

Comparison of Two Known Chromosomal Rearrangements in the $\delta\beta$ -Globin Complex with Identical DNA Breakpoints But Causing Different Hb A(2) Levels.

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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 genotype	nd	-158 C/T	-158 C/C
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	GTGTTCACTAGCAACCTCAAACAGACACC ATG GTG CAY CTG ACT CCT GAG GAG AAG	TCTGCCGTT
$\delta\beta^*$	ACACTTTCTTCTGACATAACA	GTGTTCACTAGCAACCTCAAACAGACACC ATG GTG CAC CTG ACT CCT GAG GAG AAG	TCTGCCGTT
$\delta\beta\delta\beta$	ACATTTGCTTCTGACACAAC	GTGTTCACTAGCAACCTCAAACAGACACC ATG GTG CAC CTG ACT CCT GAG GAG AAG	ACTGCTGTC
δ	ACACTTTCTTCTGACATAACA	GTGTTCACTAGCAACCTCAAACAGACACC ATG GTG CAT CTG ACT CCT GAG GAG AAG	ACTGCTGTC

Figure 1B:

