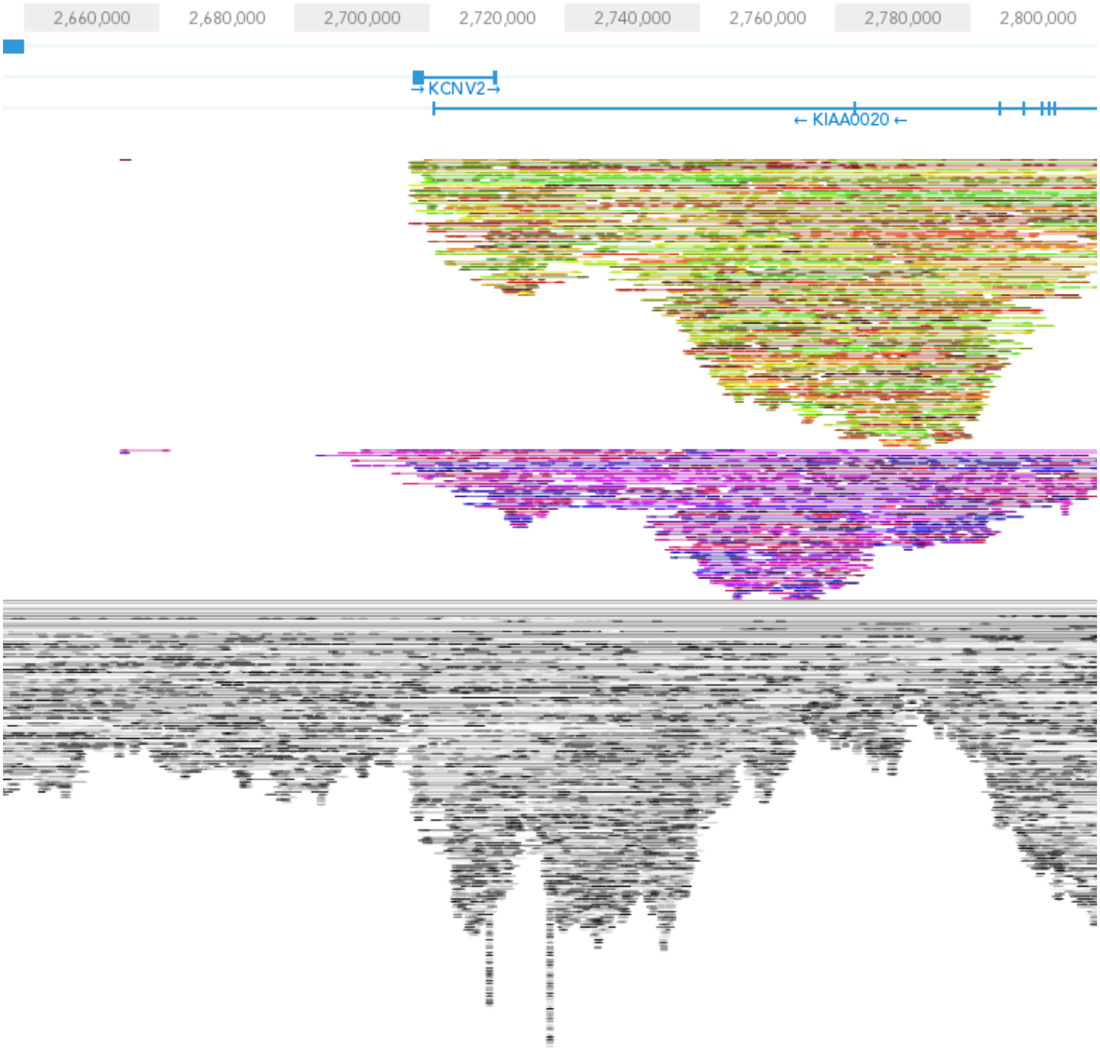
A


2,660,000
2,680,000
2,700,000
2,720,000
2,740,000
2,760,000
2,780,000
2,800,000
→ KCNV2 →
← KIAA0020 ←

B

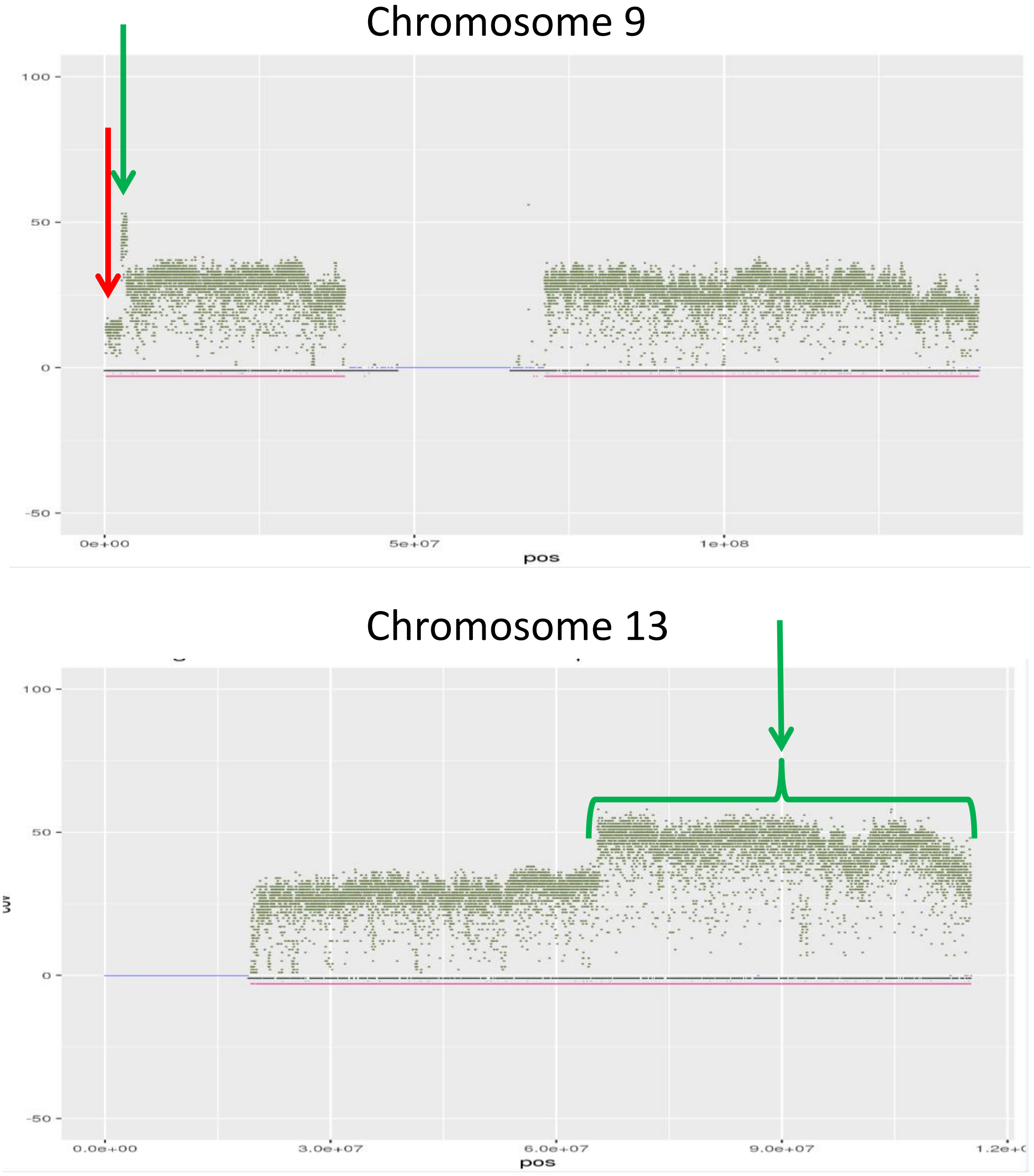


Figure S1. Loupe visualization and depth analysis of patient 2. A. Loupe visualization of the 9p terminal deletion (1-2,727,510) and duplication (2,725,389-3,625,909) junction region. Yellow linked-reads are associated with a haplotype and pink linked-reads are associated with the other. The grey linked-reads correspond to unphased linked-reads. **B**. The mean depths are calculated using Refscope, a CNRGH in house filtering strategy that only consider non-ambiguous read mapping. Red arrow indicates the deletion and green arrows indicate the duplications.

