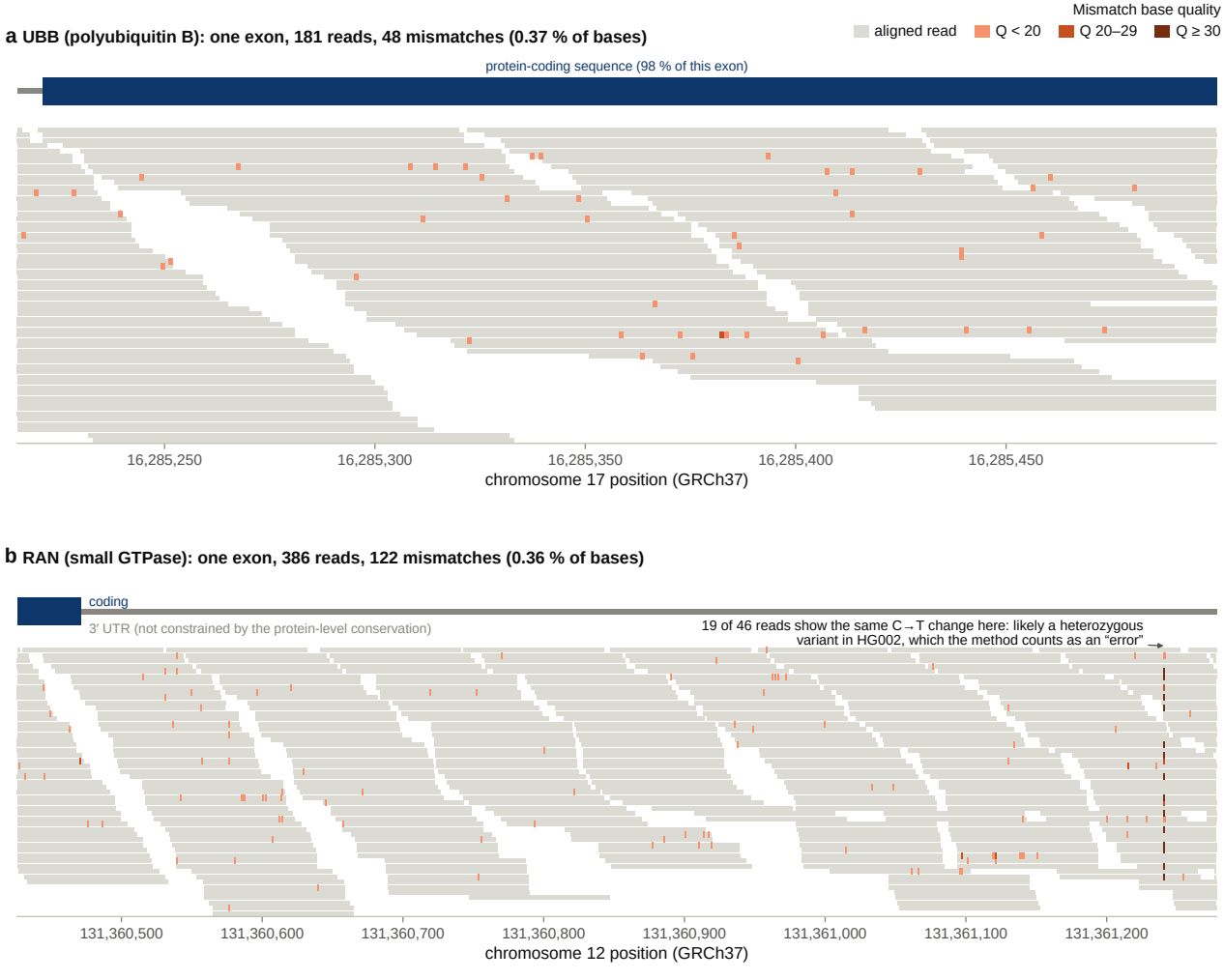
Mismatch base quality
aligned read
Q < 20
Q 20–29
Q ≥ 30
a UBB (polyubiquitin B): one exon, 181 reads, 48 mismatches (0.37 % of bases)
protein-coding sequence (98 % of this exon)
16,285,250
16,285,300
16,285,350
16,285,400
16,285,450
chromosome 17 position (GRCh37)
b RAN (small GTPase): one exon, 386 reads, 122 mismatches (0.36 % of bases)
coding
3′ UTR (not constrained by the protein-level conservation)
19 of 46 reads show the same C→T change here: likely a heterozygous variant in HG002, which the method counts as an "error"
131,360,500
131,360,600
131,360,700
131,360,800
131,360,900
131,361,000
131,361,100
131,361,200
chromosome 12 position (GRCh37)