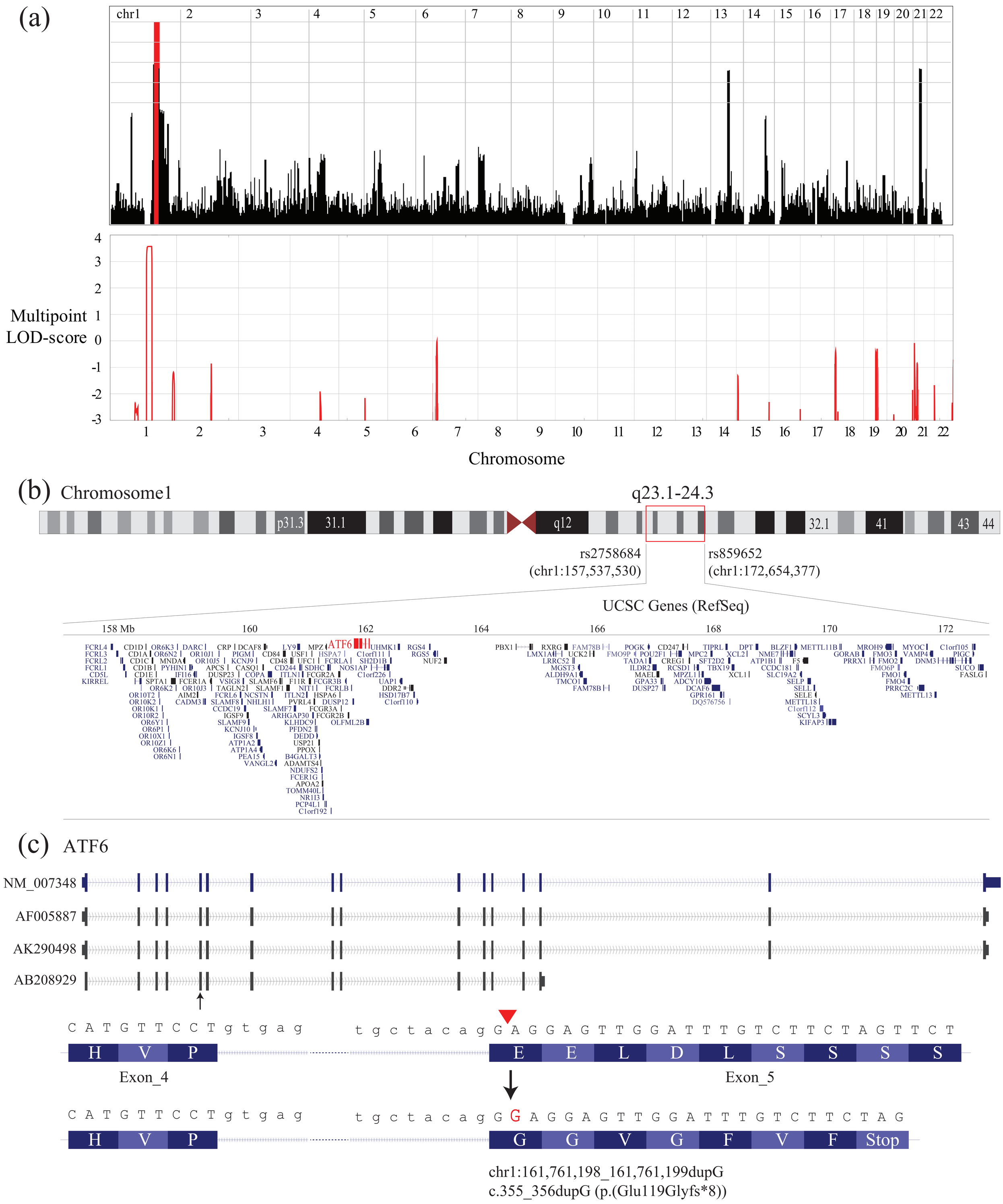
(a)
chr1 2 3 4 5 6 7 8 9 10 11 12 13 14 15 16 17 18 19 20 21 22
Multipoint LOD-score
4 3 2 1 0 -1 -2 -3
Chromosome
(b) Chromosome1
q23.1-24.3
p31.3 31.1 q12 32.1 41 43 44
rs2758684 (chr1:157,537,530)
rs859652 (chr1:172,654,377)
UCSC Genes (RefSeq)
158 Mb 160 162 164 166 168 170 172
ATF6
(c) ATF6
NM_007348
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AB208929
C A T G T T C C T g t g a g
t g c t a c a g G A G G A G T T G G A T T T G T C T T C T A G T T C T
H V P
E E L D L S S S S
Exon_4
Exon_5
C A T G T T C C T g t g a g
t g c t a c a g G G A G G A G T T G G A T T T G T C T T C T A G
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G G V G F V F Stop
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c.355_356dupG (p.(Glu119Glyfs*8))