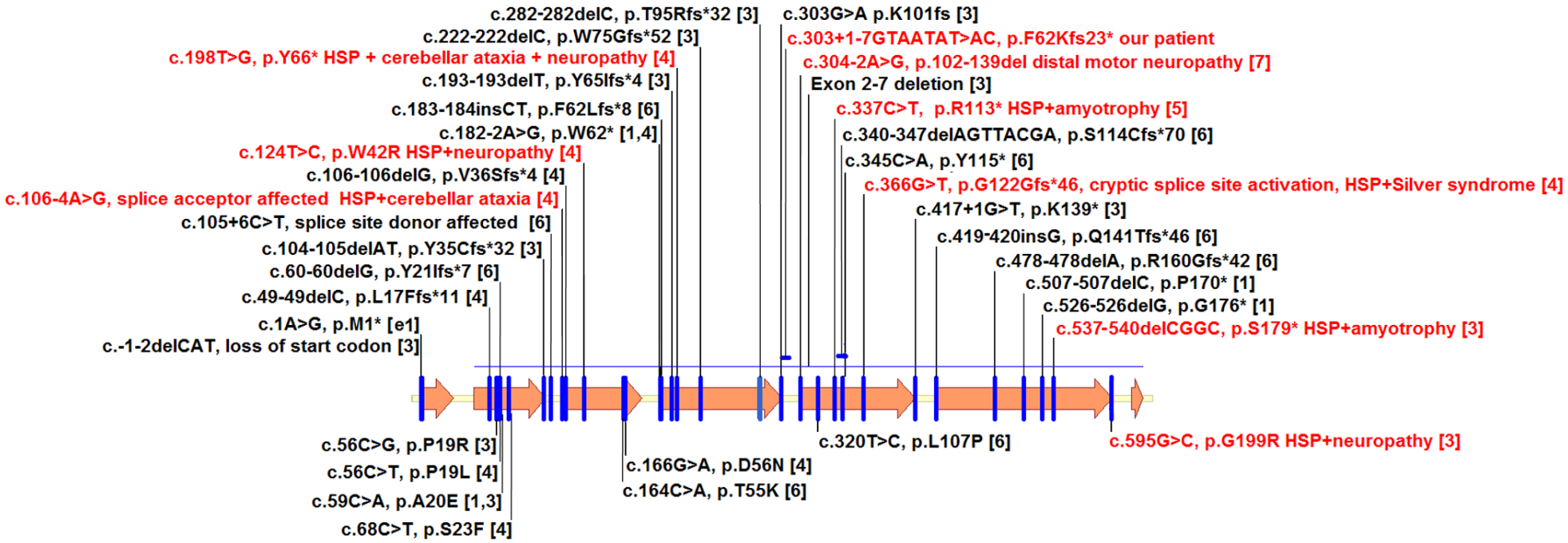

Figure e-1: Coding exons of the *REEP1* gene with annotated mutations that have been described in the literature. The references are provided between square brackets. The numbering of the mutations refers to the RefSeq NM_0022912 sequence. The exons are drawn to scale, but the introns were removed. The majority of mutations (depicted above the exons), are truncating splice site and nonsense mutations, as well as InDeletions. Missense mutations are depicted below the exons. Mutations **in black** cause a **pure HSP phenotype**, while mutations **highlighted in red** have a more **complex phenotype**. There is no mutational hotspot on the gene.

e REFERENCES

e1 Bot ST de, Veldink JH, Vermeer S, et al. ATL1 and REEP1 mutations in hereditary and sporadic upper motor neuron syndromes. J Neurol. 2013 Mar 1;260(3):869–875.