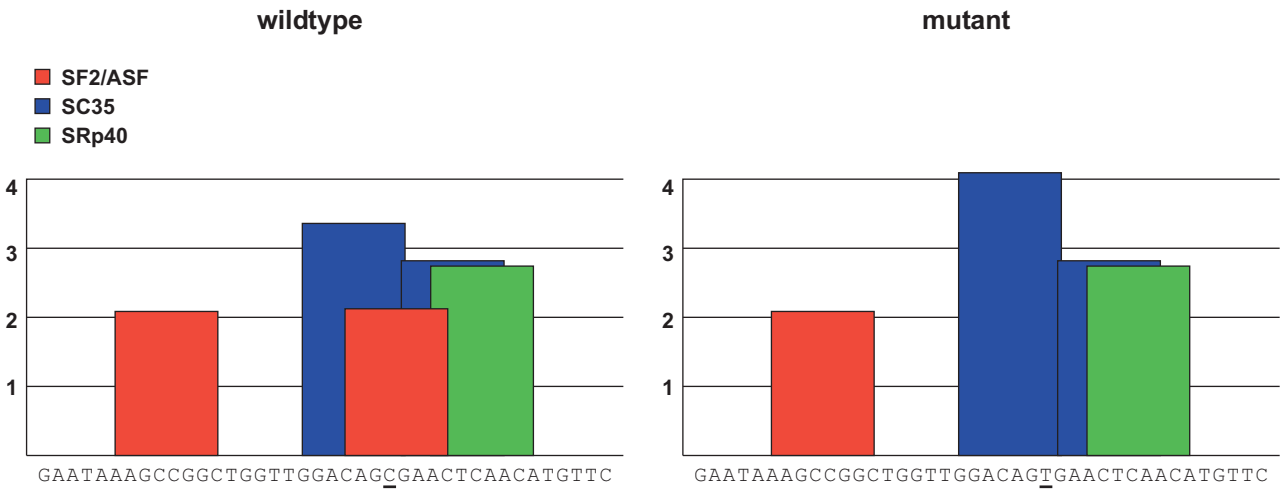


A

c.2229C>T
↓
tgccactaca aggtaacctt ttctctgcag GAATAAAGCC GGCTGGTTGG ACAGCGAACT CAACATGTTC ACCCACCAGT ACCTGCAGCC CAGCAGATAG

B



Splicing factor	Position	Binding site	Score (wildtype/mutant)
SF2/ASF	6-12	AAGCCGG	2.12 / 2.12
SF2/ASF	22-28	CAGC <u>G</u> AA	2.17 / --
SC35	19-26	GGACAGC <u>G</u>	3.41 / 4.05
SC35	26-33	GA <u>A</u> CTCAA	2.85 / 2.85
SRp40	28-34	ACTCAA <u>C</u>	2.82 / 2.82