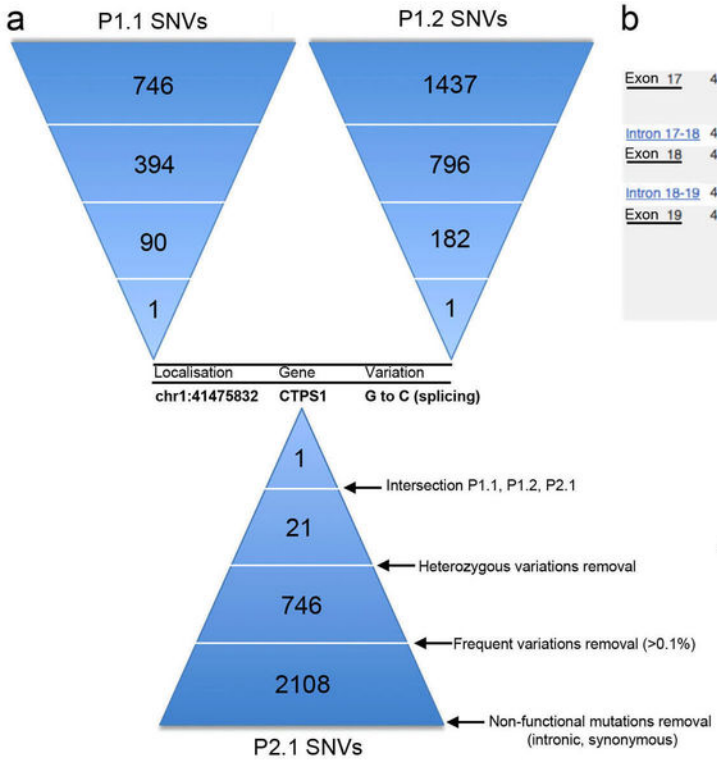
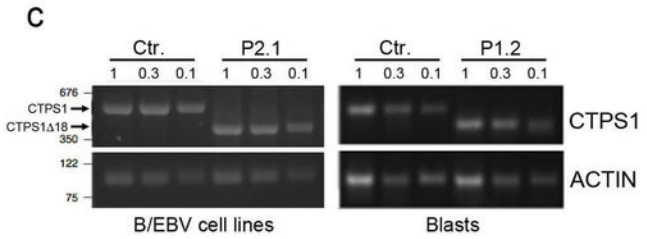

a
P1.1 SNVs
P1.2 SNVs
746
394
90
1
1437
796
182
1
Localisation
Gene
Variation
chr1:41475832
CTPS1
G to C (splicing)
1
21
746
2108
Intersection P1.1, P1.2, P2.1
Heterozygous variations removal
Frequent variations removal (>0.1%)
Non-functional mutations removal
(intronic, synonymous)
P2.1 SNVs

b
Exon 17 41,475,117 41,475,261 ATCATCCCTTTTTTGTTGGGGTTCAGTACCACCCTGAGTTCCTGTCCAGGCCTATCAAGC
CCTCCCCACCATACTTTGGCCTCCTCCTGGCCTCTGTGGGGCGGCTCTCACATTACCTCC
AGAAAGGCTGCAGGCTCTCACCCAG
Intron 17-18 41,475,262 41,475,832 gtaggcgcactctttgcttcagtaa.....tgcgtaaaccatctgaattctacag
Exon 18 41,475,833 41,475,926 GGACACCTATAGTGACAGGAGTGGAAGCAGCTCCCCTGACTCTGAAATCACCGAACTGAA
GTTTCCATCAATAAATCATGACTGATCTTGTAGC
Intron 18-19 41,475,927 41,477,329 gtaagtggtactttaaagttttagt.....ccacttttttttttctttaaacag
Exon 19 41,477,330 41,478,235 GGATGATTCTTCAAGAGACCGTTCAAACTTGGGTAGAGTTTACAGCTCTGACTTTACACT
CGGCTTTGGAGACTTTCTTTAAATTATGTTTTTATTAAGATTATTTTATTATGCGGAAAG
GTATTTGGGAAACTTGTCACTTGCATGTCCCATCACGTGTACTGGCTCCTCTGTGGTGTC
TGCCTGTTGCGTGACACTCTCCTTGCAGTTCTTGAGTTGCGGCAGAACATCGCGATGGGA
ACCGATGGTGGGTGGGGCTGCAGAGTGCCCCATCGGTCACCTTGTTTCTCAACTACCTCG
CATCATTGCAGATGCTAGCGGGTTGCCTGTCGCTTTCCCTTGGATACCTAGACCGTTATA
AAGTGTGCCACATGGACTTACCGAGCATGGAGAGAGGATTTTAGCTAGGATTTGAACACT


c
Ctr.
P2.1
Ctr.
P1.2
1 0.3 0.1 1 0.3 0.1
1 0.3 0.1 1 0.3 0.1
676
CTPS1
CTPS1Δ18
350
122
75
CTPS1
ACTIN
B/EBV cell lines
Blasts