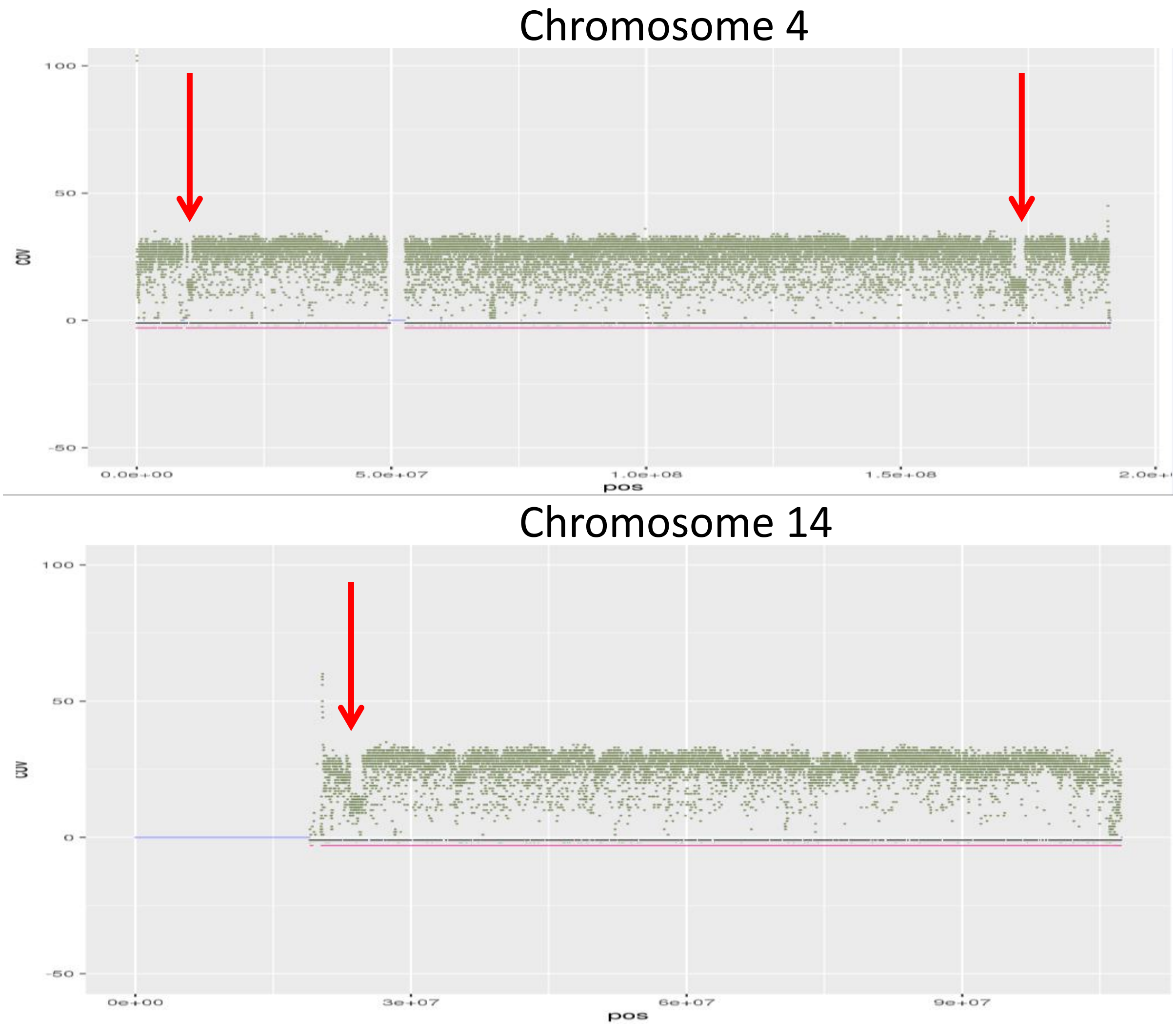


Figure S2. Depth analysis of patient 4. The mean depths are calculated using Refscope. Red arrows indicate the three deletions detected by array-CGH.

