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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:

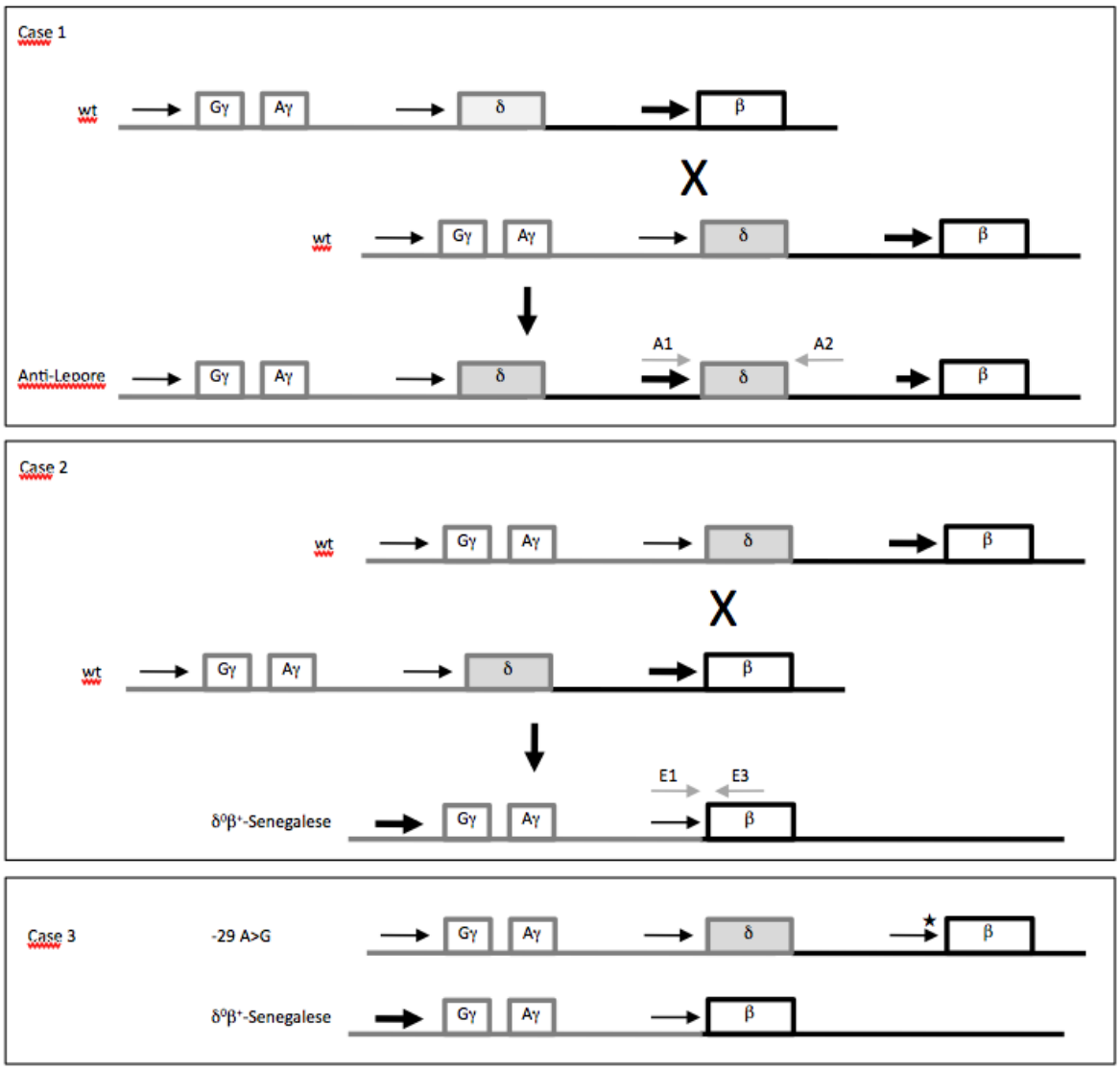
Parameters	**Case 1**	**Case 2**	**Case 3**
Age-sex	50-M	44-F	34-M
RBC (10^{12}/L)	5.3	4.6	4.6
Hb (g/l)	153	108	102
MCV (fl)	86.8	74.7	67
MCH (pg)	28.9	23.5	22
Ferritin (mg/l)	nd	39.2	182
HbA_2	17	2.1	1.9
HbF	0.5	14.2	74
Gγ promoter	nd	-158 C/T	-158 C/C
genotype	δ-βδ-β	$\delta^0\beta^+$	$\delta^0\beta^+$ / -29A>G
Alpha deletions (GAP-PCR)	αα/αα	αα/αα	αα/αα

nd: not determined

Figure 1A:

	cap site	initiation codon	
β	ACATTTGCTTCTGACACAACT	GTGTTCACTAGCAACCTCAAACAGACACC ATG GTG CAY CTG ACT CCT GAG GAG AAG	TCTGCCGTT
$\delta^0\beta^+$	*ACACTTTCTTCTGACATAACA*	GTGTTCACTAGCAACCTCAAACAGACACC ATG GTG CAC CTG ACT CCT GAG GAG AAG	TCTGCCGTT
δ-βδ–β	ACATTTGCTTCTGACACAACT	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:


Case 1
wt
Gγ
Aγ
δ
β
X
wt
Gγ
Aγ
δ
β
A1
A2
Anti-Lepore
Gγ
Aγ
δ
δ
β
Case 2
wt
Gγ
Aγ
δ
β
X
wt
Gγ
Aγ
δ
β
E1
E3
δ⁰β⁺-Senegalese
Gγ
Aγ
β
Case 3
-29 A>G
Gγ
Aγ
δ
β
δ⁰β⁺-Senegalese
Gγ
Aγ
β