**Supplementary Figure 4: Investigation of PABPC1 and PABPC3 sequences**

**A: PABPC3 sequencing reads alignment with PABPC1 and PABPC3 reference sequence**

**Reference sequences:**

**PABPC3**: TCAGAACCATGCTGCATACTATCCTCCTAGCCAAATTGCTCGACTAAGACCAAGTCCTCGCTGGACTGCTCAGGGTGCCAGACCTCATCCATTCCAAAATAAGCCCAGTGCTATCCGCCCA

**PABPC1**: TCAGAACCGTGCTGCATACTATCCTCCTAGCCAAATTGCTCAACTAAGACCAAGTCCTCGCTGGACTGCTCAGGGTGCCAGACCTCATCGTAAGTCTTTTTTCTTTTAATTAAGGTTGGGGATTGAG

**Exome sequencing alignment**:

 TCAGAACCATGCTGCATACTATCCTCCTAGCCAAATTGCTCAGCTAAGACCAAGTCCTCGCTGGACTGCTCA 41=1X28=2S 8=1X61=2S

 TCCTCCTAGCCAAATTGCTCAGCTAAGACCAAGTCCTCGCTGGACTGCTCAGGGTGCCAGACCTCATCCATT 20=2X50= 21=1X46=4S

 CCTCCTAGCCAAATTGCTCAGCTAAGACCAAGTCCTCGCTGGACTGCTCAGGGTGCCAGACCTCATCCATTC 19=2X51= 20=1X46=5S

 CTCCTAGCCAAATTGCTCAGCTAAGACCAAGTCCTCGCTGGACTGCTCAGGGTGCCAGACCTCATCCATTCC 18=2X52= 19=1X46=6S

 CTCCTAGCCAAATTGCTCAGCTAAGACCAAGTCCTCGCTGGACTGCTCAGGGTGCCAGACCTCATCCATTCC 18=2X52= 19=1X46=6S

 TCCTAGCCAAATTGCTCAGCTAAGACCAAGTCCTCGCTGGACTGCTCAGGGTGCCAGACCTCATCCATTCCA 17=2X53= 18=1X46=7S

 AGCCAAATTGCTCAGCTAAGACCAAGTCCTCGCTGGACTGCTCAGGGTGCCAGACCTCATCCATTCCAAAAT 13=2X57= 14=1X46=11S

 AGCCAAATTGCTCAGCTAAGACCAAGTCCTCGCTGGACTGCTCAGGGTGCCAGACCTCATCCATTCCAAAAT 13=2X57= 14=1X46=11S

 AGCCAAATTGCTCAGCTAAGACCAAGTCCTCGCTGGACTGCTCAGGGTGCCAGACCTCATCCATTCCAAAAT 13=2X57= 14=1X46=11S

 AGCCAAATTGCTCAGCTAAGACCAAGTCCTCGCTGGACTGCTCAGGGTGCCAGACCTCATCCATTCCAAAAT 13=2X57= 14=1X46=11S

 AGCCAAATTGCTCAGCTAAGACCAAGTCCTCGCTGGACTGCTCAGGGTGCCAGACCTCATCCATTCCAAAAT 13=2X57= 14=1X46=11S

 CCAAATTGCTCAGCTAAGACCAAGTCCTCGCTGGACTGCTCAGGGTGCCAGACCTCATCCATTCCAAAATAT 11=2X58=1S 12=1X46=13S

 CCAAATTGCTCAGCTAAGACCAAGTCCTCGCTGGACTGCTCAGGGTGCCAGACCTCATCCATTCCAAAATAT 11=2X58=1S 12=1X46=13S

 CCAAATTGCTCAGCTAAGACCAAGTCCTCGCTGGACTGCTCAGGGTGCCAGACCTCATCCATTCCAAAATAT 11=2X58=1S 12=1X46=13S

 CAAATTGCTCAGCTAAGACCAAGTCCTCGCTGGACTGCTCAGGGTGCCAGACCTCATCCATTCCAAAATATG 10=2X58=2S 11=1X46=14S

 AAATTGCTCAGCTAAGACCAAGTCCTCGCTGGACTGCTCAGGGTGCCAGACCTCATCCATTCCAAAATATGC 9=2X58=3S 10=1X46=15S

 GCTCAGCTAAGACCAAGTCCTCGCTGGACTGCTCAGGGTGCCAGACCTCATCCATTCCAAAATATGCCCGGT 4=2X58=1X4=3S 5=1X46=20S

 GCTCAGCTAAGACCAAGTCCTCGCTGGACTGCTCAGGGTGCCAGACCTCATCCATTCCAAAATATGCCCGGT 4=2X58=1X4=3S 5=1X46=20S

 TCAGCTAAGACCAAGTCCTCGCTGGACTGCTCAGGGTGCCAGACCTCATCCATTCCAAAATATGCCCGGTGC 2=2X58=1X4=1X4= 4S46=22S

 TCAGCTAAGACCAAGTCCTCGCTGGACTGCTCAGGGTGCCAGACCTCATCCATTCCAAAATATGCCCGGTGC 2=2X58=1X4=1X4= 4S46=22S

 AGCTAAGACCAAGTCCTCGCTGGACTGCTCAGGGTGCCAGACCTCATCCATTCCAAAATATGCCCGGTGCTA 2X58=1X4=1X6= 2S46=24S

**B: PABPC3 dinucleotide change resulting in R -->Q amino acid change in the protein sequence.**

‘Mutated’ peptide sequence tested: Y Y P P S Q I A **Q** L

**Wild-type sequence: Mutated sequence:**

|tac|tat|cct|cct|agc|caa|att|gct|c**ga**|cta| |tac|tat|cct|cct|agc|caa|att|gct|c**ag**|cta|

 Y Y P P S Q I A R L Y Y P P S Q I A Q L