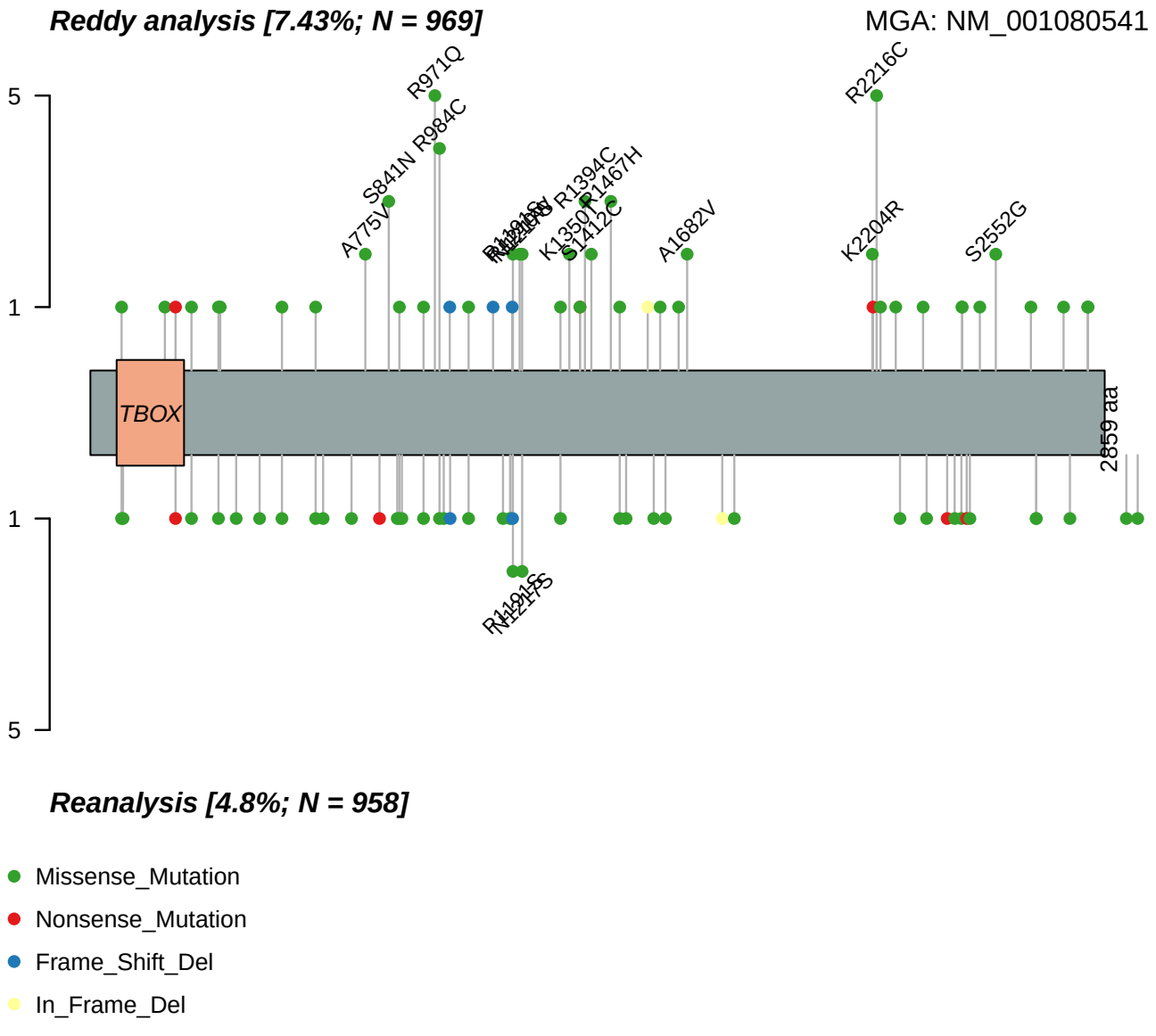
Reddy analysis [7.43%; N = 969]
MGA: NM_001080541
R971Q
R2216C
R984C
S841N
A775V
R1394C
R1467H
K1350R
S1412C
A1682V
K2204R
S2552G
TBOX
2859 aa
5
1
1
5
Reanalysis [4.8%; N = 958]
Missense_Mutation
Nonsense_Mutation
Frame_Shift_Del
In_Frame_Del