A.


Chromosome X
100
50
0
-50
0.0e+00
5.0e+07
1.0e+08
1.5e+08
pos

B.


133,020,000
133,025,000
133,030,000
133,035,000
133,040,000
133,045,000
133,050,000
133,055,000
133,060,000
133,065,000
133,070,000
133,075,000
133,080,000
133,085,000
133,090,00
← GPC3 ←

C.

Figure S3. Loupe visualization and depth analysis of patient 5. A. The mean depths are calculated using Refscope. Green arrow indicates the duplication. **B and C.** Loupe visualization focused on the deletion (B) and duplication (C).

g.[chr1:pter_7130532::chrX:49826909_cen_qter]

chr1(+)

TTGAAAAATGGGCGTGATACTGGTGGTCGCTGTTGTCTTCCCGCAGCGATAGAATGAGGCTCTGCAGTGATTTCTTTTCCTTCCAAAGTCGAAAGCTCCT

der(X)

TTGAAAAATGGGCGTGATACTGGTGGTCGCTGTTGTCTTCCCGCAGCGATATTCAGGAAACCCATCTCACGTGCAGAGACACACATAGGCTCAAAATAAA

chrX (+)

GACACAGACTGGCAAGTTGGATAAAGAGTCAAGACCCATCAGTGTGCTGTATTCAGGAAACCCATCTCACGTGCAGAGACACACATAGGCTCAAAATAAA

g.[chrX:pter_49826908::chr1:7130533_cen_qter]

chrX (+)

CACAGACTGGCAAGTTGGATAAAGAGTCAAGACCCATCAGTGTGCTGTATTCAGGAAACCCATCTCACGTGCAGAGACACACATAGGCTCAAAATAAAAG

der(1)

CACAGACTGGCAAGTTGGATAAAGAGTCAAGACCCATCAGTGTGCTGTATAGAATGAGGCTCTGCAGTGATTTCTTTTCCTTCCAAAGTCGAAAGCTCCT

chr1(+)

TTGAAAAATGGGCGTGATACTGGTGGTCGCTGTTGTCTTCCCGCAGCGATAGAATGAGGCTCTGCAGTGATTTCTTTTCCTTCCAAAGTCGAAAGCTCCT

Figure S4. Sanger sequencing of patient 7.

