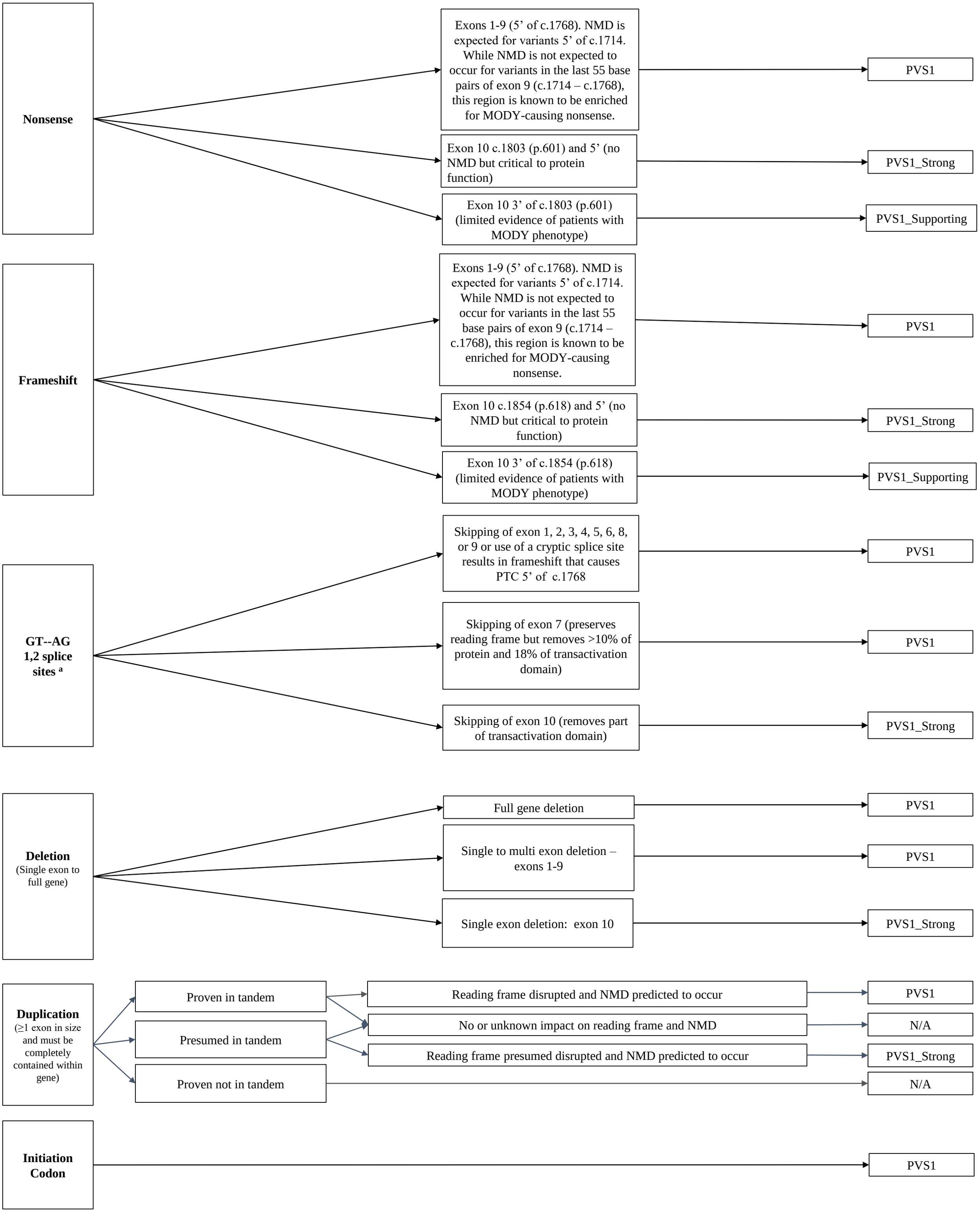
Nonsense
Exons 1-9 (5’ of c.1768). NMD is expected for variants 5’ of c.1714. While NMD is not expected to occur for variants in the last 55 base pairs of exon 9 (c.1714 – c.1768), this region is known to be enriched for MODY-causing nonsense.
PVS1
Exon 10 c.1803 (p.601) and 5’ (no NMD but critical to protein function)
PVS1_Strong
Exon 10 3’ of c.1803 (p.601) (limited evidence of patients with MODY phenotype)
PVS1_Supporting
Frameshift
Exons 1-9 (5’ of c.1768). NMD is expected for variants 5’ of c.1714. While NMD is not expected to occur for variants in the last 55 base pairs of exon 9 (c.1714 – c.1768), this region is known to be enriched for MODY-causing nonsense.
PVS1
Exon 10 c.1854 (p.618) and 5’ (no NMD but critical to protein function)
PVS1_Strong
Exon 10 3’ of c.1854 (p.618) (limited evidence of patients with MODY phenotype)
PVS1_Supporting
GT--AG 1,2 splice sites a
Skipping of exon 1, 2, 3, 4, 5, 6, 8, or 9 or use of a cryptic splice site results in frameshift that causes PTC 5’ of c.1768
PVS1
Skipping of exon 7 (preserves reading frame but removes >10% of protein and 18% of transactivation domain)
PVS1
Skipping of exon 10 (removes part of transactivation domain)
PVS1_Strong
Deletion
(Single exon to full gene)
Full gene deletion
PVS1
Single to multi exon deletion – exons 1-9
PVS1
Single exon deletion: exon 10
PVS1_Strong
Duplication
(≥1 exon in size and must be completely contained within gene)
Proven in tandem
Presumed in tandem
Proven not in tandem
Reading frame disrupted and NMD predicted to occur
PVS1
No or unknown impact on reading frame and NMD
N/A
Reading frame presumed disrupted and NMD predicted to occur
PVS1_Strong
N/A
Initiation Codon
PVS1