

SOLiD4 RAW DATA



12.179 variants



Filter1: variants which fulfill these 4 conditions

- 1- placed in exonic or splicing regions
- 2- NRA frequency ≤ 0.01 en 1000g2012 database
- 3- %NRA ≤ 4 in our study group
- 4- functional significance (indels, stopgain, splicing, missense)



219 variants



Filter2: variants which fulfill 3 of these 4 conditions:

- 1- coverage ≥ 20
- 2- %NRA ≥ 30
- 3- MQV ≥ 15 for both alleles
- 4- (reference MQV - variant MQV) ≤ 5



77 variants



SANGER
confirmation