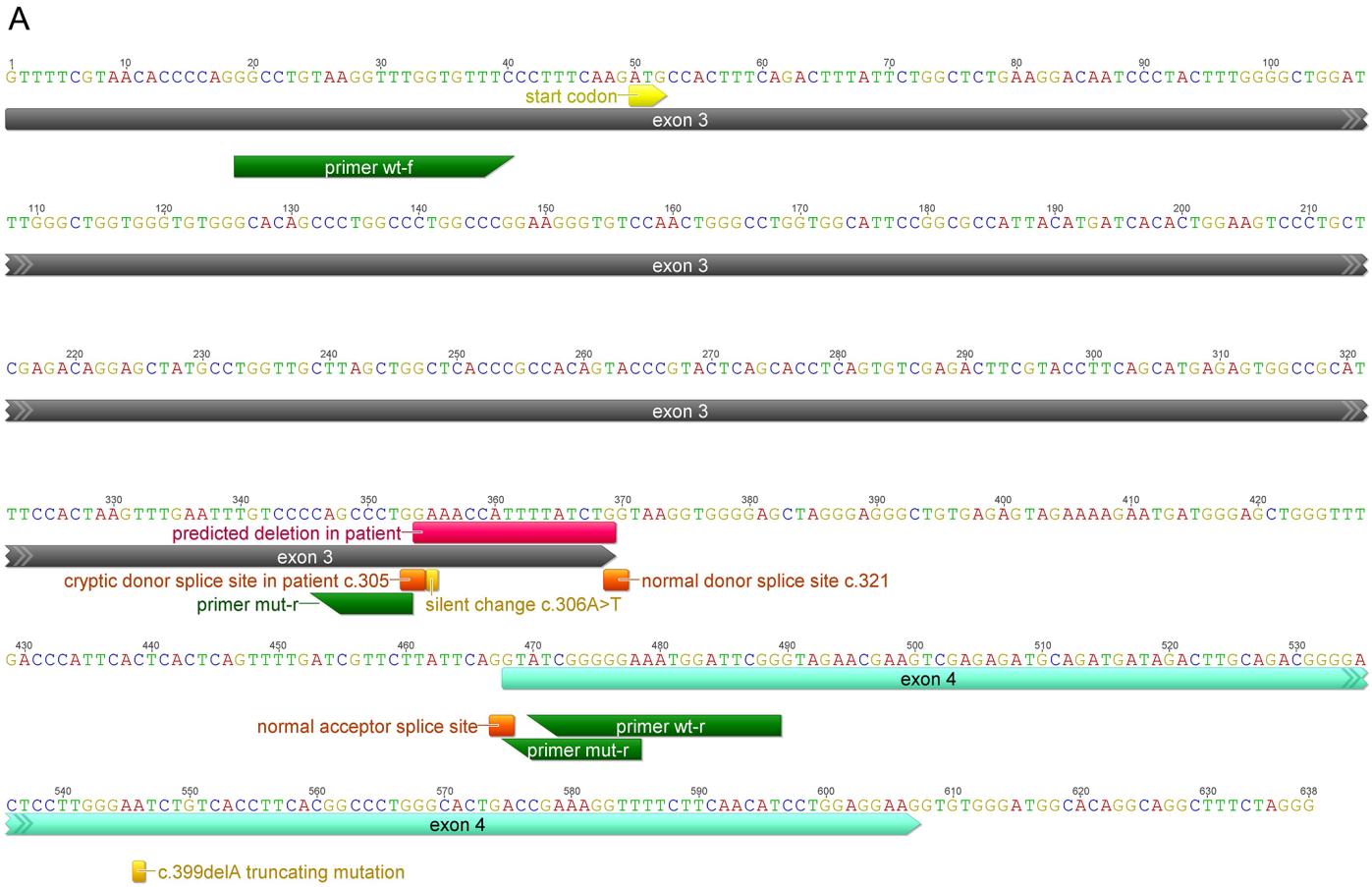
A
GTTTTCGTAACACCCCAGGGCCTGTAAGGTTTGGTGTTTCCCTTTCAAGATGCCACTTTCAGACTTTATTCTGGCTCTGAAGGACAATCCCTACTTTGGGGCTGGAT
start codon
exon 3
primer wt-f
TTGGGCTGGTGGGTGTGGGCACAGCCCTGGCCCTGGCCCGGAAGGGTGTCCAACTGGGCCTGGTGGCATTCCGGCGCCATTACATGATCACACTGGAAGTCCCTGCT
exon 3
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exon 3
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predicted deletion in patient
exon 3
cryptic donor splice site in patient c.305
normal donor splice site c.321
primer mut-r
silent change c.306A>T
GACCCATTCACTCACTCAGTTTTGATCGTTCTTATTCAGGTATCGGGGGAAATGGATTCGGGTAGAACGAAGTCGAGAGATGCAGATGATAGACTTGCAGACGGGGA
exon 4
normal acceptor splice site
primer wt-r
primer mut-r
CTCCTTGGGAATCTGTCACCTTCACGGCCCTGGGCACTGACCGAAAGGTTTTCTTCAACATCCTGGAGGAAGGTGTGGGATGGCACAGGCAGGCTTTCTAGGG
exon 4
c.399delA truncating mutation
B
exon 3
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P Y F G A G F G L V G V G T A L A L A R K G V Q L G L V A F R
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exon 3
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R H Y M I T L E V P A R D R S Y A W L L S W L T R H S T R T Q
GCGGTAATGTACTAGTGTGACCTTCAGGGACGAGCTCTGTCCTCGATACGGACCAACGAATCGACCGAGTGGGCGGTGTCATGGGCATGAGTC
nonsense amino acids
exon 3
exon 4
CACCTCAGTGTCGAGACTTCGTACCTTCAGCATGAGAGTGGCCGCATTTCCACTAAGTTTGAATTTGTCCCCAGCCCTGGTATCGGGGGAA
H L S V E T S Y L Q H E S G R I S T K F E F V P S P G I G G
GTGGAGTCACAGCTCTGAAGCATGGAAGTCGTACTCTCACCGGCGTAAAGGTGATTCAAACTTAAACAGGGGTCGGGACCATAGCCCCCTT
primer mut-r