

Amplification of the *GAA* cDNA:

c1F-ATGAGGCAGCAGGTAGGACAG
c1R-AACTGGTCCGCAAAGAACAG
c2F-CACCGTCCCCACTCTACAG
c2R-TGGAAGTCAGCCACCATGTC
c3F-GCCGCTGATTGGGAAGGTAT
c3R-GTCTGCAGGTCGTACCATGT
c4F-ACTCCTCCCCCACCTCTACA
c4R-ACTGCTCTCCCATCAACAGC

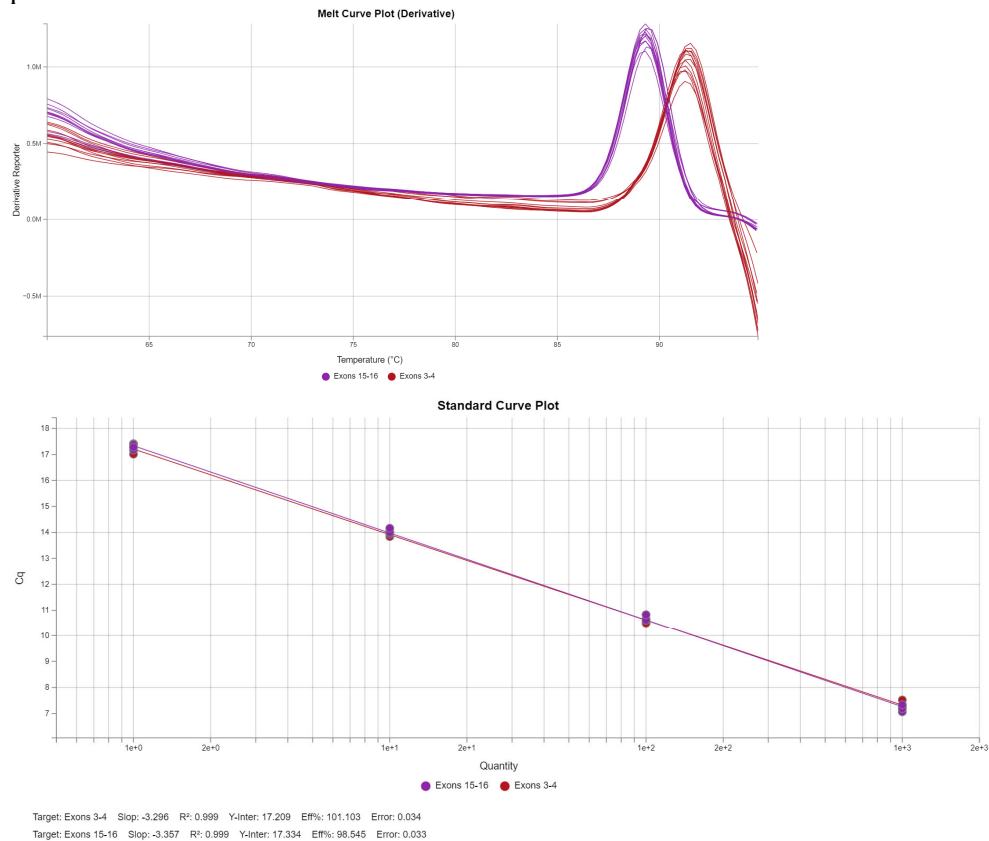
qPCR for the *GAA* expression:

exons 3-4:

q3F-CACCGTCCCCACTCTACAG
q3R-AACTGGTCCGCAAAGAACAG

exons 15-16:

q15F-ACTCCTCCCCCACCTCTACA
q16R-GGTCCACAGTCCAGGTGCTA



qPCR for the chimeric mRNA isoform expression:

GAA exons 14-15:

q14F-CGACGTCTGCGGCTTCCT
q15R-CCTGGGGCAGACTGAGCA

GAA exon 15-TE:

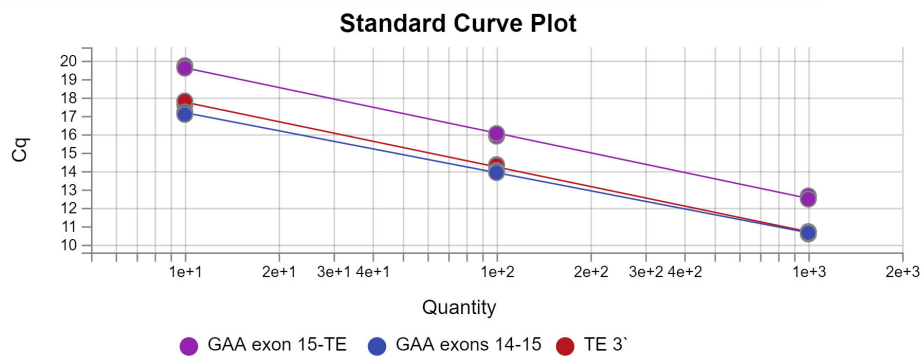
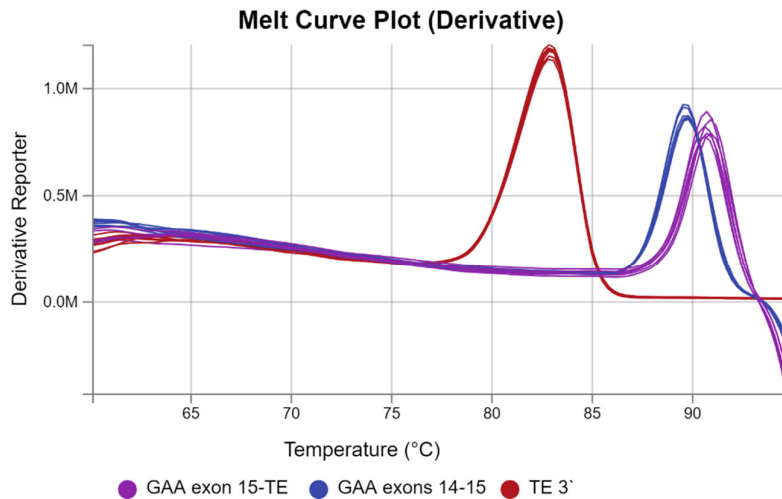
q15F-CTCTACACACTGTTCCACCAGG

