

## Supplementary data: SNPs in genes with copy number variation: A question of specificity

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The bases at equivalent positions of the duplicon(s) for each SNP are shown in table 1 for *HBA1* and table 2 (a, b) for *PSORS1* and *GHI*.

**Table 1.** SNPs of haemoglobin:  $\alpha$ -locus 1 (NCBI Build 126).

SNP ID	Nucleotide change	Location	Wild type bases				
			<i>HbA1</i>	<i>HbA2</i>	<i>HbZ</i>	<i>HbQ1</i>	<i>HbM</i>
<b>rs28928888</b>	T>C	exon 1	T	T	<u>C</u>	T	<u>C</u>
<b>rs28928885</b>	C>G	exon 1	C	C	<u>G</u>	<u>G</u>	A
<b>rs1799896</b>	G>T	exon 1	G	G	<u>G</u>	<u>G</u>	<u>T</u>
<b>rs11548605</b>	C>T	exon 1	C	C	C	C	C
<b>rs28928880</b>	A>G	exon 1	A	A	<u>G</u>	<u>G</u>	<u>G</u>
<b>rs28928886</b>	G>C	exon 2	G	G	<u>G</u>	<u>G</u>	<u>G</u>
<b>rs28928883</b>	A>C	exon 2	A	A	A	A	A
<b>rs11548604</b>	C>T	exon 2	C	C	C	G	C
<b>rs28928878</b>	G>A	exon 2	G	G	G	G	G
<b>rs28928887</b>	G>C	exon 2	G	G	G	<u>C</u>	G
<b>rs1060339</b>	C>A	exon 2	C	C	C	<u>C</u>	G
<b>rs3180281</b>	C>G	exon 2	C	C	A	A	A
rs28928882	C>A	exon 2	C	C	C	C	C
rs28928875	G>C/A	exon 2	G	G	G	G	G
<b>rs3208520</b>	C>A	exon 2	C	C	C	C	C
rs28928876	C>T	exon 2	C	C	C	C	C
<b>rs1061009</b>	C>A	exon 2	C	C	C	C	C
<b>rs17407508</b>	C>T	exon 2	C	C	C	C	C
rs28928879	A>G	exon 2	A	A	A	A	A
<b>rs28928877</b>	A>G	exon 2	A	A	C	A	C
rs28928884	C>T	exon 3	C	C	C	C	C
<b>rs17419748</b>	C>T	exon 3	C	C	C	C	C
<b>rs17135333</b>	C>T	exon 3	C	C	C	C	C
<b>rs1058069</b>	C>A	exon 3	C	C	G	C	C
<b>rs3209323*</b>	C>T	exon 3	C	C	C	C	<u>T</u>
rs28928881	G>T	exon 3	G	G	G	G	G
<b>rs1058334</b>	C>A	exon 3	C	C	C	C	C
<b>rs17135334</b>	C>G	exon 3	C	C	C	C	–
<b>rs3180978</b>	C>T	exon 3	C	C	<u>T</u>	C	C

The SNPs withdrawn in Build 127 are in bold. The potential PSVs are italicized and underlined.

\*Same as rs17134763 of HBA2; ‘–’ base is absent in HBM at the equivalent position.

**Table 2 a.** SNPs of *PSORS1* (HLA-C; NCBI Build 127).

SNPs	Heterozygosity (NCBI)	Nucleotide change	Location	Wild type bases	
				HLA-C	HLA-B
<b>rs7767581</b>	0.407	C>G	exon 1	C	<u>G</u>
rs2308538	0.442	G>C/T	exon 2	G	G
rs11547357	0.476	A>C	exon 2	A	A
rs16895963	0.500	G>C/T	exon 2	G	G
rs1050428	0.500	G>C/T	exon 2	G	G
rs16868214	0.499	C>T	exon 2	C	C
rs1050420	0.493	G>A	exon 2	G	G
rs17416870	0.495	C>T	exon 2	C	C
rs17408553	0.498	T>G	exon 2	T	T
<b>rs9264664</b>	0.410	C>T	intron 2	C	C
rs1131115	0.487	G>C/T	exon 3	G	G
rs2308575	0.493	C>G/A	exon 3	C	C
rs2308592	0.500	A>G/T	exon 3	A	C
rs17413671	0.476	A>G	exon 3	A	A
<b>rs9264647</b>	0.407	A>G	intron 3	A	<u>G</u>
<b>rs9264645</b>	0.400	G>A	intron 3	G	<u>A</u>
<b>rs9264638</b>	0.483	G>A	intron 3	G	<u>G</u>
<b>rs9264636</b>	0.407	C>T	intron 3	C	<u>C</u>
<b>rs9264608</b>	0.405	A>G	intron 5	A	<u>G</u>
<b>rs9264606</b>	0.400	A>C	intron 5	A	<u>A</u>
<b>rs9264603</b>	0.415	G>A	intron 5	G	<u>G</u>
<b>rs9264602</b>	0.405	G>A	intron 5	G	<u>G</u>
<b>rs9264601</b>	0.455	G>A	intron 5	G	<u>G</u>
<b>rs9264594</b>	0.421	A>G	intron 7	A	<u>A</u>
<b>rs2001181</b>	0.412	C>T	intron 7	C	<u>C</u>
rs1094	0.500	C>T	exon 8	C	C
<b>rs1130586</b>	0.500	G>A	exon 8	G	<u>G</u>
rs1130552	0.484	T>C	exon 8	T	T
<b>rs1049281</b>	0.453	T>C	exon 8	T	<u>C</u>

The SNPs genotyped in HAPMAP are in bold. The potential PSVs are italicized and underlined.

**Table 2 b.** SNPs of growth hormone 1 (*GHI*; NCBI Build 127).

SNP ID	Heterozygosity (NCBI)	Nucleotide change	Location	Wild type bases				
				GHI	GHI	CSH1	CSHL1	CSH2
rs9282699	0.5	A>G	exon 1	A	A	A	<u>G</u>	A
rs6172	0.5	A>C	exon 1	A	A	A	<u>C</u>	A
rs9282698	0.5	T>C	intron 1	T	T	T	<u>C</u>	T
rs41295029	0.5	T>C	intron 1	T	T	<u>C</u>	<u>C</u>	<u>C</u>
rs41295031	0.5	G>C	intron 2	G	G	G	G	G
rs41295033	0.5	A>T	intron 2	A	A	G	G	<u>T</u>
rs41295035	0.5	G>A	intron 2	G	G	G	G	<u>A</u>
rs41295037	0.5	A>C	intron 3	A	A	<u>C</u>	C	<u>C</u>
rs41295039	0.5	C>G	intron 3	C	C	<u>G</u>	G	<u>G</u>
rs41295041	0.5	C>T	exon 4	C	C	C	T	C
rs41295043	0.5	G>A	exon 4	G	G	G	G	G
rs41295245	0.5	C>T	intron 4	C	C	C	C	C
rs2665802	0.5	T>A	intron 4	T	T	T	T	T
rs41295247	0.5	C>T	intron 4	C	C	C	C	C
rs41295249	0.5	T>G	intron 4	T	T	T	T	<u>G</u>
rs41295251	0.5	A>G	intron 4	A	A	<u>G</u>	<u>G</u>	<u>G</u>
rs41295253	0.5	C>A	exon 5	C	C	G	<u>A</u>	C
rs41295255	0.5	G>T	exon 5	G	G	G	<u>T</u>	G

SNPs shown in this table are not genotyped in HapMap. The potential PSVs are italicized and underlined.