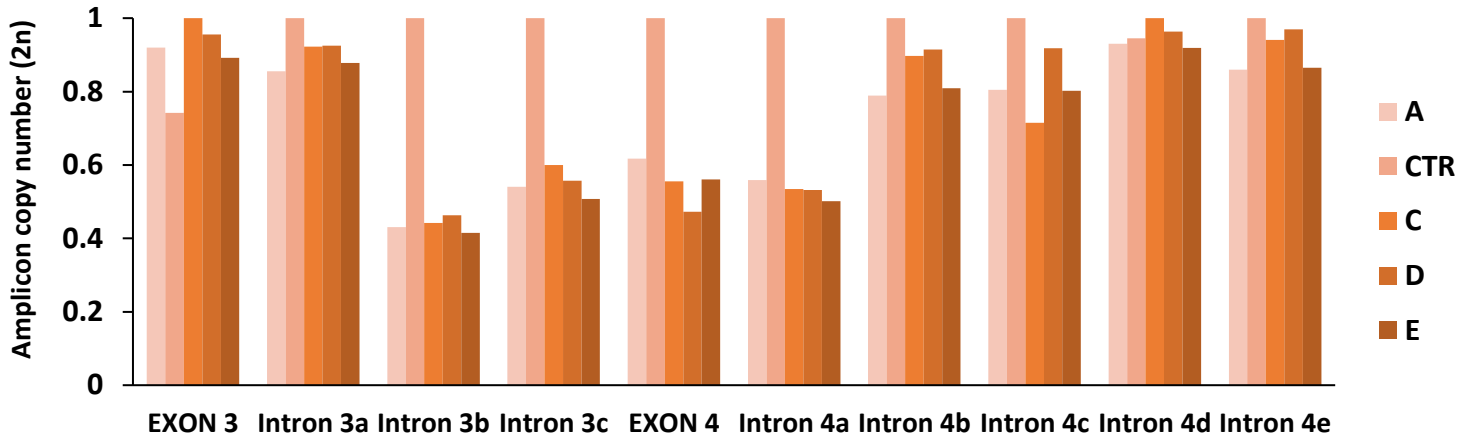
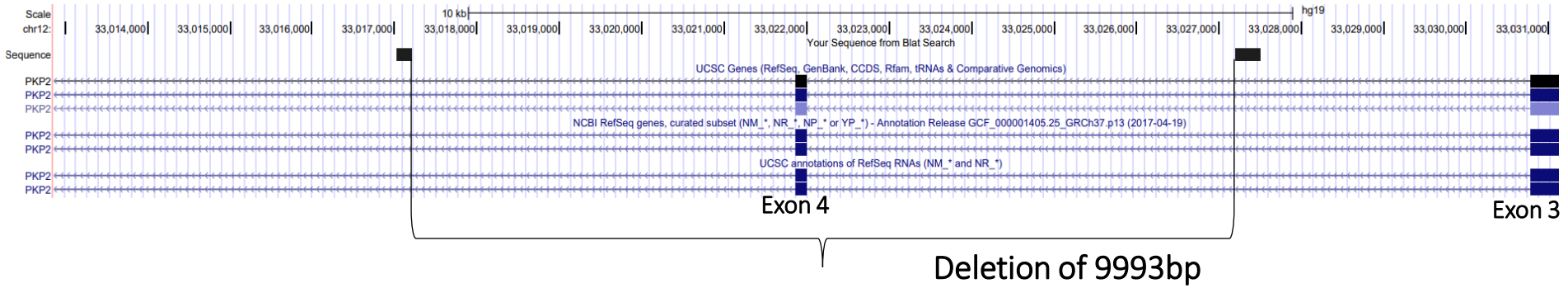
A


Amplicon copy number (2n)
1
0.8
0.6
0.4
0.2
0
EXON 3
Intron 3a
Intron 3b
Intron 3c
EXON 4
Intron 4a
Intron 4b
Intron 4c
Intron 4d
Intron 4e
A
CTR
C
D
E

B


Scale
chr12:
10 kb
hg19
33,014,000
33,015,000
33,016,000
33,017,000
33,018,000
33,019,000
33,020,000
33,021,000
33,022,000
33,023,000
33,024,000
33,025,000
33,026,000
33,027,000
33,028,000
33,029,000
33,030,000
33,031,000
Sequence
Your Sequence from Blat Search
UCSC Genes (RefSeq, GenBank, CCDS, Rfam, tRNAs & Comparative Genomics)
PKP2
PKP2
PKP2
NCBI RefSeq genes, curated subset (NM_*, NR_*, NP_* or YP_*) - Annotation Release GCF_000001405.25_GRCh37.p13 (2017-04-19)
PKP2
PKP2
UCSC annotations of RefSeq RNAs (NM_* and NR_*)
PKP2
PKP2
Exon 4
Exon 3
Deletion of 9993bp

C


p.Asn346Leufs*12
Plakophilin-2 wildtype
1
2
3
4
5
6
7
8
8 ARM domains
Predicted Plakophilin-2 mutated
1

**Fig. S1:** Characterization of *PKP2* mutation. A) By quantitative PCR, we identified the breakpoints of *PKP2* mutation within intron 3 and intron 4. A value of 0.4 to 0.6 was indicative of the amplicon deletion. B) By Sanger sequencing, a deletion of 9993 bp was identified. C) Representation at protein level of the heterozygous deletion effect. It is predicted to insert a premature stop codon within the first ARM repeat, leading to a truncated protein.