TP5`R-TGAGGCAGGAGAATCAGGCA

TE 3`:

TP3`F-CCTCTCTAAACCAAGAGACCAC

TP3`R-GTTCAATTTGCCATTGTCCAGG



Target: TE 3' Slope: -3.524 R²: 0.999 Y-Inter: 21.271 Eff%: 92.217 Error: 0.053

Target: GAA exon 15-TE Slope: -3.55 R²: 0.999 Y-Inter: 23.164 Eff%: 91.296 Error: 0.062

Target: GAA exons 14-15 Slope: -3.263 R²: 0.999 Y-Inter: 20.439 Eff%: 102.503 Error: 0.035

qPCR for the *GAA* expression:

EIF4A3q_F-TGGCCCAGACATCTGTAGAAA
EIF4A3q_R-AGAAAGAGCGGGAGTCCAT

Bisulfite sequencing of *GAA*:

exon 1:

bis1F-GATTTTTTAAATGTTGGTTGTTTT
bis1R-AACTAAAAATCTAAACTCAAACCTCC
bis1.2F-TTTTGGGAATAATTGTGAGTTATGGAGTATA
bis1.2R-CCTCAACTTCCCAACTAAAAAACC

exon 3-4:

bis3F-TTGGATTAGGATTATTTTGTGGAAT
bis4R-CTCCAATCTCCAAAACAAACAACAC

5' of intron 15:

bis15.1F-TGGTTTAGATAGAGGTAATTGTGTT
bis15.1R-ACCTACTAAATAAATAAAAAACCCC

3' of intron 15:

bis15.2F-TAGGAAATAGGATAGGGTAGAGTTG
bis15.2R-ATTCCAACAAATAAAAATCAATACC

Gene specific primers for RACE:

Race1F-CCTACTGGGAAGTGAGGAAAC
Race2F-TTACTGTGCAAGCTGGGATT
Race3F-CCTTTCAAGACCTAGTTTACTAAC

Consensus sequence of the TE amplicon

TCCCGTCTCCCTCTCCCTCTCCCGTCTCCCTCTCCCTCTCCCGTCTCCCTCTCCCTCTC
CCGNNNNNNNNNNCCCTCTCCCTCTCCCTCTCCCTCTCCCTCTCCCTCTCCCTCTCCCT
CTCCCTCTCCGTCTCCGTCTCCGTCTCCCGTCTCCCTCCACGGTCTCCCTCTCAAGCC
GAGCCAAAGCTGGACGGTACTGCTGCCATCTCGGCTCACTGCAACCTCCCTGCCTGA
TTCTCCTGCCTCAGCCTGCCGAGTGCTGCGATTGCAGGCACGCGCCGCCACGCCTG
ACTGGTTTTTCGGTTTTTTTTTTTGGTGGAGATGGGGTTTCGCTGTGTTGGCCGGGCTGCT
CTCCAGCTCCTAACCGCGAGTGATCCGCCAGCCTCGGCCTCCCGAGGTGCCGGGATT
GCAGATGAAGTCTCGTTCACTCAGTGCTCAATGGTGCCAGGCTGGAGTGCAGTGGC
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GCCTCTTCCCCGCCGCCATCCCATCTAGGAAGTGAGGAGCGTCTCTGCCCCGCCGCC
CATCGTCTGAGATGTGGGGAGCACCTCTGCCCCACCGCCCTGTCTGGGATGTGAGGA
GCGCCTCTGCTGGGCCGCAACCCTGTCTGGGAGGTGAGGAGTGTCTCTGCCCCGCCGCC
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GCCGCCCTACTGGGAAGTGAGGAGCCCTCTGCCCCGCCAGCCGCCCGCCAGG
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CCCCCTGCCTGGCCAGCCGCCCGTCCGGGAGGTGAGGGGCGCCTCTGCCCCGCCGCC
CCCCTACTGGGAAGTGAGGACCCCTCTGCCCCGCCAGCCGCCCGTCCGGGAGGGA
GGTGGGGGGGGTTCAGCACCCCGCCCGGCCAGCCGCCCGTCCGGGAGGGAGGTGGG
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GGGAGGGAGGTGGGGGGGGTTCAGCACCCCGCCCGGCCAGCCGCCCGTCCGGGAG
GGAGGTGGGGGGNTCAGCCCCCTGCCTGGCCAGCCGCCCGTCCGGGAGGTGAGGG
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CTGCCTGGCCAGCCGCCCGTCCGGGAGGGCGGTGGGGGGGTTCAGCCCCCGCCCG
GCCAGCCGCCCATCTGGGAGGTGAGGGGCACTTCTGCCGGGCCGCCCTACTGGG
AAGTGAGGAGCCCTCTGCCCCGCCACGACCCCGTCTGGGAGGTGTGCCAGCGGC
TCATTGGGGATGGGCCATGATGACAATGGCGGTTTTGTGGAATAGAAAGGCGGGAA
GGGTGGGGAAAAAATTGAGAAATCGGATGGTTGCTGGGTCTGTGTGGATAGAAGTA

[illegible]