

Supplemental Table 1. Germline mutations of *SDHB*, *SDHC* and *SDHD* found among the 578 eligible HNP patients

Gene	cDNA	Protein	Exon	Carriers	Ref	
SDHB	c.3G>A	p.Met1?	1	1	new	
	c. 21delC	p.Ser8ProfsX1	1	1	(1)	
	c.49delA	p.Thr17ProfsX60	1	1	new	
	c.73_76delGCCT	p.Ala25ProfsX51	2	1	new	
	c.79C>A	p.Arg27X	2	1	(2)	
	c.136C>G	p.Arg46Gly	2	2	(3)	
	c.137G>A	p.Arg46Gln	2	5	(4)	
	c.141G>A	p.Trp47X	2	1	(5)	
	c.166_170del CCTCA	p.Pro56delTyrfsX5	2	2	(1)	
	c.238A>G	p.Lys80Glu	3	1	new	
	c.268C>T	p.Arg90X	3	2	(6)	
	c. 269G>A	p.Arg90Gln	3	1	new	
	c.277T>C	p.Cys93Arg	3	2	(7)	
	c.287-2A>G	aberrant splicing	4	2	(1)	
	c.293G>A	p.Cys98Tyr	4	1	(8)	
	c.312insCACTGCA	p.Ile105HisfsX16	4	1	(9)	
	c.328A>C	p.Thr110Pro	4	1	new	
	c.423+1G>A	aberrant splicing	4	1	(10)	
	c.540G>A	aberrant splicing	5	1	(9)	
	c.541-2A>G	aberrant splicing	6	1	(11)	
	c.549_552delTACinsATACAG	p.Tyr184delinsIleGln	6	1	new	
	c.557G>A	p.Cys186Tyr	6	1	(9)	
	c.589C>T	p.Pro197Ser	6	2	(9)	
	c.649C>T	p.Arg217Cys	7	2	(12)	
	c.650G>T	p.Arg217Leu	7	1	new	
	c.688C>T	p.Arg230Cys	7	2	(13)	
	c.689G>A	p.Arg230His	7	2	(13)	
	c.689G>T	p.Arg230Leu	7	2	new	
	c.718_719delCT	p.Leu240IlefsX15	7	1	new	
	c.724C>A	p.Arg242Ser*	7	2	new	
	c.724C>T	p.Arg242Cys	7	2	(14)	
	c.725G>A	p.Arg242His	7	3	(1)	
	c.736A>T	p.Ile246Phe	7	1	new	
	c.761C>T	p.Pro254Leu	7	1	(9)	
	c.765+1G>A	aberrant splicing	7	1	(1)	
	c.778G>C	p.Gly260Arg	8	2	new	
	c.780delG	p.Ala262LeufsX8	8	1	new	
		Deletion Exon 1			2	*
		Deletion Exon 1-8			1	*
		Deletion Exon 3			2	*
	Deletion Exon 5			1	*	
	Duplication Exon 3			1	*	

Gene	cDNA	Protein	Exon	Carriers	Ref
SDHC	c.1A>G	p.Met1?	1	1	(15)
	c.2T>A	p.Met1?	1	1	new
	c.3G>A	p.Met1?	1	2	(16)
	c.23dupA	p.His8GlnfsX12	2	1	new
	c.39C>A	p.Cys13X	2	1	(15)
	c.43C>T	p.Arg15X	2	1	(17)
	c.78-2A>G	aberrant splicing	3	1	new
	c.148C>T	p.Arg50Cys	3	1	new
	c.166A>T	p.Ile56Phe	3	1	new
	c.173T>C	p.Ile58Thr	3	1	new
	c.210C>G	p.Cys70Trp	4	1	new
	c.214C>T	p.Arg72Cys	4	2	(15)
	c.218insA	p.Thr74HisfsX24	4	1	new
	c.224G>A	p.Gly75Asp	4	2	new
	c.253_255dupTTT	p.Phe85dup	5	1	new
	c.397C>T	p.Arg133X	5	3	(17)
	c.405+1G>T	aberrant splicing	5	1	(18)
	c.439C>T	p.Gln147X	5	1	(15)
	Deletion Exon 4-6				2
Deletion Exon 5				1	*

Gene	cDNA	Protein	Exon	Carriers	Ref
SDHD	c.2T>A	p.M1?	1	1	new
	c.14G>A	p.Trp5X	1	8	(3)
	c.33C>A	p.Cys11X	1	6	(3)
	c.33C>T	-	1	2	new
	c.36_37delTG	p.Ala13ProfsX55	1	1	(3)
	c.49C>T	p.Arg17X	1	1	new
	c.50G>T	p.Arg17Leu	1	1	(9)
	c.52+2T>G	aberrant splicing	1	1	(19)
	c.53-2A>G	aberrant splicing	2	3	new
	c.55dupT	p.Leu19SerfsX50	2	1	new
	c.64C>T	p.Arg22X	2	1	(20)
	c.106C>T	p.Gln36X	2	1	(21)
	c.112C>T	p.Arg38X	2	3	(21)
	c.118A>G	p.Ile40Val	2	1	new
	c.120_127delCCCAGAAT	p.Ile40MetfsX26	2	1	(9)
	c.129G>A	p.Trp43X	2	2	(22)
	c.168_169delTT	p.Ser57TrpfsX11	2	1	(23)
	c.170-1G>T	aberrant splicing	3	5	(24)
	c.184_185insTC	p.Ala62ValfsX25	3	1	new
	c.191_192delTC	p.Leu64ProfsX4	3	2	(25)
	c.206_218del13 bp	p.Glu69ValfsX13	3	1	(1)
	c.208A>G	p.Arg70Gly	3	1	(20)
	c.230T>G	p.Leu77Arg	3	1	new
	c.233_242del10 bp	p.Ile81ProfsX2	3	1	new

Gene	cDNA	Protein	Exon	Carriers	Ref	
SDHD	c.242C>T	p.Pro81Leu	3	9	(21)	
	c.252T>G	p.Tyr84X	3	2	(1)	
	c.274G>T	p.Arg92Tyr	3	1	(21)	
	c.276_278delCTA	p.Tyr93del	3	1	(25)	
	c.302T>C	p.Leu101Pro	3	1	new	
	c.314+1G>C	aberrant splicing	3	1	new	
	c.317delG	p.Gly106AlafsX29	4	1	new	
	c.317G>T	p.Gly106Val	4	2	new	
	c.325C>T	p.Gln109X	4	1	(26)	
	c.334_337delACTG	p.Asp113MetfsX21	4	1	(6)	
	c.337_340delGACT	p.Asp113MetfsX22	4	5	(27)	
	c.341A>G	p.Tyr114Cys	4	10	(27)	
	c.341_342delAT	p.Tyr114CysfsX76	4	1	(8)	
	c.367G>A	p.Ala123Thr	4	1	new	
	c.370delG	p.Ala124ProfsX11	4	1	new	
	c.386_387insT	p.Leu129PhefsX62	4	1	(7)	
	c.408delT	p.Phe136LeufsX25	4	1	new	
	c.443G>T	p.Gly148Val	4	1	(8)	
	Deletion Exons 1-2				1	*
	Deletion Exons 1-4				2	*
Deletion Exons 3				1	*	
Deletion Exons 3-4				2	*	