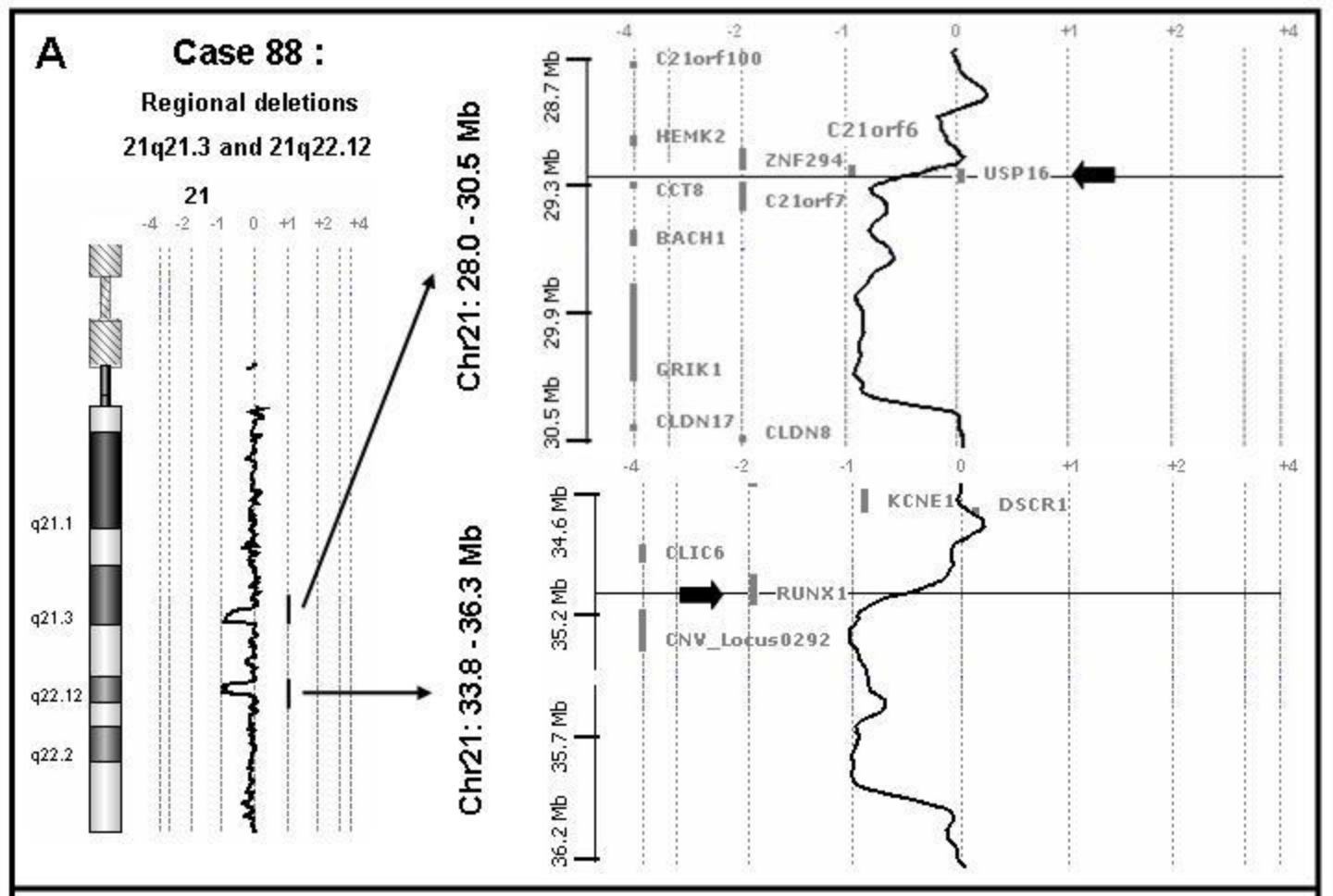
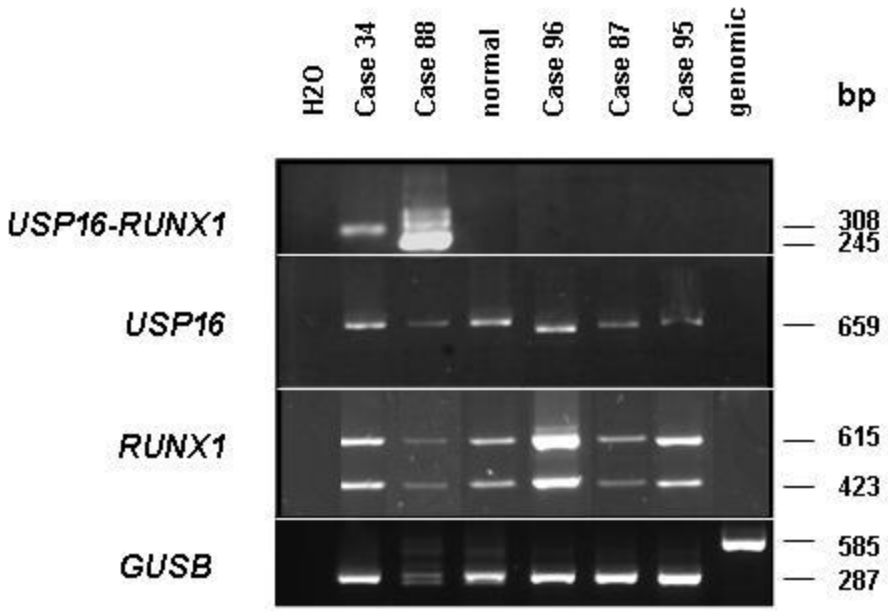

A
Case 88 :
Regional deletions
21q21.3 and 21q22.12
21
-4 -2 -1 0 +1 +2 +4
q21.1
q21.3
q22.12
q22.2
Chr21: 28.0 - 30.5 Mb
Chr21: 33.8 - 36.3 Mb
28.7 Mb
29.3 Mb
29.9 Mb
30.5 Mb
C21orf100
HEMK2
ZNF294
C21orf6
USP16
CCT8
C21orf7
BACH1
GRIK1
CLDN17
CLDN8
34.6 Mb
35.2 Mb
35.7 Mb
36.2 Mb
KCNE1
DSCR1
CLIC6
RUNX1
CNV_Locus0292


B
H2O
Case 34
Case 88
normal
Case 96
Case 87
Case 95
genomic
bp
USP16-RUNX1
USP16
RUNX1
GUSB
308
245
659
615
423
585
287