

# Supplementary Figure S1.

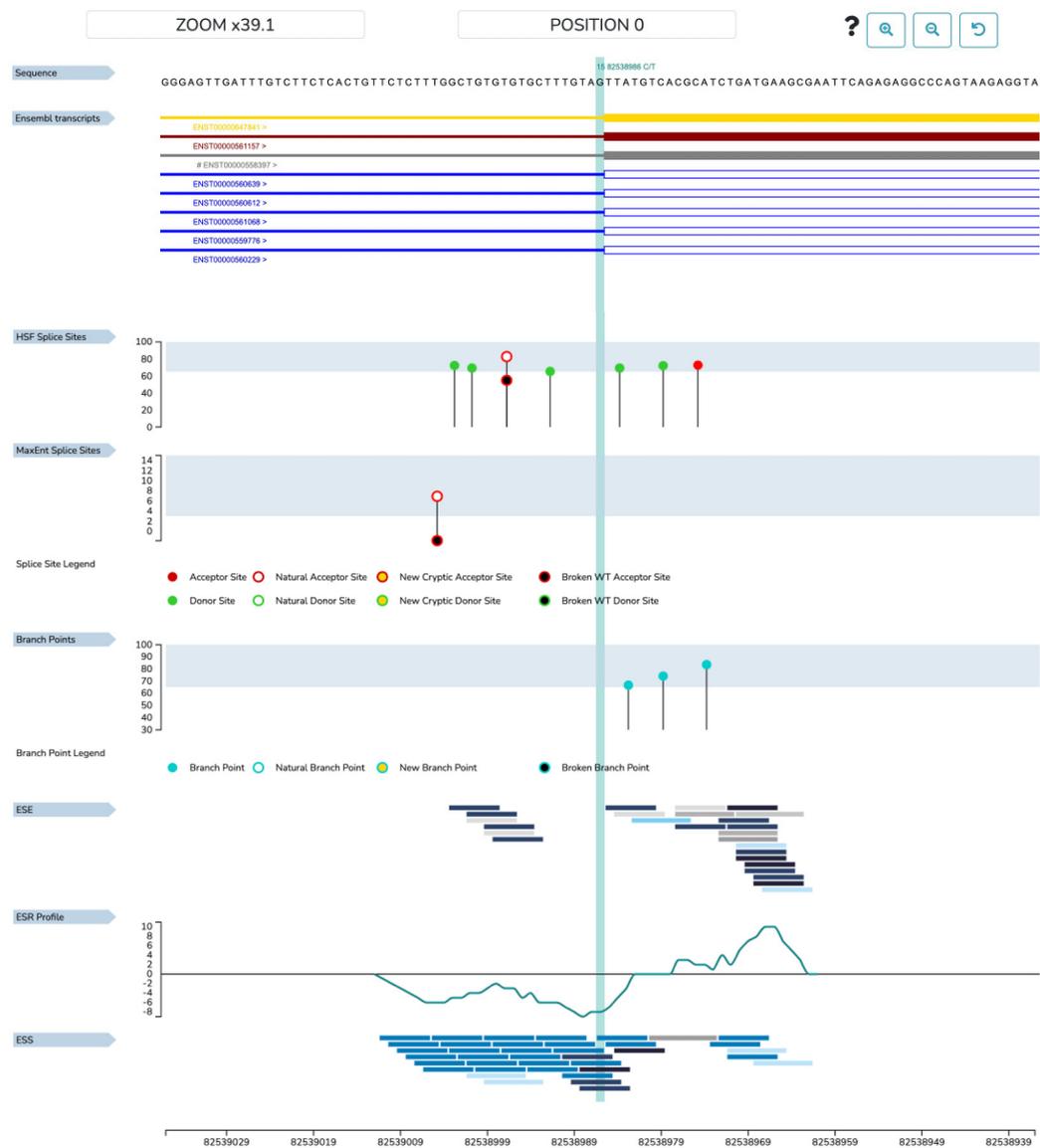
## (A) Mutation analysis for *RPS17* variant

RPS17 ENST00000558397 (6 exons) [Display Settings](#)

Per page: 5

Mutation	HGVS	Predict Impact
● 15 82538986 C/T	ENST00000558397.1:c.156-1G>A	<ul style="list-style-type: none"> <li>Broken WT Acceptor Site : Alteration of the WT Acceptor site, most probably affecting splicing (HSF)</li> <li>Broken WT Acceptor Site : Alteration of the WT Acceptor site, most probably affecting splicing (MaxEnt)</li> </ul>

« 1 » 1 mutations for the selected gene.



## (B) Mutation analysis for *RPS26* variant

RPS26 ENST00000356464 (5 exons) Display Settings

Per page: 5

Mutation	HGVS	Predict Impact
● 12 56042602 GG/G	ENST00000356464.10:c.181+1 del	<ul style="list-style-type: none"> <li>• New Donor splice site : Activation of a cryptic Donor site. Potential alteration of splicing (HSF)</li> <li>• New Donor splice site : Activation of a cryptic Donor site. Potential alteration of splicing (MaxEnt)</li> <li>• Broken WT Donor Site : Alteration of the WT Donor site, most probably affecting splicing (HSF)</li> <li>• Broken WT Donor Site : Alteration of the WT Donor site, most probably affecting splicing (MaxEnt)</li> <li>• New Donor splice site : Activation of a cryptic Donor site. Potential alteration of splicing (HSF)</li> <li>• New Donor splice site : Activation of a cryptic Donor site. Potential alteration of splicing (HSF)</li> </ul>

« ‹ 1 › » 1 mutations for the selected gene.

