524	INTRON52	160	ATACATTTTTAAATCAATTCAGGGCTTATATAGTTGCAAAGCAT GCATTGATGGGTGTG
DMDE53A	31697665	EXON53	101	GAATCAGTGGGATGAAGTACAAGAACACCTTCAGAACCGGAGG CAACAGTT
DMDE53B	31697571	EXON53	101	GAAGAAGCTGAGCAGGTCTTAGGACAGGCCAGCAAGCTTG AGTCATG
DMDE53C	31697504	EXON53	128	TCCCTATACAGTAGATGCAATCCAAAAGAAAATCACAGAAAACC AAGTTAGTATCAAAGATACCTTT
DMDE54A	31676246	EXON54	148	AAAATCTATAGCAGTTGGCCAAAGACCTCCGCCAGTGGCAGAC AAATGTAGA
DMDE54B	31676194	EXON54	136	GGCAAATGACTTGGCCCTGAAACTTCTCCGGGATTATTCTGCAG ATG
DMDE55A	31645922	EXON55	110	ACTCATAGATTACTGCAACAGTTCCCCCTGGACCTGGAAAAGTT TCTTGCCT
DMDE55B	31645878	EXON55	104	CTTGCTGGCTTACAGAAGCTGAAACAACCTGCCAATGTCCTACA GGATGC
DMDE56A	31525570	EXON56	151	CATATTCTTCTCTCTGCTGTCCTGTAGGACCTCCAAGGTGAAAT TGAAGCTCAC
DMDE56B	31525357	INTRON56	145	AGACAATGAGGAAAATTTGGCCATTTTAATTCATTTGTGGCCTT TTTGCTCC
DMDE57A	31515061	EXON57	139	TGGTACGCTGCTGTTCTTTTTCAGGTCCCATTTGGAAGCCAGTTC TGA
DMDE57B	31514920	EXON57	148	GGCGACTTTCAGCAGTTCAGAAGCAGAACGATGTACATAGGG TAGGACATT

DMDE58A	31497139	EXON58	122	TGAGACTGTACGAATATTTCTGACAGAGCAGCCTTTGGAAGGA CTAGAGAACTCTACCAGG
DMDE59A	31496227	EXON59	110	GACTCTCTCCAAGATCACCTCGAGAAAGTCAAGGTACCGTCTAC TTCTTTGCTTCA
DMDE60A	31462736	EXON60	101	TGACTATTGCACACAGGCACTTCGAGGAGAAATTGCGCCTCTG AAAGA
DMDE61A	31366781	INTRON60	122	TTTAAATTGTTCCCTCATTATATAGAATGAGAGAACATCATTCTC TCCTTTTCCTCCCAGG
DMDE61B	31366701	EXON61	98	TGCATGAAGCCCACAGGGACTTTGGTCCAGCATCTCAGCACTTT C
DMDE62A	31341761	EXON62	98	CTTTTCAGCGTCTGTCCAGGGTCCCTGGGAGAGGCCATCTCGC
DMDE63A	31279074	EXON63	151	CCCAAAATGACAGAGCTCTACCAGTCTTTAGGTAAGGACATGG CCATGTTTCCTCC
DMDE64A	31241200	EXON64	151	AATGTCAGATTCTCAGCTTATAGGACTGCCATGAAACTCCGAAG ACTGCAGAAGG
DMDE65A	31227775	EXON65	136	AGCTGCATGTGATGCCTTGGACCAGCACAACTCAAGCAAAAT G
DMDE66A	31224749	EXON66	110	GGGAGGATCCGTGCTCTGTCTTTTAAACTGGCATCATTCCCT GTGTAAA
DMDE67A	31222185	EXON67	98	GGATTTTGTGACCAGCGCAGGCTGGGCCTCCTTCTGCATGATT
DMDE68A	31200863	EXON68	157	TGTAACATCTGCAAAGAGTGTCCAATCATTGGATTGAGGTATTA GGAACCAAAAAAAAA
DMDE69A	31198486	INTRON69	98	CCATGGTGAATATTGCACTCCGGTAAGTTTGACGCCAGCCTGA CG
DMDE70A	31196782	INTRON70	115	GTCTTAGAGGGGGACAACATGGAAACGTGAGTAGTAGCAAAAG CAGAACACACTCTTG
DMDE71A	31196077	EXON71	148	CATTTTTGTTTTGCAGTCCCGTTACTCTGATCAACTTCTGGCCA GTAGATTCTG
DMDE72A	31191652	INTRON72	116	TCACGCATTGAACATTATGCTAGCAGGTATGAGACTAGTTGTAT GCCAGGCAAATATTG
DMDE73A	31190467	EXON73	128	TCTTATCTAAATGATAGCATCTCTCTAATGAGAGCATGTAAGT ATCCCATCTCTTTTACAAAATGTTCC
DMDE74A	31187558	INTRON74	122	GAATCCTAGCAGATCTTGAGGAAGAAAACAGGTGAGTTTTCTTT CTAGCTTTGTCATTGGTATG
DMDE75A	31165427	EXON75	101	GGAAGACCACAATAAACAGCTGGAGTCACAGTTACACAGGCTA AGGCAGCT
DMDE76A	31164430	EXON76	136	CCTATGCTGCTCCGAGTGGTTGGCAGTCAAACCTTCGGACTCCAT GG
DMDE77A	31152217	INTRON77	166	AACTCAACAACCTCTCCCTAGTTCAAGAGGTAAGCTCCAATAC CTAGAAGGGACTCAGATT
DMDE78A	31144757	INTRON78	136	CCCCTGAAAAGCCAATGAGAGAGGTTAGTGAGATTCAGGCTCA CGGCC

DMDE79A 31140037 EXON79 159 TTTCTCTTGTTTTCCAGGACACAATGTAGGAAGTCTTTCCACA  
TGGCAGATGATT

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**A****B**

**FIGURE S1. CNVplex results of proband 1 (A) and proband 2 (B).**



**A****B**

**FIGURE S2.** Whole exome sequencing results of proband 1 (A) and proband 2 (B).