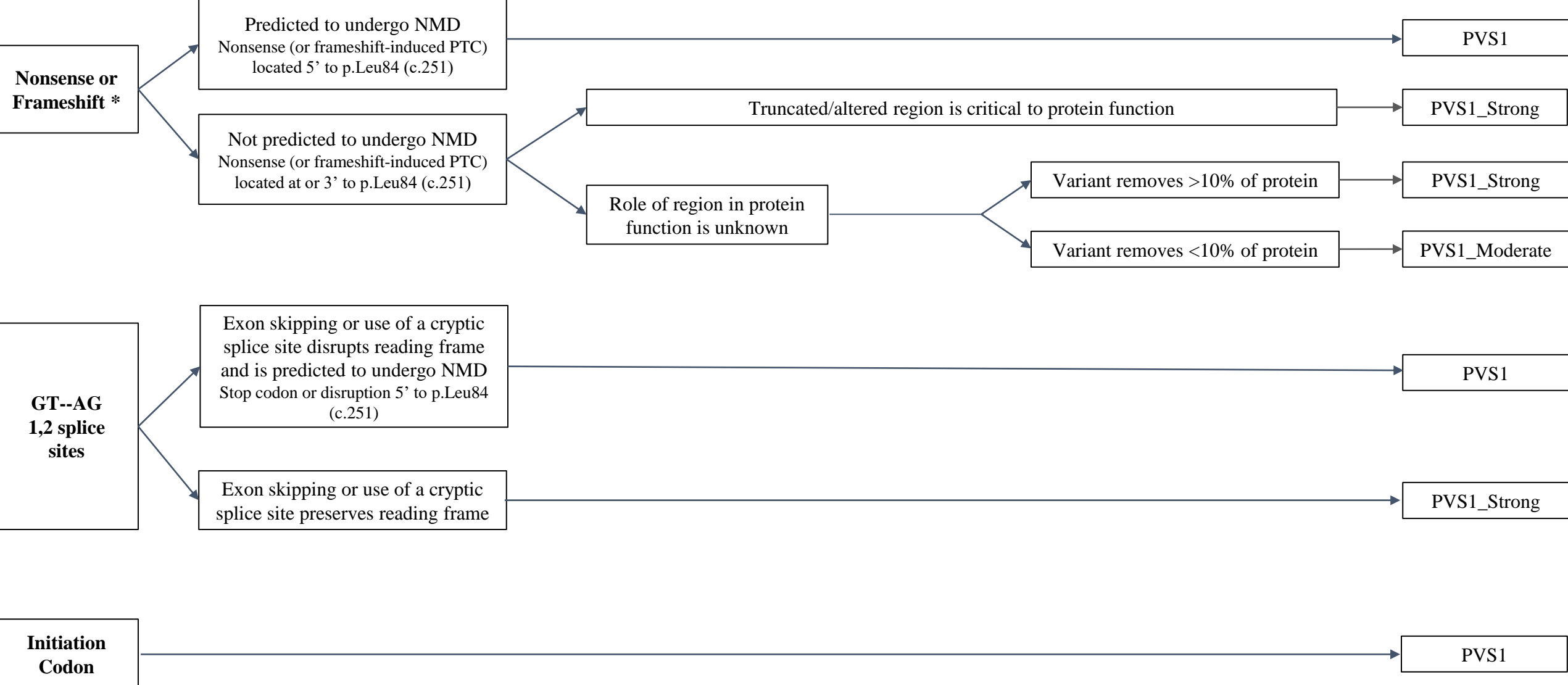


HBA2-specific recommendations for application of PVS1. Adapted from Tayoun et al. 2018



* The borders between NMD-activating and NMD-resistant nonsense mutations in *HBA2* and *HBA1* are undefined (DOI: [10.1016/j.biocel.2017.07.014](https://doi.org/10.1016/j.biocel.2017.07.014)).

Follow recommendations published by Tayoun et al. 2018, whereby NMD prediction is based on the premature termination codon not occurring in the 3' most exon or the 3'-most 50 bp of the penultimate exon.