

Supplementary Table 4 **Primer sequences and mutation detection**

Allele-specific primers				
Mutation	Allele	Forward primer	Reverse primer	Detection
del8809–13035 (p1)	wt	12602–12622: TCATCCCTGTAGCATTGTTTCG	16348–16328: GACTGTAATGTGCTATGTACG	MseI
	del	8282–8301: CCCCTCTAGAGCCCCTGTA		
del12103–14411 (p2)	wt	13122–13140: CTTCCACCCCCTAGCAGAA	148–129: TGAGGCAGGAATCAAAGACA	mm-PCR
	del	11911–11934: CACGTTCTCCTGATCAAATATCAC		
del10311–15571 (p7)	wt	15544–15562: CCACATCAAGCCCGAATGA	148–129: TGAGGCAGGAATCAAAGACA	SphI+ BanII
	del	9140–9163: CTGTCGCCTTAATCCAAGCCTACG		
del9098–14651 (p8)	wt	14588–14611: CCCCCATAAATAGGAGAAGGCTTA	148–129: TGAGGCAGGAATCAAAGACA	SphI+ BanII
	del	8282–8301: CCCCTCTAGAGCCCCTGTA		
del10198–13755 (p9)	wt	13122–13140: CTTCCACCCCCTAGCAGAA	148–129: TGAGGCAGGAATCAAAGACA	SphI+ BanII
	del	9921–9940: GCCTGATACTGGCATTTTGT		
del11032–13232 (p10)	wt	13122–13140: CTTCCACCCCCTAGCAGAA	148–129: TGAGGCAGGAATCAAAGACA	SphI+ BanII
	del	10542–10565: CACCTCATATCCTCCCTACTATGC		
del8426–14138 (t1)	wt	13122–13140: CTTCCACCCCCTAGCAGAA	148–129: TGAGGCAGGAATCAAAGACA	mm-PCR
	del	8282–8301: CCCCTCTAGAGCCCCTGTA		
A3243G(MELAS) (p3–5)	wt	16166–16185: ACCCAATCCACATCAAAC	3267–3243: TTAAGTTTTATGCGATTACCGG <u>C</u> CT	mm-PCR
	mut		3267–3243: TTAAGTTTTATGCGATTACCGG <u>C</u> CC	
A185G (p3, p4, p11–16)	A	164–185: CACCTACGTTCAATATTACAGA	265–246: AGCGGCTGTGCAGACATTCA	mm-PCR
	G	164–185: CACCTACGTTCAATATTACAGG		