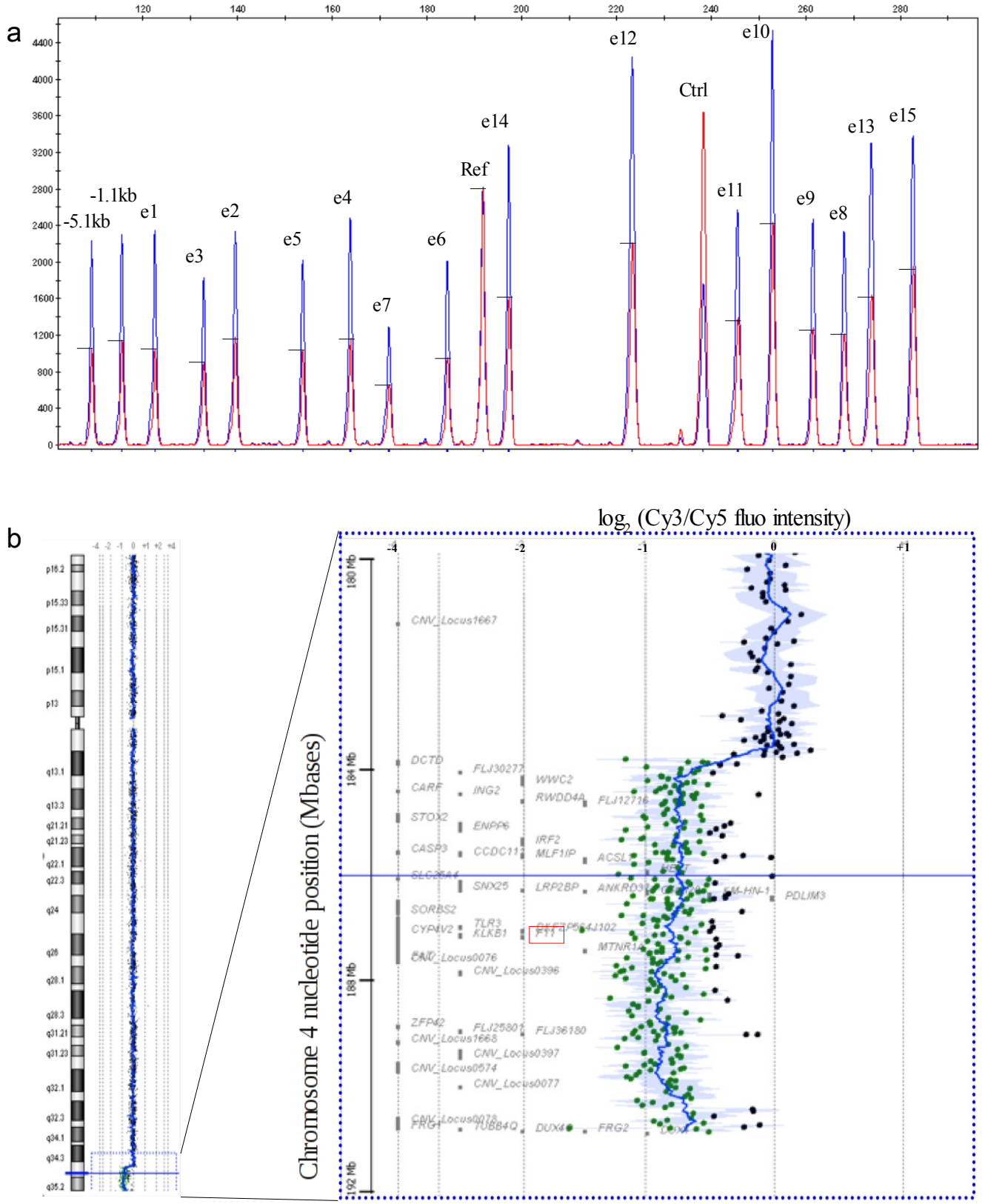
a
-5.1kb
-1.1kb
e1
e2
e3
e4
e5
e6
e7
Ref
e14
e12
Ctrl
e10
e11
e9
e8
e13
e15
b
log2 (Cy3/Cy5 fluo intensity)
Chromosome 4 nucleotide position (Mbases)
180 Mb
184 Mb
188 Mb
192 Mb
CNV_Locus1667
DCTD
FLJ30277
CARF
ING2
WWC2
RWDD4A
FLJ12716
STOX2
ENPP6
CASP3
CCDC11
IRF2
MLF1IP
ACSL1
SNX25
LRP2BP
PDLIM3
SORBS2
TLR3
KLKB1
F11
MTNR1A
CNV_Locus0076
CNV_Locus0396
ZFP42
FLJ25801
FLJ36180
CNV_Locus1668
CNV_Locus0397
CNV_Locus0574
CNV_Locus0077
FRG1
TUBB4Q
DUX4
FRG2

Figure 1
Delineation of the large heterozygous 4qTer deletion. **(a)** Systematic screening of heterozygous rearrangements at the *F11* locus by QFM-PCR. The QFM-PCR electropherogram of patient XXIV/41 is overlaid with that of a normal control following normalization against the reference diploid *F2* amplicon located on chromosome 11 (Ref). Another control amplicon located in the *F9* gene on chromosome X (control) was used to validate the quantitative analysis by determining the sex based on chromosome X dosage. All peaks corresponding to sequences in the *F11* gene show a two-fold reduction (horizontal bars) of their height, indicating a heterozygous deletion of the whole gene. **(b)** The large deletion of 4qTer revealed by arrayCGH. The $\log_2$ of the Cy3/Cy5 fluorescence intensity ratio is plotted against the chromosome 4 nucleotide position (hg18). Grey dots indicate copy number losses (log2 ratio~-1), while the black dots indicate no copy number change. More than 7 Mb of sequences are involved in this deletion, and the *F11* gene (outlined in red) is located in the middle.