Suppl. Table 1. All *HIF2A* and *HIF1A* gene variants identified in this study

Gene	Exon	Nucleotide	Protein	Type	SNPdb ID	Number of events*	Het**
HIF2A	2	c.41G>A	p.R14K	missense	rs73926269	2	2
HIF2A	2	c.212C>A	p.S71Y	missense	Unreported	1	1
HIF2A	3	c.218-9C>T		intronic	rs75984373		
HIF2A	3	c.218-7insT		intronic	rs66811540		
HIF2A	3	c.218-11C>A_T		intronic	rs202210836		
HIF2A	3	c.218-10-11insCT		intronic	rs142684263		
HIF2A	3	c.218-13C>G		intronic	rs200435589		
HIF2A	3	c.218-11C>G		intronic	rs202210336*		
HIF2A	3	c.218-55C>A		intronic	rs6743991	199	95
HIF2A	6	c.779+8G>T		intronic	Unreported	1	1
HIF2A	8	c.993C>T	p.N331N	silent	Unreported	1	1
HIF2A	9	c.1035-7C>G		intronic	rs7557402	265	111
HIF2A	9	c.1249+38C>T		intronic	rs77794056	7	7
HIF2A	9	c.1113C>T	p.N371N	silent	rs149518347	2	2
HIF2A	10	c.1443+51C>T		intronic	rs181412376	1	1
HIF2A	10	c.1443+61A>G		intronic	rs17035085	28	28
HIF2A	10	c.1350G>C	p.E450D	missense	Unreported	1	1
HIF2A	10	c.1281T>C	p.Y427Y	silent	Unreported	1	1
HIF2A	11	c.1554+48G>C		intronic	rs7598371	230	134
HIF2A	11	c.1444-137A>G		intronic	rs7571218	301	49
HIF2A	12	c.1591C>T	p.P531S	missense	Favier et al	1	1
HIF2A	12	c.1592C>T	p.P531L	missense	Unreported	1	1
HIF2A	12	c.1591C>A	p.P531T	missense	Unreported	1	1
HIF2A	12	c.1908T>C	p.N636N	silent	rs35606117	9	9
HIF2A	12	c.1737G>A	p.P579P	silent	rs184760160	1	1
HIF2A	12	c.1833C>T	p.A611A	silent	rs41281469	1	1
HIF2A	15	c.2288-26G>A		intronic	rs17035089	8	8
HIF2A	15	c.2353C>A	p.P785T	missense	rs61518065	10	8
HIF2A	15	c.2296A>C	p.T766P	missense	rs59901247	19	13
HIF2A	15	c.2461+27C>G		intronic	Unreported	3	3
HIF2A	15	c.2461+41G>A		intronic	Unreported	7	7
HIF2A	15	c.2403A>C	p.P801P	silent	Unreported	1	1
HIF2A	15	c.2511C>T	p.P837P	silent	rs35795449	5	5
HIF2A	16	c.2613+31C>A		intronic	Unreported	1	1
HIF1A	9	c.1744C>T	p.P582S	missense	rs11549465	32	30
HIF1A	9	c.1762G>A	p.A588T	missense	rs11549467	2	2
HIF1A	12	c.1800A>T	p.T600T	silent	rs61755645	2	2
HIF1A	12	c.2093+30_2093+31delTC		intronic	rs72248236	2	1
HIF1A	12	c.2093+57T>C		intronic	rs192696339	1	1
HIF1A	12	c.2093+99T>C		intronic	rs4902080	123	14

Het=heterozygosity; m=nonsynonymous missense mutation; s=synonymous missense mutation; i=intronic; **counted by allele (total n=478 for HIF2A and n=258 for HIF1A); **same position as rs202210336, but distinct nucleotide change. Variants highlighted in yellow were predicted to be pathogenic by SIFT or Polyphen 2 algorithms and were further evaluated.