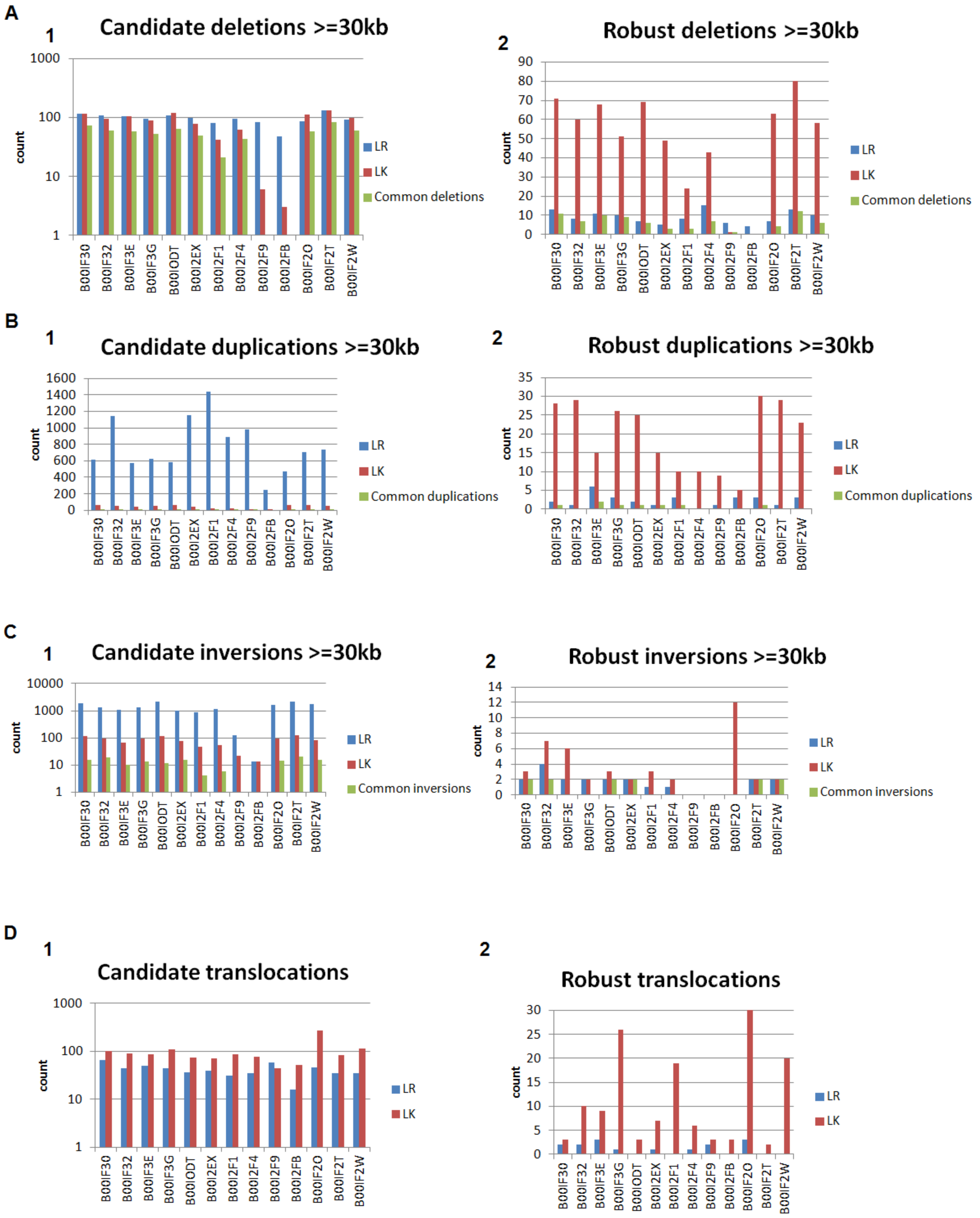

A
1
Candidate deletions >=30kb
1000
100
10
1
count
LR
LK
Common deletions
2
Robust deletions >=30kb
90
80
70
60
50
40
30
20
10
0
count
LR
LK
Common deletions
B
1
Candidate duplications >=30kb
1600
1400
1200
1000
800
600
400
200
0
count
LR
LK
Common duplications
2
Robust duplications >=30kb
35
30
25
20
15
10
5
0
count
LR
LK
Common duplications
C
1
Candidate inversions >=30kb
10000
1000
100
10
1
count
LR
LK
Common inversions
2
Robust inversions >=30kb
14
12
10
8
6
4
2
0
count
LR
LK
Common inversions
D
1
Candidate translocations
1000
100
10
1
count
LR
LK
2
Robust translocations
30
25
20
15
10
5
0
count
LR
LK
B00IF30
B00IF32
B00IF3E
B00IF3G
B00IODT
B00I2EX
B00I2F1
B00I2F4
B00I2F9
B00I2FB
B00IF2O
B00IF2T
B00IF2W

Figure S5 LinkedSV and LongRanger SV call comparisons. This figure represents the counts of candidate, robust and common calls of deletions (**A**), duplications (**B**) and inversions (**C**) identified by linkedSV (LK) and LongRanger (LR). A log-10 scale is used for candidates calls. In Fig. D-2 the y axis was cut; the count of robust translocations in B00IF2O is equal to 155.