

Table 1:

Parameters	Case 1	Case 2	Case 3
Age-sex	50-M	44-F	34-M
RBC ($10^{12}/L$)	5.3	4.6	4.6
Hb (g/l)	153	108	102
MCV (fl)	86.8	74.7	67
MCH (pg)	28.9	23.5	22
Ferritin (mg/l)	nd	39.2	182
HbA ₂	17	2.1	1.9
HbF	0.5	14.2	74
G γ promoter	nd	-158 C/T	-158 C/C
genotype	δ - β δ - β	$\delta^0\beta^+$	$\delta^0\beta^+$ / -29A>G
Alpha deletions (GAP-PCR)	$\alpha\alpha/\alpha\alpha$	$\alpha\alpha/\alpha\alpha$	$\alpha\alpha/\alpha\alpha$

nd: not determined

Figure 1A:

	cap site	initiation codon	
β	ACATTTGCTTCTGACACAAC	<u>GTGTTCACTAGCAACCTCAAACAGACACC ATG GTG CAY CTG ACT CCT GAG GAG AAG</u>	TCTGCCGTT
$\delta^{\circ}\beta^*$	ACACTTCTTCTGACATAACA	<u>GTGTTCACTAGCAACCTCAAACAGACACC ATG GTG CAC CTG ACT CCT GAG GAG AAG</u>	TCTGCCGTT
$\delta\beta\delta-\beta$	ACATTTGCTTCTGACACAAC	<u>GTGTTCACTAGCAACCTCAAACAGACACC ATG GTG CAC CTG ACT CCT GAG GAG AAG</u>	ACTGCTGTC
δ	ACACTTCTTCTGACATAACA	<u>GTGTTCACTAGCAACCTCAAACAGACACC ATG GTG CAT CTG ACT CCT GAG GAG AAG</u>	ACTGCTGTC

Figure 1B:

