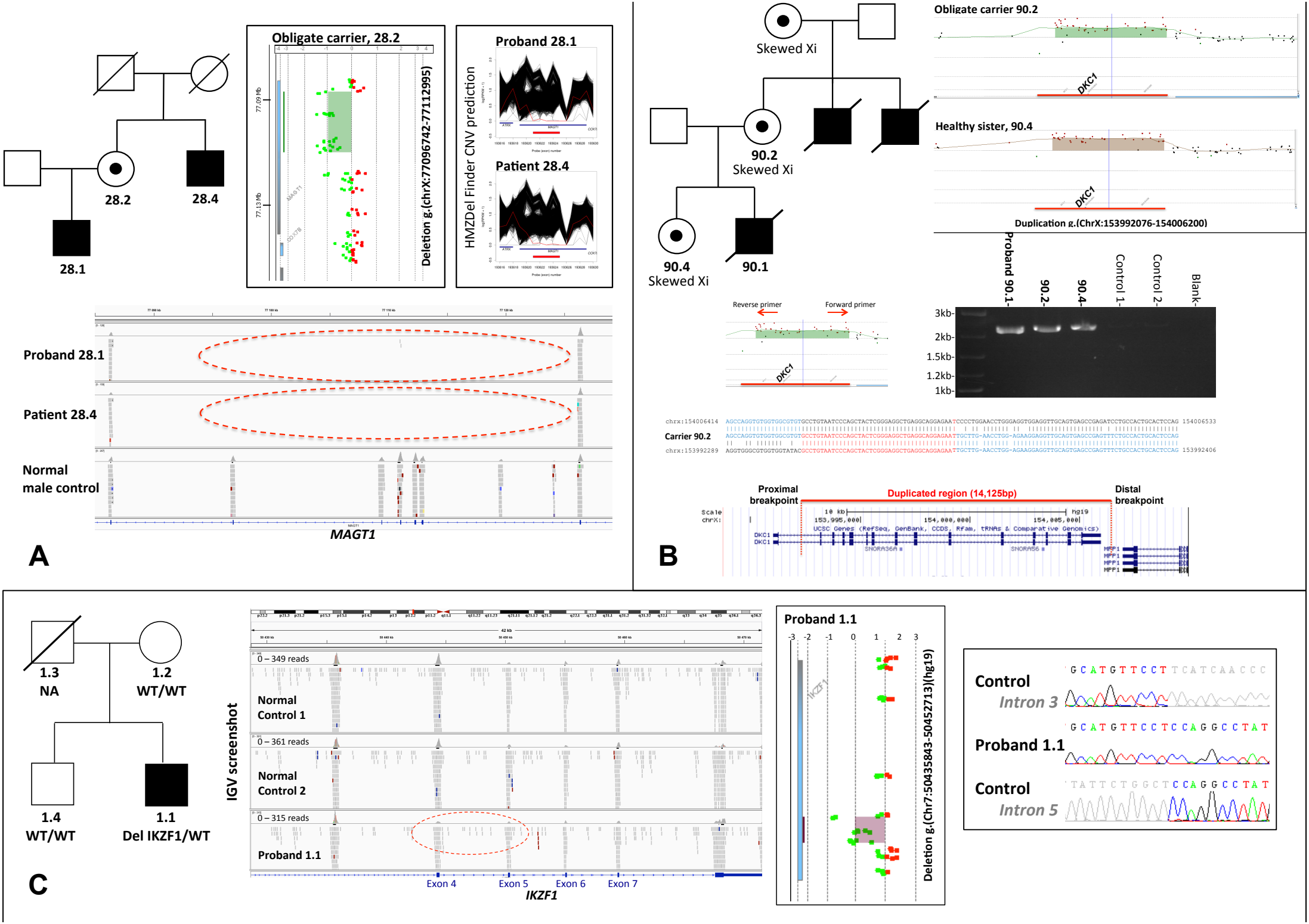
28.2
28.4
28.1
Obligate carrier, 28.2
Deletion g.(chrX:77096742-77112995)
Proband 28.1
Patient 28.4
HMZDel Finder CNV prediction
Proband 28.1
Patient 28.4
Normal male control
MAGT1
A
Skewed Xi
90.2 Skewed Xi
90.4 Skewed Xi
90.1
Obligate carrier 90.2
Healthy sister, 90.4
DKC1
Duplication g.(ChrX:153992076-154006200)
Proband 90.1-
90.2-
90.4-
Control 1-
Control 2-
Blank-
3kb-
2kb-
1.5kb-
1.2kb-
1kb-
Reverse primer
Forward primer
Carrier 90.2
Proximal breakpoint
Duplicated region (14,125bp)
Distal breakpoint
B
1.3 NA
1.2 WT/WT
1.4 WT/WT
1.1 Del IKZF1/WT
IGV screenshot
0 – 349 reads
Normal Control 1
0 – 361 reads
Normal Control 2
0 – 315 reads
Proband 1.1
Exon 4
Exon 5
Exon 6
Exon 7
IKZF1
Proband 1.1
Deletion g.(Chr7:50435843-50452713)(hg19)
Control
Intron 3
Proband 1.1
Control
Intron 5
C