

Indel-SNP

Pipeline steps

All variants: 36,140,217



1. Min. variant read count (x=3).



2. Indels only.

1,193,870



3. Indel size >3 bp.

208,460



4. Critical indel presence: all HD-16.

7,199



5. Identify indel-SNP pairs in HD-16:
“critical” indel coupled to marker SNP.

13,301 Pairs (6,640 critical indels)



6. Critical indel presence/absence check.

12,753



7. Indel-SNP association check.

Candidates submitted: 2,937

The Indel-SNP Association Check

(+) Ref: the Indel form different from the Reference

cYsN (+ -) = 0 must be in at least 2 samples reads sum ≥ 3
cYsY (+ +) reads sum ≥ 3
cNsN (- -)
cNsY (- +)

(-) Ref: the Indel form identical to the Reference

cYsN (+ -)
cYsY (+ +)
cNsN (- -) = 0 must be in at least 2 samples reads sum ≥ 3
cNsY (- +) reads sum ≥ 3

This filter is found to be the strongest constraint. It eliminates the majority of the candidates.

Note:

c : Critical Indel; s : Marker SNP

**A less stringent setting is used:
2 samples → 1 samples**